INTERGEN	TEST LİSTESİ		Doküman No INT.KLT.LST.17	Yürürlük Tarihi 01.08.2017	Revizyon No 01	Revizyon Tarihi 18.05.2020	Güncelleme Tarihi 14.02.2023	NOT: Merkezimizde çalışan tüm testler Pazar günü hariç tüm günler çalışılmaktadır.						Fiyatlara KDV Dahil Değildir.
Hastalık/ Test adı	Disease/ Test name	Gen/ Gene	Alternatif gen adı/ Alternative gene name	Yöntem/ Method	Gerekli materyal	Akreditasyon Kapsamında	Test süresi (En kısa)/ Turn Around Time-TAT (Shortest)	Test süresi (En uzun)/ Turn Around Time-TAT (Longest)	Test süresi (En kısa)/ Turn Around Time-TAT (Shortest)-PRENATAL	Test süresi (En uzun)/ Turn Around Time-TAT (Longest)-PRENATAL	Ortalama Test Süresi (Gün)/ Expected Reporting Date (Days)	Prenatal/ Postnatal/ Preimplantasyon	Hastalık Bilgisi/ Disease Info	Sample
(Adenin Fosforibosiltransferaz Eksikliği) (ACYP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	(Adenine phosphoribosyltransferase deficiency) (ACYP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACYP2	ACYP2, ACYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prader-Willi sendromu (PWS), Angelman sendromu (AS) (Delesyon + Metilasyon) (PWS/AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2) (MLPA)	(PWS), Angelman syndrome (AS) (deletion + methylation) (PWS / AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2)	PWS/AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2	.	MLPA	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample / EDTA'lı kemik iliği
Prader-Willi sendromu (PWS), Angelman sendromu (AS) (Delesyon + Metilasyon) (PWS/AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2) (MLPA) (Prenatal)	(PWS), Angelman syndrome (AS) (deletion + methylation) (PWS / AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2) (MLPA) (Prenatal)	PWS/AS region (15q11-13), MKRN3, MAGEL2, NDN, SNRPN, UBE3A, ATP10A, GABRB3, OCA2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
[Asetilasyon, Yavaş], 243400, Otozomal Resesif (NAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Acetylation, slow], 243400, Autosomal recessive (NAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAT2	NAT2, AAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Alfa-Aktinin-3 Eksikliği] (ACTN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Alpha-actinin-3 deficiency] (ACTN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN3	ACTN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[AMP Deaminaz Eksikliği, Eritrositik], 612874, Otozomal Resesif (Adenosin Monofosfat Deaminaz Eksikliği) (AMPD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[AMP deaminase deficiency, erythrocytic], 612874, Autosomal recessive (Adenosine monophosphate deaminase deficiency) (AMPD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMPD3	AMPD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Analjezi, Kappa-Opioid Reseptörü Agonistine Bağlı, Kadınlarla Özgül], 613098 (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Analgesia from kappa-opioid receptor agonist, female-specific], 613098 (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Anjiyotensin I-Dönüştürücü Enzim, Benign Serum Yüksekmesi] (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Angiotensin I-converting enzyme, benign serum increase] (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Anhaptoglobinemi], 614081 (HP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Anhaptoglobinemia], 614081 (HP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HP	HP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Aquaporin-1 Eksikliği] (AQP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Aquaporin-1 deficiency] (AQP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP1	AQP1, CHIP28, CO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Aksiller Koku, Varyasyon], 117800, Otozomal Dominant (ABCC11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Axillary odor, variation in], 117800, Autosomal dominant (ABCC11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC11	ABCC11, MRP8, EWWD, WW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Beta-Glikopiranozit Tatma] (TAS2R16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Beta-glycopyranoside tasting] (TAS2R16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAS2R16	TAS2R16, T2R16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Bilirubin, Serum Düzeyi, QTL1], 601816 (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Bilirubin, serum level of, QTL1], 601816 (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gilbert syndrome (GILBS) [MIM:143500]: Occurs as a consequence of reduced bilirubin transferase activity and is often detected in young adults with vague non-specific complaints. {ECO:0000269|PubMed:11013440, ECO:0000269|PubMed:12139570, ECO:0000269|PubMed:7715297, ECO:0000269|PubMed:9627603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Birbeck Granül Eksikliği], 613393 (CD207 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Birbeck granule deficiency], 613393 (CD207 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD207	CD207, LANGERIN, CLEC4K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Birbeck granule deficiency (BIRGD) [MIM:613393]: A condition characterized by the absence of Birbeck granules in epidermal Langerhans cells. Despite the lack of Birbeck granules, Langerhans cells are present in normal numbers and have normal morphologic characteristics and antigen-presenting capacity. {ECO:0000269|PubMed:15816828}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Kan Grubu Cromer], 613793 (CD55 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group Cromer], 613793 (CD55 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD55	CD55, DAF, CROM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu GIL], 607457 (AQP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group GIL], 607457 (AQP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP3	AQP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, ABO Sistemi], 616093 (ABO geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, ABO system], 616093 (ABO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABO	ABO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Auberger Sistemi], 111200 (BCAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Auberger system], 111200 (BCAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAM	LU, AU, BCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Colton], 110450 (AQP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Colton], 110450 (AQP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP1	AQP1, CHIP28, CO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Diego], 110500 (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Diego], 110500 (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Dombrock], 616060 (ART4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Dombrock], 616060 (ART4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ART4	ART4, DO, DOK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Duffy Sistemi], 110700, Otozomal Dominant (ACKR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Duffy system], 110700, Autosomal dominant (ACKR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACKR1	DARC, FY, GPD, WBCQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Froese], 601551 (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Froese], 601551 (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Gerbich], 616089 (Sirtma) (GYPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Gerbich], 616089 (Matela) (GYPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYPC	GYPC, GE, GPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Globosid Sistemi], 615021 (B3GALNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, globoside system], 615021 (B3GALNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GALNT1	B3GALT3, GLCT3, GLOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, I], 110800, Otozomal Dominant (GCNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, I], 110800, Autosomal dominant (GCNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCNT2	GCNT2, I, CTRCT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Hint Sistemi], 609027 (CD44 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Indian system], 609027 (CD44 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD44	CD44, MDU2, MDU3, MIC4, IN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, John-Milton-Hagen Sistemi], 614745 (SEMA7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, John-Milton-Hagen system], 614745 (SEMA7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEMA7A	SEMA7A, SEMAL, SEMAK1, CDW108, JMH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Kell], 110900 (KEL geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Kell], 110900 (KEL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KEL	KEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Kidd], 111000 (SLC14A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Kidd], 111000 (SLC14A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC14A1	SLC14A1, JK, UTE, UT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Knops Sistemi], 607486 (CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Knops system], 607486 (CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR1	CR1, C3BR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Landsteiner-Wiener], 111250 (ICAM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Landsteiner-Wiener], 111250 (ICAM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ICAM4	ICAM4, CD242, LW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Langereis Sistemi], 111600 (ABCB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Langereis system], 111600 (ABCB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB6	ABCB6, MTABC3, MCOPCB7, LAN, DUH3, PSHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Lewis] (FUT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Lewis] (FUT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT3	FUT3, LE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Lüteryen Null], 247420, Otozomal Resesif (BCAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Lutheran null], 247420, Autosomal recessive (BCAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAM	LU, AU, BCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Lüteryen Sistemi], 111200 (BCAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Lutheran system], 111200 (BCAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAM	LU, AU, BCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, MN] (GYPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, MN] (GYPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYPA	GYPA, MN, GPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, OK], 111380 (BSG geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, OK], 111380 (BSG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSG	BSG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, P1Pk Sistemi, S Fenotipi], 111400 (A4GALT geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, P1Pk system, p phenotype], 111400 (A4GALT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	A4GALT	A4GALT, P1PK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, P1Pk Sistemi, P Fenotipi], 111400 (A4GALT geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, P1Pk system, P phenotype], 111400 (A4GALT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	A4GALT	A4GALT, P1PK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, P1PK Sistemi, P(k) Fenotipi], 111400 (B3GALNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, P1PK system, P(k) phenotype], 111400 (B3GALNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GALNT1	B3GALT3, GLCT3, GLOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Radin], 111620 (ERMAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Radin], 111620 (ERMAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERMAP	ERMAP, SC, RD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Ralph], 179620 (CD151 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Raph], 179620 (CD151 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD151	CD151, PETA3, SFA1, MER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Rhesus], 111690 (RHCE geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Rhesus], 111690 (RHCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHCE	RHCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Scianna Sistemi], 111750 (ERMAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Scianna system], 111750 (ERMAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERMAP	ERMAP, SC, RD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Ss] (GYPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Ss] (GYPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYPB	GYPB, SS, MNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Swann], 601550 (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Swann], 601550 (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Vel Sistemi], 615264, Otozomal Resesif (SMIM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Vel system], 615264, Autosomal recessive (SMIM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMIM1	SMIM1, VEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Waldner], 112010 (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Waldner], 112010 (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Wright], 112050 (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Wright], 112050 (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Grubu, Yt Sistemi], 112100 (ACHE geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood group, Yt system], 112100 (ACHE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACHE	ACHE, YT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Basıncı Düzenleme QTL], 145500, Multifaktöriyel (SELE geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood pressure regulation QTL], 145500, Multifactorial (SELE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SELE	SELE, ELAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kan Basıncı Düzenleme QTL], 181500, Multifaktöriyel (RGS5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood pressure regulation QTL], 145500, Multifactorial (RGS5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RGS5	RGS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

[Kan Basıncı Düzenleme QTL] 145500, Multifaktöriyel (ATP1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Blood pressure regulation QTL], 145500, Multifactorial (ATP1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1B1	ATP1B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Vücut Kitle İndeksi QTL16], 613444 (Distal 16p11.2 Mikrodelesyon Sendromu) (2200)	[Body mass index QTL16], 613444 (Distal 16p11.2 microdeletion syndrome) (440)	.	BMIQ16, DEL16p11.2, C16DEL.p11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
[Bombay Fenotipi] (FUT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Bombay phenotype] (FUT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT2	FUT2, SE, B12QTL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Bombay Fenotipi], 616754, Otozomal Resesif (FUT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Bombay phenotype], 616754, Autosomal recessive (FUT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT1	FUT1, H, HH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kemik Mineral Yoğunluğu QTL 15], 613418, Otozomal Resesif, Otozomal Dominant (MIR2861 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Bone mineral density QTL 15], 613418, Autosomal recessive, Autosomal dominant (MIR2861 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIR2861	MIR2861, MIRN2861, BMND15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kemik Mineral Yoğunluğu Değişkenliği 1], 601884, Otozomal Dominant (Otozomal Dominant Osteosklerоz, Worth Tipi) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Bone mineral density variability 1], 601884, Autosomal dominant (Autosomal dominant osteosclerosis, Worth type) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otozomal Dominant, 615082 [C3HEX, Özellği Koku] (OR2J3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[C3HEX, ability to smell], 615082, Autosomal dominant (OR2J3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OR2J3	OR2J3, C3HEXS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Çölotriosidaz Eksikliği], 614122, Otozomal Resesif (CHT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Chitotriosidase deficiency], 614122, Autosomal recessive (CHT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHIT1	CHIT, CHITD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kolostrum Salgılanması, Varyasyon], 117800, Otozomal Dominant (ABCC11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Colostrum secretion, variation in], 117800, Autosomal dominant (ABCC11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC11	ABCC11, MRP8, EWWD, WW	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Dopamin-Beta-Hidroksilaz Aktivitesi Seviyeleri, Plazma] (Dopamin Beta-Hidroksilaz Eksikliği) (DBH geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Dopamine-beta-hydroxylase activity levels, plasma] (Dopamine beta-hydroxylase deficiency) (DBH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DBH	DBH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dopamine beta-hydroxylase deficiency (DBH deficiency) [MIM:223360]: Characterized by profound deficits in autonomic and cardiovascular function, but apparently only subtle signs, if any, of central nervous system dysfunction. {ECO:0000269	PubMed:1857564}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Disalbüminemik Hipertiroksinemi], 615999 (ALB geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Dysalbuminemic hyperthyroxinemia], 615999 (ALB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALB	ALB, ANALBA, FDAH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperthyroxinemia, familial dysalbuminemic (FDAH) [MIM:615999]: A disorder characterized by abnormally elevated levels of total serum thyroxine (T4) in euthyroid patients. It is due to abnormal serum albumin that binds T4 with enhanced affinity. {ECO:0000269|PubMed:7852505, ECO:0000269|PubMed:8048949, ECO:0000269|PubMed:9329347, ECO:0000269|PubMed:9589637}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Distranstiretinemik Hipertiroksinemi], 145680, Otozomal Dominant (TTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Dystransthyretinemic hyperthyroxinemia], 145680, Autosomal dominant (TTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTR	TTR, PALB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis, transthyretin-related (AMYL-TTR) [MIM:105210]: A hereditary generalized amyloidosis due to transthyretin amyloid deposition. Protein fibrils can form in different tissues leading to amyloid polyneuropathies, amyloidotic cardiomyopathy, carpal tunnel syndrome, systemic senile amyloidosis. The disease includes leptomeningeal amyloidosis that is characterized by primary involvement of the central nervous system. Neuropathologic examination shows amyloid in the walls of leptomeningeal vessels, in pia arachnoid, and subpial deposits. Some patients also develop vitreous amyloid deposition that leads to visual impairment (oculeptomeningeal amyloidosis). Clinical features include seizures, stroke-like episodes, dementia, psychomotor deterioration, variable amyloid deposition in the vitreous humor. {ECO:0000269|PubMed:10036587, ECO:0000269|PubMed:10071047, ECO:0000269|PubMed:10211412, ECO:0000269|PubMed:10436378, ECO:0000269|PubMed:10439117, ECO:0000269|PubMed:10611950, ECO:0000269|PubMed:10627135, ECO:0000269|PubMed:10694917, ECO:0000269|PubMed:10842705, ECO:0000269|PubMed:10842718, ECO:0000269|PubMed:10882995, ECO:0000269|PubMed:11445644, ECO:0000269|PubMed:11866053, ECO:0000269|PubMed:12050338, ECO:0000269|PubMed:12557757, ECO:0000269|PubMed:12771253, ECO:0000269|PubMed:1301926, ECO:0000269|PubMed:1351039,	EDTA Blood Sample
[Kulak Kiri, Islak / Kuru], 117800, Otozomal Dominant (ABCC11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Earwax, wet/dry], 117800, Autosomal dominant (ABCC11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC11	ABCC11, MRP8, EWWD, WW	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Ecullizumab, Zayıf Tepki], 615749, Otozomal Dominant (C5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Ecullizumab, poor response to], 615749, Autosomal dominant (C5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C5	C5, C5D, ECLZB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Eozinofil Peroksidaz Eksikliği], 261500, Otozomal Resesif (EPX geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Eosinophil peroxidase deficiency], 261500, Autosomal recessive (EPX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPX	EPX, EPXD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Eosinophil peroxidase deficiency (EPXD) [MIM:261500]: A rare abnormality without clinical symptoms characterized by decreased or absent peroxidase activity and decreased volume of the granule matrix in eosinophils. {ECO:0000269	PubMed:7809065}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Eritrositoz, Ailesel, 1] 133100, Otozomal Dominant (Primer Ailesel Polisitemi) (EPOR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Erythrocytosis, familial, 1], 133100, Autosomal dominant (Primary familial polycythemia) (EPOR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPOR	EPOR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythrocytosis, familial, 1 (ECYT1) [MIM:133100]: An autosomal dominant disorder characterized by increased serum red blood cell mass, elevated hemoglobin and hematocrit, hypersensitivity of erythroid progenitors to erythropoietin, erythropoietin low serum levels, and no increase in platelets nor leukocytes. It has a relatively benign course and does not progress to leukemia. {ECO:0000269|PubMed:8174675, ECO:0000269|PubMed:8506290, ECO:0000269|PubMed:8608241}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Ezetimib, Yanıtsızlığı] (NPC1L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Ezetimibe, nonresponse to] (NPC1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPC1L1	NPC1L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Açlık Plazma Glikoz Seviyesi QTL 5], 613463 (GCKR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Fasting plasma glucose level QTL 5], 613463 (GCKR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCKR	GCKR, GKRP, FGQTL5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Fleck Retina, Ailesel Benign], 228980, Otozomal Resesif (Ailesel Benign Flecked Retina) (PLA2G5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Fleck retina, familial benign], 228980, Autosomal recessive (Familial benign flecked retina) (PLA2G5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G5	PLA2G5, FRFB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fleck retina, familial benign (FRFB) [MIM:228980]: An autosomal recessive condition associated with a distinctive retinal appearance and no apparent visual or electrophysiologic deficits. Affected individuals are asymptomatic, but fundus examination reveals a striking pattern of diffuse, yellow-white, fleck-like lesions extending to the far periphery of the retina but sparing the foveal region. {ECO:0000269	PubMed:22171713}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Fruktosuri], 229800, Otozomal Resesif (Esansiyel Früktozüri) (KHK geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Fructosuria], 229800, Autosomal recessive (Essential fructosuria) (KHK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KHK	KHK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Gama-Glutamiltransferaz, Aileseł Yüksek Serum], 137181 (GGT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Gamma-glutamyltransferase, familial high serum], 137181 (GGT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GGT2	GGT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Gilbert Sendromu], 143500, Otozomal Resesif (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Gilbert syndrome], 143500, Autosomal recessive (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gilbert syndrome (GILBS) [MIM:143500]: Occurs as a consequence of reduced bilirubin transferase activity and is often detected in young adults with vague non-specific complaints. {ECO:0000269|PubMed:11013440, ECO:0000269|PubMed:12139570, ECO:0000269|PubMed:7715297, ECO:0000269|PubMed:9627603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Gliserol Kantitatif Özellik Eğrisi], 614411, Otozomal Resesif (AQP7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Glycerol quantitative trait locus], 614411, Autosomal recessive (AQP7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP7	AQP7, GLYCQTL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Glyoksalaz 2 Eksikliği], 614033, Otozomal Dominant (HAGH geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Glyoxalase II deficiency], 614033, Autosomal dominant (HAGH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HAGH	HAGH, GLO2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Saç Morfolojisi 1, Saç Kalınlığı], 612630 (Hipohidrotik Ektodermal Displazi) (EDAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hair morphology 1, hair thickness], 612630 (Hypohidrotic ectodermal dysplasia) (EDAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDAR	EDAR, DL, ED3, EDA3, HRM1, ECTD10A, ECTD10B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (ECTD10A) [MIM:129490]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal dominant condition characterized by hypotrichosis, abnormal or missing teeth, and hypohidrosis due to the absence of sweat glands. {ECO:0000269|PubMed:10431241, ECO:0000269|PubMed:18231121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Hemoglobin, Yüksek İrtifa Adaptasyon], 609070, Otozomal Dominant (EGLN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hemoglobin, high altitude adaptation], 609070, Autosomal dominant (EGLN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGLN1	EGLN1, PHD2, HIFPH2, C1orf12, ZMYND6, SM20, ECYT3, HALAH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Herediter Alfa Feto Protein Persistansı], 615970, Otozomal Dominant (Kalıtsal Alfa Fetoprotein Persistansı) (AFP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hereditary persistence of alpha-fetoprotein], 615970, Autosomal dominant (Hereditary persistence of alpha-fetoprotein) (AFP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFP	AFP, HPAFP, AFPD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-fetoprotein deficiency (AFPD) [MIM:615969]: A benign condition characterized by undetectable AFP levels in the amniotic fluid. Affected individuals are asymptomatic and present normal development. {ECO:0000269|PubMed:15280901, ECO:0000269|PubMed:18854864}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Herediter Fetal Hemoglobin Persistansı], 613566 (Fetal Hemoglobin Persistansı-Orak Hücre Hastalığı Sendromu) (KLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hereditary persistence of fetal hemoglobin], 613566 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (KLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDN44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

[Hex A Yalancı Eksikliği], 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (HEXA geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hex A pseudodeficiency], 272800, Autosomal recessive (Tay-Sachs disease) (HEXA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEXA	HEXA, TSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM2-ganglosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14568483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Hex A Yalancı Eksikliği], 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (MLPA)	[Hex A pseudodeficiency], 272800, Autosomal recessive (Tay-Sachs disease) (MLPA)	HEXA	HEXA, TSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GM2-ganglosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14568483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Yüksek Yoğunluklu Lipoprotein (HDL) Kolesterol Seviyesi QTL 10], 143470, Otozomal Dominant (CETP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL 10], 143470, Autosomal dominant (CETP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CETP	CETP, HDLCQ10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperalphalipoproteinemia 1 (HALP1) [MIM:143470]: A condition characterized by high levels of high density lipoprotein (HDL) and increased HDL cholesterol levels. {ECO:0000269|PubMed:12091484, ECO:0000269|PubMed:2215607, ECO:0000269|PubMed:8408659}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Yüksek Dansiteli Lipoprotein Kolesterol Seviyesi QTL 11] (Ailesel Lipoprotein Lipaz Eksikliği) (LPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL 11] (Familial lipoprotein lipase deficiency) (LPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPL	LPL, LIPD, HDLCQ11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lipoprotein lipase deficiency (LPL deficiency) [MIM:238600]: Recessive disorder usually manifesting in childhood. On a normal diet, patients often present with abdominal pain, hepatosplenomegaly, lipemia retinalis, eruptive xanthomata, and massive hypertriglyceridemia, sometimes complicated with acute pancreatitis. {ECO:0000269|PubMed:10660334, ECO:0000269|PubMed:10787434, ECO:0000269|PubMed:11068186, ECO:0000269|PubMed:11099402, ECO:0000269|PubMed:11134145, ECO:0000269|PubMed:11441134, ECO:0000269|PubMed:12204001, ECO:0000269|PubMed:12641539, ECO:0000269|PubMed:12966036, ECO:0000269|PubMed:1400331, ECO:0000269|PubMed:1479292, ECO:0000269|PubMed:14984478, ECO:0000269|PubMed:15185149, ECO:0000269|PubMed:1521525, ECO:0000269|PubMed:15256764, ECO:0000269|PubMed:15877202, ECO:0000269|PubMed:1598907, ECO:0000269|PubMed:1619366, ECO:0000269|PubMed:1639392, ECO:0000269|PubMed:1674945, ECO:0000269|PubMed:1702428, ECO:0000269|PubMed:1730727, ECO:0000269|PubMed:1752947, ECO:0000269|PubMed:1907278, ECO:0000269|PubMed:1969408, ECO:0000269|PubMed:1975597,	EDTA Blood Sample
[Yüksek Yoğunluklu Lipoprotein Kolesterol Seviyesi QTL 12], 612797 (LIPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL 12], 612797 (LIPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPC	LIPC, HL, LIPH, HDLCQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic lipase deficiency (HL deficiency) [MIM:614025]: A disorder characterized by elevated levels of beta-migrating very low density lipoproteins, and abnormally triglyceride-rich low and high density lipoproteins. {ECO:0000269|PubMed:10660332, ECO:0000269|PubMed:1301939}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Yüksek Dansiteli Lipoprotein (HDL) Kolesterol Seviyesi QTL 8] (VNN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL 8] (VNN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VNN1	VNN1, HDLCQ8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Yüksek Dansiteli Lipoprotein (HDL) Kolesterol Seviyesi QTL 9] (PLTP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL 9] (PLTP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLTP	PLTP, HDLCQ9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Yüksek Yoğunluklu Lipoprotein (HDL) Kolesterol Düzeyi QTL6], 610762 (SCARB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High density lipoprotein cholesterol level QTL6], 610762 (SCARB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCARB1	SCARB1, CD36L1, CLA1, HDLQTL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Yüksek Molekül Ağırlıklı Kininojen Eksikliği], 228960, Otozomal Resesif (Konjenital Yüksek Molekül Ağırlıklı Kininojen Eksikliği)) (KNG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[High molecular weight kininogen deficiency], 228960, Autosomal recessive (Congenital high-molecular-weight kininogen deficiency) (KNG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KNG1	KNG1, KNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: High molecular weight kininogen deficiency (HMWK deficiency) [MIM:228960]: Autosomal recessive coagulation defect. Patients with HMWK deficiency do not have a hemorrhagic tendency, but they exhibit abnormal surface-mediated activation of fibrinolysis. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Histidinemia], 235800, Otozomal Dominant (Histidinemi) (HAL geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Histidinemia], 235800, Autosomal dominant (Histidinemia) (HAL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HAL	HAL, HSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Histidinemia (HSTID) [MIM:235800]: Autosomal recessive disease characterized by increased histidine and histamine as well as decreased urocanic acid in body fluids. {ECO:0000269|PubMed:15806399}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Hiperfenilalaninemi, PKU Olmayan, Hafif], 261600, Otozomal Resesif (Fenilketonüri) (PAH geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hyperphenylalaninemia, non-PKU mild], 261600, Autosomal recessive (Phenylketonuria) (PAH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAH	PAH, PKU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Hiperfenilalaninemi, PKU Olmayan, Hafif], 261600, Otozomal Resesif (Fenilketonüri) (MLPA)	[Hyperphenylalaninemia, non-PKU mild], 261600, Autosomal recessive (Phenylketonuria) (MLPA)	PAH	PAH, PKU1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
[Hiperproglukagonemi] (GCG geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hyperproglucagonemia] (GCG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCG	GCG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Hiperproreninemi] (Renal Tübüler Disgenezisi) (REN geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hyperproreninemia] (Renal tubular dysgenesis) (REN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REN	REN, HNFJ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Hiposeruloplazminemi, Kalıtsal], 604290, Otozomal Resesif (Aseruloplazminemi) (CP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hypoceruloplasminemia, hereditary], 604290, Autosomal recessive (Aceruloplasminemia) (CP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CP	CP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Ceruloplasmin levels are decreased in Wilson disease, in which copper cannot be incorporated into ceruloplasmin in liver because of defects in the copper-transporting ATPase 2.	EDTA Blood Sample
[Hipohaptoglobinemi], 614081 (HP geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Hypohaptoglobinemia], 614081 (HP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HP	HP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anhaptoglobinemia (AHP) [MIM:614081]: A condition characterized by the absence of the serum glycoprotein haptoglobin. Serum levels of haptoglobin vary among normal persons: levels are low in the neonatal period and in the elderly, differ by population, and can be influenced by environmental factors, such as infection. Secondary hypohaptoglobinemia can occur as a consequence of hemolysis, during which haptoglobin binds to free hemoglobin. Congenital haptoglobin deficiency is a risk factor for anaphylactic non-hemolytic transfusion reactions. {ECO:0000269|PubMed:14999562}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[IgE Düzeyleri QTL], 147050, Otozomal Dominant (PHF11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[IgE levels QTL], 147050, Autosomal dominant (PHF11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHF11	PHF11, NYREN34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Ige, Yüksek Miktar], 147050, Otozomal Dominant (IL21R geni) (Tüm Gen Dizi Analizi) (Postnatal)	[IgE, elevated level of], 147050, Autosomal dominant (IL21R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL21R	IL21R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Igg Reseptörü I, Fagositik, Ailesel Eksikliği] (FCGR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	[IgG receptor I, phagocytic, familial deficiency of] (FCGR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR1A	FCGR1A, IGFR1, CD64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[IMPDH2'nin Erzim Aktivitesi, Farklılaşma] (IMPDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[IMPDH2 enzyme activity, variation in] (IMPDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPDH2	IMPDH2, IMPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Inosin Trifosfat Eksikliği], 613850 (ITPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Inosine triphosphatase deficiency], 613850 (ITPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPA	ITPA, EIEE35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Interlökin 6, Serum Seviyesi, QTL], 614752 (IL6R geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Interleukin 6, serum level of, QTL], 614752 (IL6R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6R	IL6R, IL6RQ, IL6Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

[İnterlökin-6 Reseptörü, QTL, Çözünür, Serum Seviyesi], 614689 (IL6R gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Interleukin-6 receptor, soluble, serum level of, QTL], 614689 (IL6R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6R	IL6R, IL6RQ, IL5Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Junior Kan Grubu Sistemi], 614490 (ABCG2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Junior blood group system], 614490 (ABCG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCG2	ABCG2, BCRP, ABCP, UAQTL1, GOUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kallikrein, İdrar Aktivitesinde Azalma], 615953 (KLK1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Kallikrein, decreased urinary activity of], 615953 (KLK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLK1	KLK1, KLKR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kininojenden Eksikliği], 228960, Otozomal Resesif (KNG1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Kininogen deficiency], 228960, Autosomal recessive (KNG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KNG1	KNG1, KNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: High molecular weight kininogen deficiency (HMWK deficiency) [MIM:228960]: Autosomal recessive coagulation defect. Patients with HMWK deficiency do not have a hemorrhagic tendency, but they exhibit abnormal surface-mediated activation of fibrinolysis. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Laktat Dehidrojenaz-B Eksikliği], 614128; LDHBD (Glikojen Depo Hastalığı, Laktat Dehidrojenaz Eksikliğine Bağlı) (LDHB gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Lactate dehydrogenase-B deficiency], 614128; LDHBD (Glycogen storage disease due to lactate dehydrogenase deficiency) (LDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDHB	LDHB, LDHBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lactate dehydrogenase B deficiency (LDHBD) [MIM:614128]: A condition with no deleterious effects on health. LDHBD is of interest to laboratory medicine mainly because it can cause misdiagnosis in those disorders in which elevation of serum LDH is expected. {ECO:0000269|PubMed:10211631, ECO:0000269|PubMed:11509017, ECO:0000269|PubMed:1597525, ECO:0000269|PubMed:2334429, ECO:0000269|PubMed:8314553, ECO:0000269|PubMed:8462975, ECO:0000269|PubMed:8611651, ECO:0000269|PubMed:9929983, ECO:0000269|Ref.19}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Düşük Yoğunluklu Lipoprotein (LDL) Kolesterol Seviyesi QTL 3], (HMGCR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Low density lipoprotein cholesterol level QTL 3] (HMGCR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGCR	HMGCR, LDLCQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Düşük Yoğunluklu Lipoprotein (LDL) Kolesterol Seviyesi QTL6], 613589, Otozomal Dominant (SORT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Low density lipoprotein cholesterol level (QTL6], 613589, Autosomal dominant (SORT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SORT1	SORT1, NT3, LDLCQ6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Maleotrombositopeni) (CD36 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Macrothrombocytopenia] (CD36 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD36	CD36, CHDS7, BDPLT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Platelet glycoprotein IV deficiency (PG4D) [MIM:608404]: A disorder characterized by macrothrombocytopenia without notable hemostatic problems and bleeding tendency. Platelet glycoprotein IV deficiency can be divided into 2 subgroups. The type I phenotype is characterized by platelets and monocytes/macrophages exhibiting complete CD36 deficiency. The type II phenotype lacks the surface expression of CD36 in platelets, but expression in monocytes/macrophages is near normal. {ECO:0000269|PubMed:11950861, ECO:0000269|PubMed:7533783}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients also have postprandial hypertriglyceridemia, insulin resistance and hypertension increasing atherosclerotic risk. {ECO:0000269|PubMed:18753675}.	EDTA Blood Sample
[Sıtma, Direnç], 611162 (Sıtma) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Malaria, resistance to], 611162 (Malaria) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Bellek, Gelişitirilmiş, QTL], 615602 (WWC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Memory, enhanced, QTL], 615602 (WWC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WWC1	WWC1, KIBRA, KIAA0869, MEMRYQTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Yenilik Arayışı Kişilik], 601696,Otozomal Dominant (DRD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Novelty seeking personality], 601696, Autosomal dominant (DRD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD4	DRD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Obezite, Direnç] (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Obesity, resistance to] (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CIMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Pentosuriya], 260800, Otozomal Resesif; PNTSU (Pentozüri) (DCXR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Pentosuria], 260800, Autosomal recessive; PNTSU (Pentosuria) (DCXR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCXR	DCXR, P34H, PNTSU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Feniltiokarbamid Tatmal], 171200, Otozomal Dominant (TAS2R38 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Phenylthiocarbamide tasting], 171200, Autosomal dominant (TAS2R38 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAS2R38	TAS2R38, T2R61, PTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Fosfohidroksilizinüri], 615011 (PHYKPL gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Phosphohydroxylysinuria], 615011 (PHYKPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHYKPL	PHYKPL, AGXT2L2, PHLU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phosphohydroxylysinuria (PHLU) [MIM:615011]: A condition characterized by elevated phosphohydroxylysine in the urine. There is no clinical phenotype associated with this finding other than the urinary metabolites. {ECO:0000269|PubMed:23242558}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Plasenta Laktojen Eksikliği] (CSH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Placental lactogen deficiency] (CSH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSH1	CSH1, CSA, PL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Plazma Glikoz, 2 Saat, QTL 2], 137241 (GIPR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Plasma glucose, 2-hour, QTL 2], 137241 (GIPR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GIPR	GIPR, PGQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Prematüre Kromatid Ayrımı Özelliği], 176430, Otozomal Dominant (BUB1B gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Premature chromatid separation trait], 176430, Autosomal dominant (BUB1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BUB1B	BUB1B, BUBR1, MVA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature chromatid separation trait (PCS) [MIM:176430]: Consists of separate and splayed chromatids with discernible centromeres and involves all or most chromosomes of a metaphase. It is found in up to 2% of metaphases in cultured lymphocytes from approximately 40% of normal individuals. When PCS is present in 5% or more of cells, it is known as the heterozygous PCS trait and has no obvious phenotypic effect, although some have reported decreased fertility. Inheritance is autosomal dominant. {ECO:0000269|PubMed:16411201}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
[Protein Z Eksikliği], 614024 (PROZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Protein Z deficiency], 614024 (PROZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROZ	PROZ, PZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kalp Atış Hızı], 607276 (ADRB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Resting heart rate], 607276 (ADRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB1	ADRB1, ADRB1R, RHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Rh-Negatif Kan Tipi] (RHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Rh-negative blood type] (RHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHD	RHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Sarkozinemi], 268900, Otozomal Resesif; SARCOS (Sarkozinemi) (SARDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Sarcosinemia], 268900, Autosomal recessive; SARCOS (Sarcosinemia) (SARDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SARDH	SARDH, SARD, SAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sarcosinemia (SARCOS) [MIM:268900]: A metabolic disorder characterized by an increased concentration of sarcosine in plasma and an increased excretion of sarcosine in urine. Sarcosinemia is most probably a benign condition without significant clinical problems. Some reports have associated sarcosinemia with mental retardation and neurologic problems. {ECO:0000269|PubMed:10444331, ECO:0000269|PubMed:22825317}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Sedoheptulokinase Eksikliği], 617213, Otozomal Resesif (Izole Sedoheptulokinase Eksikliği) (SHPK geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Sedoheptulokinase deficiency], 617213, Autosomal recessive (Isolated sedoheptulokinase deficiency) (SHPK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHPK	SHPK, CARKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Kısa Uyku], 612975, Otozomal Dominant (BHLHE41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Short sleeper], 612975, Autosomal dominant (BHLHE41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BHLHE41	BHLHE41, BHLHB3, DEC2, SHARP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 1, Sarışın / Kahverengi Saç], 227220, Otozomal Resesif (HERC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 1, blond/brown hair], 227220, Autosomal recessive (HERC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HERC2	HERC2, SHEP1, MRT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 1, Sarışın / Kahverengi Saç], 227220, Otozomal Resesif (Okulokütanöz Albinizm Tip 2) (OCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 1, blond/brown hair], 227220, Autosomal recessive (Oculocutaneous albinism type 2) (OCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCA2	OCA2, P, PED, D15S12, BOCA, EYCL3, HCL3, SHEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 1, Mavi / Mavi Olmayan Gözler], 227220, Otozomal Resesif (OCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220, Autosomal recessive (OCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCA2	OCA2, P, PED, D15S12, BOCA, EYCL3, HCL3, SHEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 1, Mavi / Mavi Olmayan Gözler], 227220, Otozomal Resesif (HERC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220, Autosomal recessive (HERC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HERC2	HERC2, SHEP1, MRT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 10, Sarı / Kahverengi Saç], 612267 (TPCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 10, blond/brown hair], 612267 (TPCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPCN2	TPCN2, TPC2, SHEP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 2, Sarışın / Açık Ten], 266300, Otozomal Resesif (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 2, blond hair/fair skin], 266300, Autosomal recessive (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 2, Kızıl Saç / Açık Ten], 266300, Otozomal Resesif (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 2, red hair/fair skin], 266300, Autosomal recessive (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 3, Mavi / Yeşil Gözler], 601800 (Oküler Albinizm-Konjenital Sensorinöral Sağırlık) (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (Ocular albinism with congenital sensorineural deafness) (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 3, Mavi / Yeşil Gözler], 601800 (Oküler Albinizm-Konjenital Sensorinöral Sağırlık) (MLPA)	[Skin/hair/eye pigmentation 3, blue/green eyes], 601800 (Ocular albinism with congenital sensorineural deafness) (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	20	60	.	.	.	.	30	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 3, Açık /Koyu / Çilli Deri], 601800 (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 3, Açık /Koyu / Çilli Deri], 601800 (MLPA)	[Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800 (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	20	60	.	.	.	.	30	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 4, Sarışın / Koyu Deri], 113750, Otozomal Resesif (Okulokütanöz Albinizm Tip 6) (SLC24A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 4, fair/dark skin], 113750, Autosomal recessive (Oculocutaneous albinism type 6) (SLC24A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A5	SLC24A5, NCKX5, SHEP4, OCA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 5, Siyah / Siyah-Olmayan Saç], 227240, Otozomal Resesif (SLC45A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240, Autosomal recessive (SLC45A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC45A2	SLC45A2, MATP, AIM1, SHEP5, OCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

[Deri / Saç / Göz Pigmentasyonu 5, Koyu / Açık Ten], 227240, Otozomal Resesif (SLC45A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 5, dark/fair skin], 227240, Autosomal recessive (SLC45A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC45A2	SLC45A2, MATP, AIM1, SHEP5, OCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 5, Açık / Koyu Gözler], 227240, Otozomal Resesif (SLC45A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 5, dark/light eyes], 227240, Autosomal recessive (SLC45A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC45A2	SLC45A2, MATP, AIM1, SHEP5, OCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 6, Sarışın / Kahverengi Saç], 210750, Otozomal Resesif (SLC24A4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750, Autosomal recessive (SLC24A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A4	SLC24A4, NCKX4, SHEP6, AI2A5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 6, Mavi / Yeşil Gözler], 210750, Otozomal Resesif (SLC24A4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 6, blue/green eyes], 210750, Autosomal recessive (SLC24A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A4	SLC24A4, NCKX4, SHEP6, AI2A5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 7, Sarı / Kahverengi Saç], 611664 (Ailesel Progresif Hiper ve Hipopigmentasyon) (KITLG gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 (Familial progressive hyper- and hypopigmentation) (KITLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KITLG	KITLG, MGF, SF, SCF, SHEP7, FPHH, DCUA, DFNA69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 9, Kahverengi / Kahverengi Olmayan Gözler], 611742 (ASIP gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742 (ASIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASIP	ASIP, AGTIL, SHEP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu 9, Koyu / Açık Saç], 611742 (ASIP gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation 9, dark/light hair], 611742 (ASIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASIP	ASIP, AGTIL, SHEP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu, Varyasyon, 11 (Melanezyen Sarışın Saç)], 612271 (TYRP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271 (TYRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYRP1	TYRP1, CAS2, GP75, SHEP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Deri / Saç / Göz Pigmentasyonu, Varyasyon, 8], 611724 (IRF4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Skin/hair/eye pigmentation, variation in, 8], 611724 (IRF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF4	IRF4, LSIRF, SHEP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Sprint Performansı] (ACTN3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Sprinting performance] (ACTN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN3	ACTN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Statinler, Zayıf Kolesterol Düşürücü Etki (HMGCR gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Statins, attenuated cholesterol lowering by] (HMGCR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGCR	HMGCR, LDLCQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Süperoksit Dismutaz, Yüksek Hücre Dışı] (SOD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Superoxide dismutase, elevated extracellular] (SOD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOD3	SOD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Troksin Bağlayıcı Globulin QTL], 300932 (SERPINA7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Thyroxine-binding globulin QTL], 300932 (SERPINA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA7	TBG, TBGQTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroxine-binding globulin deficiency (TBG deficiency) [MIM:314200]: Mutations in the SERPINA7 gene can result as a whole spectrum of deficiencies, characterized by either reduced or increased TBG levels in the serum. Patients show, respectively, reduced or elevated protein-bound iodine but are euthyroid. (ECO:0000269|PubMed:1294376, ECO:0000269|PubMed:1515456, ECO:0000269|PubMed:1901689, ECO:0000269|PubMed:1906047, ECO:0000269|PubMed:2155256, ECO:0000269|PubMed:2501669). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
[Transferin Serum Seviyesi QTL2'nin], 614193 (HFE gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Transferin serum level QTL2], 614193 (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Ürik Asit Konsantrasyonu, Serum, QTL1], 138900, Otozomal Dominant (ABCG2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Uric acid concentration, serum, QTL1], 138900, Autosomal dominant (ABCG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCG2	ABCG2, BCRP, ABCP, UAQTL1, GOUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Ürik Asit Konsantrasyonu, Serum, QTL4], 612671, Otozomal Dominant (SLC17A3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[Uric acid concentration, serum, QTL4], 612671, Autosomal dominant (SLC17A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A3	SLC17A3, NPT4, UAQTL4, GOUT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
[Beyaz Kan Hücre Sayısı QTL], 611862 (ACKR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	[White blood cell count QTL], 611862 (ACKR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACKR1	DARC, FY, GPD, WBCQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
10p subtel (FISH)	10p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
10p subtel (FISH) (Prenatal)	10p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
10q subtel (FISH)	10q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
10q subtel (FISH) (Prenatal)	10q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
11o subtel (FISH)	11o subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
11p subtel (FISH) (Prenatal)	11p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
11q subtel (FISH)	11q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
11q subtel (FISH) (Prenatal)	11q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
11q22.3 Delesyonu (ATM) (FISH)	11q22.3 (ATM Deletion) (FISH)	11q22.3	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
11q23 Delesyonu (FISH)	11q23 Deletion (FISH)	11q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)	*	10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
11q23 Delesyonu (FISH) (Prenatal)	11q23 Deletion (FISH) (Prenatal)	11q23	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
12 alfa satellit (FISH)	12 alpha satellite (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
12 alfa satellit (FISH) (Prenatal)	12 alpha satellite (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
12p subtel (FISH)	12p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
12p subtel (FISH) (Prenatal)	12p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
12q subtel (FISH)	12q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
12q subtel (FISH) (Prenatal)	12q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
13q subtel (FISH)	13q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
13q subtel (FISH) (Prenatal)	13q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
13q14.2 Delesyonu (RB1) (FISH)	13q14.2 Deletion (RB1) (FISH)	13q14.2	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
13q14.2 Delesyonu (RB1) (FISH) (Prenatal)	13q14.2 Deletion (RB1) (FISH) (Prenatal)	13q14.2	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
14q subtel (FISH)	14q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
14q subtel (FISH) (Prenatal)	14q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
15 alfa satellit (FISH)	15 alpha satellite (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
15 alfa satellit (FISH) (Prenatal)	15 alpha satellite (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
15q subtel (FISH)	15q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
15q subtel (FISH) (Prenatal)	15q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
16p subtel (FISH)	16p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
16p subtel (FISH) (Prenatal)	16p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
16q subtel (FISH)	16q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
16q subtel (FISH) (Prenatal)	16q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17-Alfa-Hidroksilaz / 17,20-Liyaz Eksikliği, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP17A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	17-alpha-hydroxylase/17,20-lyase deficiency, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late -onset (NC or LOH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

17-Alfa-Hidroksilaz / 17,20-Liyaz Eksikliği, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (MLPA)	17-alpha-hydroxylase/17,20-lyase deficiency, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
17-Alfa-Hidroksilaz / 17,20-Liyaz Eksikliği, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP17A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	17-alpha-hydroxylase/17,20-lyase deficiency, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17-Alfa-Hidroksilaz / 17,20-Liyaz Eksikliği, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (Prenatal) (MLPA)	17-alpha-hydroxylase/17,20-lyase deficiency, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (Prenatal) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17-Beta-Hidroksi Steroid Dehidrojenaz 10 Eksikliği, 300438, X'e Bağlı Dominant (HSD10 Hastalığı) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438, X-linked dominant (HSD10 disease) (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
17-Beta-Hidroksi Steroid Dehidrojenaz 10 Eksikliği, 300438, X'e Bağlı Dominant (HSD10 Hastalığı) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	17-beta-hydroxysteroid dehydrogenase X deficiency, 300438, X-linked dominant (HSD10 disease) (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17,20-Liyaz Eksikliği, İzole, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP17A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	17,20-lyase deficiency, isolated, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
17,20-Liyaz Eksikliği, İzole, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (MLPA)	17,20-lyase deficiency, isolated, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

17,20-Liyaz Eksikliği, İzole, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP17A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	17,20-lyase deficiency, isolated, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17,20-Liyaz Eksikliği, İzole, 202110, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (Prenatal) (MLPA)	17,20-lyase deficiency, isolated, 202110, Autosomal recessive (Congenital adrenal hyperplasia) (Prenatal) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17p subtel (FISH)	17p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
17p subtel (FISH) (Prenatal)	17p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith Magenis sendromu (17p11.2 mikrodelesyonu) (FISH)	17p11.2 deletion syndrome (FISH)	17p11.2	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Smith Magenis sendromu (17p11.2 mikrodelesyonu) (Prenatal) (FISH)	17p11.2 deletion syndrome (FISH) (Prenatal)	17p11.2	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miller-Dieker syndrome /17p13.3 Delesyon Sendromu / 1izole lizensefali/ subkortikal band heterotopisi (FISH)	17p13.3 deletion syndrome (FISH)	17p13.3	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Miller-Dieker syndrome /17p13.3 Delesyon Sendromu / 1izole lizensefali/ subkortikal band heterotopisi (FISH) (Prenatal)	17p13.3 deletion syndrome (FISH) (Prenatal)	17p13.3	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17q subtel (FISH)	17q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
17q subtel (FISH) (Prenatal)	17q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
18p subtel (FISH)	18p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
18p subtel (FISH) (Prenatal)	18p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
18q subtel (FISH)	18q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
18q subtel (FISH) (Prenatal)	18q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
19p subtel (FISH)	19p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
19p subtel (FISH) (Prenatal)	19p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
E2A (TCF3) (19p13.3) Breakapart (FISH)	E2A (TCF3) (19p13.3) Breakapart (FISH)	19p13	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
19q subtel (FISH)	19q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
19q subtel (FISH) (Prenatal)	19q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
1p subtel (FISH)	1p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
1p subtel (FISH) (Prenatal)	1p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
1p/ 19q Delesyon Analizi (FISH)	1p/19q deletion analysis (FISH)	.	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinde doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
1q36 sendromu (FISH)	1q36 syndrome (FISH)	1q36	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
1q36 sendromu (FISH) (Prenatal)	1q36 syndrome (FISH) (Prenatal)	1q36	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
1q subtel (FISH)	1q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
1q subtel (FISH) (Prenatal)	1q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
2-Aminoadipik 2-Oksoadipik Asidüri, 204750, Otozomal Resesif; AMOXAD (2-Aminoadipik 2-Oksoadipik Asidüri) (DHTKD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	2-aminoadipic 2-oxoadipic aciduria, 204750, Autosomal recessive; AMOXAD (2-aminoadipic 2-oxoadipic aciduria) (DHTKD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHTKD1	DHTKD1, KIAA1630, AMOXAD, CMT2Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 2-aminoadipic 2-oxoadipic aciduria (AMOXAD) [MIM:204750]: A metabolic disorder characterized by increased levels of 2- oxoadipate and 2-hydroxyadipate in the urine, and elevated 2- aminoadipate in the plasma. Patients can have mild to severe intellectual disability, muscular hypotonia, developmental delay, ataxia, and epilepsy. Most cases are asymptomatic. (ECO:0000269	PubMed:23141293). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
2-Aminoadipik 2-Oksoadipik Asidüri, 204750, Otozomal Resesif; AMOXAD (2-Aminoadipik 2-Oksoadipik Asidüri) (DHTKD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	2-aminoadipic 2-oxoadipic aciduria, 204750, Autosomal recessive; AMOXAD (2-aminoadipic 2-oxoadipic aciduria) (DHTKD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHTKD1	DHTKD1, KIAA1630, AMOXAD, CMT2Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 2-aminoadipic 2-oxoadipic aciduria (AMOXAD) [MIM:204750]: A metabolic disorder characterized by increased levels of 2- oxoadipate and 2-hydroxyadipate in the urine, and elevated 2- aminoadipate in the plasma. Patients can have mild to severe intellectual disability, muscular hypotonia, developmental delay, ataxia, and epilepsy. Most cases are asymptomatic. (ECO:0000269	PubMed:23141293). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
2-Metilbutirilglisinüri, 610006, Otozomal Resesif (2-Metilbutiril-CoA Dehidrogenaz Eksikliği) (ACADSB geni) (Tüm Gen Dizi Analizi) (Postnatal)	2-methylbutyrylglycinuria, 610006, Autosomal recessive (2-methylbutyryl-CoA dehydrogenase deficiency) (ACADSB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACADSB	ACADSB, SBCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
2-Metilbutirilglisinüri, 610006, Otozomal Resesif (2-Metilbutiril-CoA Dehidrogenaz Eksikliği) (ACADSB geni) (Tüm Gen Dizi Analizi) (Prenatal)	2-methylbutyrylglycinuria, 610006, Autosomal recessive (2-methylbutyryl-CoA dehydrogenase deficiency) (ACADSB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACADSB	ACADSB, SBCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
2,4-Dienoyl-CoA Redüktaz Eksikliği, 616034, Otozomal Resesif; DECRD (Progresif Ensefalopati-Lökodistrofi, DECR Eksikliğine Bağlı) (NADK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	2,4-dienoyl-CoA reductase deficiency, 616034, Autosomal recessive; DECRD (Progressive encephalopathy with leukodystrophy due to DECR deficiency) (NADK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NADK2	NADK2, C5orf33, DECRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
2,4-Dienoyl-CoA Redüktaz Eksikliği, 616034, Otozomal Resesif; DECRD (Progresif Ensefalopati-Lökodistrofi, DECR Eksikliğine Bağlı) (NADK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	2,4-dienoyl-CoA reductase deficiency, 616034, Autosomal recessive; DECRD (Progressive encephalopathy with leukodystrophy due to DECR deficiency) (NADK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NADK2	NADK2, C5orf33, DECRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
20p subtel (FISH)	20p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
20p subtel (FISH) (Prenatal)	20p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
20q subtel (FISH)	20q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
20q subtel (FISH) (Prenatal)	20q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
20q Delesyonu (20q12-20q13.1) (FISH)	20q Deletion (20q12-20q13.1) (FISH)	20q12	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
21q subtel (FISH)	21q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
21q subtel (FISH) (Prenatal)	21q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
22q subtel (FISH)	22q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
22q subtel (FISH) (Prenatal)	22q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
22q11 Mikrodelesyonu/ DiGeorge/ VCFS Sendrom (FISH)	22q11 microdeletion/ DiGeorge / VCFS syndrome (FISH)	22q11	.	FISH	Heparinli Kan (2-4 ml)	*	10	30	.	.	20	Postnatal	.	Heparinized blood sample
22q11 Mikrodelesyonu/ DiGeorge/ VCFS Sendrom (FISH) (Prenatal)	22q11 microdeletion/ DiGeorge / VCFS syndrome (FISH) (Prenatal)	22q11	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
2o subtel (FISH)	2o subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
2p subtel (FISH) (Prenatal)	2p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
2q subtel (FISH)	2q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample

2q subtel (FISH) (Prenatal)	2q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Hidroksi-3-Metilglutaril-CoA Liyaz Eksikliği, HMGCLD (HMG-CoA Liyaz Eksikliği, 246450, Otozomal Resesif) (3-Hidroksi-3-Metilglutarik Asiduri) (HMGCL geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY; HMGCLD (HMG-CoA lyase deficiency, 246450, Autosomal recessive) (3-hydroxy-3-methylglutaric aciduria) (HMGCL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGCL	HMGCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Hidroksi-3-Metilglutaril-CoA Liyaz Eksikliği, HMGCLD (HMG-CoA Liyaz Eksikliği, 246450, Otozomal Resesif) (3-Hidroksi-3-Metilglutarik Asiduri) (HMGCL geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-HYDROXY-3-METHYLGLUTARYL-CoA LYASE DEFICIENCY; HMGCLD (HMG-CoA lyase deficiency, 246450, Autosomal recessive) (3-hydroxy-3-methylglutaric aciduria) (HMGCL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HMGCL	HMGCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Hidroksiasİl-CoA Dehidrojenaz Yetmezliği, 231530, Otozomal Resesif (Hiperinsülinizm, Kısa Zincirli 3-Hydroxyacyl-CoA Dehidrojenaz Yetmezliğine Bağlı) (HADH geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530, Autosomal recessive (Hyperinsulinism due to short chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADH	HADHSC, SCHAD, HHF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3-alpha-hydroxyacyl-CoA dehydrogenase deficiency (HADH deficiency) [MIM:231530]: A metabolic disorder with various clinical presentations including hypoglycemia, hepatoencephalopathy, myopathy or cardiomyopathy, and in some cases sudden death. (ECO:0000269	Ref.13). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
3-Hidroksiasİl-CoA Dehidrojenaz Yetmezliği, 231530, Otozomal Resesif (Hiperinsülinizm, Kısa Zincirli 3-Hydroxyacyl-CoA Dehidrojenaz Yetmezliğine Bağlı) (HADH geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-hydroxyacyl-CoA dehydrogenase deficiency, 231530, Autosomal recessive (Hyperinsulinism due to short chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADH	HADHSC, SCHAD, HHF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3-alpha-hydroxyacyl-CoA dehydrogenase deficiency (HADH deficiency) [MIM:231530]: A metabolic disorder with various clinical presentations including hypoglycemia, hepatoencephalopathy, myopathy or cardiomyopathy, and in some cases sudden death. (ECO:0000269	Ref.13). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Hidroksibütiril-CoA Hidrolaz Eksikliği, 250620, Otozomal Resesif; HIBCHD (Nörodejenerasyon, 3-Hidroksibütiril-CoA Hidrolaz Eksikliğine Bağlı) (HIBCH geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-hydroxyisobutyryl-CoA hydrolase deficiency, 250620, Autosomal recessive; HIBCHD (Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency) (HIBCH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HIBCH	HIBCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Hidroksibütiril-CoA Hidrolaz Eksikliği, 250620, Otozomal Resesif; HIBCHD (Nörodejenerasyon, 3-Hidroksibütiril-CoA Hidrolaz Eksikliğine Bağlı) (HIBCH geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-hydroxyisobutyryl-CoA hydrolase deficiency, 250620, Autosomal recessive; HIBCHD (Neurodegeneration due to 3-hydroxyisobutyryl-CoA hydrolase deficiency) (HIBCH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HIBCH	HIBCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-M Sendromu 1, 273750, Otozomal Resesif; 3M1 (3M Sendromu) (CUL7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-M syndrome 1, 273750, Autosomal recessive; 3M1 (3M syndrome) (CUL7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CUL7	CUL7, 3M1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3M syndrome 1 (3M1) [MIM:273750]: An autosomal recessive disorder characterized by severe pre- and postnatal growth retardation, facial dysmorphism, large head circumference, and normal intelligence and endocrine function. Skeletal changes include long slender tubular bones and tall vertebral bodies. (ECO:0000269|PubMed:16142236, ECO:0000269|PubMed:17675530, ECO:0000269|PubMed:23018678). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
3-M Sendromu 1, 273750, Otozomal Resesif; 3M1 (3M Sendromu) (CUL7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-M syndrome 1, 273750, Autosomal recessive; 3M1 (3M syndrome) (CUL7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CUL7	CUL7, 3M1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3M syndrome 1 (3M1) [MIM:273750]: An autosomal recessive disorder characterized by severe pre- and postnatal growth retardation, facial dysmorphism, large head circumference, and normal intelligence and endocrine function. Skeletal changes include long slender tubular bones and tall vertebral bodies. (ECO:0000269|PubMed:16142236, ECO:0000269|PubMed:17675530, ECO:0000269|PubMed:23018678). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-M Sendromu 2, 612921; 3M2 (3M Sendromu) (OBSL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-M syndrome 2, 612921; 3M2 (3M syndrome) (OBSL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OBSL1	OBSL1, KIAA0657, 3M2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-M Sendromu 2, 612921; 3M2 (3M Sendromu) (OBSL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-M syndrome 2, 612921; 3M2 (3M syndrome) (OBSL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OBSL1	OBSL1, KIAA0657, 3M2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-M Sendromu 3, 614205, Otozomal Resesif; 3M3 (3M Sendromu) (CCDC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-M syndrome 3, 614205, Autosomal recessive; 3M3 (3M syndrome) (CCDC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC8	CCDC8, 3M3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-M Sendromu 3, 614205, Otozomal Resesif; 3M3 (3M Sendromu) (CCDC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-M syndrome 3, 614205, Autosomal recessive; 3M3 (3M syndrome) (CCDC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC8	CCDC8, 3M3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilkrotonil-CoA Karboksilaz 1 Eksikliği, 210200, Otozomal Resesif; MCC1D (3 -Metilkrotonilglisinüri 1) (3-Metilkrotonil-CoA Karboksilaz Eksikliği) (MCCC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200, Autosomal recessive; MCC1D (3 -methylcrotonylglycinuria 1) (3-methylcrotonyl-CoA carboxylase deficiency) (MCCC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCCC1	MCCC1, MCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3-methylcrotonoyl-CoA carboxylase 1 deficiency (MCC1D) [MIM:210200]: An autosomal recessive disorder of leucine catabolism. The phenotype is variable, ranging from neonatal onset with severe neurological involvement to asymptomatic adults. There is a characteristic organic aciduria with massive excretion of 3- hydroxyisovaleric acid and 3-methylcrotonylglycine, usually in combination with a severe secondary carnitine deficiency. (ECO:0000269|PubMed:11170888, ECO:0000269|PubMed:11181649, ECO:0000269|PubMed:11406611, ECO:0000269|PubMed:16010683, ECO:0000269|PubMed:17968484, ECO:0000269|PubMed:21071250, ECO:0000269|PubMed:22150417, ECO:0000269|PubMed:22264772, ECO:0000269|PubMed:22642865, ECO:0000269|Ref.6). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
3-Metilkrotonil-CoA Karboksilaz 1 Eksikliği, 210200, Otozomal Resesif; MCC1D (3 -Metilkrotonilglisinüri 1) (3-Metilkrotonil-CoA Karboksilaz Eksikliği) (MCCC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200, Autosomal recessive; MCC1D (3 -methylcrotonylglycinuria 1) (3-methylcrotonyl-CoA carboxylase deficiency) (MCCC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCCC1	MCCC1, MCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3-methylcrotonoyl-CoA carboxylase 1 deficiency (MCC1D) [MIM:210200]: An autosomal recessive disorder of leucine catabolism. The phenotype is variable, ranging from neonatal onset with severe neurological involvement to asymptomatic adults. There is a characteristic organic aciduria with massive excretion of 3- hydroxyisovaleric acid and 3-methylcrotonylglycine, usually in combination with a severe secondary carnitine deficiency. (ECO:0000269|PubMed:11170888, ECO:0000269|PubMed:11181649, ECO:0000269|PubMed:11406611, ECO:0000269|PubMed:16010683, ECO:0000269|PubMed:17968484, ECO:0000269|PubMed:21071250, ECO:0000269|PubMed:22150417, ECO:0000269|PubMed:22264772, ECO:0000269|PubMed:22642865, ECO:0000269|Ref.6). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilkrotonil-CoA Karboksilaz 2 Eksikliği, 210210, Otozomal Resesif; MCC2D (3 -Metilkrotonilglisinüri 2) (3-Metilkrotonil-CoA Karboksilaz Eksikliği) (MCCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210, Autosomal recessive; MCC2D (3 -methylcrotonylglycinuria 2) (3-methylcrotonyl-CoA carboxylase deficiency) (MCCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCCC2	MCCC2, MCCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3-methylcrotonoyl-CoA carboxylase 2 deficiency (MCC2D) [MIM:210210]: An autosomal recessive disorder of leucine catabolism. The phenotype is variable, ranging from neonatal onset with severe neurological involvement to asymptomatic adults. There is a characteristic organic aciduria with massive excretion of 3- hydroxyisovaleric acid and 3-methylcrotonylglycine, usually in combination with a severe secondary carnitine deficiency. (ECO:0000269|PubMed:11170888, ECO:0000269|PubMed:11181649, ECO:0000269|PubMed:11406611, ECO:0000269|PubMed:16010683, ECO:0000269|PubMed:17968484, ECO:0000269|PubMed:21071250, ECO:0000269|PubMed:22150417, ECO:0000269|PubMed:22264772, ECO:0000269|PubMed:22642865). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
3-Metilkrotonil-CoA Karboksilaz 2 Eksikliği, 210210, Otozomal Resesif; MCC2D (3 -Metilkrotonilglisinüri 2) (3-Metilkrotonil-CoA Karboksilaz Eksikliği) (MCCC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210, Autosomal recessive; MCC2D (3 -methylcrotonylglycinuria 2) (3-methylcrotonyl-CoA carboxylase deficiency) (MCCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCCC2	MCCC2, MCCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3-methylcrotonoyl-CoA carboxylase 2 deficiency (MCC2D) [MIM:210210]: An autosomal recessive disorder of leucine catabolism. The phenotype is variable, ranging from neonatal onset with severe neurological involvement to asymptomatic adults. There is a characteristic organic aciduria with massive excretion of 3- hydroxyisovaleric acid and 3-methylcrotonylglycine, usually in combination with a severe secondary carnitine deficiency. (ECO:0000269|PubMed:11170888, ECO:0000269|PubMed:11181649, ECO:0000269|PubMed:11406611, ECO:0000269|PubMed:16010683, ECO:0000269|PubMed:17968484, ECO:0000269|PubMed:21071250, ECO:0000269|PubMed:22150417, ECO:0000269|PubMed:22264772, ECO:0000269|PubMed:22642865). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asidüri, Sağırlık, Ensefalopati ve Leigh Benzeri Sendrom, 614739, Otozomal Resesif; MEGDEL (MEGDEL Sendromu) (SERAC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739, Autosomal recessive; MEGDEL (MEGDEL syndrome) (SERAC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERAC1	SERAC1, MEGDEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Metilglutakonik Asidüri, Sağırlık, Ensefalopati ve Leigh Benzeri Sendrom, 614739, Otozomal Resesif; MEGDEL (MEGDEL Sendromu) (SERAC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome, 614739, Autosomal recessive; MEGDEL (MEGDEL syndrome) (SERAC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERAC1	SERAC1, MEGDEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asitüri, Tip 1, 250950, Otozomal Resesif; MGCA1 (3 -Metilglutakonik Asidüri Tip 1) (AUH geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria, type I, 250950, Autosomal recessive; MGCA1 (3-methylglutaconic aciduria type 1) (AUH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AUH	AUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3-methylglutaconic aciduria 1 (MGA1) [MIM:250950]: An inborn error of leucine metabolism. It leads to an autosomal recessive syndrome with variable clinical phenotype, ranging from delayed speech development to severe psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia. MGA1 can be distinguished from other forms of MGA by the pattern of metabolite excretion: 3-methylglutaconic acid levels are higher than those detected in other forms, whereas methylglutaric acid levels are usually only slightly elevated and there is a high level of 3-hydroxyisovaleric acid excretion (not present in other MGA forms). [ECO:0000269	PubMed:12655555]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

3-Metilglutakonik Asitüri, Tip 1, 250950, Otozomal Resesif; MGCA1 (3-Metilglutakonik Asidüri Tip 1) (AUH geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria, type I, 250950, Autosomal recessive; MGCA1 (3-methylglutaconic aciduria type 1) (AUH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AUH	AUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3-methylglutaconic aciduria 1 (MGA1) [MIM:250950]: An inborn error of leucine metabolism. It leads to an autosomal recessive syndrome with variable clinical phenotype, ranging from delayed speech development to severe psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia. MGA1 can be distinguished from other forms of MGA by the pattern of metabolite excretion: 3-methylglutaconic acid levels are higher than those detected in other forms, whereas methylglutaric acid levels are usually only slightly elevated and there is a high level of 3-hydroxyisovaleric acid excretion (not present in other MGA forms). {ECO:0000269|PubMed:12655553}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asitüri, Tip 3, 258501, Otozomal Resesif; MGCA3 (3-Metilglutakonik Asidüri Tip 3) (OPA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria, type III, 258501, Autosomal recessive; MGCA3 (3-methylglutaconic aciduria type 3) (OPA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA3	OPA3, MGA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Metilglutakonik Asitüri, Tip 3, 258501, Otozomal Resesif; MGCA3 (3-Metilglutakonik Asidüri Tip 3) (OPA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria, type III, 258501, Autosomal recessive; MGCA3 (3-methylglutaconic aciduria type 3) (OPA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPA3	OPA3, MGA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asidüri, Tip 5, 610198, Otozomal Resesif; MGCA5 (Dilate Kardiyomiyopati-Ataksi) (DNAJC19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria, type V, 610198, Autosomal recessive; MGCA5 (Dilated cardiomyopathy with ataxia) (DNAJC19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC19	DNAJC19, TIM14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Metilglutakonik Asidüri, Tip 5, 610198, Otozomal Resesif; MGCA5 (Dilate Kardiyomiyopati-Ataksi) (DNAJC19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria, type V, 610198, Autosomal recessive; MGCA5 (Dilated cardiomyopathy with ataxia) (DNAJC19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNAJC19	DNAJC19, TIM14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asidüri, Tip 7, Katarakt, Nörolojik Tutulum ve Nötropeni, 616271, Otozomal Resesif; MEGCANN (3-Metilglutakonik Asidüri Tip 7) (CLPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271, Autosomal recessive; MEGCANN (3-methylglutaconic aciduria type 7) (CLPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLPB	CLPB, SKD3, MEGCANN, MGCA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Metilglutakonik Asidüri, Tip 7, Katarakt, Nörolojik Tutulum ve Nötropeni, 616271, Otozomal Resesif; MEGCANN (3-Metilglutakonik Asidüri Tip 7) (CLPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria, type VII, with cataracts, neurologic involvement and neutropenia, 616271, Autosomal recessive; MEGCANN (3-methylglutaconic aciduria type 7) (CLPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLPB	CLPB, SKD3, MEGCANN, MGCA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3-Metilglutakonik Asitüri, Tip 8, 617248, Otozomal Resesif (Geç Başlangıçlı Parkinson Hastalığı) (HTRA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3-methylglutaconic aciduria, type VIII, 617248, Autosomal recessive (Young-onset Parkinson disease) (HTRA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA2	HTRA2, OMI, PARK13, PRSS25, MGCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3-Metilglutakonik Asitüri, Tip 8, 617248, Otozomal Resesif (Geç Başlangıçlı Parkinson Hastalığı) (HTRA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3-methylglutaconic aciduria, type VIII, 617248, Autosomal recessive (Young-onset Parkinson disease) (HTRA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HTRA2	HTRA2, OMI, PARK13, PRSS25, MGCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3MC Sendromu 1, 257920, Otozomal Resesif (Michels Sendromu) (MASP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3MC syndrome 1, 257920, Autosomal recessive; 3MC1 (Michels syndrome) (MASP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MASP1	MASP1, CRARF, 3MC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3MC syndrome 1 (3MC1) [MIM:257920]: A disorder characterized by facial dysmorphism that includes hypertelorism, blepharophimosis, blepharoptosis and highly arched eyebrows, cleft lip and/or palate, craniosynostosis, learning disability and genital, limb and vesicorenal anomalies. The term 3MC syndrome includes Carnevale, Mingarelli, Malpuech, and Michels syndromes. {ECO:0000269|PubMed:21258343}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
3MC Sendromu 1, 257920, Otozomal Resesif (Michels Sendromu) (Prenatal) (MASP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3MC syndrome 1, 257920, Autosomal recessive; 3MC1 (Michels syndrome) (MASP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MASP1	MASP1, CRARF, 3MC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3MC syndrome 1 (3MC1) [MIM:257920]: A disorder characterized by facial dysmorphism that includes hypertelorism, blepharophimosis, blepharoptosis and highly arched eyebrows, cleft lip and/or palate, craniosynostosis, learning disability and genital, limb and vesicorenal anomalies. The term 3MC syndrome includes Carnevale, Mingarelli, Malpuech, and Michels syndromes. {ECO:0000269|PubMed:21258343}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3MC Sendromu 2, 265050, Otozomal Resesif; 3MC2 (Carnevale Sendromu) (COLEC11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3MC syndrome 2, 265050, Autosomal recessive; 3MC2 (Carnevale syndrome) (COLEC11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COLEC11	COLEC11, CLK1, 3MC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3MC Sendromu 2, 265050, Otozomal Resesif; 3MC2 (Carnevale Sendromu) (Prenatal) (COLEC11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3MC syndrome 2, 265050, Autosomal recessive; 3MC2 (Carnevale syndrome) (COLEC11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COLEC11	COLEC11, CLK1, 3MC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3MC Sendromu 3, 3MC3 (Malpuech Sendromu) (Malpuech Sendromu) (COLEC10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	3MC SYNDROME 3; 3MC3 (Malpuech syndrome) (Malpuech syndrome) (COLEC10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COLEC10	COLEC10, CLC13, 3MC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
3MC Sendromu 3, 3MC3 (Malpuech Sendromu) (Malpuech Sendromu) (Prenatal) (COLEC10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	3MC SYNDROME 3; 3MC3 (Malpuech syndrome) (Malpuech syndrome) (COLEC10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COLEC10	COLEC10, CLC13, 3MC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3p subtel (FISH)	3p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
3p subtel (FISH) (Prenatal)	3p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3p Sendromu, 613792, Otozomal Dominant (Distal Monozomi 3p) (2200)	3p- syndrome, 613792, Autosomal dominant (Distal monosomy 3p) (440)	.	DEL3pterp25, C3DEL pterp25	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
3p Sendromu, 613792, Otozomal Dominant (Distal Monozomi 3p) (Prenatal)	3p- syndrome, 613792, Autosomal dominant (Distal monosomy 3p) (Prenatal)	.	DEL3pterp25, C3DEL pterp25	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
3q subtel (FISH)	3q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
3q subtel (FISH) (Prenatal)	3q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu 1, 400045 (46,XX Overtestiküler Cinsiyet Farklılaşma Bozukluğu) (SRY geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XX sex reversal 1, 400045 (46,XX ovotesticular disorder of sex development) (SRY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Mullerian structures are normal. {ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570825, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47}. Note=The disease is caused by	EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu 1, 400045 (46,XX Overtestiküler Cinsiyet Farklılaşma Bozukluğu) (SRY geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XX sex reversal 1, 400045 (46,XX ovotesticular disorder of sex development) (SRY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Mullerian structures are normal. {ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570825, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47}. Note=The disease is caused by	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

46,XX Cinsiyet Uyumsuzluğu 1; SRXX1 (46,XX Overtestiküler Cinsiyet Farklılaşma Bozukluğu) (SRY geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XX SEX REVERSAL 1; SRXX1 (46,XX ovotesticular disorder of sex development) (SRY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Muellerian structures are normal. (ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570829, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47). Note=The disease is caused by	EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu 1; SRXX1 (46,XX Overtestiküler Cinsiyet Farklılaşma Bozukluğu) (SRY geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XX SEX REVERSAL 1; SRXX1 (46,XX ovotesticular disorder of sex development) (SRY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Muellerian structures are normal. (ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570829, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47). Note=The disease is caused by	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu, 278850, Otozomal Dominant (46,XX Testiküler Cinsiyet Farklılaşma Bozukluğu) (Kromozom 17q24 Duplikasyon Sendromu) (46,XX Testiküler Cinsel Farklılaşma Bozukluğu) (MLPA)	46,XX sex reversal 2, 278850, Autosomal dominant (46,XX testicular disorder of sex development) (CHROMOSOME 17q24 DUPLICATION SYNDROME) (46,XX testicular disorder of sex development) (MLPA)	SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu, 278850, Otozomal Dominant (46,XX Testiküler Cinsiyet Farklılaşma Bozukluğu) (Kromozom 17q24 Duplikasyon Sendromu) (46,XX Testiküler Cinsel Farklılaşma Bozukluğu) (Prenatal)	46,XX sex reversal 2, 278850, Autosomal dominant (46,XX testicular disorder of sex development) (CHROMOSOME 17q24 DUPLICATION SYNDROME) (46,XX testicular disorder of sex development) (Prenatal)	.	SRXX2, DUP17q24.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XX Cinsiyet Uyumsuzluğu 3, 300833, X'e Bağlı Dominant (46,XX Testiküler Cinsiyet Farklılaşma Bozukluğu) (Kromozom Xq26 Delesyon Sendromu) (46,XX Testiküler Cinsel Farklılaşma Bozukluğu) (2200)	46,XX sex reversal 3, 300833, X-linked dominant (46,XX testicular disorder of sex development) (CHROMOSOME Xq26 DELETION SYNDROME) (46,XX testicular disorder of sex development) (440)	.	SRXX3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
46,XX Cinsiyet Uyumsuzluğu 3, 300833, X'e Bağlı Dominant (46,XX Testiküler Cinsiyet Farklılaşma Bozukluğu) (Kromozom Xq26 Delesyon Sendromu) (46,XX Testiküler Cinsel Farklılaşma Bozukluğu) (Prenatal)	46,XX sex reversal 3, 300833, X-linked dominant (46,XX testicular disorder of sex development) (CHROMOSOME Xq26 DELETION SYNDROME) (46,XX testicular disorder of sex development) (Prenatal)	.	SRXX3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XX, Cinsiyet Uyumsuzluğu, Parsiyel-Böbrek, Adrenal, Akciğer Displazisi; SERKAL (SERKAL Sendromu) (WNT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS, ADRENALS, AND LUNGS; SERKAL (SERKAL syndrome) (WNT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT4	WNT4, SERKAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
46,XX, Cinsiyet Uyumsuzluğu, Parsiyel-Böbrek, Adrenal, Akciğer Displazisi; SERKAL (SERKAL Sendromu) (SERKAL Sendromu) (MLPA)	46,XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS, ADRENALS, AND LUNGS; SERKAL (SERKAL syndrome) (SERKAL syndrome) (MLPA)	WNT4	WNT4, SERKAL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
46,XX, Cinsiyet Uyumsuzluğu, Parsiyel-Böbrek, Adrenal, Akciğer Displazisi; SERKAL (SERKAL Sendromu) (Prenatal) (WNT4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS, ADRENALS, AND LUNGS; SERKAL (SERKAL syndrome) (WNT4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT4	WNT4, SERKAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XX, Cinsiyet Uyumsuzluğu, Parsiyel-Böbrek, Adrenal, Akciğer Displazisi; SERKAL (SERKAL Sendromu) (SERKAL Sendromu) (Prenatal) (MLPA)	46,XX SEX REVERSAL WITH DYSGENESIS OF KIDNEYS, ADRENALS, AND LUNGS; SERKAL (SERKAL syndrome) (SERKAL syndrome) (Prenatal) (MLPA)	WNT4	WNT4, SERKAL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Parsiyel Gonadal Disgenezis, Minifasiküler Nöropati, 607080 (46,XY Gonadal Disgenezi - Motor ve Duyusal Nöropati Sendromu) (DHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome) (DHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHH	DHH, SRXY7, GDXYM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 46,XY sex reversal 7 (SRXY7) [MIM:233420]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. SRXY7 patients have no functional gonads. (ECO:0000269	PubMed:15356051). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
46,XY Parsiyel Gonadal Disgenezis, Minifasiküler Nöropati, 607080 (46,XY Gonadal Disgenezi - Motor ve Duyusal Nöropati Sendromu) (DHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (46,XY gonadal dysgenesis-motor and sensory neuropathy syndrome) (DHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHH	DHH, SRXY7, GDXYM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 7 (SRXY7) [MIM:233420]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. SRXY7 patients have no functional gonads. (ECO:0000269	PubMed:15356051). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 1, 400044; SRXY1 (46,XY Komplet Gonadal Disgenezisi) (SRY geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 1, 400044; SRXY1 (46,XY complete gonadal dysgenesis) (SRY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Muellerian structures are normal. (ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570829, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47). Note=The disease is caused by	EDTA Blood Sample

46,XY Cinsiyet Uyumsuzluğu 1, 400044; SRXY1 (46,XY Komplet Gonatal Disgenezisi) (SRY geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 1, 400044; SRXY1 (46,XY complete gonadal dysgenesis) (SRY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRY	SRY, TDF, TDY, SRXX1, SRXY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Muellerian structures are normal. {ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570829, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47}. Note=The disease is caused by	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 10, 616425, Otozomal Dominant (46,XY Komplet Gonatal Disgenezis) (46,XY Komplet Gonadal Disgenezis)(Kromozom 17q24 delesyon sendromu) (2200)	46,XY sex reversal 10, 616425, Autosomal dominant (46,XY complete gonadal dysgenesis) (46,XY complete gonadal dysgenesis) (CHROMOSOME 17q24 DELETION SYNDROME) (440)	.	SRXY10	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	60	Postnatal		EDTA Blood Sample and Heparinized blood sample
46,XY Cinsiyet Uyumsuzluğu 10, 616425, Otozomal Dominant (46,XY Komplet Gonatal Disgenezis) (46,XY Komplet Gonadal Disgenezis)(Kromozom 17q24 delesyon sendromu) (Prenatal)	46,XY sex reversal 10, 616425, Autosomal dominant (46,XY complete gonadal dysgenesis) (46,XY complete gonadal dysgenesis) (CHROMOSOME 17q24 DELETION SYNDROME) (Prenatal)	.	SRXY10	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 2, Dozaj Duyarlı, 300018, X'e Bağlı (46,XY Komplet Gonadal Disgenezis) (NR0B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 2, dosage-sensitive, 300018, X-linked (46,XY complete gonadal dysgenesis) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: 46,XY sex reversal 2 (SRXY2) [MIM:300018]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. {ECO:0000269|PubMed:9486644}. Note=The disease is caused by mutations affecting the gene represented in this entry. XY individuals with a duplication of part of the short arm of the X chromosome and an intact SRY gene develop as females. The single X chromosome in these individuals does not undergo X-chromosome inactivation; therefore, these individuals presumably carry 2 active copies of genes, including the NR0B1 gene, in the duplicated region. Individuals with deletion of this region develop as males. Genes within the dosage-sensitive sex reversal region are, therefore, not essential for testis development, but, when present in a double dose, interfere with testis formation..	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 2, Dozaj Duyarlı, 300018, X'e Bağlı (46,XY Komplet Gonadal Disgenezis) (NR0B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 2, dosage-sensitive, 300018, X-linked (46,XY complete gonadal dysgenesis) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 3, 612965, Otozomal Dominant (46,XY Komplet Gonadal Disgenezis) (NR5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 3, 612965, Autosomal dominant (46,XY complete gonadal dysgenesis) (NR5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 3, 612965, Otozomal Dominant (46,XY Komplet Gonadal Disgenezis) (MLPA)	46,XY sex reversal 3, 612965, Autosomal dominant (46,XY complete gonadal dysgenesis) (MLPA)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. {ECO:0000269|PubMed:11038323}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 3, 612965, Otozomal Dominant (46,XY Komplet Gonadal Disgenezis) (NR5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 3, 612965, Autosomal dominant (46,XY complete gonadal dysgenesis) (NR5A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 3, 612965, Otozomal Dominant (46,XY Komplet Gonadal Disgenezis) (NR5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal) (MLPA)	46,XY sex reversal 3, 612965, Autosomal dominant (46,XY complete gonadal dysgenesis) (Prenatal) (MLPA)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. {ECO:0000269|PubMed:11038323}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 4, 154230 (46,XY Komplet Gonadal Disgenezis) (2200)	46,XY sex reversal 4, 154230 (46,XY complete gonadal dysgenesis) (440)	.	DEL9p24.3, C9DEL p24.3, SRXY4	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
46,XY Cinsiyet Uyumsuzluğu 4, 154230 (46,XY Komplet Gonadal Disgenezis) (Prenatal)	46,XY sex reversal 4, 154230 (46,XY complete gonadal dysgenesis) (Prenatal)	.	DEL9p24.3, C9DEL p24.3, SRXY4	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 5, 613080, Otozomal Resesif; SRXY5 (46,XY Komplet Gonadal Disgenezis) (CBX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 5, 613080, Autosomal recessive; SRXY5 (46,XY complete gonadal dysgenesis) (CBX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBX2	CBX2, M33, SRXY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 5, 613080, Otozomal Resesif; SRXY5 (46,XY Komplet Gonadal Disgenezis) (CBX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 5, 613080, Autosomal recessive; SRXY5 (46,XY complete gonadal dysgenesis) (CBX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CBX2	CBX2, M33, SRXY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 5 (SRXY5) [MIM:613080]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. {ECO:0000269|PubMed:19561780}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 6, 613762, Otozomal Dominant; SRXY6 (46,XY Komplet Gonadal Disgenezis) (MAP3K1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 6, 613762, Autosomal dominant; SRXY6 (46,XY complete gonadal dysgenesis) (MAP3K1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP3K1	MAP3K1, MEKK1, MEKK, SRXY6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: 46,XY sex reversal 6 (SRXY6) [MIM:613762]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. {ECO:0000269|PubMed:21129722}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 6, 613762, Otozomal Dominant; SRXY6 (46,XY Komplet Gonadal Disgenezis) (MAP3K1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 6, 613762, Autosomal dominant; SRXY6 (46,XY complete gonadal dysgenesis) (MAP3K1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP3K1	MAP3K1, MEKK1, MEKK, SRXY6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 6 (SRXY6) [MIM:613762]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. {ECO:0000269|PubMed:21129722}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 7, 233420, Otozomal Resesif; SRXY7 (46,XY Komplet Gonadal Disgenezis) (DHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 7, 233420, Autosomal recessive; SRXY7 (46,XY complete gonadal dysgenesis) (DHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHH	DHH, SRXY7, GDXYM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: 46,XY sex reversal 7 (SRXY7) [MIM:233420]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. SRXY7 patients have no functional gonads. {ECO:0000269|PubMed:15356051}. Note=The disease may be caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 7, 233420, Otozomal Resesif; SRXY7 (46,XY Komplet Gonadal Disgenezis) (DHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 7, 233420, Autosomal recessive; SRXY7 (46,XY complete gonadal dysgenesis) (DHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHH	DHH, SRXY7, GDXYM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 7 (SRXY7) [MIM:233420]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. SRXY7 patients have no functional gonads. {ECO:0000269|PubMed:15356051}. Note=The disease may be caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 8, 614279, Otozomal Resesif; SRXY8 (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (AKR1C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 8, 614279, Autosomal recessive; SRXY8 (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKR1C2	AKR1C2, DDH2, DD2, HAKRD, SRXY8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 8, 614279, Otozomal Resesif; SRXY8 (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (AKR1C2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 8, 614279, Autosomal recessive; SRXY8 (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AKR1C2	AKR1C2, DDH2, DD2, HAKRD, SRXY8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 8, Modifiye Edici, 614279, Otozomal Resesif (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (AKR1C4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 8, modifier of, 614279, Autosomal recessive (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKR1C4	AKR1C4, CHDR, CDR, HAKRA, DD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: 46,XY sex reversal 8 (SRXY8) [MIM:614279]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. {ECO:0000269|PubMed:21802064}. Note=The gene represented in this entry may act as a disease modifier. A splicing mutation resulting in loss of AKR1C4 exon 2 has been found in affected individuals carrying a causative mutation in AKR1C2 (PubMed:21802064). These patients manifest a more severe disease phenotype than individuals only carrying mutations in AKR1C2. {ECO:0000269|PubMed:21802064}..	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 8, Modifiye Edici, 614279, Otozomal Resesif (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (AKR1C4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 8, modifier of, 614279, Autosomal recessive (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AKR1C4	AKR1C4, CHDR, CDR, HAKRA, DD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: 46,XY sex reversal 8 (SRXY8) [MIM:614279]: A disorder of sex development. Affected individuals have a 46,XY karyotype but present as phenotypically normal females. {ECO:0000269|PubMed:21802064}. Note=The gene represented in this entry may act as a disease modifier. A splicing mutation resulting in loss of AKR1C4 exon 2 has been found in affected individuals carrying a causative mutation in AKR1C2 (PubMed:21802064). These patients manifest a more severe disease phenotype than individuals only carrying mutations in AKR1C2. {ECO:0000269|PubMed:21802064}..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 9, 616067, Otozomal Dominant (46,XY Parsiyel Gonadal Disgenezis) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	46,XY sex reversal 9, 616067, Autosomal dominant (46,XY partial gonadal dysgenesis) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
46,XY Cinsiyet Uyumsuzluğu 9, 616067, Otozomal Dominant (46,XY Parsiyel Gonadal Disgenezis) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	46,XY sex reversal 9, 616067, Autosomal dominant (46,XY partial gonadal dysgenesis) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
4p subtel (FISH)	4p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	20	Postnatal	.	Heparinized blood sample
4p subtel (FISH) (Prenatal)	4p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
4p16.3 Delesyon Sendromu/ Wolf - Hirschhorn Sendromu (FISH)	4p16.3 deletion syndrome/ Wolf-Hirschhorn syndrome (FISH)	4p16.3	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	20	Postnatal	.	Heparinized blood sample
4p16.3 Delesyon Sendromu/ Wolf - Hirschhorn Sendromu (FISH) (Prenatal)	4p16.3 deletion syndrome/ Wolf-Hirschhorn syndrome (FISH) (Prenatal)	4p16.3	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
4q subtel (FISH)	4q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	20	Postnatal	.	Heparinized blood sample
4q subtel (FISH) (Prenatal)	4q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
5 - FU (5 - Florouracil/ Toksisitesi) (DPD geni- Ati 2A-IVS14+1G-A ve *3,*4,*5A,*7,*9,*10,*12,*13, M166V, R86tH, D949V alelleri) (SnapShot analizi) (DPYD geni) (Dizi Analizi) (Postnatal)	5 fluorouracil toxicity (DPD gene - Alel 2A-IVS14+1G-A and *3,*4,*5A,*7,*9,*10,*12,*13, M166V, R886H, D949V allels) (SnapShot analysis) (DPYD gene) (Sequence Analysis) (Postnatal)	DPYD	DPYD, DPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample

5-Florouarasil Toksisitesi, 274270, Otozomal Resesif (Dihidropirimidin Dehidrojenaz Eksikliği) (DPYD gen) (Tüm Gen Dizi Analizi) (Postnatal)	5-fluorouracil toxicity, 274270, Autosomal recessive (Dihydropyrimidine dehydrogenase deficiency) (DPYD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPYD	DPYD, DPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
5-Oksoprolinaz Eksikliği, 260005, Otozomal Resesif, Otozomal Dominant, OPLAHD (5-Oksoprolinaz Eksikliği) (OPLAH gen) (Tüm Gen Dizi Analizi) (Postnatal)	5-oxoprolinase deficiency, 260005, Autosomal recessive, Autosomal dominant, OPLAHD (5-oxoprolinase deficiency) (OPLAH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPLAH	OPLAH, OPLAHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 5-oxoprolinase deficiency (OPLAHD) [MIM:260005]: A disorder characterized by calcium oxalate/carbonate urolithiasis, and excessive urinary 5-oxo-L-proline. Affected individuals have recurrent episodes of vomiting, diarrhea, and abdominal pain. {ECO:0000269	PubMed:21651516}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
5-Oksoprolinaz Eksikliği, 260005, Otozomal Resesif, Otozomal Dominant, OPLAHD (5-Oksoprolinaz Eksikliği) (OPLAH gen) (Tüm Gen Dizi Analizi) (Prenatal)	5-oxoprolinase deficiency, 260005, Autosomal recessive, Autosomal dominant, OPLAHD (5-oxoprolinase deficiency) (OPLAH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPLAH	OPLAH, OPLAHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 5-oxoprolinase deficiency (OPLAHD) [MIM:260005]: A disorder characterized by calcium oxalate/carbonate urolithiasis, and excessive urinary 5-oxo-L-proline. Affected individuals have recurrent episodes of vomiting, diarrhea, and abdominal pain. {ECO:0000269	PubMed:21651516}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
5p Delesyon Sendromu/ Cri du chat/ Cat cry (FISH)	5p deletion syndrome/ Cri du chat/ Cat cry (FISH)	5p	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
5p Delesyon Sendromu/ Cri du chat/ Cat cry (FISH) (Prenatal)	5p deletion syndrome/ Cri du chat/ Cat cry (FISH) (Prenatal)	5p	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
5p subtel (FISH)	5p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
5p subtel (FISH) (Prenatal)	5p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
5q subtel (FISH)	5q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
5q subtel (FISH) (Prenatal)	5q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
5q31 Delesyonu (FISH)	5q31 Deletion (EGR1) (FISH)	5q31	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
6p subtel (FISH)	6p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
6p subtel (FISH) (Prenatal)	6p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
6q subtel (FISH)	6q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
6q subtel (FISH) (Prenatal)	6q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
7p subtel (FISH)	7p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
7p subtel (FISH) (Prenatal)	7p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
7q subtel (FISH)	7q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
7q subtel (FISH) (Prenatal)	7q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Williams-Beuren sendromu / 7q11.23 Delesyon Sendromu (FISH)	7q11.23 deletion syndrome (FISH)	7q11.23	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Williams-Beuren sendromu / 7q11.23 Delesyon Sendromu (FISH) (Prenatal)	7q11.23 deletion syndrome (FISH) (Prenatal)	7q11.23	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
7q31 Delesyonu (7q31-7q22) (FISH)	7q31 Deletion (7q31-7q22) (FISH)	7q31	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
8 Trizomisi/ Monozomisi (FISH)	8 Trisomy/ Monosomy (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
8 Trizomisi/ Monozomisi (FISH) (Prenatal)	8 Trisomy/ Monosomy (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
8p subtel (FISH)	8p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
8p subtel (FISH) (Prenatal)	8p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
8q subtel (FISH)	8q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
8q subtel (FISH) (Prenatal)	8q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
9p subtel (FISH)	9p subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
9p subtel (FISH) (Prenatal)	9p subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
9q subtel (FISH)	9q subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
9q subtel (FISH) (Prenatal)	9q subtel (FISH) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aarskog-Scott Sendromu, 305400, X'e Bağlı; AAS (Aarskog-Scott Sendromu) (FGD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Aarskog-Scott syndrome, 305400, X-linked recessive; AAS (Aarskog-Scott syndrome) (FGD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGD1	FGD1, FGDY, AAS, MRXS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aarskog-Scott syndrome (AAS) [MIM:305400]: A rare multisystemic disorder characterized by disproportionately short stature, and by facial, skeletal and urogenital anomalies. Some patients manifest mental retardation, attention deficit disorder and hyperactivity. {ECO:0000269|PubMed:10930571, ECO:0000269|PubMed:11093277, ECO:0000269|PubMed:14560308}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Abakavir Hipersensitivitesi, Yatkınlık (HLA-B geni) (HLA Analizi/ HLA Analysis) (Postnatal)	Abacavir hypersensitivity, susceptibility to (HLA-B gene) (HLA Analizi/ HLA Analysis) (Postnatal)	HLA-B	HLA-B, SPDA1	HLA Analizi/ HLA Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCD Sendromu, 600501, Otozomal Resesif (ABCD Sendromu) (EDNRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	ABCD syndrome, 600501, Autosomal recessive (ABCD syndrome) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCD Sendromu, 600501, Otozomal Resesif (ABCD Sendromu) (EDNRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	ABCD syndrome, 600501, Autosomal recessive (ABCD syndrome) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Abdominal Obezite-Metabolik Sendrom 3, 615812, Otozomal Dominant (DYRK1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Abdominal obesity-metabolic syndrome 3, 615812, Autosomal dominant (DYRK1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYRK1B	DYRK1B, MIRK, AOMS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Abdominal obesity-metabolic syndrome 3 (AOMS3) [MIM:615812]: A form of abdominal obesity-metabolic syndrome, a disorder characterized by abdominal obesity, high triglycerides, low levels of high density lipoprotein cholesterol, high blood pressure, and elevated fasting glucose levels. AOMS3 is characterized by early-onset coronary artery disease, central obesity, hypertension, and diabetes. {ECO:0000269	PubMed:24827035}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Abetalipoproteinemi, 200100, Otozomal Resesif; ABL (Abetalipoproteinemi) (MTTP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Abetalipoproteinemia, 200100, Autosomal recessive; ABL (Abetalipoproteinemia) (MTTP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTTP	MTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Abetalipoproteinemia (ABL) [MIM:200100]: An autosomal recessive disorder of lipoprotein metabolism. Affected individuals produce virtually no circulating apolipoprotein B-containing lipoproteins (chylomicrons, VLDL, LDL, lipoprotein(a)). Malabsorption of the antioxidant vitamin E occurs, leading to spinocerebellar and retinal degeneration. {ECO:0000269|PubMed:10679949, ECO:0000269|PubMed:10946006, ECO:0000269|PubMed:22236406, ECO:0000269|PubMed:23475612, ECO:0000269|PubMed:25108285, ECO:0000269|PubMed:26224785, ECO:0000269|PubMed:8939939}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Abetalipoproteinemi, 200100, Otozomal Resesif; ABL (Abetalipoproteinemi) (MTTP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Abetalipoproteinemia, 200100, Autosomal recessive; ABL (Abetalipoproteinemia) (MTTP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTTP	MTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Abetalipoproteinemia (ABL) [MIM:200100]: An autosomal recessive disorder of lipoprotein metabolism. Affected individuals produce virtually no circulating apolipoprotein B-containing lipoproteins (chylomicrons, VLDL, LDL, lipoprotein(a)). Malabsorption of the antioxidant vitamin E occurs, leading to spinocerebellar and retinal degeneration. {ECO:0000269|PubMed:10679949, ECO:0000269|PubMed:10946006, ECO:0000269|PubMed:22236406, ECO:0000269|PubMed:23475612, ECO:0000269|PubMed:25108285, ECO:0000269|PubMed:26224785, ECO:0000269|PubMed:8939939}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ablefaron-Makrostomi Sendromu, 200110, Otozomal Dominant; AMS (Ablefaron Makrostomi Sendromu) (TWIST2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ablepharon-macrostomia syndrome, 200110, Autosomal dominant; AMS (Ablepharon macrostomia syndrome) (TWIST2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST2	TWIST2, DERMO1, SETLSS, FFDD3, BBRSAY, AMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ablepharon-macrostomia syndrome (AMS) [MIM:200110]: A congenital ectodermal dysplasia characterized by absent eyelids, macrostomia, microtia, redundant skin, sparse hair, dysmorphic nose and ears, variable abnormalities of the nipples, genitalia, fingers, and hands, largely normal intellectual and motor development, and poor growth. {ECO:0000269	PubMed:26119818}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Düşük Materyali Kromozom Analizi	Abortion- Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Merkezimizden temin edilen transport besi yeri içinde/ Acil durumlarda steril serum fizyolojik içinde	*	15	45	.	.	21	Postnatal	.	Transport Medium suppliedfrom our center/ In urgency in Sterile Serum Physiologic
Abruptio Plasenta, Yatkınlık (MTHFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Abruptio placentae, susceptibility to (MTHFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFD1	MTHFD, MTHFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Abruzzo Erickson Sendromu, 302905, X'e Bağlı; ABERS (Abruzzo-Erickson Sendromu) (TBX22 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Abruzzo-Erickson syndrome, 302905, X-linked; ABERS (Abruzzo-Erickson syndrome) (TBX22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX22	TBX22, CPX, ABERS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Abruzzo-Erickson syndrome (ABERS) [MIM:302905]: A disease characterized by cleft palate, coloboma, hypospadias, deafness, short stature, and radial synostosis. {ECO:0000269	PubMed:22784330}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Abruzzo Erickson Sendromu, 302905, X'e Bağlı; ABERS (Abruzzo-Erickson Sendromu) (TBX22 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Abruzzo-Erickson syndrome, 302905, X-linked; ABERS (Abruzzo-Erickson syndrome) (TBX22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX22	TBX22, CPX, ABERS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Abruzzo-Erickson syndrome (ABERS) [MIM:302905]: A disease characterized by cleft palate, coloboma, hypospadias, deafness, short stature, and radial synostosis. {ECO:0000269	PubMed:22784330}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Akampomelik Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acampomelic campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akampomelik Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (MLPA)	Acampomelic campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akampomelik Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acampomelic campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akampomelik Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (Prenatal) (MLPA)	Acampomelic campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (Prenatal) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ACAT2 Eksikliği, 614055, Sporadik Vakalar (ACAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACAT2 deficiency, 614055, Isolated cases (ACAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAT2	ACAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAT2 Eksikliği, 614055, Sporadik Vakalar (ACAT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ACAT2 deficiency, 614055, Isolated cases (ACAT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACAT2	ACAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akatalasemi, 614097 (Akatalazemi) (CAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acatalasemia, 614097 (Acatalasemia) (CAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAT	CAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acatalasemia (ACATLAS) [MIM:614097]: A metabolic disorder characterized by a total or near total loss of catalase activity in red cells. It is often associated with ulcerating oral lesions. {ECO:0000269	PubMed:2308162}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akatalasemi, 614097 (Akatalazemi) (CAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acatalasemia, 614097 (Acatalasemia) (CAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAT	CAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Acatalasemia (ACATLAS) [MIM:614097]: A metabolic disorder characterized by a total or near total loss of catalase activity in red cells. It is often associated with ulcerating oral lesions. {ECO:0000269	PubMed:2308162}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hızlandırılmış Tümör Oluşumu, Yatkınlık, 614401 (MDM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Accelerated tumor formation, susceptibility to, 614401 (MDM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MDM2	MDM2, ACTFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hızlandırılmış Tümör Oluşumu, Yatkınlık, 614401 (MLPA)	Accelerated tumor formation, susceptibility to, 614401 (MLPA)	MDM2	MDM2, ACTFS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Note=Seems to be amplified in certain tumors (including soft tissue sarcomas, osteosarcomas and gliomas). A higher frequency of splice variants lacking p53 binding domain sequences was found in late-stage and high-grade ovarian and bladder carcinomas. Four of the splice variants show loss of p53 bindin...	EDTA Blood Sample
ACE Delesyon - İnsersiyon Polimorfizmi (DI)	ACE deletion insertion polymorphism	.	.	PCR/ Jel Elektroforezi	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Asetil-CoA Karboksilaz Eksikliği, 613933, Otozomal Resesif (ACACA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acetyl-CoA carboxylase deficiency, 613933, Autosomal recessive (ACACA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACACA	ACACA, ACAC, ACC1, ACACAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acetyl-CoA carboxylase 1 deficiency (ACACAD) [MIM:613933]: An inborn error of de novo fatty acid synthesis associated with severe brain damage, persistent myopathy and poor growth. {ECO:0000269	PubMed:31144432}. Note=The disease is caused by mutations affecting the gene represented in this entry...	EDTA Blood Sample
Asetil-CoA Karboksilaz Eksikliği, 613933, Otozomal Resesif (ACACA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acetyl-CoA carboxylase deficiency, 613933, Autosomal recessive (ACACA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACACA	ACACA, ACAC, ACC1, ACACAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Acetyl-CoA carboxylase 1 deficiency (ACACAD) [MIM:613933]: An inborn error of de novo fatty acid synthesis associated with severe brain damage, persistent myopathy and poor growth. {ECO:0000269	PubMed:31144432}. Note=The disease is caused by mutations affecting the gene represented in this entry...	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akalazya-Addisonianism-Alacrimia Sendromu, 231550, Otozomal Resesif; AAAS (Triple A Sendromu) (AAAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achalasia-addisonianism-alacrimia syndrome, 231550, Autosomal recessive; AAAS (Triple A syndrome) (AAAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AAAS	AAAS, AAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akalazya-Addisonianism-Alacrimia Sendromu, 231550, Otozomal Resesif; AAAS (Triple A Sendromu) (AAAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Achalasia-addisonianism-alacrimia syndrome, 231550, Autosomal recessive; AAAS (Triple A syndrome) (AAAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AAAS	AAAS, AAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Açeiropodi, 200500, Otozomal Resesif; ACHP (Açeiropod) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acheiropody, 200500, Autosomal recessive; ACHP (Acheiropodia) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Açeiropodi, 200500, Otozomal Resesif; ACHP (Açeiropod) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acheiropody, 200500, Autosomal recessive; ACHP (Acheiropodia) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondrogenezis Ib, 600972, Otozomal Resesif; ACG1B (Akondrogenezis) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achondrogenesis Ib, 600972, Autosomal recessive; ACG1B (Achondrogenesis) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akondrogenezis Ib, 600972, Otozomal Resesif; ACG1B (Akondrogenezis) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Achondrogenesis Ib, 600972, Autosomal recessive; ACG1B (Achondrogenesis) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondrogenezis, Tip 1A, 200600, Otozomal Resesif; ACG1A (Akondrogenezis) (TRIP11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achondrogenesis, type IA, 200600, Autosomal recessive; ACG1A (Achondrogenesis) (TRIP11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIP11	TRIP11, TRIP230, CEV14, ACG1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondrogenesis 1A (ACG1A) [MIM:200600]: A form of achondrogenesis type 1, a lethal form of chondrodysplasia characterized by deficient ossification in the lumbar vertebrae and absent ossification in the sacral, pubic and ischial bones and clinically by stillbirth or early death. In addition to severe micromelia, there is a disproportionately large cranium due to marked edema of soft tissues. {ECO:0000269	PubMed:20089971}. Note=The disease is caused by mutations affecting the gene represented in this entry...	EDTA Blood Sample
Akondrogenezis, Tip 1A, 200600, Otozomal Resesif; ACG1A (Akondrogenezis) (TRIP11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Achondrogenesis, type IA, 200600, Autosomal recessive; ACG1A (Achondrogenesis) (TRIP11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIP11	TRIP11, TRIP230, CEV14, ACG1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Achondrogenesis 1A (ACG1A) [MIM:200600]: A form of achondrogenesis type 1, a lethal form of chondrodysplasia characterized by deficient ossification in the lumbar vertebrae and absent ossification in the sacral, pubic and ischial bones and clinically by stillbirth or early death. In addition to severe micromelia, there is a disproportionately large cranium due to marked edema of soft tissues. {ECO:0000269	PubMed:20089971}. Note=The disease is caused by mutations affecting the gene represented in this entry...	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondrogenezis, Tip 2 Veya Hipokondrogenezis, 200610, Otozomal Dominant; ACG2 (Akondrogenezis) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achondrogenesis, type II or hypochondrogenesis, 200610, Autosomal dominant; ACG2 (Achondrogenesis) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akondrogenezis, Tip 2 Veya Hipokondrogenezis, 200610, Otozomal Dominant; ACG2 (Akondrogenezis) (MLPA)	Achondrogenesis, type II or hypochondrogenesis, 200610, Autosomal dominant; ACG2 (Achondrogenesis) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Akondrogenezis, Tip 2 Veya Hipokondrogenezis, 200610, Otozomal Dominant; ACG2 (Akondrogenezis) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Achondrogenesis, type II or hypochondrogenesis, 200610, Autosomal dominant; ACG2 (Achondrogenesis) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondrogenezis, Tip 2 Veya Hipokondrogenezis, 200610, Otozomal Dominant; ACG2 (Akondrogenezis) (Prenatal) (MLPA)	Achondrogenesis, type II or hypochondrogenesis, 200610, Autosomal dominant; ACG2 (Achondrogenesis) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondroplazi, 100800, Otozomal Dominant, ACH (FGFR3 Geni - G1138A ve G1138C Mutasyonları) (Akondroplazi) (FGFR3 geni) (Dizi Analizi) (Postnatal)	Achondroplasia, 100800, Autosomal dominant; ACH (FGFR3 Gene - G1138A and G1138C Mutations) (Achondroplasia) (FGFR3 gene) (Dizi Analizi) (Postnatal)	FGFR3	FGFR3, ACH	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akondroplazi, 100800, Otozomal Dominant, ACH (FGFR3 Geni - G1138A ve G1138C Mutasyonları) (Akondroplazi) (FGFR3 geni) (Dizi Analizi) (Prenatal)	Achondroplasia, 100800, Autosomal dominant; ACH (FGFR3 Gene - G1138A and G1138C Mutations) (Achondroplasia) (Prenatal) (FGFR3 gene) (Dizi Analizi) (Prenatal)	FGFR3	FGFR3, ACH	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondroplazi, Şiddetli, Gelişme Geriliği ve Akantozis Nigrikans; Saldan (Ağır Akondroplazi-Gelişimsel Gecikme-Akantozis Nigrikans Sendromu (SADDAN Sendromu)) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACHONDROPLASIA, SEVERE, WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN (Severe achondroplasia-developmental delay-acanthosis nigricans syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akondroplazi, Şiddetli, Gelişme Geriliği ve Akantozis Nigrikans; Saldan (Ağır Akondroplazi-Gelişimsel Gecikme-Akantozis Nigrikans Sendromu (SADDAN Sendromu)) (MLPA)	ACHONDROPLASIA, SEVERE, WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN (Severe achondroplasia-developmental delay-acanthosis nigricans syndrome) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akondroplazi, Şiddetli, Gelişme Geriliği ve Akantozis Nigrikans; Saldan (Ağır Akondroplazi-Gelişimsel Gecikme-Akantozis Nigrikans Sendromu (SADDAN Sendromu)) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ACHONDROPLASIA, SEVERE, WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN (Severe achondroplasia-developmental delay-acanthosis nigricans syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akondroplazi, Şiddetli, Gelişme Geriliği ve Akantozis Nigrikans; Saldan (Ağır Akondroplazi-Gelişimsel Gecikme-Akantozis Nigrikans Sendromu (SADDAN Sendromu)) (Prenatal) (MLPA)	ACHONDROPLASIA, SEVERE, WITH DEVELOPMENTAL DELAY AND ACANTHOSIS NIGRICANS; SADDAN (Severe achondroplasia-developmental delay-acanthosis nigricans syndrome) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromatopsi 2, 216900, Otozomal Resesif; ACHM2 (Akromatopsi) (CNGA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achromatopsia 2, 216900, Autosomal recessive; ACHM2 (Achromatopsia) (CNGA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNGA3	CNGA3, CNG3, ACHM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achromatopsia 2 (ACHM2) [MIM:216900]: An ocular stationary disorder due to the absence of functioning cone photoreceptors in the retina. It is characterized by total colorblindness, low visual acuity, photophobia and nystagmus. {ECO:0000269|PubMed:11536077, ECO:0000269|PubMed:14757870, ECO:0000269|PubMed:15712225, ECO:0000269|PubMed:18521937, ECO:0000269|PubMed:24903488, ECO:0000269|PubMed:9662398}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akromatopsi 3, 262300, Otozomal Resesif; ACHM3 (Akromatopsi) (CNGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achromatopsia 3, 262300, Autosomal recessive; ACHM3 (Achromatopsia) (CNGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNGB3	CNGB3, ACHM3, ACHM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achromatopsia 3 (ACHM3) [MIM:262300]: An ocular stationary disorder due to the absence of functioning cone photoreceptors in the retina. It is characterized by total colorblindness, low visual acuity, photophobia and nystagmus. Achromatopsia type 3 patients manifest severe myopia. {ECO:0000269|PubMed:10888875, ECO:0000269|PubMed:10958649, ECO:0000269|PubMed:12357335, ECO:0000269|PubMed:14757870, ECO:0000269|PubMed:15657609, ECO:0000269|PubMed:15712225}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Akromatopsi 4, 613856; ACHM4 (Akromatopsi) (GNAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achromatopsia 4, 613856; ACHM4 (Achromatopsia) (GNAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAT2	GNAT2, ACHM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akromatopsi 6, 610024, Otozomal Resesif, Otozomal Dominant (Akromatopsi) (PDE6H geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achromatopsia 6, 610024, Autosomal recessive, Autosomal dominant (Achromatopsia) (PDE6H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6H	PDE6H, RCD3, ACHM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone dystrophy, retinal 3A (RCD3A) [MIM:610024]: A rare form of cone dystrophy associated with supernormal rod responses. The disorder is characterized by reduced visual acuity, photoaversion, night blindness, and abnormal color vision. At an early age, the retina shows subtle depigmentation at the macula and, later, more obvious areas of atrophy. {ECO:0000269	PubMed:15629837}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Akromatopsi 7, 616517, Otozomal Resesif; ACHM7 (Akromatopsi) (ATF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Achromatopsia 7, 616517, Autosomal recessive; ACHM7 (Achromatopsia) (ATF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATF6	ATF6, ACHM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Asit-Labil Alt-Birim, Eksikliği, 615961; ACLSD (Kısa Boy, Primer Asit-Labil Alt Birim Eksikliği) (IGFALS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acid-labile subunit, deficiency of, 615961; ACLSD (Short stature due to primary acid-labile subunit deficiency) (IGFALS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGFALS	IGFALS, ALS, ACLSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akne Inversa, Ailesel, 1, 142690, Otozomal Dominant (NCSTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acne inversa, familial, 1, 142690, Autosomal dominant (NCSTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NCSTN	NCSTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acne inversa, familial, 1 (ACNINV1) [MIM:142690]: A chronic relapsing inflammatory disease of the hair follicles characterized by recurrent draining sinuses, painful skin abscesses, and disfiguring scars. Manifestations typically appear after puberty. {ECO:0000269|PubMed:20929727, ECO:0000269|PubMed:21430703, ECO:0000269|PubMed:21405993}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Akne Inversa, Ailesel, 2, 613736, Otozomal Dominant (PSENEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acne inversa, familial, 2, 613736, Autosomal dominant (PSENEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSENEN	PSENEN, PEN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akne Inversa, Ailesel, 3, 613737, Otozomal Dominant (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acne inversa, familial, 3, 613737, Autosomal dominant (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akrokallozal Sendrom, 200990, Otozomal Resesif; ACLS (Akrokallozal Sendrom) (KIF7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrocallosal syndrome, 200990, Autosomal recessive; ACLS (Acrocallosal syndrome) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akrokallozal Sendrom, 200990, Otozomal Resesif; ACLS (Akrokallozal Sendrom) (KIF7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrocallosal syndrome, 200990, Autosomal recessive; ACLS (Acrocallosal syndrome) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrokapitoferomoral Displazi, 607778, Otozomal Resesif; ACFD (Akrokapitoferomoral Displazi) (IHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrocapitoferomoral dysplasia, 607778, Autosomal recessive; ACFD (Acrocapitoferomoral dysplasia) (IHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IHH	IHH, BDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akrokapitoferomoral Displazi, 607778, Otozomal Resesif; ACFD (Akrokapitoferomoral Displazi) (IHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrocapitoferomoral dysplasia, 607778, Autosomal recessive; ACFD (Acrocapitoferomoral dysplasia) (IHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IHH	IHH, BDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrodermatit Enteropati, 201100, Otozomal Resesif; AE2 (Akrodermatitis Enteropatika) (SLC39A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrodermatitis enteropathica, 201100, Autosomal recessive; AE2 (Acrodermatitis enteropathica) (SLC39A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC39A4	SLC39A4, ZIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akrodermatit Enteropati, 201100, Otozomal Resesif; AE2 (Akrodermatitis Enteropatika) (SLC39A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrodermatitis enteropathica, 201100, Autosomal recessive; AE2 (Acrodermatitis enteropathica) (SLC39A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC39A4	SLC39A4, ZIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrodizostoz 1, e Hormon Direnci, 101800, Otozomal Dominant; ACRDYS1 (Akrodizostozis) (PRKAR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrodysostosis 1, with or without hormone resistance, 101800, Autosomal dominant; ACRDYS1 (Acrodysostosis) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrodysostosis 1, with or without hormone resistance (ACRDYS1) [MIM:101800]: A form of skeletal dysplasia characterized by short stature, severe brachydactyly, facial dysostosis, and nasal hypoplasia. Affected individuals often have advanced bone age and obesity. Laboratory studies show resistance to multiple hormones, including parathyroid, thyrotropin, calcitonin, growth hormone-releasing hormone, and gonadotropin. However, not all patients show endocrine abnormalities. {ECO:0000269|PubMed:21651393, ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:22723333, ECO:0000269|PubMed:23043190, ECO:0000269|PubMed:23425300}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Akrodizostoz 1, ± Hormon Direnci, 101800, Otozomal Dominant; ACRDYS1 (Akrodizostozis) (PRKAR1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrodysostosis 1, with or without hormone resistance, 101800, Autosomal dominant; ACRDYS1 (Acrodysostosis) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acrodysostosis 1, with or without hormone resistance (ACRDYS1) [MIM:101800]: A form of skeletal dysplasia characterized by short stature, severe brachydactyly, facial dysostosis, and nasal hypoplasia. Affected individuals often have advanced bone age and obesity. Laboratory studies show resistance to multiple hormones, including parathyroid, thyrotropin, calcitonin, growth hormone-releasing hormone, and gonadotropin. However, not all patients show endocrine abnormalities. {ECO:0000269|PubMed:21651393, ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:22723333, ECO:0000269|PubMed:23043190, ECO:0000269|PubMed:23425300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrodizostoz 2, ± Hormon Direnci, 614613, Otozomal Dominant; ACRDYS2 (Akrodizostozis) (PDE4D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrodysostosis 2, with or without hormone resistance, 614613, Autosomal dominant; ACRDYS2 (Acrodysostosis) (PDE4D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE4D	PDE4D, DPDE3, STRK1, ACRDYS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrodysostosis 2, with or without hormone resistance (ACRDYS2) [MIM:614613]: A pleiotropic disorder characterized by skeletal, endocrine, and neurological abnormalities. Skeletal features include brachycephaly, midface hypoplasia with a small upturned nose, brachydactyly, and lumbar spinal stenosis. Endocrine abnormalities include hypothyroidism and hypogonadism in males and irregular menses in females. Developmental disability is a common finding but is variable in severity and can be associated with significant behavioral problems. {ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:23033274, ECO:0000269|PubMed:23043190}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akrodizostoz 2, ± Hormon Direnci, 614613, Otozomal Dominant; ACRDYS2 (Akrodizostozis) (PDE4D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrodysostosis 2, with or without hormone resistance, 614613, Autosomal dominant; ACRDYS2 (Acrodysostosis) (PDE4D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDE4D	PDE4D, DPDE3, STRK1, ACRDYS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acrodysostosis 2, with or without hormone resistance (ACRDYS2) [MIM:614613]: A pleiotropic disorder characterized by skeletal, endocrine, and neurological abnormalities. Skeletal features include brachycephaly, midface hypoplasia with a small upturned nose, brachydactyly, and lumbar spinal stenosis. Endocrine abnormalities include hypothyroidism and hypogonadism in males and irregular menses in females. Developmental disability is a common finding but is variable in severity and can be associated with significant behavioral problems. {ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:23033274, ECO:0000269|PubMed:23043190}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrofasiyal Disostoz 1, Nager Tipi, 154400, Otozomal Dominant; AFD1 (Nager Sendromu) (SF3B4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrofacial dysostosis 1, Nager type, 154400, Autosomal dominant; AFD1 (Nager syndrome) (SF3B4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SF3B4	SF3B4, SF3B49, SAP49, AFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrofacial dysostosis 1, Nager type (AFD1) [MIM:154400]: A form of acrofacial dysostosis, a group of disorders which are characterized by malformation of the craniofacial skeleton and the limbs. The major facial features of AFD1 include downslanted palpebral fissures, midface retrusion, and micrognathia, the latter of which often requires the placement of a tracheostomy in early childhood. Limb defects typically involve the anterior (radial) elements of the upper limbs and manifest as small or absent thumbs, triphalangeal thumbs, radial hypoplasia or aplasia, and radioulnar synostosis. Phocomelia of the upper limbs and, occasionally, lower-limb defects have also been reported. {ECO:0000269	PubMed:22541558}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akrofasiyal Disostoz 1, Nager Tipi, 154400, Otozomal Dominant; AFD1 (Nager Sendromu) (SF3B4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrofacial dysostosis 1, Nager type, 154400, Autosomal dominant; AFD1 (Nager syndrome) (SF3B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SF3B4	SF3B4, SF3B49, SAP49, AFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acrofacial dysostosis 1, Nager type (AFD1) [MIM:154400]: A form of acrofacial dysostosis, a group of disorders which are characterized by malformation of the craniofacial skeleton and the limbs. The major facial features of AFD1 include downslanted palpebral fissures, midface retrusion, and micrognathia, the latter of which often requires the placement of a tracheostomy in early childhood. Limb defects typically involve the anterior (radial) elements of the upper limbs and manifest as small or absent thumbs, triphalangeal thumbs, radial hypoplasia or aplasia, and radioulnar synostosis. Phocomelia of the upper limbs and, occasionally, lower-limb defects have also been reported. {ECO:0000269	PubMed:22541558}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrofasiyal Disostoz, Cincinnati, OH Tipi, 616462, Otozomal Dominant; AFDCIN (Koanal Atrezi-Işitme Kaybı-Kalp Defekti-Kraniyofasiyal Dismorfizm Sendromu (Bum-Mckeown Sendromu)) (POLR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrofacial dysostosis, Cincinnati type, 616462, Autosomal dominant; AFDCIN (Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome) (Bum-McKeown syndrome) (POLR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR1A	POLR1A, RPA194, AFDCIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrofacial dysostosis, Cincinnati type (AFDCIN) [MIM:616462]: A form of acrofacial dysostosis, a group of disorders which are characterized by malformation of the craniofacial skeleton and, in some patients, the limbs. {ECO:0000269	PubMed:25913037}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akrofasiyal Disostoz, Cincinnati, OH Tipi, 616462, Otozomal Dominant; AFDCIN (Koanal Atrezi-Işitme Kaybı-Kalp Defekti-Kraniyofasiyal Dismorfizm Sendromu (Bum-Mckeown Sendromu)) +POLR1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acrofacial dysostosis, Cincinnati type, 616462, Autosomal dominant; AFDCIN (Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome) (POLR1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR1A	POLR1A, RPA194, AFDCIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acrofacial dysostosis, Cincinnati type (AFDCIN) [MIM:616462]: A form of acrofacial dysostosis, a group of disorders which are characterized by malformation of the craniofacial skeleton and, in some patients, the limbs. {ECO:0000269	PubMed:25913037}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akrokeratozis Verruciformis, 101900, Otozomal Dominant; AKV (Akrokeratozis Verruciformis, Hopf Tipi) (ATP2A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acrokeratosis verruciformis, 101900, Autosomal dominant; AKV (Acrokeratosis verruciformis of Hopf) (ATP2A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2A2	ATP2A2, ATP2B, DAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrokeratosis verruciformis (AKV) [MIM:101900]: A localized disorder of keratinization, which is inherited as an autosomal dominant trait. Its onset is early in life with multiple flat-topped, flesh-colored papules on the hands and feet; punctate keratoses on the palms and soles, with varying degrees of nail involvement. The histopathology shows a distinctive pattern of epidermal features with hyperkeratosis, hypergranulosis and acanthosis together with papillomatosis. These changes are frequently associated with circumscribed elevations of the epidermis that are said to resemble church spires. There are no features of dyskeratosis or acantholysis, the typical findings in lesions of Darier disease. {ECO:0000269	PubMed:12542527}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akromegali. Somatik, 102200 (Akromegali) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromegaly, somatic, 102200 (Acromegaly) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Akromelik Frontonazal Disostoz, 603671, Otozomal Dominant; AFND (Akromelik Frontonazal Displazi) (ZSWIM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromelic frontonasal dysostosis, 603671, Autosomal dominant; AFND (Acromelic frontonasal dysplasia) (ZSWIM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZSWIM6	ZSWIM6, KIAA1577, AFND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acromelic frontonasal dysostosis (AFND) [MIM:603671]: A rare variant form of frontonasal dysplasia, an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism, broadening of the nasal root, median facial cleft affecting the nose and/or upper lip and palate, unilateral or bilateral clefting of the alae nasi, lack of formation of the nasal tip, anterior cranium bifidum occultum, a V-shaped or widow's peak frontal hairline. AFND is characterized by the association of frontonasal malformations with various combinations of polydactyly, tibial hypoplasia, epibulbar dermoid, encephalocele, corpus callosum agenesis and Dandy-Walker malformation. {ECO:0000269	PubMed:25105228}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akromelik Frontonazal Disostoz, 603671, Otozomal Dominant; AFND (Akromelik Frontonazal Displazi) (ZSWIM6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acromelic frontonasal dysostosis, 603671, Autosomal dominant; AFND (Acromelic frontonasal dysplasia) (ZSWIM6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZSWIM6	ZSWIM6, KIAA1577, AFND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acromelic frontonasal dysostosis (AFND) [MIM:603671]: A rare variant form of frontonasal dysplasia, an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism, broadening of the nasal root, median facial cleft affecting the nose and/or upper lip and palate, unilateral or bilateral clefting of the alae nasi, lack of formation of the nasal tip, anterior cranium bifidum occultum, a V-shaped or widow's peak frontal hairline. AFND is characterized by the association of frontonasal malformations with various combinations of polydactyly, tibial hypoplasia, epibulbar dermoid, encephalocele, corpus callosum agenesis and Dandy-Walker malformation. {ECO:0000269	PubMed:25105228}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromezomelik Displazi, Demirhan Tipi, 609441, Otozomal Resesif (Brakidaktili Tip A2) (BMPR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromesomelic dysplasia, Demirhan type, 609441, Autosomal recessive (Brachydactyly type A2) (BMPR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1B	BMPR1B, ALK6, AMDD, BDA2, BDA1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akromezomelik Displazi, Demirhan Tipi, 609441, Otozomal Resesif (Brakidaktili Tip A2) (BMPR1B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acromesomelic dysplasia, Demirhan type, 609441, Autosomal recessive (Brachydactyly type A2) (BMPR1B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMPR1B	BMPR1B, ALK6, AMDD, BDA2, BDA1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromezomelik Displazi, Hunter-Thompson Tipi, 201250, Otozomal Resesif; AMDH (Akromezomelik Displazi, Hunter-Thomson Tipi) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromesomelic dysplasia, Hunter-Thompson type, 201250, Autosomal recessive; AMDH (Acromesomelic dysplasia, Hunter-Thomson type) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akromezomelik Displazi, Hunter-Thompson Tipi, 201250, Otozomal Resesif; AMDH (Akromezomelik Displazi, Hunter-Thomson Tipi) (GDF5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acromesomelic dysplasia, Hunter-Thompson type, 201250, Autosomal recessive; AMDH (Acromesomelic dysplasia, Hunter-Thomson type) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromezomelik Displazi, Maroteaux Tipi, 602875, Otozomal Resesif; AMDM (Akromezomelik Displazi, Maroteaux Tipi) (NPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromesomelic dysplasia, Maroteaux type, 602875, Autosomal recessive; AMDM (Acromesomelic dysplasia, Maroteaux type) (NPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acromesomelic dysplasia, Maroteaux type (AMDM) [MIM:602875]: An autosomal recessive acromesomelic chondrodysplasia. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs and hand/foot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knob-like fingers). AMDM is characterized by axial skeletal involvement with wedging of vertebral bodies. In AMDM all skeletal elements are present but show abnormal rates of linear growth. {ECO:0000269	PubMed:15146390}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Akromezomelik Displazi, Maroteaux Tipi, 602875, Otozomal Resesif; AMDM (Akromezomelik Displazi, Maroteaux Tipi) (NPR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acromesomelic dysplasia, Maroteaux type, 602875, Autosomal recessive; AMDM (Acromesomelic dysplasia, Maroteaux type) (NPR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	DISEASE: Acromesomelic dysplasia, Maroteaux type (AMDM) [MIM:602875]: An autosomal recessive acromesomelic chondrodysplasia. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs and hand/foot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knob-like fingers). AMDM is characterized by axial skeletal involvement with wedging of vertebral bodies. In AMDM all skeletal elements are present but show abnormal rates of linear growth. (ECO:0000269	PubMed:15146390). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromikrik Displazi, 102370, Otozomal Dominant; ACMICD (Akromikrik Displazi) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Acromicric dysplasia, 102370, Autosomal dominant; ACMICD (Acromicric dysplasia) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. (ECO:0000269	PubMed:21683323). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akromikrik Displazi, 102370, Otozomal Dominant; ACMICD (Akromikrik Displazi) (MLPA)	Acromicric dysplasia, 102370, Autosomal dominant; ACMICD (Acromicric dysplasia) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	.	20	60	.	.	30	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. (ECO:0000269	PubMed:21683323). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akromikrik Displazi, 102370, Otozomal Dominant; ACMICD (Akromikrik Displazi) (Prenatal) (FBN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Acromicric dysplasia, 102370, Autosomal dominant; ACMICD (Acromicric dysplasia) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. (ECO:0000269	PubMed:21683323). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akromikrik Displazi, 102370, Otozomal Dominant; ACMICD (Akromikrik Displazi) (Prenatal) (MLPA)	Acromicric dysplasia, 102370, Autosomal dominant; ACMICD (Acromicric dysplasia) (Prenatal) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. (ECO:0000269	PubMed:21683323). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ACTH Bağımsız Makronodüler Adrenal Hiperplazi 2, 615954, Otozomal Dominant; Somatik Mutasyon (Cushing Sendromu, Makronodüler Adrenal Hiperplaziye Bağlı) (ARMC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACTH-independent macronodular adrenal hyperplasia 2, 615954, Autosomal dominant, Somatic mutation (Cushing syndrome due to macronodular adrenal hyperplasia) (ARMC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARMC5	ARMC5, AIMAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde	.	30	120	.	.	60	Postnatal	DISEASE: ACTH-independent macronodular adrenal hyperplasia 2 (AIMAH2) [MIM:615954]: A form of macronodular adrenal hyperplasia characterized by multiple, bilateral, non-pigmented, benign, adrenocortical nodules. It results in excessive production of cortisol leading to ACTH-independent Cushing syndrome. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. (ECO:0000269|PubMed:24283234, ECO:0000269|PubMed:24601692, ECO:0000269|PubMed:24905064). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample/ Biopsy of affected site-transport media
ACTH Bağımsız Makronodüler Adrenal Hiperplazi, 219080, Sporadik Vakalar (Cushing Sendromu, Makronodüler Adrenal Hiperplaziye Bağlı) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACTH-independent macronodular adrenal hyperplasia, 219080, Isolated cases(Cushing syndrome due to macronodular adrenal hyperplasia) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
ACTH Bağımsız Makronodüler Adrenal Hiperplazi, 219080, Sporadik Vakalar (Cushing Sendromu, Makronodüler Adrenal Hiperplazive Bağlı) (MLPA)	ACTH-independent macronodular adrenal hyperplasia, 219080, Isolated cases(Cushing syndrome due to macronodular adrenal hyperplasia) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Akut lenfoblastik lösemi (ALL), KML (IKZF1 7p12.2) (MLPA)	Acute lymphoblastic leukemia (ALL), KML (IKZF1 7p12.2) (MLPA)	IKZF1 7p12.2	.	MLPA	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample / EDTA'lı kemik iliği
Açil-CoA Dehidrojenaz, Orta-Zincirli, Eksikliği; ACADMD (MCAD Eksikliği) (Orta Zincirli Açil-CoA Dehidrojenaz Eksikliği) (ACADM geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF; ACADMD (MCAD deficiency) (Medium chain acyl-CoA dehydrogenase deficiency) (ACADM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACADM	ACADM, MCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acyl-CoA dehydrogenase medium-chain deficiency (ACADMD) [MIM:201450]: An inborn error of mitochondrial fatty acid beta- oxidation which causes fasting hypoglycemia, hepatic dysfunction and encephalopathy, often resulting in death in infancy. (ECO:0000269|PubMed:10767181, ECO:0000269|PubMed:11349232, ECO:0000269|PubMed:11409868, ECO:0000269|PubMed:11486912, ECO:0000269|PubMed:1671131, ECO:0000269|PubMed:1684086, ECO:0000269|PubMed:1902818, ECO:0000269|PubMed:2251268, ECO:0000269|PubMed:2393404, ECO:0000269|PubMed:2394825, ECO:0000269|PubMed:7603790, ECO:0000269|PubMed:7929823, ECO:0000269|PubMed:8198141, ECO:0000269|PubMed:9158144). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Açil-CoA Dehidrojenaz, Orta-Zincirli, Eksikliği; ACADMD (MCAD Eksikliği) (Orta Zincirli Açil-CoA Dehidrojenaz Eksikliği) (ACADM geni) (Tüm Gen Dizi Analizi) (Prenatal)	ACYL-CoA DEHYDROGENASE, MEDIUM-CHAIN, DEFICIENCY OF; ACADMD (MCAD deficiency) (Medium chain acyl-CoA dehydrogenase deficiency) (ACADM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACADM	ACADM, MCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acyl-CoA dehydrogenase medium-chain deficiency (ACADMD) [MIM:201450]: An inborn error of mitochondrial fatty acid beta- oxidation which causes fasting hypoglycemia, hepatic dysfunction and encephalopathy, often resulting in death in infancy. (ECO:0000269|PubMed:10767181, ECO:0000269|PubMed:11349232, ECO:0000269|PubMed:11409868, ECO:0000269|PubMed:11486912, ECO:0000269|PubMed:1671131, ECO:0000269|PubMed:1684086, ECO:0000269|PubMed:1902818, ECO:0000269|PubMed:2251268, ECO:0000269|PubMed:2393404, ECO:0000269|PubMed:2394825, ECO:0000269|PubMed:7603790, ECO:0000269|PubMed:7929823, ECO:0000269|PubMed:8198141, ECO:0000269|PubMed:9158144). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Açil-CoA Dehidrojenaz, Kısa Zincirli, Eksikliği; ACADSD (SCAD Eksikliği) (Kısa Zincirli Açil-CoA Dehidrojenaz Eksikliği) (ACADS geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF; ACADSD (SCAD deficiency)(Short chain acyl-CoA dehydrogenase deficiency) (ACADS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACADS	ACADS, SCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acyl-CoA dehydrogenase short-chain deficiency (ACADSD) [MIM:201470]: An inborn error of mitochondrial fatty acid beta- oxidation resulting in acute acidosis and muscle weakness in infants, and a form of lipid-storage myopathy in adults. (ECO:0000269|PubMed:11134486, ECO:0000269|PubMed:1692038, ECO:0000269|PubMed:9499414). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Açil-CoA Dehidrojenaz, Kısa Zincirli, Eksikliği; ACADSD (SCAD Eksikliği) (Kısa Zincirli Açil-CoA Dehidrojenaz Eksikliği) (Kısa Zincirli Açil-CoA Dehidrojenaz Yetmezliği) (ACADS geni) (Tüm Gen Dizi Analizi) (Prenatal)	ACYL-CoA DEHYDROGENASE, SHORT-CHAIN, DEFICIENCY OF; ACADSD (SCAD deficiency)(Short chain acyl-CoA dehydrogenase deficiency) (ACADS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACADS	ACADS, SCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acyl-CoA dehydrogenase short-chain deficiency (ACADSD) [MIM:201470]: An inborn error of mitochondrial fatty acid beta- oxidation resulting in acute acidosis and muscle weakness in infants, and a form of lipid-storage myopathy in adults. (ECO:0000269|PubMed:11134486, ECO:0000269|PubMed:1692038, ECO:0000269|PubMed:9499414). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Açil-CoA Dehidrojenaz, Çok Uzun Zincirli, Eksikliği; ACADVLD (VLCAD Eksikliği) (Çok Uzun Zincirli Açil-CoA Dehidrojenaz Eksikliği) (ACADVL geni) (Tüm Gen Dizi Analizi) (Postnatal)	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD (VLCAD deficiency) (Very long-chain acyl-CoA dehydrogenase deficiency) (ACADVL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACADVL	ACADVL, VLCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acyl-CoA dehydrogenase very long-chain deficiency (ACADVLD) [MIM:201475]: An inborn error of mitochondrial fatty acid beta-oxidation which leads to impaired long-chain fatty acid beta-oxidation. It is clinically heterogeneous, with three major phenotypes: a severe childhood form characterized by early onset, high mortality and high incidence of cardiomyopathy, a milder childhood form with later onset, characterized by hypoketotic hypoglycemia, low mortality and rare cardiomyopathy; an adult form, with isolated skeletal muscle involvement, rhabdomyolysis and myoglobinuria, usually triggered by exercise or fasting. (ECO:0000269|PubMed:10077518, ECO:0000269|PubMed:8554073, ECO:0000269|PubMed:9546340). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Açil-CoA Dehidrojenaz, Çok Uzun Zincirli, Eksikliği; ACADVLD (VLCAD Eksikliği) (Çok Uzun Zincirli Açil-CoA Dehidrojenaz Eksikliği) (ACADVL geni) (Tüm Gen Dizi Analizi) (Prenatal)	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF; ACADVLD (VLCAD deficiency) (Very long chain acyl-CoA dehydrogenase deficiency) (ACADVL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACADVL	ACADVL, VLCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acyl-CoA dehydrogenase very long-chain deficiency (ACADVLD) [MIM:201475]: An inborn error of mitochondrial fatty acid beta-oxidation which leads to impaired long-chain fatty acid beta-oxidation. It is clinically heterogeneous, with three major phenotypes: a severe childhood form characterized by early onset, high mortality and high incidence of cardiomyopathy; a milder childhood form with later onset, characterized by hypoketotic hypoglycemia, low mortality and rare cardiomyopathy; an adult form, with isolated skeletal muscle involvement, rhabdomyolysis and myoglobinuria, usually triggered by exercise or fasting. {ECO:0000269|PubMed:10077518, ECO:0000269|PubMed:8554073, ECO:0000269|PubMed:9546340}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adams-Oliver Sendromu 1, 100300, Otozomal Dominant; AOS1 (Adams-Oliver Sendromu) (ARHGAP31 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 1, 100300, Autosomal dominant; AOS1 (Adams-Oliver syndrome) (ARHGAP31 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGAP31	ARHGAP31, CDGAP, KIAA1204, AOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adams-Oliver syndrome 1 (AOS1) [MIM:100300]: A disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:21565291}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adams-Oliver Sendromu 1, 100300, Otozomal Dominant; AOS1 (Adams-Oliver Sendromu) (ARHGAP31 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 1, 100300, Autosomal dominant; AOS1 (Adams-Oliver syndrome) (ARHGAP31 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARHGAP31	ARHGAP31, CDGAP, KIAA1204, AOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adams-Oliver syndrome 1 (AOS1) [MIM:100300]: A disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:21565291}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adams-Oliver Sendromu 2, 614219, Otozomal Resesif; AOS2 (Adams-Oliver Sendromu) (DOCK6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 2, 614219, Autosomal recessive; AOS2 (Adams-Oliver syndrome) (DOCK6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOCK6	DOCK6, KIAA1395, AOS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adams-Oliver syndrome 2 (AOS2) [MIM:614219]: A disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:21820096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adams-Oliver Sendromu 2, 614219, Otozomal Resesif; AOS2 (Adams-Oliver Sendromu) (DOCK6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 2, 614219, Autosomal recessive; AOS2 (Adams-Oliver syndrome) (DOCK6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOCK6	DOCK6, KIAA1395, AOS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adams-Oliver syndrome 2 (AOS2) [MIM:614219]: A disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:21820096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adams-Oliver Sendromu 3, 614814, Otozomal Dominant; AOS3 (Adams-Oliver Sendromu) (RBPJ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 3, 614814, Autosomal dominant; AOS3 (Adams-Oliver syndrome) (RBPJ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBPJ	RBPJ, RBPSUH, IGKJRB1, AOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adams-Oliver syndrome 3 (AOS3) [MIM:614814]: An autosomal dominant form of Adams-Oliver syndrome, a disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. AOS3 patients manifest characteristic vertex scalp defects and terminal limb defects, but without congenital heart defects, other associated defects, or immune defects. {ECO:0000269|PubMed:22883147}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adams-Oliver Sendromu 3, 614814, Otozomal Dominant; AOS3 (Adams-Oliver Sendromu) (RBPJ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 3, 614814, Autosomal dominant; AOS3 (Adams-Oliver syndrome) (RBPJ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBPJ	RBPJ, RBPSUH, IGKJRB1, AOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adams-Oliver syndrome 3 (AOS3) [MIM:614814]: An autosomal dominant form of Adams-Oliver syndrome, a disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. AOS3 patients manifest characteristic vertex scalp defects and terminal limb defects, but without congenital heart defects, other associated defects, or immune defects. {ECO:0000269|PubMed:22883147}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adams-Oliver Sendromu 4, 615297, Otozomal Resesif; AOS4 (Adams-Oliver Sendromu) (EOGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 4, 615297, Autosomal recessive; AOS4 (Adams-Oliver syndrome) (EOGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EOGT	EOGT, EOGT1, C3orf64, AOS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adams-Oliver syndrome 4 (AOS4) [MIM:615297]: A form of Adams-Oliver syndrome, a disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:23522784}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adams-Oliver Sendromu 4, 615297, Otozomal Resesif; AOS4 (Adams-Oliver Sendromu) (EOGT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 4, 615297, Autosomal recessive; AOS4 (Adams-Oliver syndrome) (EOGT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EOGT	EOGT, EOGT1, C3orf64, AOS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adams-Oliver syndrome 4 (AOS4) [MIM:615297]: A form of Adams-Oliver syndrome, a disorder characterized by the congenital absence of skin (aplasia cutis congenita) in combination with transverse limb defects. Aplasia cutis congenita can be located anywhere on the body, but in the vast majority of the cases, it is present on the posterior parietal region where it is often associated with an underlying defect of the parietal bones. Limb abnormalities are typically limb truncation defects affecting the distal phalanges or entire digits (true ectrodactyly). Only rarely, metatarsals/metacarpals or more proximal limb structures are also affected. Apart from transverse limb defects, syndactyly, most commonly of second and third toes, can also be observed. The clinical features are highly variable and can also include cardiovascular malformations, brain abnormalities and vascular defects such as cutis marmorata and dilated scalp veins. {ECO:0000269|PubMed:23522784}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adams-Oliver Sendromu 5, 616028, Otozomal Dominant; AOS5 (Adams-Oliver Sendromu) (NOTCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 5, 616028, Autosomal dominant; AOS5 (Adams-Oliver syndrome) (NOTCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH1	NOTCH1, TAN1, AOSS, AOVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adams-Oliver Sendromu 5, 616028, Otozomal Dominant; AOS5 (Adams-Oliver Sendromu) (NOTCH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 5, 616028, Autosomal dominant; AOS5 (Adams-Oliver syndrome) (NOTCH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOTCH1	NOTCH1, TAN1, AOSS, AOVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Adams-Oliver Sendromu 6, 616589, Otozomal Dominant; AOS6 (Adams-Oliver Sendromu) (DLL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adams-Oliver syndrome 6, 616589, Autosomal dominant; AOS6 (Adams-Oliver syndrome) (DLL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLL4	DLL4, AOS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adams-Oliver Sendromu 6, 616589, Otozomal Dominant; AOS6 (Adams-Oliver Sendromu) (DLL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adams-Oliver syndrome 6, 616589, Autosomal dominant; AOS6 (Adams-Oliver syndrome) (DLL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLL4	DLL4, AOS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adenin Fosforibosiltransferaz Eksikliği, 614723, Otozomal Resesif; APRTD (Adenin Fosforibosiltransferaz Eksikliği) (APRT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenine phosphoribosyltransferase deficiency, 614723, Autosomal recessive; APRTD (Adenine phosphoribosyltransferase deficiency) (APRT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APRT	APRT, APRTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adenine phosphoribosyltransferase deficiency (APRTD) [MIM:614723]: An enzymatic deficiency that can lead to urolithiasis and renal failure. Patients have 2,8-dihydroxyadenine (DHA) urinary stones. {ECO:0000269|PubMed:11243733, ECO:0000269|PubMed:1353080, ECO:0000269|PubMed:15571218, ECO:0000269|PubMed:1746557, ECO:0000269|PubMed:21635362, ECO:0000269|PubMed:3343350, ECO:0000269|PubMed:3680503, ECO:0000269|PubMed:7915931}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adenin Fosforibosiltransferaz Eksikliği, 614723, Otozomal Resesif; APRTD (Adenin Fosforibosiltransferaz Eksikliği) (APRT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adenine phosphoribosyltransferase deficiency, 614723, Autosomal recessive; APRTD (Adenine phosphoribosyltransferase deficiency) (APRT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	APRT	APRT, APRTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adenine phosphoribosyltransferase deficiency (APRTD) [MIM:614723]: An enzymatic deficiency that can lead to urolithiasis and renal failure. Patients have 2,8-dihydroxyadenine (DHA) urinary stones. {ECO:0000269|PubMed:11243733, ECO:0000269|PubMed:1353080, ECO:0000269|PubMed:15571218, ECO:0000269|PubMed:1746557, ECO:0000269|PubMed:21635362, ECO:0000269|PubMed:3343350, ECO:0000269|PubMed:3680503, ECO:0000269|PubMed:7915931}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akciğer Adenokarsinomu, Somatik, 211980 (PRKN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenocarcinoma of lung, somatic, 211980 (PRKN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKN	PRKN, PARK2, PDJ, LPRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Akciğer Adenokarsinomu, Somatik, 211980 (BRAF V600E - 599 ve 601. kodonların mutasyonları - Ekzon 15 mutasyonları) (BRAF geni) (Dizi Analizi) (Postnatal)	Adenocarcinoma of lung, somatic, 211980 (BRAF V600E - 599 ve 601. codons mutations - Exon 15 mutations) (BRAF gene) (Sequence Analysis) (Postnatal)	BRAF	BRAF	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal		Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adenokarsinoma, Kolon, Somatik (RAD54L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenocarcinoma, colonic, somatic (RAD54L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD54L	RAD54L, HRS4, HRAD54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adenokarsinoma, Over, Somatik, 167000 (PRKN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenocarcinoma, ovarian, somatic, 167000 (PRKN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKN	PRKN, PARK2, PDJ, LPRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adenoma, Periampuller, Somatik (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenoma, periampullary, somatic (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adenomlar, Multipl Kolorektal, 608456, Otozomal Resesif (MUTYH İlişkili Ailenin Allesel Adenomatoz Polipozi) (MUTYH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenomas, multiple colorectal, 608456, Autosomal recessive (MUTYH-related attenuated familial adenomatous polyposis) (MUTYH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUTYH	MUTYH, MYH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adenomlar, Multipl Kolorektal, 608456, Otozomal Resesif (MUTYH İlişkili Ailenin Allesel Adenomatoz Polipozi) (MLPA)	Adenomas, multiple colorectal, 608456, Autosomal recessive (MUTYH-related attenuated familial adenomatous polyposis) (MLPA)	MUTYH	MUTYH, MYH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Adenomlar, Tükürük Bezi Pleomorfik, Somatik, 181030 (Tükürük Bezlerinin Benign Epitelyal Tümörü) (PLAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenomas, salivary gland pleomorphic, somatic, 181030 (Benign epithelial tumor of salivary glands) (PLAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAG1	PLAG1, SGPA, PSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adenomatöz Polipozis Koli, 175100, Otozomal Dominant; FAP1 (Ailesel Adenomatöz Polipozis) (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenomatous polyposis coli, 175100, Autosomal dominant; FAP1 (Familial adenomatous polyposis) (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adenomatöz Polipozis Koli, 175100, Otozomal Dominant; FAP1 (Ailesel Adenomatöz Polipozis) (MLPA)	Adenomatous polyposis coli, 175100, Autosomal dominant; FAP1 (Familial adenomatous polyposis) (MLPA)	APC	APC, GS, FPC, BTPS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Adenozin Deaminaz Eksikliği, Parsiyel, 102700, Otozomal Resesif, Somatik Mozaik (Ağır Kombine İmmun Yetmezlik, Adenozin Deaminaz Eksikliğine Bağlı) (ADA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenosine deaminase deficiency, partial, 102700, Autosomal recessive, Somatic mosaicism (Severe combined immunodeficiency due to adenosine deaminase deficiency) (ADA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADA	ADA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T-cell-negative/B-cell-negative/NK-cell-negative due to adenosine deaminase deficiency (ADASCID) [MIM:102700]: An autosomal recessive disorder accounting for about 50% of non-X-linked SCIDs. SCID refers to a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients with SCID present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell- mediated cellular immunity due to a defect in T-cell development. ADA deficiency has been diagnosed in chronically ill teenagers and adults (late or adult onset). Population and newborn screening programs have also identified several healthy individuals with normal immunity who have partial ADA deficiency. {ECO:0000269|PubMed:10200056, ECO:0000269|PubMed:1284479, ECO:0000269|PubMed:2166947, ECO:0000269|PubMed:2783588, ECO:0000269|PubMed:3182793, ECO:0000269|PubMed:3839802, ECO:0000269|PubMed:6208479, ECO:0000269|PubMed:7599635, ECO:0000269|PubMed:8227344, ECO:0000269|PubMed:8299233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adenozin Deaminaz Eksikliği, Parsiyel, 102700, Otozomal Resesif, Somatik Mozaik (Ağır Kombine İmmun Yetmezlik, Adenozin Deaminaz Eksikliğine Bağlı) (ADA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adenosine deaminase deficiency, partial, 102700, Autosomal recessive, Somatic mosaicism (Severe combined immunodeficiency due to adenosine deaminase deficiency) (ADA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADA	ADA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T-cell-negative/B-cell-negative/NK-cell-negative due to adenosine deaminase deficiency (ADASCID) [MIM:102700]: An autosomal recessive disorder accounting for about 50% of non-X-linked SCIDs. SCID refers to a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients with SCID present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell- mediated cellular immunity due to a defect in T-cell development. ADA deficiency has been diagnosed in chronically ill teenagers and adults (late or adult onset). Population and newborn screening programs have also identified several healthy individuals with normal immunity who have partial ADA deficiency. {ECO:0000269|PubMed:10200056, ECO:0000269|PubMed:1284479, ECO:0000269|PubMed:2166947, ECO:0000269|PubMed:2783588, ECO:0000269|PubMed:3182793, ECO:0000269|PubMed:3839802, ECO:0000269|PubMed:6208479, ECO:0000269|PubMed:7599635, ECO:0000269|PubMed:8227344, ECO:0000269|PubMed:8299233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adenozin Trifosfat Yüksekliği, Eritrositler, 102900, Otozomal Dominant (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (PKLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenosine triphosphate, elevated, of erythrocytes, 102900, Autosomal dominant (Hemolytic anemia due to red cell pyruvate kinase deficiency) (PKLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKLR	PKLR, PK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269|PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Adenozin Trifosfat Yüksekliği, Eritrositler, 102900, Otozomal Dominant (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (MLPA)	Adenosine triphosphate, elevated, of erythrocytes, 102900, Autosomal dominant (Hemolytic anemia due to red cell pyruvate kinase deficiency) (MLPA)	PKLR	PKLR, PK1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269|PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Adenilosüksinaz Eksikliği, 103050, Otozomal Resesif; ADSLD (Adenilosüksinat Liyaz Eksikliği) (ADSL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adenylosuccinase deficiency, 103050, Autosomal recessive; ADSLD (Adenylosuccinate lyase deficiency) (ADSL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADSL	ADSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adenilosüksinaz Eksikliği, 103050, Otozomal Resesif; ADSLD (Adenilosüksinat Liyaz Eksikliği) (ADSL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adenylosuccinase deficiency, 103050, Autosomal recessive; ADSLD (Adenylosuccinate lyase deficiency) (ADSL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADSL	ADSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adermatoglifi, 136000, Otozomal Dominant; ADERM (Izole Konjenital Adermatoglifi) (SMARCAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adermatoglyphia, 136000, Autosomal dominant; ADERM (Isolated congenital adermatoglyphia) (SMARCAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCAD1	SMARCAD1, KIAA1122, ETL1, HEL1, ADERM, BASNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adiponektin Eksikliği, 612556 (ADIPOQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adiponectin deficiency, 612556 (ADIPOQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADIPOQ	ADIPOQ, APM1, GBP28, ADIPQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adrenal Adenom, Somatik (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal adenoma, somatic (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adrenal Kortikal Karsinom, 202300, Otozomal Resesif; ADCC (Adrenokortikal Karsinom) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal cortical carcinoma, 202300, Autosomal recessive; ADCC (Adrenocortical carcinoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adrenal Kortikal Karsinom, 202300, Otozomal Resesif; ADCC (Adrenokortikal Karsinom) (MLPA)	Adrenal cortical carcinoma, 202300, Autosomal recessive; ADCC (Adrenocortical carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 11-Beta-Hidroksilaz Eksikliğine Bağlı, 202010, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP11B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010, Autosomal recessive (Congenital adrenal hyperplasia) (CYP11B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B1	CYP11B1, P450C11, FHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 11-Beta-Hidroksilaz Eksikliğine Bağlı, 202010, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP11B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010, Autosomal recessive (Congenital adrenal hyperplasia) (CYP11B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP11B1	CYP11B1, P450C11, FHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

17-Alfa-Hidroksilaz Eksikliğine Bağlı Adrenal Hiperplazi, Konjenital (Konjenital Adrenal Hiperplazi) (CYP17A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
17-Alfa-Hidroksilaz Eksikliğine Bağlı Adrenal Hiperplazi, Konjenital (Konjenital Adrenal Hiperplazi) (MLPA)	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY (Congenital adrenal hyperplasia) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
17-Alfa-Hidroksilaz Eksikliğine Bağlı Adrenal Hiperplazi, Konjenital (Konjenital Adrenal Hiperplazi) (CYP17A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY (Congenital adrenal hyperplasia) (CYP17A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP17A1	CYP17A1, CYP17, P450C17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
17-Alfa-Hidroksilaz Eksikliğine Bağlı Adrenal Hiperplazi, Konjenital (Konjenital Adrenal Hiperplazi) (Prenatal) (MLPA)	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO 17-ALPHA-HYDROXYLASE DEFICIENCY (Congenital adrenal hyperplasia) (Prenatal) (MLPA)	CYP17A1	CYP17A1, CYP17, P450C17	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 5 (AH5) [MIM:202110]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10720067, ECO:0000269|PubMed:11549685, ECO:0000269|PubMed:11836339, ECO:0000269|PubMed:12466376, ECO:0000269|PubMed:14671162, ECO:0000269|PubMed:1515452, ECO:0000269|PubMed:1714904, ECO:0000269|PubMed:1740503, ECO:0000269|PubMed:19793597, ECO:0000269|PubMed:24140098, ECO:0000269|PubMed:24498484, ECO:0000269|PubMed:25650406, ECO:0000269|PubMed:2808364, ECO:0000269|PubMed:8027220, ECO:0000269|PubMed:8245018, ECO:0000269|PubMed:8345056, ECO:0000269|PubMed:8396144, ECO:0000269|PubMed:8550762, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP21A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (CYP21A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP21A2	CYP21A2, CYP21, CA21H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:1406699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460,	EDTA Blood Sample

Adrenal Hiperplazi, Konjenital, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (MLPA)	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (MLPA)	CYP21A2	CYP21A2, CYP21, CA21H	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:14066699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP21A2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (CYP21A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP21A2	CYP21A2, CYP21, CA21H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:14066699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (Prenatal) (MLPA)	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (Prenatal) (MLPA)	CYP21A2	CYP21A2, CYP21, CA21H	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOAH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:14066699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 3-Beta-Hidroksi Steroid Dehidrojenaz 2 Eksikliğine Bağlı, 201810, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (HSD3B2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810, Autosomal recessive (Congenital adrenal hyperplasia) (HSD3B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD3B2	HSD3B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Mild HSD3B2 deficiency in hyperandrogenic females is associated with characteristic traits of polycystic ovary syndrome, such as insulin resistance and luteinizing hormone hypersecretion. (ECO:0000269	PubMed:14764797).;	EDTA Blood Sample
Adrenal Hiperplazi, Konjenital, 3-Beta-Hidroksi Steroid Dehidrojenaz 2 Eksikliğine Bağlı, 201810, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (HSD3B2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810, Autosomal recessive (Congenital adrenal hyperplasia) (HSD3B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD3B2	HSD3B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Mild HSD3B2 deficiency in hyperandrogenic females is associated with characteristic traits of polycystic ovary syndrome, such as insulin resistance and luteinizing hormone hypersecretion. (ECO:0000269	PubMed:14764797).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal hipoplazi, konjenital, 300200, X'e bağlı resesif (lizole gliserol kinaz eksikliği) (NR0B1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal hypoplasia, congenital, 300200, X-linked recessive (isolated glycerol kinase deficiency) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adrenal hipoplazi, konjenital, 300200, X'e bağlı resesif (lizole gliserol kinaz eksikliği) (NR0B1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenal hypoplasia, congenital, 300200, X-linked recessive (isolated glycerol kinase deficiency) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Hipoplazi, Konjenital, AHC (Somatomegalik Konjenital Adrenal Hipoplazi) (NR0B1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	ADRENAL HYPOPLASIA, CONGENITAL; AHC (Cytomegalic congenital adrenal hypoplasia) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adrenal Hipoplazi, Konjenital, AHC (Somatomegalik Konjenital Adrenal Hipoplazi) (MLPA)	ADRENAL HYPOPLASIA, CONGENITAL; AHC (Cytomegalic congenital adrenal hypoplasia) (MLPA)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Adrenal Hipoplazi, Konjenital, AHC (Somatomegalik Konjenital Adrenal Hipoplazi) (NR0B1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	ADRENAL HYPOPLASIA, CONGENITAL; AHC (Cytomegalic congenital adrenal hypoplasia) (NR0B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Hipoplazi, Konjenital, AHC (Somatomegalik Konjenital Adrenal Hipoplazi) (Prenatal) (MLPA)	ADRENAL HYPOPLASIA, CONGENITAL; AHC (Cytomegalic congenital adrenal hypoplasia) (Prenatal) (MLPA)	NR0B1	DAX1, AHC, AHX, NR0B1, SRXY2	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenal Yetmezlik, Konjenital, 46,XY Cinsiyet Uyumsuzluğu, Parsiyel Veya Komplet, 613743 (46,XY Cinsiyet Farklılaşma Bozukluğu- CYP11A1 Eksikliği Sendromuna Bağlı Adrenal Yetmezlik) (CYP11A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete, 613743 (46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency syndrome) (CYP11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11A1	CYP11A1, P450SCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal insufficiency, congenital, with 46,XY sex reversal (AICSR) [MIM:613743]: A rare disorder that can present as acute adrenal insufficiency in infancy or childhood. ACTH and plasma renin activity are elevated and adrenal steroids are inappropriately low or absent; the 46,XY patients have female external genitalia, sometimes with clitoromegaly. The phenotypic spectrum ranges from prematurity, complete underandrogenization, and severe early-onset adrenal failure to term birth with clitoromegaly and later-onset adrenal failure. Patients with congenital adrenal insufficiency do not manifest the massive adrenal enlargement typical of congenital lipoid adrenal hyperplasia. (ECO:0000269|PubMed:11502818, ECO:0000269|PubMed:12161514, ECO:0000269|PubMed:16705068, ECO:0000269|PubMed:18182448, ECO:0000269|PubMed:19116240). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adrenal Yetmezlik, Konjenital, 46,XY Cinsiyet Uyumsuzluğu, Parsiyel Veya Komplet, 613743 (46,XY Cinsiyet Farklılaşma Bozukluğu- CYP11A1 Eksikliği Sendromuna Bağlı Adrenal Yetmezlik) (CYP11A1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenal insufficiency, congenital, with 46,XY sex reversal, partial or complete, 613743 (46,XY disorder of sex development-adrenal insufficiency due to CYP11A1 deficiency syndrome) (CYP11A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP11A1	CYP11A1, P450SCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal insufficiency, congenital, with 46,XY sex reversal (AICSR) [MIM:613743]: A rare disorder that can present as acute adrenal insufficiency in infancy or childhood. ACTH and plasma renin activity are elevated and adrenal steroids are inappropriately low or absent; the 46,XY patients have female external genitalia, sometimes with clitoromegaly. The phenotypic spectrum ranges from prematurity, complete underandrogenization, and severe early-onset adrenal failure to term birth with clitoromegaly and later-onset adrenal failure. Patients with congenital adrenal insufficiency do not manifest the massive adrenal enlargement typical of congenital lipoid adrenal hyperplasia. (ECO:0000269|PubMed:11502818, ECO:0000269|PubMed:12161514, ECO:0000269|PubMed:16705068, ECO:0000269|PubMed:18182448, ECO:0000269|PubMed:19116240). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenokortikal Yetersizlik (NR5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenocortical insufficiency (NR5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, AD4BP, POF7, SRXY3, SPGF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. (ECO:0000269	PubMed:11038323). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Adrenokortikal Yetersizlik (MLPA)	Adrenocortical insufficiency (MLPA)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, AD4BP, POF7, SRXY3, SPGF8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. (ECO:0000269	PubMed:11038323). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Adrenokortikal Tümör, Somatik (PRKAR1A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenocortical tumor, somatic (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Adrenokortikotropik Hormonu Eksikliği, 201400, Otozomal Resesif (Konjenital İzole ACTH Eksikliği) (TBX19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenocorticotropic hormone deficiency, 201400, Autosomal recessive (Congenital isolated ACTH deficiency) (TBX19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX19	TBX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: ACTH deficiency, isolated (IAD) [MIM:201400]: A disorder that is characterized by adrenal insufficiency symptoms, such as weight loss, lack of appetite (anorexia), weakness, nausea, vomiting and low blood pressure (hypotension). The pituitary hormone ACTH is decreased or absent, and other cortisol and other steroid hormone levels in the blood are abnormally low. {ECO:0000269|PubMed:11290323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Adrenokortikotropik Hormonu Eksikliği, 201400, Otozomal Resesif (Konjenital İzole ACTH Eksikliği) (TBX19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenocorticotropic hormone deficiency, 201400, Autosomal recessive (Congenital isolated ACTH deficiency) (TBX19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX19	TBX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: ACTH deficiency, isolated (IAD) [MIM:201400]: A disorder that is characterized by adrenal insufficiency symptoms, such as weight loss, lack of appetite (anorexia), weakness, nausea, vomiting and low blood pressure (hypotension). The pituitary hormone ACTH is decreased or absent, and other cortisol and other steroid hormone levels in the blood are abnormally low. {ECO:0000269|PubMed:11290323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenoleukodistrofi, 300100, X'e Bağlı; ALD (X'e Bağlı Adrenoleukodistrofi) (ABCD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenoleukodystrophy, 300100, X-linked recessive; ALD (X-linked adrenoleukodystrophy) (ABCD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCD1	ABCD1, ALD, AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenoleukodystrophy (ALD) [MIM:300100]: A peroxisomal metabolic disorder characterized by progressive multifocal demyelination of the central nervous system and by peripheral adrenal insufficiency (Addison disease). It results in mental deterioration, corticospinal tract dysfunction, and cortical blindness. Different clinical manifestations exist like: cerebral childhood ALD (CALD), adult cerebral ALD (ACALD), adrenomyeloneuropathy (AMN) and 'Addison disease only' (ADO) phenotype. {ECO:0000269|PubMed:10369742, ECO:0000269|PubMed:10480364, ECO:0000269|PubMed:10737980, ECO:0000269|PubMed:10980539, ECO:0000269|PubMed:11438993, ECO:0000269|PubMed:11810273, ECO:0000269|PubMed:15643618, ECO:0000269|PubMed:21700483, ECO:0000269|PubMed:21889498, ECO:0000269|PubMed:7581394, ECO:0000269|PubMed:7717396, ECO:0000269|PubMed:7825602, ECO:0000269|PubMed:7849723, ECO:0000269|PubMed:7904210, ECO:0000269|PubMed:8040304, ECO:0000269|PubMed:8566952, ECO:0000269|PubMed:8651290, ECO:0000269|PubMed:9452087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Adrenoleukodistrofi, 300100, X'e Bağlı; ALD (X'e Bağlı Adrenoleukodistrofi) (ABCD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenoleukodystrophy, 300100, X-linked recessive; ALD (X-linked adrenoleukodystrophy) (ABCD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCD1	ABCD1, ALD, AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenoleukodystrophy (ALD) [MIM:300100]: A peroxisomal metabolic disorder characterized by progressive multifocal demyelination of the central nervous system and by peripheral adrenal insufficiency (Addison disease). It results in mental deterioration, corticospinal tract dysfunction, and cortical blindness. Different clinical manifestations exist like: cerebral childhood ALD (CALD), adult cerebral ALD (ACALD), adrenomyeloneuropathy (AMN) and 'Addison disease only' (ADO) phenotype. {ECO:0000269|PubMed:10369742, ECO:0000269|PubMed:10480364, ECO:0000269|PubMed:10737980, ECO:0000269|PubMed:10980539, ECO:0000269|PubMed:11438993, ECO:0000269|PubMed:11810273, ECO:0000269|PubMed:15643618, ECO:0000269|PubMed:21700483, ECO:0000269|PubMed:21889498, ECO:0000269|PubMed:7581394, ECO:0000269|PubMed:7717396, ECO:0000269|PubMed:7825602, ECO:0000269|PubMed:7849723, ECO:0000269|PubMed:7904210, ECO:0000269|PubMed:8040304, ECO:0000269|PubMed:8566952, ECO:0000269|PubMed:8651290, ECO:0000269|PubMed:9452087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Adrenomiyelonöropati, Yetişkin, 300100, X'e Bağlı Resesif; ALD (X'e Bağlı Adrenoleukodistrofi) (ABCD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adrenomyeloneuropathy, adult, 300100, X-linked recessive; ALD (X-linked adrenoleukodystrophy) (ABCD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCD1	ABCD1, ALD, AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenoleukodystrophy (ALD) [MIM:300100]: A peroxisomal metabolic disorder characterized by progressive multifocal demyelination of the central nervous system and by peripheral adrenal insufficiency (Addison disease). It results in mental deterioration, corticospinal tract dysfunction, and cortical blindness. Different clinical manifestations exist like: cerebral childhood ALD (CALD), adult cerebral ALD (ACALD), adrenomyeloneuropathy (AMN) and 'Addison disease only' (ADO) phenotype. {ECO:0000269|PubMed:10369742, ECO:0000269|PubMed:10480364, ECO:0000269|PubMed:10737980, ECO:0000269|PubMed:10980539, ECO:0000269|PubMed:11438993, ECO:0000269|PubMed:11810273, ECO:0000269|PubMed:15643618, ECO:0000269|PubMed:21700483, ECO:0000269|PubMed:21889498, ECO:0000269|PubMed:7581394, ECO:0000269|PubMed:7717396, ECO:0000269|PubMed:7825602, ECO:0000269|PubMed:7849723, ECO:0000269|PubMed:7904210, ECO:0000269|PubMed:8040304, ECO:0000269|PubMed:8566952, ECO:0000269|PubMed:8651290, ECO:0000269|PubMed:9452087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Adrenomiyelonöropati, Yetişkin, 300100, X'e Bağlı Resesif; ALD (X'e Bağlı Adrenoleukodistrofi) (ABCD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Adrenomyeloneuropathy, adult, 300100, X-linked recessive; ALD (X-linked adrenoleukodystrophy) (ABCD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCD1	ABCD1, ALD, AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenoleukodystrophy (ALD) [MIM:300100]: A peroxisomal metabolic disorder characterized by progressive multifocal demyelination of the central nervous system and by peripheral adrenal insufficiency (Addison disease). It results in mental deterioration, corticospinal tract dysfunction, and cortical blindness. Different clinical manifestations exist like: cerebral childhood ALD (CALD), adult cerebral ALD (ACALD), adrenomyeloneuropathy (AMN) and 'Addison disease only' (ADO) phenotype. {ECO:0000269|PubMed:10369742, ECO:0000269|PubMed:10480364, ECO:0000269|PubMed:10737980, ECO:0000269|PubMed:10980539, ECO:0000269|PubMed:11438993, ECO:0000269|PubMed:11810273, ECO:0000269|PubMed:15643618, ECO:0000269|PubMed:21700483, ECO:0000269|PubMed:21889498, ECO:0000269|PubMed:7581394, ECO:0000269|PubMed:7717396, ECO:0000269|PubMed:7825602, ECO:0000269|PubMed:7849723, ECO:0000269|PubMed:7904210, ECO:0000269|PubMed:8040304, ECO:0000269|PubMed:8566952, ECO:0000269|PubMed:8651290, ECO:0000269|PubMed:9452087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kataraktsız Yetişkin I Fenotipi, 110800, Otozomal Dominant (GCNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Adult i phenotype without cataract, 110800, Autosomal dominant (GCNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCNT2	GCNT2, Ii, CTRCT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 13, with adult i phenotype (CTRCT13) [MIM:116700]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT13 is associated with the rare adult i phenotype, in which adult red blood cells are rich in i antigen and contain low levels of I antigen. {ECO:0000269|PubMed:11739194}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
ADULT Sendromu, 103285, Otozomal Dominant (ADULT Sendromu) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ADULT syndrome, 103285, Autosomal dominant (ADULT syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269|PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
ADULT Sendromu, 103285, Otozomal Dominant (ADULT Sendromu) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ADULT syndrome, 103285, Autosomal dominant (ADULT syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269|PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gelişmiş Uyku Fazı Sendromu, Ailesel, 1, 604348, Otozomal Dominant (Ailesel İlerlemiş Uyku Fazı Sendromu) (PER2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Advanced sleep phase syndrome, familial, 1, 604348, Autosomal dominant (Familial advanced sleep-phase syndrome) (PER2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PER2	PER2, FASPS1, KIAA0347	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Advanced sleep phase syndrome, familial, 1 (FASPS1) [MIM:604348]: A disorder characterized by very early sleep onset and offset. Individuals are 'morning larks' with a 4 hours advance of the sleep, temperature and melatonin rhythms. {ECO:0000269|PubMed:11737563}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Geciktmiş Uyku Fazı Sendromu, Ailesel, 3, 616882, Otozomal Dominant (Ailesel İlerlemiş Uyku Faz Sendromu) (PER3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Advanced sleep phase syndrome, familial, 3, 616882, Autosomal dominant (Familial advanced sleep-phase syndrome) (PER3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PER3	PER3, FASPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İleri Uyku Faz Sendromu, Ailesel, 2, 615224, Otozomal Dominant (Ailesel İlerlemiş Uyku Faz Sendromu) (CSNK1D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Advanced sleep-phase syndrome, familial, 2, 615224, Autosomal dominant (Familial advanced sleep-phase syndrome) (CSNK1D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSNK1D	CSNK1D, ASPS, FASPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Advanced sleep phase syndrome, familial, 2 (FASPS2) [MIM:615224]: A disorder characterized by very early sleep onset and offset. Individuals are 'morning larks' with a 4 hours advance of the sleep, temperature and melatonin rhythms. {ECO:0000269|PubMed:15800623, ECO:0000269|PubMed:23639092}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aerodigestiv Kanal Kanseri, Sıkamöz Hücreli, Alkole Bağlı, Konuyucu (ADH1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aerodigestive tract cancer, squamous cell, alcohol-related, protection against (ADH1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADH1B	ADH1B, ADH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (FGG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (FGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269	PubMed:8097046}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (FGG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (FGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Ailesel Afibrinojenemi) (FGB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Familial afibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Afibrinojenemi, Konjenital, 202400, Otozomal Resesif (Ailesel Afibrinojenemi) (FGB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Afibrinogenemia, congenital, 202400, Autosomal recessive (Familial afibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 1, 601495, Otozomal Resesif, AGM1 (özle Agamaglobulinemi) (IGHM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 1, 601495, Autosomal recessive, AGM1 (Isolated agammaglobulinemia) (IGHM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGHM	IGHM, MU, AGM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agammaglobulinemia 1, autosomal recessive (AGM1) [MIM:601495]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:8890099}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Agamaglobulinemi 1, 601495, Otozomal Resesif, AGM1 (özle Agamaglobulinemi) (IGHM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 1, 601495, Autosomal recessive, AGM1 (Isolated agammaglobulinemia) (IGHM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGHM	IGHM, MU, AGM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agammaglobulinemia 1, autosomal recessive (AGM1) [MIM:601495]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:8890099}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 2, 613500, Otozomal Resesif, AGM2 (özle Agamaglobulinemi) (IGLL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 2, 613500, Autosomal recessive, AGM2 (Isolated agammaglobulinemia) (IGLL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGLL1	IGLL1, IGD, IGL5, VPREB2, AGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Agamaglobulinemi 2, 613500, Otozomal Resesif, AGM2 (özle Agamaglobulinemi) (IGLL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 2, 613500, Autosomal recessive, AGM2 (Isolated agammaglobulinemia) (IGLL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGLL1	IGLL1, IGD, IGL5, VPREB2, AGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 3, 613501, Otozomal Resesif, AGM3 (özle Agamaglobulinemi) (CD79A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 3, 613501, Autosomal recessive, AGM3 (Isolated agammaglobulinemia) (CD79A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD79A	CD79A, IGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agammaglobulinemia 3, autosomal recessive (AGM3) [MIM:613501]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269|PubMed:10525050, ECO:0000269|PubMed:11920841}. Note=The disease is caused by mutations affecting the gene represented in this entry. Two different mutations, one at the splice donor site of intron 2 and the other at the splice acceptor site for exon 3, have been identified. Both mutations give rise to a truncated protein.;	EDTA Blood Sample
Agamaglobulinemi 3, 613501, Otozomal Resesif, AGM3 (özle Agamaglobulinemi) (CD79A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 3, 613501, Autosomal recessive, AGM3 (Isolated agammaglobulinemia) (CD79A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD79A	CD79A, IGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agammaglobulinemia 3, autosomal recessive (AGM3) [MIM:613501]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269|PubMed:10525050, ECO:0000269|PubMed:11920841}. Note=The disease is caused by mutations affecting the gene represented in this entry. Two different mutations, one at the splice donor site of intron 2 and the other at the splice acceptor site for exon 3, have been identified. Both mutations give rise to a truncated protein.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 4, 613502, Otozomal Resesif, AGM4 (özle Agamaglobulinemi) (BLNK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 4, 613502, Autosomal recessive, AGM4 (Isolated agammaglobulinemia) (BLNK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLNK	BLNK, SLP65, AGM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agammaglobulinemia 4, autosomal recessive (AGM4) [MIM:613502]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:10583958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Agamaglobulinemi 4, 613502, Otozomal Resesif, AGM4 (özle Agamaglobulinemi) (BLNK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 4, 613502, Autosomal recessive, AGM4 (Isolated agammaglobulinemia) (BLNK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BLNK	BLNK, SLP65, AGM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agammaglobulinemia 4, autosomal recessive (AGM4) [MIM:613502]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:10583958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 5, 613506, Otozomal Dominant, AGM5 (özle Agamaglobulinemi) (LRRC8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 5, 613506, Autosomal dominant, AGM5 (Isolated agammaglobulinemia) (LRRC8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRRC8A	LRRC8A, KIAA1437, AGM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agammaglobulinemia 5, autosomal dominant (AGM5) [MIM:613506]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:14660746}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving LRRC8 has been found in a patient with congenital agammaglobulinemia. Translocation t(9;20)(q33.2;q13.2). The translocation truncates the LRRC8 gene, resulting in deletion of the eighth, ninth, and half of the seventh LRR domains.;	EDTA Blood Sample
Agamaglobulinemi 5, 613506, Otozomal Dominant, AGM5 (özle Agamaglobulinemi) (LRRC8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 5, 613506, Autosomal dominant, AGM5 (Isolated agammaglobulinemia) (LRRC8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRRC8A	LRRC8A, KIAA1437, AGM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agammaglobulinemia 5, autosomal dominant (AGM5) [MIM:613506]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269	PubMed:14660746}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving LRRC8 has been found in a patient with congenital agammaglobulinemia. Translocation t(9;20)(q33.2;q13.2). The translocation truncates the LRRC8 gene, resulting in deletion of the eighth, ninth, and half of the seventh LRR domains.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Agamaglobulinemi 6, 612692, Otozomal Resesif; AGM6 (İzole Agamaglobulinemi) (CD79B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 6, 612692, Autosomal recessive; AGM6 (Isolated agammaglobulinemia) (CD79B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD79B	CD79B, IGB, B29, AGM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agammaglobulinemia 6, autosomal recessive (AGM6) [MIM:612692]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269|PubMed:17675462}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Agamaglobulinemi 6, 612692, Otozomal Resesif; AGM6 (İzole Agamaglobulinemi) (CD79B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 6, 612692, Autosomal recessive; AGM6 (Isolated agammaglobulinemia) (CD79B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD79B	CD79B, IGB, B29, AGM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agammaglobulinemia 6, autosomal recessive (AGM6) [MIM:612692]: A primary immunodeficiency characterized by profoundly low or absent serum antibodies and low or absent circulating B-cells due to an early block of B-cell development. Affected individuals develop severe infections in the first years of life. {ECO:0000269|PubMed:17675462}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 7, Otozomal Resesif, 615214, Otozomal Resesif; AGM7 (İzole Agamaglobulinemi) (PIK3R1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 7, autosomal recessive, 615214, Autosomal recessive; AGM7 (Isolated agammaglobulinemia) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Agamaglobulinemi 7, Otozomal Resesif, 615214, Otozomal Resesif; AGM7 (İzole Agamaglobulinemi) (PIK3R1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 7, autosomal recessive, 615214, Autosomal recessive; AGM7 (Isolated agammaglobulinemia) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi 8, Otozomal Dominant, 616941, Otozomal Dominant; AGM8 (İzole Agamaglobulinemi) (TCF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia 8, autosomal dominant, 616941, Autosomal dominant; AGM8 (Isolated agammaglobulinemia) (TCF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCF3	TCF3, E2A, AGM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Agamaglobulinemi 8, Otozomal Dominant, 616941, Otozomal Dominant; AGM8 (İzole Agamaglobulinemi) (TCF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia 8, autosomal dominant, 616941, Autosomal dominant; AGM8 (Isolated agammaglobulinemia) (TCF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCF3	TCF3, E2A, AGM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi ve İzole Hormonu Eksikliği, 307200, X'e Bağlı (Non-Akiz İzole Büyüme Hormonu Eksikliği) (BTK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia and isolated hormone deficiency, 307200, X-linked recessive (Non-acquired isolated growth hormone deficiency) (BTK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BTK	BTK, AGMX1, IMD1, XLA, AT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. {ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678660, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633429, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9280283}.	EDTA Blood Sample
Agamaglobulinemi ve İzole Hormonu Eksikliği, 307200, X'e Bağlı (Non-Akiz İzole Büyüme Hormonu Eksikliği) (BTK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia and isolated hormone deficiency, 307200, X-linked recessive (Non-acquired isolated growth hormone deficiency) (BTK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BTK	BTK, AGMX1, IMD1, XLA, AT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. {ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678660, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633429, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9280283}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Agamaglobulinemi, 300755, X'e Bağlı 1, X'e Bağlı; XLA (X'e Bağlı Agamaglobulinemi) (BTK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agammaglobulinemia, X-linked 1, 300755, X-linked recessive; XLA (X-linked agammaglobulinemia) (BTK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BTK	BTK, AGMX1, IMD1, XLA, AT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. {ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678660, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633429, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9280283}.	EDTA Blood Sample
Agamaglobulinemi, 300755, X'e Bağlı 1, X'e Bağlı; XLA (X'e Bağlı Agamaglobulinemi) (BTK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agammaglobulinemia, X-linked 1, 300755, X-linked recessive; XLA (X-linked agammaglobulinemia) (BTK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BTK	BTK, AGMX1, IMD1, XLA, AT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. {ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678660, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633429, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9280283}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korpus Kallosum Agenezisi, Periferik Nöropati, 218000, Otozomal Resesif; ACCPN (Korpus Kallozum Agenezisi-Nöronopati Sendromu) (SLC12A6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agenesis of the corpus callosum with peripheral neuropathy, 218000, Autosomal recessive; ACCPN (Corpus callosum agenesis-neuronopathy syndrome) (SLC12A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC12A6	SLC12A6, KCC3A, KCC3B, KCC3, ACCPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with peripheral neuropathy (ACCPN) [MIM:218000]: A disease that is characterized by severe progressive sensorimotor neuropathy, mental retardation, dysmorphic features and complete or partial agenesis of the corpus callosum. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Korpus Kallosum Agenezisi, Periferik Nöropati, 218000, Otozomal Resesif; ACCPN (Korpus Kallozum Agenezisi-Nöronopati Sendromu) (SLC12A6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agenesis of the corpus callosum with peripheral neuropathy, 218000, Autosomal recessive; ACCPN (Corpus callosum agenesis-neuronopathy syndrome) (SLC12A6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC12A6	SLC12A6, KCC3A, KCC3B, KCC3, ACCPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with peripheral neuropathy (ACCPN) [MIM:218000]: A disease that is characterized by severe progressive sensorimotor neuropathy, mental retardation, dysmorphic features and complete or partial agenesis of the corpus callosum. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Agnati-Otosefali Kompleks, 202650, Otozomal Resesif, Otozomal Dominant; AGOTC (Agnati-Holoprozensefali-Situs İnversus Sendromu) (PRRX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Agnathia-otocephaly complex, 202650, Autosomal recessive, Autosomal dominant; AGOTC (Agnathia-holoprosencephaly-situs inversus syndrome) (PRRX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRRX1	PRRX1, PMX1, PHOX1, AGOTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agnathia-otocephaly complex (AGOTC) [MIM:202650]: A rare condition characterized by mandibular hypoplasia or agnathia, ventromedial auricular malposition (melotia) and/or auricular fusion (synotia), and microstomia with oroglossal hypoplasia or aglossia. Holoprosencephaly is the most commonly identified association, but skeletal, genitourinary, and cardiovascular anomalies, and situs inversus have been reported. The disorder is almost always lethal. {ECO:0000269	PubMed:21294718}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Agnati-Otosefali Kompleks, 202650, Otozomal Resesif, Otozomal Dominant; AGOTC (Agnati-Holoprozensefali-Situs İnversus Sendromu) (PRRX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Agnathia-otocephaly complex, 202650, Autosomal recessive, Autosomal dominant; AGOTC (Agnathia-holoprosencephaly-situs inversus syndrome) (PRRX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRRX1	PRRX1, PMX1, PHOX1, AGOTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agnathia-otocephaly complex (AGOTC) [MIM:202650]: A rare condition characterized by mandibular hypoplasia or agnathia, ventromedial auricular malposition (melotia) and/or auricular fusion (synotia), and microstomia with oroglossal hypoplasia or aglossia. Holoprosencephaly is the most commonly identified association, but skeletal, genitourinary, and cardiovascular anomalies, and situs inversus have been reported. The disorder is almost always lethal. {ECO:0000269	PubMed:21294718}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ATIC Eksikliğine Bağlı AICA-Riboziduri, 608688, Otozomal Resesif (AICA-Riboziddir) (ATIC geni) (Tüm Gen Dizi Analizi) (Postnatal)	AICA-ribosiduria due to ATIC deficiency, 608688, Autosomal recessive (AICA-ribosiduria) (ATIC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATIC	ATIC, PURH, AICAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: AICAR transformylase/IMP cyclohydrolase deficiency (AICAR) [MIM:608688]: A neurologically devastating inborn error of purine biosynthesis. Patients excrete massive amounts of AICA- riboside in the urine and accumulate AICA-ribotide and its derivatives in erythrocytes and fibroblasts. AICAR causes profound mental retardation, epilepsy, dysmorphic features and congenital blindness. {ECO:0000269	PubMed:15114530}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
ATIC Eksikliğine Bağlı AICA-Riboziduri, 608688, Otozomal Resesif (AICA-Riboziddir) (ATIC geni) (Tüm Gen Dizi Analizi) (Prenatal)	AICA-ribosiduria due to ATIC deficiency, 608688, Autosomal recessive (AICA-ribosiduria) (ATIC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATIC	ATIC, PURH, AICAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: AICAR transformylase/IMP cyclohydrolase deficiency (AICAR) [MIM:608688]: A neurologically devastating inborn error of purine biosynthesis. Patients excrete massive amounts of AICA- riboside in the urine and accumulate AICA-ribotide and its derivatives in erythrocytes and fibroblasts. AICAR causes profound mental retardation, epilepsy, dysmorphic features and congenital blindness. {ECO:0000269	PubMed:15114530}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 1, Dominant ve Resesif, 225750, Otozomal Resesif, Otozomal Dominant; AGS1 (Aicardi-Goutieres Sendromu) (TREX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750, Autosomal recessive, Autosomal dominant; AGS1 (Acardi-Goutières syndrome) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 1, Dominant ve Resesif, 225750, Otozomal Resesif, Otozomal Dominant; AGS1 (Aicardi-Goutieres Sendromu) (TREX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 1, dominant and recessive, 225750, Autosomal recessive, Autosomal dominant; AGS1 (Acardi-Goutières syndrome) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 2, 610181, Otozomal Resesif; AGS2 (Aicardi-Goutieres Sendromu) (RNASEH2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 2, 610181, Autosomal recessive; AGS2 (Acardi-Goutières syndrome) (RNASEH2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASEH2B	RNASEH2B, DLEU8, FLJ11712, AGS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 2, 610181, Otozomal Resesif; AGS2 (Aicardi-Goutieres Sendromu) (RNASEH2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 2, 610181, Autosomal recessive; AGS2 (Acardi-Goutières syndrome) (RNASEH2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNASEH2B	RNASEH2B, DLEU8, FLJ11712, AGS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 3, 610329, Otozomal Resesif; AGS2 (Aicardi-Goutieres Sendromu) (RNASEH2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 3, 610329, Autosomal recessive; AGS2 (Acardi-Goutières syndrome) (RNASEH2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASEH2C	RNASEH2C, AYP1, FLJ20974, AGS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 3, 610329, Otozomal Resesif; AGS2 (Aicardi-Goutieres Sendromu) (RNASEH2C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 3, 610329, Autosomal recessive; AGS2 (Acardi-Goutières syndrome) (RNASEH2C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNASEH2C	RNASEH2C, AYP1, FLJ20974, AGS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 4, 610333, Otozomal Resesif; AGS4 (Aicardi-Goutieres Sendromu) (RNASEH2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 4, 610333, Autosomal recessive; AGS4 (Acardi-Goutières syndrome) (RNASEH2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASEH2A	RNASEH2A, RNH4A, AGS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aicardi-Goutieres syndrome 4 (AGS4) [MIM:610333]: A form of Aicardi-Goutieres syndrome, a genetically heterogeneous disease characterized by cerebral atrophy, leukoencephalopathy, intracranial calcifications, chronic cerebrospinal fluid (CSF) lymphocytosis, increased CSF alpha-interferon, and negative serologic investigations for common prenatal infection. Clinical features as thrombocytopenia, hepatosplenomegaly and elevated hepatic transaminases along with intermittent fever may erroneously suggest an infective process. Severe neurological dysfunctions manifest in infancy as progressive microcephaly, spasticity, dystonic posturing and profound psychomotor retardation. Death often occurs in early childhood. {ECO:0000269|PubMed:16845400, ECO:0000269|PubMed:17846997, ECO:0000269|PubMed:20131292}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aicardi-Goutieres Sendromu 4, 610333, Otozomal Resesif; AGS4 (Aicardi-Goutieres Sendromu) (RNASEH2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 4, 610333, Autosomal recessive; AGS4 (Acardi-Goutières syndrome) (RNASEH2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNASEH2A	RNASEH2A, RNH4A, AGS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aicardi-Goutieres syndrome 4 (AGS4) [MIM:610333]: A form of Aicardi-Goutieres syndrome, a genetically heterogeneous disease characterized by cerebral atrophy, leukoencephalopathy, intracranial calcifications, chronic cerebrospinal fluid (CSF) lymphocytosis, increased CSF alpha-interferon, and negative serologic investigations for common prenatal infection. Clinical features as thrombocytopenia, hepatosplenomegaly and elevated hepatic transaminases along with intermittent fever may erroneously suggest an infective process. Severe neurological dysfunctions manifest in infancy as progressive microcephaly, spasticity, dystonic posturing and profound psychomotor retardation. Death often occurs in early childhood. {ECO:0000269|PubMed:16845400, ECO:0000269|PubMed:17846997, ECO:0000269|PubMed:20131292}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 5, 612952, Otozomal Resesif; AGS5 (Aicardi-Goutieres Sendromu) (SAMHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 5, 612952, Autosomal recessive; AGS5 (Acardi-Goutières syndrome) (SAMHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAMHD1	SAMHD1, AGS5, DCIP, CHBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aicardi-Goutieres syndrome 5 (AGS5) [MIM:612952]: A form of Aicardi-Goutieres syndrome, a genetically heterogeneous disease characterized by cerebral atrophy, leukoencephalopathy, intracranial calcifications, chronic cerebrospinal fluid (CSF) lymphocytosis, increased CSF alpha-interferon, and negative serologic investigations for common prenatal infection. Clinical features as thrombocytopenia, hepatosplenomegaly and elevated hepatic transaminases along with intermittent fever may erroneously suggest an infective process. Severe neurological dysfunctions manifest in infancy as progressive microcephaly, spasticity, dystonic posturing and profound psychomotor retardation. Death often occurs in early childhood. {ECO:0000269|PubMed:19525956, ECO:0000269|PubMed:20131292, ECO:0000269|PubMed:20842748}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 5, 612952, Otozomal Resesif; AGS5 (Aicardi-Goutieres Sendromu) (SAMHD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 5, 612952, Autosomal recessive; AGS5 (Acardi-Goutières syndrome) (SAMHD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SAMHD1	SAMHD1, AGS5, DCIP, CHBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aicardi-Goutieres syndrome 5 (AGS5) [MIM:612952]: A form of Aicardi-Goutieres syndrome, a genetically heterogeneous disease characterized by cerebral atrophy, leukoencephalopathy, intracranial calcifications, chronic cerebrospinal fluid (CSF) lymphocytosis, increased CSF alpha-interferon, and negative serologic investigations for common prenatal infection. Clinical features as thrombocytopenia, hepatosplenomegaly and elevated hepatic transaminases along with intermittent fever may erroneously suggest an infective process. Severe neurological dysfunctions manifest in infancy as progressive microcephaly, spasticity, dystonic posturing and profound psychomotor retardation. Death often occurs in early childhood. {ECO:0000269|PubMed:19525956, ECO:0000269|PubMed:20131292, ECO:0000269|PubMed:20842748}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 6, 615010, Otozomal Resesif; AGS6 (Aicardi-Goutieres Sendromu) (ADAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 6, 615010, Autosomal recessive; AGS6 (Acardi-Goutières syndrome) (ADAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAR	ADAR, DRADA, DSH, DSRAD, IFI4, G1P1, AGS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 6, 615010, Otozomal Resesif; AGS6 (Aicardi-Goutieres Sendromu) (ADAR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 6, 615010, Autosomal recessive; AGS6 (Acardi-Goutières syndrome) (ADAR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADAR	ADAR, DRADA, DSH, DSRAD, IFI4, G1P1, AGS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aicardi-Goutieres Sendromu 7, 615846, Otozomal Dominant; AGS7 (Aicardi-Goutieres Sendromu) (IFIH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aicardi-Goutieres syndrome 7, 615846, Autosomal dominant; AGS7 (Acardi-Goutières syndrome) (IFIH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFIH1	IFIH1, MDA5, AGS7, SGMRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aicardi-Goutieres Sendromu 7, 615846, Otozomal Dominant; AGS7 (Aicardi-Goutieres Sendromu) (IFIH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aicardi-Goutieres syndrome 7, 615846, Autosomal dominant; AGS7 (Acardi-Goutières syndrome) (IFIH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFIH1	IFIH1, MDA5, AGS7, SGMRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
AIDS, Genlerinş / Hızlı Progresyon, 609423 (KIR3DL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	AIDS, delayed/rapid progression to, 609423 (KIR3DL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIR3DL1	KIR3DL1, NKAT3, NKB1, AMB11, KIR3DS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIDS, Hızlı Progresyon, 609423 (IFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	AIDS, rapid progression to, 609423 (IFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIDS, Direnç, 609423 (CXCL12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	AIDS, resistance to, 609423 (CXCL12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CXCL12	CXCL12, SDF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIDS, Yavaş Progresyon, 609423 (IL4R geni) (Tüm Gen Dizi Analizi) (Postnatal)	AIDS, slow progression to, 609423 (IL4R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL4R	IL4R, IL4RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIDS, Yavaş Progresyon, 609423 (CXCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	AIDS, slow progression to, 609423 (CXCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CXCR1	CXCR1, IL8RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Al-Gazali-Bakalinova Sendromu, 607131, Otozomal Resesif; AGBK (Multipl Epifizyal Displazi, Al-Gazali Tipi) (KIF7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Al-Gazali-Bakalinova syndrome, 607131, Autosomal recessive; AGBK (Multiple epiphyseal dysplasia, Al-Gazali type) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Al-Gazali-Bakalinova Sendromu, 607131, Otozomal Resesif; AGBK (Multipl Epifizyal Displazi, Al-Gazali Tipi) (KIF7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Al-Gazali-Bakalinova syndrome, 607131, Autosomal recessive; AGBK (Multiple epiphyseal dysplasia, Al-Gazali type) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Al-Raqad Sendromu, 616459, Otozomal Resesif; ARS (DCPS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Al-Raqad syndrome, 616459, Autosomal recessive; ARS (DCPS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCPS	DCPS, HINT5, DCS1, ARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Al-Raqad syndrome (ARS) [MIM:616459]: A syndrome characterized by delayed psychomotor development, moderate to severe intellectual disability, poor or absent speech, microcephaly, congenital hypotonia, and severe growth delay. (ECO:0000269|PubMed:25701870, ECO:0000269|PubMed:25712129). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Al-Raqad Sendromu, 616459, Otozomal Resesif; ARS (DCPS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Al-Raqad syndrome, 616459, Autosomal recessive; ARS (DCPS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCPS	DCPS, HINT5, DCS1, ARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Al-Raqad syndrome (ARS) [MIM:616459]: A syndrome characterized by delayed psychomotor development, moderate to severe intellectual disability, poor or absent speech, microcephaly, congenital hypotonia, and severe growth delay. (ECO:0000269|PubMed:25701870, ECO:0000269|PubMed:25712129). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alakrima, Akalia, Mental Retardasyon Sendromu, 615510, Otozomal Resesif; AAMR (Triple A Sendromu) (GMPPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alacrima, achalasia, and mental retardation syndrome, 615510, Autosomal recessive; AAMR (Triple A syndrome) (GMPPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GMPPA	GMPPA, AAMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alakrima, Akalia, Mental Retardasyon Sendromu, 615510, Otozomal Resesif; AAMR (Triple A Sendromu) (GMPPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alacrima, achalasia, and mental retardation syndrome, 615510, Autosomal recessive; AAMR (Triple A syndrome) (GMPPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GMPPA	GMPPA, AAMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alagille Sendromu 1, 118450, Otozomal Dominant; ALGS1 (Alagille Sendromu) (JAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alagille syndrome 1, 118450, Autosomal dominant; ALGS1 (Alagille syndrome) (JAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAG1	JAG1, AGS1, AHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. (ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alagille Sendromu 1, 118450, Otozomal Dominant; ALGS1 (Alagille Sendromu) (MLPA)	Alagille syndrome 1, 118450, Autosomal dominant; ALGS1 (Alagille syndrome) (MLPA)	JAG1	JAG1, AGS1, AHD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. (ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alagille Sendromu 1, 118450, Otozomal Dominant; ALGS1 (Alagille Sendromu) (JAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alagille syndrome 1, 118450, Autosomal dominant; ALGS1 (Alagille syndrome) (JAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JAG1	JAG1, AGS1, AHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. (ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alagille Sendromu 1, 118450, Otozomal Dominant; ALGS1 (Alagille Sendromu) (Prenatal) (MLPA)	Alagille syndrome 1, 118450, Autosomal dominant; ALGS1 (Alagille syndrome) (Prenatal) (MLPA)	JAG1	JAG1, AGS1, AHD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. (ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alagille Sendromu 2, 610205, Otozomal Dominant; ALGS2 (Alagille Sendromu) (NOTCH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alagille syndrome 2, 610205, Autosomal dominant; ALGS2 (Alagille syndrome) (NOTCH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH2	NOTCH2, AGS2, HJCYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hajdu-Cheney syndrome (HJCYS) [MIM:102500]: A rare skeletal disorder characterized by the association of facial anomalies, acro-osteolysis, general osteoporosis, insufficient ossification of the skull, and periodontal disease (premature loss of permanent teeth). Other features include cleft palate, congenital heart defects, polycystic kidneys, orthopedic problems and anomalies of the genitalia, intestines and eyes. (ECO:0000269|PubMed:21378985, ECO:0000269|PubMed:21378989). Note=The disease is caused by mutations affecting the gene represented in this entry. NOTCH2 mutations associated with Hajdu- Cheney syndrome cluster to the last coding exon of the gene. This suggests that the mutant mRNA products may escape nonsense- mediated decay and the resulting truncated NOTCH2 proteins act in a gain-of-function manner.	EDTA Blood Sample
Alagille Sendromu 2, 610205, Otozomal Dominant; ALGS2 (Alagille Sendromu) (NOTCH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alagille syndrome 2, 610205, Autosomal dominant; ALGS2 (Alagille syndrome) (NOTCH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOTCH2	NOTCH2, AGS2, HJCYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hajdu-Cheney syndrome (HJCYS) [MIM:102500]: A rare skeletal disorder characterized by the association of facial anomalies, acro-osteolysis, general osteoporosis, insufficient ossification of the skull, and periodontal disease (premature loss of permanent teeth). Other features include cleft palate, congenital heart defects, polycystic kidneys, orthopedic problems and anomalies of the genitalia, intestines and eyes. (ECO:0000269|PubMed:21378985, ECO:0000269|PubMed:21378989). Note=The disease is caused by mutations affecting the gene represented in this entry. NOTCH2 mutations associated with Hajdu- Cheney syndrome cluster to the last coding exon of the gene. This suggests that the mutant mRNA products may escape nonsense- mediated decay and the resulting truncated NOTCH2 proteins act in a gain-of-function manner.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aland Island Göz Hastalığı, 300600, X'e Bağlı (Aland Island Göz Hastalığı) (CACNA1F geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aland Island eye disease, 300600, X-linked (Aland Islands eye disease) (CACNA1F gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1F	CACNA1F, CSNB2, CORDX3, CSNB2A, AIED, OA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alazami Sendromu, 615071, Otozomal Resesif; ALAZS (Mikrosefalik Primordial Dwarfizm, Alazami Tipi) (LARP7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alazami syndrome, 615071, Autosomal recessive; ALAZS (Microcephalic primordial dwarfism, Alazami type) (LARP7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARP7	LARP7, PIP7S, ALAZS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alazami syndrome (ALAZS) [MIM:615071]: A syndromic form of primordial dwarfism, a condition characterized by severe growth restriction that has its onset in utero, and results in short stature and undersize. ALAZS patients manifest severe intellectual disability and distinct facial features including malar hypoplasia, deep-set eyes, broad nose, short philtrum, and macrostomia. Some patients have non-specific and inconsistent skeletal findings, for example, scoliosis and mild epiphyseal changes in the proximal phalanges, but no frank dysplasia. (ECO:0000269|PubMed:21937992, ECO:0000269|PubMed:22865833). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alazami Sendromu, 615071, Otozomal Resesif; ALAZS (Mikrosefalik Primordial Dwarfizm, Alazami Tipi) (LARP7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alazami syndrome, 615071, Autosomal recessive; ALAZS (Microcephalic primordial dwarfism, Alazami type) (LARP7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARP7	LARP7, PIP7S, ALAZS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alazami syndrome (ALAZS) [MIM:615071]: A syndromic form of primordial dwarfism, a condition characterized by severe growth restriction that has its onset in utero, and results in short stature and undersize. ALAZS patients manifest severe intellectual disability and distinct facial features including malar hypoplasia, deep-set eyes, broad nose, short philtrum, and macrostomia. Some patients have non-specific and inconsistent skeletal findings, for example, scoliosis and mild epiphyseal changes in the proximal phalanges, but no frank dysplasia. (ECO:0000269|PubMed:21937992, ECO:0000269|PubMed:22865833). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alazami-Yuan Sendromu, 617126, Otozomal Resesif; ALYUS (TAF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alazami-Yuan syndrome, 617126, Autosomal recessive; ALYUS (TAF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF6	TAF6, TAF2E, TAFII80, ALYUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alazami-Yuan Sendromu, 617126, Otozomal Resesif; ALYUS (TAF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alazami-Yuan syndrome, 617126, Autosomal recessive; ALYUS (TAF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAF6	TAF6, TAF2E, TAFII80, ALYUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Albinizm, Kahverengi Okülokutanöz, 203200, Otozomal Resesif (Okulokütanöz Albinizm Tip 2) (OCA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, brown oculocutaneous, 203200, Autosomal recessive (Oculocutaneous albinism type 2) (OCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCA2	OCA2, P, PED, D15S12, BOCA, EYCL3, HCL3, SHEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 2 (OCA2) [MIM:203200]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is reduced in skin, hair, and eyes. Although affected infants may appear at birth to have complete absence of melanin pigment, most patients acquire small amounts of pigment with age. Visual anomalies include decreased acuity and nystagmus. The phenotype is highly variable. The hair of affected individuals may turn darker with age, and pigmented nevi or freckles may be seen. African and African American individuals may have yellow hair and blue-gray or hazel irides. One phenotypic variant, 'brown OCA,' has been described in African and African American populations and is characterized by light brown hair and skin color and gray to tan irides. {ECO:0000269|PubMed:10649493, ECO:0000269|PubMed:10671067, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:12713581, ECO:0000269|PubMed:12727022, ECO:0000269|PubMed:12876664, ECO:0000269|PubMed:17385796, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:7762554, ECO:0000269|PubMed:7874125, ECO:0000269|PubMed:9259203}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 1A, 203100, Otozomal Resesif, OCA1A (Okulokütanöz Albinizm Tip 1) (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type IA, 203100, Autosomal recessive, OCA1A (Oculocutaneous albinism type 1) (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11295837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:15146472, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1899321, ECO:0000269|PubMed:1943686, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 1A, 203100, Otozomal Resesif, OCA1A (Okulokütanöz Albinizm Tip 1) (MLPA)	Albinism, oculocutaneous, type IA, 203100, Autosomal recessive, OCA1A (Oculocutaneous albinism type 1) (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11295837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:15146472, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1899321, ECO:0000269|PubMed:1943686, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 1B, 606952; OCA1B (Okulokütanöz Albinizm Tip 1) (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type IB, 606952; OCA1B (Oculocutaneous albinism type 1) (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11295837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:15146472, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1899321, ECO:0000269|PubMed:1943686, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 1B, 606952; OCA1B (Okulokütanöz Albinizm Tip 1) (MLPA)	Albinism, oculocutaneous, type IB, 606952; OCA1B (Oculocutaneous albinism type 1) (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11295837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:15146472, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1899321, ECO:0000269|PubMed:1943686, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 2, 203200, Otozomal Resesif, OCA2 (Okulokütanöz Albinizm Tip 2) (OCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type II, 203200, Autosomal recessive, OCA2 (Oculocutaneous albinism type 2) (OCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCA2	OCA2, P, PED, D15S12, BOCA, EYCL3, HCL3, SHEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 2 (OCA2) [MIM:203200]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is reduced in skin, hair, and eyes. Although affected infants may appear at birth to have complete absence of melanin pigment, most patients acquire small amounts of pigment with age. Visual anomalies include decreased acuity and nystagmus. The phenotype is highly variable. The hair of affected individuals may turn darker with age, and pigmented nevi or freckles may be seen. African and African American individuals may have yellow hair and blue-gray or hazel irides. One phenotypic variant, 'brown OCA,' has been described in African and African American populations and is characterized by light brown hair and skin color and gray to tan irides. {ECO:0000269|PubMed:10649493, ECO:0000269|PubMed:10671067, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:12713581, ECO:0000269|PubMed:12727022, ECO:0000269|PubMed:12876664, ECO:0000269|PubMed:17385796, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:7762554, ECO:0000269|PubMed:7874125, ECO:0000269|PubMed:9259203}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 2, Modifiye Edici, 203200, Otozomal Resesif (Okulokütanöz Albinizm Tip 2) (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type II, modifier of, 203200, Autosomal recessive (Oculocutaneous albinism type 2) (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Albinizm, Okülokutanöz, Tip 3, 203290, Otozomal Resesif; OCA3 (Okülokütanöz Albinizm Tip 3) (TYRP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type III, 203290, Autosomal recessive; OCA3 (Oculocutaneous albinism type 3) (TYRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYRP1	TYRP1, CAS2, GP75, SHEP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 3 (OCA3) [MIM:203290]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is reduced in skin, hair, and eyes. Tyrosinase activity is normal and patients have only moderate reduction of pigment. The eyes present red reflex on transillumination of the iris, dilution of color of iris, nystagmus and strabismus. Darker-skinned individuals have bright copper-red coloration of the skin and hair. {ECO:0000269|PubMed:16704458, ECO:0000269|PubMed:23504663}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 4, 606574; OCA4 (Oculokütanöz Albinizm Tip 4) (SLC45A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type IV, 606574; OCA4 (Oculocutaneous albinism type 4) (SLC45A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC45A2	SLC45A2, MATP, AIM1, SHEP5, OCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 4 (OCA4) [MIM:606574]: A disorder of pigmentation characterized by reduced biosynthesis of melanin in the skin, hair and eyes. Patients show reduced or lacking pigmentation associated with classic albinism ocular abnormalities, including decreased visual acuity, macular hypoplasia, optic dysplasia, atypical choroidal vessels, and nystagmus. {ECO:0000269|PubMed:11574907, ECO:0000269|PubMed:14722913, ECO:0000269|PubMed:14961451, ECO:0000269|PubMed:15656822, ECO:0000269|PubMed:17768386, ECO:0000269|PubMed:19865097, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:25703744}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 6, 113750, Otozomal Resesif (Oculokütanöz Albinizm Tip 6) (SLC24A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type VI, 113750, Autosomal recessive (Oculocutaneous albinism type 6) (SLC24A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A5	SLC24A5, NCKX5, SHEP4, OCA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 6 (OCA6) [MIM:113750]: A disorder characterized by a reduction or complete loss of melanin in the skin, hair and eyes. Patients show reduced or lacking pigmentation often accompanied by eye symptoms such as photophobia, strabismus, moderate to severe visual impairment, and nystagmus. {ECO:0000269|PubMed:23364476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Albinizm, Okülokutanöz, Tip 7, 615179, Otozomal Resesif; OCA7 (Oculokütanöz Albinizm Tipi 7) (C10orf11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Albinism, oculocutaneous, type VII, 615179, Autosomal recessive; OCA7 (Oculocutaneous albinism type 7) (C10orf11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C10orf11	C10orf11, OCA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 7 (OCA7) [MIM:615179]: A disorder of pigmentation characterized by reduced biosynthesis of melanin in the skin, hair and eyes. Patients show reduced or lacking pigmentation associated with classic albinism ocular abnormalities, including decreased visual acuity, macular hypoplasia, optic dysplasia, atypical choroidal vessels, and nystagmus. {ECO:0000269|PubMed:23385477}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alkol Bağımlılığı, Karşı Koruma, 103780, Multifaktöriyel (ADH1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol dependence, protection against, 103780, Multifactorial (ADH1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADH1C	ADH1C, ADH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alkol Bağımlılığı, Karşı Koruma, 103780, Multifaktöriyel (ADH1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol dependence, protection against, 103780, Multifactorial (ADH1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADH1B	ADH1B, ADH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alkol Bağımlılığı, Yatkınlık, 103780, Multifaktöriyel (TAS2R16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol dependence, susceptibility to, 103780, Multifactorial (TAS2R16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAS2R16	TAS2R16, T2R16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alkol Bağımlılığı, Yatkınlık, 103780, Multifaktöriyel (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol dependence, susceptibility to, 103780, Multifactorial (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alkol Bağımlılığı, Yatkınlık, 103780, Multifaktöriyel (GABRA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol dependence, susceptibility to, 103780, Multifactorial (GABRA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRA2	GABRA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alkol Yatkınlık, Akut, 610251; Otozomal Dominant (ALDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alcohol sensitivity, acute, 610251; Autosomal dominant (ALDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH2	ALDH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aldosteron/Renin Oranı Artışı (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aldosterone to renin ratio raised (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aldosteronizm, Glukokortikoid İle Düzelen, 103900, Otozomal Dominant; HALD1 (Ailesel Hiperaldosteronizm I) (CYP11B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aldosteronism, glucocorticoid-remediable, 103900, Autosomal dominant; HALD1 (Familial hyperaldosteronism type I) (CYP11B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B1	CYP11B1, P450C11, FHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.	EDTA Blood Sample
Alexander Hastalığı, 203450, Otozomal Dominant; ALXDRD (Alexander Hastalığı) (GFAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alexander disease, 203450, Autosomal dominant; ALXDRD (Alexander disease) (GFAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFAP	GFAP, ALXDRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alexander disease (ALXDRD) [MIM:203450]: A rare disorder of the central nervous system. The most common form affects infants and young children, and is characterized by progressive failure of central myelination, usually leading to death within the first decade. Infants with Alexander disease develop a leukodystrophy with macrocephaly, seizures, and psychomotor retardation. Patients with juvenile or adult forms typically experience ataxia, bulbar signs and spasticity, and a more slowly progressive course. Histologically, Alexander disease is characterized by Rosenthal fibers, homogeneous eosinophilic inclusions in astrocytes. {ECO:0000269|PubMed:11138011, ECO:0000269|PubMed:11567214, ECO:0000269|PubMed:11595337, ECO:0000269|PubMed:12034785, ECO:0000269|PubMed:12034796, ECO:0000269|PubMed:12581808, ECO:0000269|PubMed:12944715, ECO:0000269|PubMed:12975300, ECO:0000269|PubMed:15030911, ECO:0000269|PubMed:15732097, ECO:0000269|PubMed:17043438, ECO:0000269|PubMed:17805552, ECO:0000269|PubMed:17894839, ECO:0000269|PubMed:17934883, ECO:0000269|PubMed:17960815, ECO:0000269|PubMed:18004641, ECO:0000269|PubMed:18079314, ECO:0000269|PubMed:19412928, ECO:0000269|PubMed:20359319, ECO:0000269|PubMed:21917775, ECO:0000269|PubMed:23364391, ECO:0000269|PubMed:23743246}.	EDTA Blood Sample
Alexander Hastalığı, 203450, Otozomal Dominant; ALXDRD (Alexander Hastalığı) (GFAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alexander disease, 203450, Autosomal dominant; ALXDRD (Alexander disease) (GFAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFAP	GFAP, ALXDRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alexander disease (ALXDRD) [MIM:203450]: A rare disorder of the central nervous system. The most common form affects infants and young children, and is characterized by progressive failure of central myelination, usually leading to death within the first decade. Infants with Alexander disease develop a leukodystrophy with macrocephaly, seizures, and psychomotor retardation. Patients with juvenile or adult forms typically experience ataxia, bulbar signs and spasticity, and a more slowly progressive course. Histologically, Alexander disease is characterized by Rosenthal fibers, homogeneous eosinophilic inclusions in astrocytes. {ECO:0000269|PubMed:11138011, ECO:0000269|PubMed:11567214, ECO:0000269|PubMed:11595337, ECO:0000269|PubMed:12034785, ECO:0000269|PubMed:12034796, ECO:0000269|PubMed:12581808, ECO:0000269|PubMed:12944715, ECO:0000269|PubMed:12975300, ECO:0000269|PubMed:15030911, ECO:0000269|PubMed:15732097, ECO:0000269|PubMed:17043438, ECO:0000269|PubMed:17805552, ECO:0000269|PubMed:17894839, ECO:0000269|PubMed:17934883, ECO:0000269|PubMed:17960815, ECO:0000269|PubMed:18004641, ECO:0000269|PubMed:18079314, ECO:0000269|PubMed:19412928, ECO:0000269|PubMed:20359319, ECO:0000269|PubMed:21917775, ECO:0000269|PubMed:23364391, ECO:0000269|PubMed:23743246}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-talasemi (HBA 16p) (MLPA)	Alfa-thalassemia (HBA 16p) (MLPA)	HBA 16p	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Alfa-talasemi (HBA 16p) (MLPA) (Prenatal)	Alfa-thalassemia (HBA 16p) (MLPA) (Prenatal)	HBA 16p	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ALK Breakapart (2p23.2-p23.1) (FISH)	ALK Breakapart (2p23.2-p23.1) (FISH)	2p23.2-p23.1	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
NTRK1, NTRK2, NTRK3 Breakapart (FISH)	NTRK1, NTRK2, NTRK3 Breakapart (FISH)	.	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	20	.	.	10	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
ALK ile İlişkili Nöroblastoma (ALK geni) (Ekzon21 - 29) (Dizi Analizi) (Postnatal)	ALK-Related Neuroblastoma Susceptibility (ALK Gene) (Exons 21-29 - Sequence analysis) (Postnatal)	ALK	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuroblastoma 3 (NBLST3) [MIM:613014]: A common neoplasm of early childhood arising from embryonic cells that form the primitive neural crest and give rise to the adrenal medulla and the sympathetic nervous system. {ECO:0000269|PubMed:18724359, ECO:0000269|PubMed:18923523, ECO:0000269|PubMed:18923525}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Alkaptonüri, 203500, Otozomal Resesif; AKU (Alkaptonüri) (HGD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alkaptonuria, 203500, Autosomal recessive; AKU (Alkaptonuria) (HGD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HGD	HGD, AKU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALL Panel (t(11;?) (q23;?) (MLL/ ?) - t(12;21) (p13;q22) (TEL/ AML1) - E2A (TCF3) Break Apart - MYC bölgesin içeren Kromozom değişiklikleri - Igh bölgesini içeren Kromozom değişiklikleri - t(9;22) (q34;q11.2) - 4, 10, 17, Kromozom düzensizlikleri ve monozomiler - p16) (FISH)	ALL Panel (t(11;?) (q23;?) (MLL/ ?) - t(12;21) (p13;q22) (TEL/ AML1) - E2A (TCF3) Break Apart - MYC rearrangements - Igh rearrangements - t(9;22) (q34;q11.2) - 4, 10, 17, abnormalities - aneuploidies - p16) (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Allan-Herndon-Dudley Sendromu, 300523, X'e Bağlı Dominant; AHDS (Allan-Herndon-Dudley Sendromu) (SLC16A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Allan-Herndon-Dudley syndrome, 300523, X-linked dominant; AHDS (Allan-Herndon-Dudley syndrome) (SLC16A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC16A2	SLC16A2, DXS128, XPCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Allan-Herndon-Dudley Sendromu, 300523, X'e Bağlı Dominant; AHDS (Allan-Herndon-Dudley Sendromu) (SLC16A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Allan-Herndon-Dudley syndrome, 300523, X-linked dominant; AHDS (Allan-Herndon-Dudley syndrome) (SLC16A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC16A2	SLC16A2, DXS128, XPCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Allantoikaz; ALLC (ALLC geni) (Tüm Gen Dizi Analizi) (Postnatal)	ALLANTOICASE; ALLC (ALLC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALLC	ALLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Allerjik Rinit, Yatkınlık, 607154 (IL13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Allergic rhinitis, susceptibility to, 607154 (IL13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL13	IL13, ALRH, BHR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Allergic rhinitis (ALRH) [MIM:607154]: A common disease with complex inheritance characterized by mucosal inflammation caused by allergen exposure. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Alopesi Universalis, 203655, Otozomal Resesif; ALUNC (Alopesia Universalis) (HR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alopecia universalis, 203655, Autosomal recessive; ALUNC (Alopecia universalis) (HR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HR	HR, AU, MUHH1, HYPT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 4 (HYPT4) [MIM:146550]: An autosomal dominant condition characterized by reduced amount of hair, alopecia, little or no eyebrows, eyelashes or body hair, and coarse, wiry, twisted hair in early childhood. {ECO:0000269|PubMed:19122663, ECO:0000269|PubMed:24961381}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alopesi, Nörolojik Bozukluklar ve Endokrinopati Sendromu, 612079, Otozomal Resesif (ANE Sendromu) (RBM28 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079, Autosomal recessive (ANE syndrome) (RBM28 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBM28	RBM28, ANES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alopesi, Nörolojik Bozukluklar ve Endokrinopati Sendromu, 612079, Otozomal Resesif (ANE Sendromu) (RBM28 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alopecia, neurologic defects, and endocrinopathy syndrome, 612079, Autosomal recessive (ANE syndrome) (RBM28 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBM28	RBM28, ANES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa - 1 - Antitripsin Eksikliği (PiM - PiS - PiZ Alelleri) (Dizi Analizi)	Alpha-1- Antitrypsin deficiency (PiM - PiS- PiZ Allels)	SERPINA1	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alfa-1-Antikimotripsin Eksikliği (SERPINA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-1-antichymotrypsin deficiency (SERPINA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA3	SERPINA3, AACT, ACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alfa-1-Antikimotripsin Eksikliği (SERPINA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-1-antichymotrypsin deficiency (SERPINA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPINA3	SERPINA3, AACT, ACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-2-Makroglobulin Eksikliği, 614036, Otozomal Dominant; A2MD (A2M geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-2-macroglobulin deficiency, 614036, Autosomal dominant; A2MD (A2M gene) (Sequence Analysis-All Coding Exons) (Postnatal)	A2M	A2M , A2MD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alfa-2-Makroglobulin Eksikliği, 614036, Otozomal Dominant; A2MD (A2M geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-2-macroglobulin deficiency, 614036, Autosomal dominant; A2MD (A2M gene) (Sequence Analysis-All Coding Exons) (Prenatal)	A2M	A2M , A2MD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-2-Plasmin İnhibitörü Eksikliği, 262850, Otozomal Resesif (Konjenital Alfa2 Antiplazmin Eksikliği) (SERPINF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-2-plasmin inhibitor deficiency, 262850, Autosomal recessive (Congenital alpha2-antiplasmin deficiency) (SERPINF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINF2	PLI, SERPINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-2-plasmin inhibitor deficiency (APLID) [MIM:262850]: An autosomal recessive disorder resulting in severe hemorrhagic diathesis. {ECO:0000269|PubMed:10583218, ECO:0000269|PubMed:2572590}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alfa-Fetoprotein Eksikliği, 615969, Otozomal Resesif; AFPD (Kalıtsal Alfa Fetoprotein Persistansı) (AFP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-fetoprotein deficiency, 615969, Autosomal recessive; AFPD (Congenital deficiency in alpha-fetoprotein) (AFP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFP	AFP, HPAFP, AFPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-fetoprotein deficiency (AFPD) [MIM:615969]: A benign condition characterized by undetectable AFP levels in the amniotic fluid. Affected individuals are asymptomatic and present normal development. {ECO:0000269|PubMed:15280901, ECO:0000269|PubMed:18854864}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alfa-Fetoprotein Eksikliği, 615969, Otozomal Resesif; AFPD (Kalıtsal Alfa Fetoprotein Persistansı) (AFP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-fetoprotein deficiency, 615969, Autosomal recessive; AFPD (Congenital deficiency in alpha-fetoprotein) (AFP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AFP	AFP, HPAFP, AFPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-fetoprotein deficiency (AFPD) [MIM:615969]: A benign condition characterized by undetectable AFP levels in the amniotic fluid. Affected individuals are asymptomatic and present normal development. {ECO:0000269|PubMed:15280901, ECO:0000269|PubMed:18854864}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Ketoglutarat Dehidrogenaz Eksikliği, 203740, Otozomal Resesif (Oksoglutarikasitüri) (OGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-ketoglutarate dehydrogenase deficiency, 203740, Autosomal recessive (Oxoglutaricaciduria) (OGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OGDH	OGDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alfa-Ketoglutarat Dehidrogenaz Eksikliği, 203740, Otozomal Resesif (Oksoglutarikasitüri) (OGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-ketoglutarate dehydrogenase deficiency, 203740, Autosomal recessive (Oxoglutaricaciduria) (OGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OGDH	OGDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Metilasetoasetik Asidüri, 203750, Otozomal Resesif (Metilasetoasetik Asidüri) (Beta-Ketotiyolaz Eksikliği) (ACAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-methylacetoacetic aciduria, 203750, Autosomal recessive(Methylacetoacetic aciduria) (Beta-ketothiolase deficiency) (ACAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAT1	ACAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alfa-Metilasetoasetik Asidüri, 203750, Otozomal Resesif (Metilasetoasetik Asidüri) (Beta-Ketotiyolaz Eksikliği) (ACAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-methylacetoacetic aciduria, 203750, Autosomal recessive(Methylacetoacetic aciduria) (Beta-ketothiolase deficiency) (ACAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACAT1	ACAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Metil-CoA Rasemaz Eksikliği, 614307, Otozomal Resesif; AMACRD (Konjenital Safra Asidi Sentezi Defekti, Tip 5) (AMACR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-methylacyl-CoA racemase deficiency, 614307, Autosomal recessive; AMACRD (Congenital bile acid synthesis defect type 4) (AMACR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMACR	AMACR, CBAS4, AMACRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 4 (CBAS4) [MIM:214950]: A disorder characterized by the presence of trihydroxycoprostanic acid in the bile and absence of cholic acid. Patients manifest neonatal jaundice, intrahepatic cholestasis and bile duct deficiency. {ECO:0000269|PubMed:10655068, ECO:0000269|PubMed:12512044}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alfa-Metil-CoA Rasemaz Eksikliği, 614307, Otozomal Resesif; AMACRD (Konjenital Safra Asidi Sentezi Defekti, Tip 5) (AMACR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-methylacyl-CoA racemase deficiency, 614307, Autosomal recessive; AMACRD (Congenital bile acid synthesis defect type 4) (AMACR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMACR	AMACR, CBAS4, AMACRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital bile acid synthesis defect 4 (CBAS4) [MIM:214950]: A disorder characterized by the presence of trihydroxycoprostanic acid in the bile and absence of cholic acid. Patients manifest neonatal jaundice, intrahepatic cholestasis and bile duct deficiency. {ECO:0000269|PubMed:10655068, ECO:0000269|PubMed:12512044}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Talasemi Miyelodisplazi Sendromu, Somatik, 300448; ATMDS (Alfa-Talasemi-Miyelodisplastik Sendrom) (ATRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448; ATMDS (Alpha-thalassemia-myelodysplastic syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA blood sample / EDTA bone marrow
Alfa-Talasemi Miyelodisplazi Sendromu, Somatik, 300448; ATMDS (Alfa-Talasemi-Miyelodisplastik Sendrom) (ATRX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448; ATMDS (Alpha-thalassemia-myelodysplastic syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Talasemi / Mental Retardasyon Sendromu, 301040, X'e Bağlı Dominant; ATRX (Alfa Talasemi- X`e Bağlı Mental Retardasyon Sendromu) (ATRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha-thalassemia/mental retardation syndrome, 301040, X-linked dominant; ATRX (Alpha-thalassemia-X-linked intellectual disability syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alfa-Talasemi / Mental Retardasyon Sendromu, 301040, X'e Bağlı Dominant; ATRX (Alfa Talasemi- X`e Bağlı Mental Retardasyon Sendromu) (MLPA)	Alpha-thalassemia/mental retardation syndrome, 301040, X-linked dominant; ATRX (Alpha-thalassemia-X-linked intellectual disability syndrome) (MLPA)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alfa-Talasemi / Mental Retardasyon Sendromu, 301040, X'e Bağlı Dominant; ATRX (Alfa Talasemi- X`e Bağlı Mental Retardasyon Sendromu) (ATRX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alpha-thalassemia/mental retardation syndrome, 301040, X-linked dominant; ATRX (Alpha-thalassemia-X-linked intellectual disability syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa-Talasemi / Mental Retardasyon Sendromu, 301040, X'e Bağlı Dominant; ATRX (Alfa Talasemi- X`e Bağlı Mental Retardasyon Sendromu) (Prenatal) (MLPA)	Alpha-thalassemia/mental retardation syndrome, 301040, X-linked dominant; ATRX (Alpha-thalassemia-X-linked intellectual disability syndrome) (Prenatal) (MLPA)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269|PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa Talasemi / Mental Retardasyon Sendromu, Tip 1, 141750, Otozomal Dominant(Alfa-Talasemi-Mental Retardasyon Sendromu, Kromozom 16 İlişkili) (2200)	Alpha-thalassemia/mental retardation syndrome, type 1, 141750, Autosomal dominant (Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16) (440)	.	HBHR, ATR1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Alfa Talasemi / Mental Retardasyon Sendromu, Tip 1, 141750, Otozomal Dominant(Alfa-Talasemi-Mental Retardasyon Sendromu, Kromozom 16 İlişkili) (Prenatal)	Alpha-thalassemia/mental retardation syndrome, type 1, 141750, Autosomal dominant (Alpha-thalassemia-intellectual disability syndrome linked to chromosome 16) (Prenatal)	.	HBHR, ATR1	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alfa/Beta T-Hücreli Lenfopeni, Gama/Delta T-Hücre Ekspansiyonu, Ağır Sitomegalovirüs Enfeksiyonu, ve Otoimmünite, 609889 (Kombine İmmün Yetmezlik T4 B4, Parsiyel RAG1 Eksikliğine Bağlı) (RAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 (Combined immunodeficiency T+ B+ due to partial RAG1 deficiency) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHIDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. {ECO:0000269|PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Alport Sendromu, 301050, X'e Bağlı Dominant; ATS (Alport Sendromu) (COL4A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alport syndrome, 301050, X-linked dominant; ATS (Alport syndrome) (COL4A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A5	COL4A5, ATS, ASLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alport syndrome, X-linked (APSX) [MIM:301050]: A syndrome that is characterized by progressive glomerulonephritis, renal failure, sensorineural deafness, specific eye abnormalities (lenticonus and macular flecks), and glomerular basement membrane defects. The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:10094548, ECO:0000269|PubMed:10561141, ECO:0000269|PubMed:10563487, ECO:0000269|PubMed:10684360, ECO:0000269|PubMed:10862091, ECO:0000269|PubMed:11004279, ECO:0000269|PubMed:11223851, ECO:0000269|PubMed:1352287, ECO:0000269|PubMed:1363780, ECO:0000269|PubMed:1376965, ECO:0000269|PubMed:1672282, ECO:0000269|PubMed:24522658, ECO:0000269|PubMed:7599631, ECO:0000269|PubMed:7853788, ECO:0000269|PubMed:8406498, ECO:0000269|PubMed:8651292, ECO:0000269|PubMed:8651296, ECO:0000269|PubMed:8829632, ECO:0000269|PubMed:8940267, ECO:0000269|PubMed:9150741, ECO:0000269|PubMed:9452056, ECO:0000269|PubMed:9848783}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alport Sendromu, 301050, X'e Bağlı Dominant; ATS (Alport Sendromu) (MLPA)	Alport syndrome, 301050, X-linked dominant; ATS (Alport syndrome) (MLPA)	COL4A5	COL4A5, ATS, ASLN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alport syndrome, X-linked (APSX) [MIM:301050]: A syndrome that is characterized by progressive glomerulonephritis, renal failure, sensorineural deafness, specific eye abnormalities (lenticonus and macular flecks), and glomerular basement membrane defects. The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:10094548, ECO:0000269|PubMed:10561141, ECO:0000269|PubMed:10563487, ECO:0000269|PubMed:10684360, ECO:0000269|PubMed:10862091, ECO:0000269|PubMed:11004279, ECO:0000269|PubMed:11223851, ECO:0000269|PubMed:1352287, ECO:0000269|PubMed:1363780, ECO:0000269|PubMed:1376965, ECO:0000269|PubMed:1672282, ECO:0000269|PubMed:24522658, ECO:0000269|PubMed:7599631, ECO:0000269|PubMed:7853788, ECO:0000269|PubMed:8406498, ECO:0000269|PubMed:8651292, ECO:0000269|PubMed:8651296, ECO:0000269|PubMed:8829632, ECO:0000269|PubMed:8940267, ECO:0000269|PubMed:9150741, ECO:0000269|PubMed:9452056, ECO:0000269|PubMed:9848783}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alport Sendromu, 301050, X'e Bağlı Dominant; ATS (Alport Sendromu) (COL4A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alport syndrome, 301050, X-linked dominant; ATS (Alport syndrome) (COL4A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A5	COL4A5, ATS, ASLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, X-linked (APSX) [MIM:301050]: A syndrome that is characterized by progressive glomerulonephritis, renal failure, sensorineural deafness, specific eye abnormalities (lenticonus and macular flecks), and glomerular basement membrane defects. The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:10094548, ECO:0000269|PubMed:10561141, ECO:0000269|PubMed:10563487, ECO:0000269|PubMed:10684360, ECO:0000269|PubMed:10862091, ECO:0000269|PubMed:11004279, ECO:0000269|PubMed:11223851, ECO:0000269|PubMed:1352287, ECO:0000269|PubMed:1363780, ECO:0000269|PubMed:1376965, ECO:0000269|PubMed:1672282, ECO:0000269|PubMed:24522658, ECO:0000269|PubMed:7599631, ECO:0000269|PubMed:7853788, ECO:0000269|PubMed:8406498, ECO:0000269|PubMed:8651292, ECO:0000269|PubMed:8651296, ECO:0000269|PubMed:8829632, ECO:0000269|PubMed:8940267, ECO:0000269|PubMed:9150741, ECO:0000269|PubMed:9452056, ECO:0000269|PubMed:9848783}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, 301050, X'e Bağlı Dominant; ATS (Alport Sendromu) (Prenatal) (MLPA)	Alport syndrome, 301050, X-linked dominant; ATS (Alport syndrome) (Prenatal) (MLPA)	COL4A5	COL4A5, ATS, ASLN	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, X-linked (APSX) [MIM:301050]: A syndrome that is characterized by progressive glomerulonephritis, renal failure, sensorineural deafness, specific eye abnormalities (lenticonus and macular flecks), and glomerular basement membrane defects. The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:10094548, ECO:0000269|PubMed:10561141, ECO:0000269|PubMed:10563487, ECO:0000269|PubMed:10684360, ECO:0000269|PubMed:10862091, ECO:0000269|PubMed:11004279, ECO:0000269|PubMed:11223851, ECO:0000269|PubMed:1352287, ECO:0000269|PubMed:1363780, ECO:0000269|PubMed:1376965, ECO:0000269|PubMed:1672282, ECO:0000269|PubMed:24522658, ECO:0000269|PubMed:7599631, ECO:0000269|PubMed:7853788, ECO:0000269|PubMed:8406498, ECO:0000269|PubMed:8651292, ECO:0000269|PubMed:8651296, ECO:0000269|PubMed:8829632, ECO:0000269|PubMed:8940267, ECO:0000269|PubMed:9150741, ECO:0000269|PubMed:9452056, ECO:0000269|PubMed:9848783}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Dominant, 104200, Otozomal Dominant (Alport Sendromu) (COL4A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alport syndrome, autosomal dominant, 104200, Autosomal dominant (Alport syndrome) (COL4A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A3	COL4A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonus and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alport Sendromu, Otozomal Dominant, 104200, Otozomal Dominant (Alport Sendromu) (MLPA)	Alport syndrome, autosomal dominant, 104200, Autosomal dominant (Alport syndrome) (MLPA)	COL4A3	COL4A3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonus and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alport Sendromu, Otozomal Dominant, 104200, Otozomal Dominant (Alport Sendromu) (COL4A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alport syndrome, autosomal dominant, 104200, Autosomal dominant (Alport syndrome) (COL4A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A3	COL4A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonus and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Dominant, 104200, Otozomal Dominant (Alport Sendromu) (Prenatal) (MLPA)	Alport syndrome, autosomal dominant, 104200, Autosomal dominant (Alport syndrome) (Prenatal) (MLPA)	COL4A3	COL4A3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonus and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (COL4A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (COL4A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A4	COL4A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (COL4A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (COL4A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A3	COL4A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonus and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (MLPA)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (MLPA)	COL4A4	COL4A4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (COL4A4 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (COL4A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A4	COL4A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (COL4A3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (COL4A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A3	COL4A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonous and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (Prenatal) (MLPA)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (Prenatal) (MLPA)	COL4A4	COL4A4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Otozomal Resesif, 203780, Otozomal Resesif (Alport Sendromu) (Prenatal) (MLPA)	Alport syndrome, autosomal recessive, 203780, Autosomal recessive (Alport syndrome) (Prenatal) (MLPA)	COL4A3	COL4A3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome, autosomal dominant (APSAD) [MIM:104200]: A syndrome characterized by progressive glomerulonephritis, glomerular basement membrane defects, renal failure, sensorineural deafness and specific eye abnormalities (lenticonous and macular flecks). The disorder shows considerable heterogeneity in that families differ in the age of end-stage renal disease and the occurrence of deafness. {ECO:0000269|PubMed:11044206, ECO:0000269|PubMed:11134255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alport Sendromu, Mental Retardasyon, Orta Yüz Hipoplazisi ve Elliptositoz, 300194 (Alport Sendromu-Mental Retardasyon-Orta Yüz Fasiyal Hipoplazisi-Elliptositozis Sendromu) (2200)	Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis, 300194 (Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome) (440)	.	AMMEC, DELXq22.3, CXDELq22.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Alport Sendromu, Mental Retardasyon, Orta Yüz Hipoplazisi ve Elliptositoz, 300194 (Alport Sendromu-Mental Retardasyon-Orta Yüz Fasiyal Hipoplazisi-Elliptositozis Sendromu) (Prenatal)	Alport syndrome, mental retardation, midface hypoplasia, and elliptocytosis, 300194 (Alport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome) (Prenatal)	.	AMMEC, DELXq22.3, CXDELq22.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alstrom Sendromu, 203800, Otozomal Resesif; ALMS (Alström Sendromu) (ALMS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alstrom syndrome, 203800, Autosomal recessive; ALMS (Alström syndrome) (ALMS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALMS1	ALMS1, ALSS, KIAA0328	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alstrom syndrome (ALMS) [MIM:203800]: A rare autosomal recessive disorder characterized by progressive cone-rod retinal dystrophy, neurosensory hearing loss, early childhood obesity and diabetes mellitus type 2. Dilated cardiomyopathy, acanthosis nigricans, male hypogonadism, hypothyroidism, developmental delay and hepatic dysfunction can also be associated with the syndrome. {ECO:0000269|PubMed:11941369, ECO:0000269|PubMed:11941370}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alstrom Sendromu, 203800, Otozomal Resesif; ALMS (Alström Sendromu) (ALMS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alstrom syndrome, 203800, Autosomal recessive; ALMS (Alström syndrome) (ALMS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALMS1	ALMS1, ALSS, KIAA0328	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alstrom syndrome (ALMS) [MIM:203800]: A rare autosomal recessive disorder characterized by progressive cone-rod retinal dystrophy, neurosensory hearing loss, early childhood obesity and diabetes mellitus type 2. Dilated cardiomyopathy, acanthosis nigricans, male hypogonadism, hypothyroidism, developmental delay and hepatic dysfunction can also be associated with the syndrome. {ECO:0000269|PubMed:11941369, ECO:0000269|PubMed:11941370}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı 2, 614820, Otozomal Dominant; AHC2 (Çocukluk Çağı Alteman Hemiplejisi) (ATP1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alternating hemiplegia of childhood 2, 614820, Autosomal dominant; AHC2 (Alternating hemiplegia of childhood) (ATP1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı 2, 614820, Otozomal Dominant; AHC2 (Çocukluk Çağı Alteman Hemiplejisi) (MLPA)	Alternating hemiplegia of childhood 2, 614820, Autosomal dominant; AHC2 (Alternating hemiplegia of childhood) (MLPA)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı 2, 614820, Otozomal Dominant; AHC2 (Çocukluk Çağı Alteman Hemiplejisi) (ATP1A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alternating hemiplegia of childhood 2, 614820, Autosomal dominant; AHC2 (Alternating hemiplegia of childhood) (ATP1A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı 2, 614820, Otozomal Dominant; AHC2 (Çocukluk Çağı Alteman Hemiplejisi) (Prenatal) (MLPA)	Alternating hemiplegia of childhood 2, 614820, Autosomal dominant; AHC2 (Alternating hemiplegia of childhood) (Prenatal) (MLPA)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı, 104290, Otozomal Dominant; AHC1 (Çocukluk Çağı Alteman Hemiplejisi) (ATP1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alternating hemiplegia of childhood, 104290, Autosomal dominant; AHC1 (Alternating hemiplegia of childhood) (ATP1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1A2	ATP1A2, FHM2, MHP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alternating hemiplegia of childhood 1 (AHC1) [MIM:104290]: A rare syndrome of episodic hemi- or quadriplegia lasting minutes to days. Most cases are accompanied by dystonic posturing, choreoathetoid movements, nystagmus, other ocular motor abnormalities, autonomic disturbances, and progressive cognitive impairment. It is typically distinguished from familial hemiplegic migraine by infantile onset and high prevalence of associated neurological deficits that become increasingly obvious with age. {ECO:0000269	PubMed:15174025}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alteman Hemipleji, Çocukluk Çağı, 104290, Otozomal Dominant; AHC1 (Çocukluk Çağı Alteman Hemiplejisi) (ATP1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alternating hemiplegia of childhood, 104290, Autosomal dominant; AHC1 (Alternating hemiplegia of childhood) (ATP1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP1A2	ATP1A2, FHM2, MHP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alternating hemiplegia of childhood 1 (AHC1) [MIM:104290]: A rare syndrome of episodic hemi- or quadriplegia lasting minutes to days. Most cases are accompanied by dystonic posturing, choreoathetoid movements, nystagmus, other ocular motor abnormalities, autonomic disturbances, and progressive cognitive impairment. It is typically distinguished from familial hemiplegic migraine by infantile onset and high prevalence of associated neurological deficits that become increasingly obvious with age. {ECO:0000269	PubMed:15174025}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alveoler Kapiller Displazi - Pulmoner Venlerin Hizalama Bozukluğu, 1, 265380, Otozomal Dominant; ACDMPV (Konjenital Alveoler Kılcal Displazi) (FOXF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380, Autosomal dominant; ACDMPV (Congenital alveolar capillary dysplasia) (FOXF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXF1	FOXF1, FKHL5, ACDMPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alveolar capillary dysplasia with misalignment of pulmonary veins (ACDMPV) [MIM:265380]: A rare developmental disorder characterized by abnormal development of the capillary vascular system in the lungs. Histological features include failure of formation and ingrowth of alveolar capillaries, medial muscular thickening of small pulmonary arterioles with muscularization of the intraacinar arterioles, thickened alveolar walls, and anomalously situated pulmonary veins running alongside pulmonary arterioles and sharing the same adventitial sheath. Less common features include a reduced number of alveoli and a patchy distribution of the histopathologic changes. Affected infants present with respiratory distress and the disease is fatal within the newborn period. Additional features include multiple congenital anomalies affecting the cardiovascular, gastrointestinal, genitourinary, and musculoskeletal systems, as well as disruption of the normal right-left asymmetry of intrathoracic or intraabdominal organs. ACDMPV is a rare cause of persistent pulmonary hypertension of the newborn, an abnormal physiologic state caused by failure of transition of the pulmonary circulation from the high pulmonary vascular resistance of the fetus to the low pulmonary vascular resistance of the newborn. {ECO:0000269|PubMed:19500772, ECO:0000269|PubMed:23505205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Alveoler Kapiller Displazi - Pulmoner Venlerin Hizalama Bozukluğu, 1, 265380, Otozomal Dominant; ACDMPV (Konjenital Alveoler Kılcal Displazi) (FOXF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Alveolar capillary dysplasia with misalignment of pulmonary veins, 265380, Autosomal dominant; ACDMPV (Congenital alveolar capillary dysplasia) (FOXF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXF1	FOXF1, FKHL5, ACDMPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alveolar capillary dysplasia with misalignment of pulmonary veins (ACDMPV) [MIM:265380]: A rare developmental disorder characterized by abnormal development of the capillary vascular system in the lungs. Histological features include failure of formation and ingrowth of alveolar capillaries, medial muscular thickening of small pulmonary arterioles with muscularization of the intraacinar arterioles, thickened alveolar walls, and anomalously situated pulmonary veins running alongside pulmonary arterioles and sharing the same adventitial sheath. Less common features include a reduced number of alveoli and a patchy distribution of the histopathologic changes. Affected infants present with respiratory distress and the disease is fatal within the newborn period. Additional features include multiple congenital anomalies affecting the cardiovascular, gastrointestinal, genitourinary, and musculoskeletal systems, as well as disruption of the normal right-left asymmetry of intrathoracic or intraabdominal organs. ACDMPV is a rare cause of persistent pulmonary hypertension of the newborn, an abnormal physiologic state caused by failure of transition of the pulmonary circulation from the high pulmonary vascular resistance of the fetus to the low pulmonary vascular resistance of the newborn. {ECO:0000269|PubMed:19500772, ECO:0000269|PubMed:23505205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alveoler Yumuşak Parça Sarkom, 606243; ASPS (Alveolar Yumuşak Doku Sarkomu) (ASPSCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alveolar soft-part sarcoma, 606243; ASPS (Alveolar soft-tissue sarcoma) (ASPSCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPSCR1	ASPSCR1, RCC17, ASPL, ASPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving ASPSCR1 has been found in two patients with of papillary renal cell carcinoma. Translocation t(X;17)(p11.2;q25). {ECO:0000269	PubMed:11358838}.;	EDTA Blood Sample

Alzheimer Hastalığı 1, Ailesel, 104300, Otozomal Dominant (Erken Başlangıçlı) Otozomal Dominant Alzheimer Hastalığı (APP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease 1, familial, 104300, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (APP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APP	APP, AAA, CVAP, AD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 1 (AD1) [MIM:104300]: A familial early- onset form of Alzheimer disease. It can be associated with cerebral amyloid angiopathy. Alzheimer disease is a neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. (ECO:0000269|PubMed:10097173, ECO:0000269|PubMed:10631141, ECO:0000269|PubMed:10665499, ECO:0000269|PubMed:10867787, ECO:0000269|PubMed:11063718, ECO:0000269|PubMed:11528419, ECO:0000269|PubMed:12034808, ECO:0000269|PubMed:1302033, ECO:0000269|PubMed:1303239, ECO:0000269|PubMed:1303275, ECO:0000269|PubMed:1415269, ECO:0000269|PubMed:15201367, ECO:0000269|PubMed:15365148, ECO:0000269|PubMed:15668448, ECO:0000269|PubMed:1671712, ECO:0000269|PubMed:1678058, ECO:0000269|PubMed:1908231, ECO:0000269|PubMed:1925564, ECO:0000269|PubMed:1944558,	EDTA Blood Sample
Alzheimer Hastalığı 18, Yatkınlık, 615590 (Retiküle Akropigmentasyon, Kitamura Tipi) (ADAM10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease 18, susceptibility to, 615590 (Reticulate acropigmentation of Kitamura) (ADAM10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAM10	ADAM10, MADM, RAK, AD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı 9, Yatkınlık, 608907, Otozomal Dominant (ABCA7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease 9, susceptibility to, 608907, Autosomal dominant (ABCA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA7	ABCA7, ABCX, AD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı -2, 104310, Otozomal Dominant; AD2 (Early-Onset Autosomal Dominant Alzheimer Disease) (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease-2, 104310, Autosomal dominant; AD2 (Early-onset autosomal dominant Alzheimer disease) (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 2 (AD2) [MIM:104310]: A late-onset neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The APOE*4 allele is genetically associated with the common late onset familial and sporadic forms of Alzheimer disease. Risk for AD increased from 20% to 90% and mean age at onset decreased from 84 to 68 years with increasing number of APOE*4 alleles in 42 families with late onset AD. Thus APOE*4 gene dose is a major risk factor for late onset AD and, in these families, homozygosity for APOE*4 was virtually sufficient to cause AD by age 80. The mechanism by which APOE*4 participates in pathogenesis is not known.	EDTA Blood Sample
Alzheimer Hastalığı-4,606889, Otozomal Dominant (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (PSEN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease-4, 606889, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (PSEN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN2	PSEN2, AD4, STM2, CMD1V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 4 (AD4) [MIM:606889]: A familial early- onset form of Alzheimer disease. Alzheimer disease is a neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. (ECO:0000269|PubMed:10631141, ECO:0000269|PubMed:10732806, ECO:0000269|PubMed:12503161, ECO:0000269|PubMed:7638622, ECO:0000269|PubMed:7651536, ECO:0000269|PubMed:9384602). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Alzheimer Hastalığı, Geç Başlangıçlı, Yatkınlık, 104300, Otozomal Dominant (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (PLAU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, late-onset, susceptibility to, 104300, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (PLAU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAU	PLAU, URK, QPD, BDPLT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Geç Başlangıçlı, Yatkınlık, 104300, Otozomal Dominant (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, late-onset, susceptibility to, 104300, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Yatkınlık, 104300, Otozomal Dominant (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (MPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, susceptibility to, 104300, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (MPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPO	MPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Yatkınlık, 104300, Otozomal Dominant (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (A2M geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, susceptibility to, 104300, Autosomal dominant (Early-onset autosomal dominant Alzheimer disease) (A2M gene) (Sequence Analysis-All Coding Exons) (Postnatal)	A2M	A2M , A2MD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Yatkınlık, 104300, Otozomal Dominant; AD (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (HFE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, susceptibility to, 104300, Autosomal dominant; AD (Early-onset autosomal dominant Alzheimer disease) (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Tip 3, 607822, Otozomal Dominant; AD (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, type 3, 607822, Autosomal dominant; AD (Early-onset autosomal dominant Alzheimer disease) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer Hastalığı, Tip 3, Spastik Paraparezi ve Apraksi, 607822, Otozomal Dominant; AD (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822, Autosomal dominant; AD (Early-onset autosomal dominant Alzheimer disease) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alzheimer hastalığı, tip 3, spastik paraparezi ve sıra dışı plaklar, 607822, Otozomal dominant; AD (Erken başlangıçlı otozomal dominant Alzheimer hastalığı) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822, Autosomal dominant; AD (Early-onset autosomal dominant Alzheimer disease) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amegakaryositik Trombositopeni, Konjenital; Cant (Konjenital Amegakaryositik Trombositopeni) (MPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL; CAMT (Congenital amegakaryocytic thrombocytopenia) (MPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPL	MPL, TPOR, MPLV, THCYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amegakaryositik Trombositopeni, Konjenital; Cant (Konjenital Amegakaryositik Trombositopeni) (MPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	AMEGAKARYOCYTIC THROMBOCYTOPENIA, CONGENITAL; CAMT (Congenital amegakaryocytic thrombocytopenia) (MPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPL	MPL, TPOR, MPLV, THCYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amnyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Amelogenezis İmperfekta, Hipomatürasyon Tipi, 2A6, 617217, Otozomal Resesif (Amelogenesis Imperfekta) (GPR68 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217, Autosomal recessive (Amelogenesis imperfecta) (GPR68 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR68	GPR68, OGR1, AI2A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1E, 301200, X'e Bağlı Dominant; AI1E (Amelogenesis İmperfekta) (AMELX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type 1E, 301200, X-linked dominant; AI1E (Amelogenesis imperfecta) (AMELX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMELX	AMELX, AMG, AIH1, AMGX, AI1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1E (AI1E) [MIM:301200]: A X- linked defect of dental enamel formation. Teeth have only a thin layer of enamel with normal hardness. The thinness of the enamel makes the teeth appear small. (ECO:0000269|PubMed:10669095, ECO:0000269|PubMed:15111628, ECO:0000269|PubMed:7599636, ECO:0000269|PubMed:7782077, ECO:0000269|PubMed:9188994). Note=The disease is caused by mutations affecting the gene represented in this entry. ;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1A, 104530, Otozomal Dominant; AI1A (Amelogenesis İmperfekta) (LAMB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IA, 104530, Autosomal dominant; AI1A (Amelogenesis imperfecta) (LAMB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB3	LAMB3, AI1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1A (AI1A) [MIM:104530]: A form of amelogenesis imperfecta, a disorder characterized by defective enamel formation. The enamel may be hypoplastic, hypomineralized or both, and affected teeth may be discoloured, sensitive or prone to disintegration. (ECO:0000269|PubMed:23632796, ECO:0000269|PubMed:23958762). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1A, 104530, Otozomal Dominant; AI1A (Amelogenesis İmperfekta) (MLPA)	Amelogenesis imperfecta, type IA, 104530, Autosomal dominant; AI1A (Amelogenesis imperfecta) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Amelogenesis imperfecta 1A (AI1A) [MIM:104530]: A form of amelogenesis imperfecta, a disorder characterized by defective enamel formation. The enamel may be hypoplastic, hypomineralized or both, and affected teeth may be discoloured, sensitive or prone to disintegration. (ECO:0000269|PubMed:23632796, ECO:0000269|PubMed:23958762). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1B, 104500, Otozomal Dominant; AI1B (Amelogenesis İmperfekta) (ENAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IB, 104500, Autosomal dominant; AI1B (Amelogenesis imperfecta) (ENAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENAM	ENAM, AIH2, AI1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1B (AI1B) [MIM:104500]: An autosomal dominant defect of enamel formation. Clinical manifestations may be variable. Some cases present with generalized enamel hypoplasia resulting in small, smooth, yellow and widely spaced teeth (smooth hypoplastic AI). Others show horizontal rows of pits, grooves or a hypoplastic area in the enamel (local hypoplastic AI). (ECO:0000269|PubMed:11487571, ECO:0000269|PubMed:11978766, ECO:0000269|PubMed:20439930, ECO:0000269|PubMed:25789606). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Amelogenezis İmperfekta, Tip 1C, 204650, Otozomal Resesif, AI1C (Amelogenesis Imperfekta) (ENAM gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IC, 204650, Autosomal recessive; AI1C (Amelogenesis imperfecta) (ENAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENAM	ENAM, AIH2, AI1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1B (AI1B) [MIM:104500]: An autosomal dominant defect of enamel formation. Clinical manifestations may be variable. Some cases present with generalized enamel hypoplasia resulting in small, smooth, yellow and widely spaced teeth (smooth hypoplastic AI). Others show horizontal rows of pits, grooves or a hypoplastic area in the enamel (local hypoplastic AI). {ECO:0000269|PubMed:11487571, ECO:0000269|PubMed:11978766, ECO:0000269|PubMed:20439930, ECO:0000269|PubMed:25789696}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1F 616270, Otozomal Resesif; AI1F (Amelogenesis Imperfekta) (AMBN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IF, 616270, Autosomal recessive; AI1F (Amelogenesis imperfecta) (AMBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMBN	AMBN, AI1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta Tip 1G, Otozomal Resesif; AI1G (Diş Minesi-Renal Sendrom), 204690, (Amelogenesis Imperfekta-Nefrokalsinozis Sendromu) (FAM20A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690, Autosomal recessive; AI1G (Amelogenesis imperfecta-nephrocalcinosis syndrome) (FAM20A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM20A	FAM20A, AIGFS, AI1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1G (AI1G) [MIM:204690]: A disorder characterized by dental anomalies, gingival overgrowth, and nephrocalcinosis. Dental anomalies include hypoplastic amelogenesis imperfecta, intrapulpal calcifications, delay of tooth eruption, hypodontia/oligodontia, periconeal radiolucencies and unerupted teeth. {ECO:0000269|PubMed:21549343, ECO:0000269|PubMed:21990045, ECO:0000269|PubMed:23434854, ECO:0000269|PubMed:23468644, ECO:0000269|PubMed:23697977, ECO:0000269|PubMed:24196488, ECO:0000269|PubMed:24259279, ECO:0000269|PubMed:24756937, ECO:0000269|PubMed:25636655, ECO:0000269|PubMed:25789606, ECO:0000269|PubMed:25827751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 1H, 616221, Otozomal Resesif; AI1H (Amelogenesis Imperfekta) (ITGB6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IH, 616221, Autosomal recessive; AI1H (Amelogenesis imperfecta) (ITGB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB6	ITGB6, AI1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 1H (AI1H) [MIM:616221]: A disorder characterized by defective enamel formation, resulting in hypoplastic and hypomineralized tooth enamel that may be rough, pitted, and/or discolored. {ECO:0000269|PubMed:24305999, ECO:0000269|PubMed:24314098}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 2A1, 204700, Otozomal Resesif; AI2A1 (Amelogenesis Imperfekta) (KLK4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IIA1, 204700, Autosomal recessive; AI2A1 (Amelogenesis imperfecta) (KLK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLK4	KLK4, EMSP1, PRSS17, AI2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 2A2, 612529, Otozomal Resesif; AI2A2 (Amelogenesis İmperfekta) (MMP20 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IIA2, 612529, Autosomal recessive; AI2A2 (Amelogenesis imperfecta) (MMP20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP20	MMP20, AI2A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta, hypomaturation type, 2A2 (AI2A2) [MIM:612529]: A defect of enamel formation. The disorder involves both primary and secondary dentitions. The teeth have a shiny agar jelly appearance and the enamel is softer than normal. Brown pigment is present in middle layers of enamel. {ECO:0000269|PubMed:15744043}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 2A3, 613211, Otozomal Resesif; AI2A3 (Amelogenesis Imperfekta) (WDR72 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IIA3, 613211, Autosomal recessive; AI2A3 (Amelogenesis imperfecta) (WDR72 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR72	WDR72, AI2A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta, hypomaturation type, 2A3 (AI2A3) [MIM:613211]: A defect of enamel formation. The disorder involves both primary and secondary dentitions. The teeth have a shiny agar jelly appearance and the enamel is softer than normal. Brown pigment is present in middle layers of enamel. {ECO:0000269|PubMed:19853237}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 2A4, 614832, Otozomal Resesif (Amelogenesis Imperfekta) (C4orf26 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IIA4, 614832, Autosomal recessive (Amelogenesis imperfecta) (C4orf26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C4orf26	C4orf26, AI2A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 2A5, 615887, Otozomal Resesif; AI2A5 (Amelogenesis İmperfekta) (SLC24A4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IIA5, 615887, Autosomal recessive; AI2A5 (Amelogenesis imperfecta) (SLC24A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A4	SLC24A4, NCKX4, SHEP6, AI2A5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta, hypomaturation type, 2A5 (AI2A5) [MIM:615887]: A defect of enamel formation. The disorder involves both primary and secondary dentitions. The teeth have a shiny agar jelly appearance and the enamel is softer than normal. Brown pigment is present in middle layers of enamel. {ECO:0000269|PubMed:23375655, ECO:0000269|PubMed:24621671}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 3, 130900, Otozomal Dominant (Amelogenesis Imperfekta) (FAM83H gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type III, 130900, Autosomal dominant (Amelogenesis imperfecta) (FAM83H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM83H	FAM83H, AI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 3; AI3 (Amelogenesis Imperfekta) (FAM83H gen) (Tüm Gen Dizi Analizi) (Postnatal)	AMELOGENESIS IMPERFECTA, TYPE III; AI3 (Amelogenesis imperfecta) (FAM83H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM83H	FAM83H, AI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenez İmperfekta, Tip 1J, 617297, Otozomal Resesif; AI1J (Hipoplastik Amelogenezis İmperfekta) (ACPT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IJ, 617297, Autosomal recessive; AI1J (Hypoplastic amelogenesis imperfecta) (ACPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACPT	ACPT, AI1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amelogenezis İmperfekta, Tip 4, 104510, Otozomal Dominant; AI4 (Amelogenesis Imperfekta) (DLX3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amelogenesis imperfecta, type IV, 104510, Autosomal dominant; AI4 (Amelogenesis imperfecta) (DLX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLX3	DLX3, TDO, AI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 4 (AI4) [MIM:104510]: An autosomal dominant defect of enamel formation associated with enlarged pulp chambers. Enamel is thin, teeth are small and widely spaced. {ECO:0000269|PubMed:15666299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aminoasitaz 1 Eksikliği, 609924, Otozomal Resesif; ACY1D (Nörolojik Bulgular, Aminoasitaz 1 Eksikliği İle İlişkili) (ACY1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Aminoacylase 1 deficiency, 609924, Autosomal recessive; ACY1D (Neurological conditions associated with aminoacylase 1 deficiency) (ACY1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACY1	ACY1, ACY1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aminoasitaz 1 Eksikliği, 609924, Otozomal Resesif; ACY1D (Nörolojik Bulgular, Aminoasitaz 1 Eksikliği İle İlişkili) (ACY1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Aminoacylase 1 deficiency, 609924, Autosomal recessive; ACY1D (Neurological conditions associated with aminoacylase 1 deficiency) (ACY1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACY1	ACY1, ACY1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aminoglikozid İle İlişkili Sağırlık (A1555G) (mtDNA 12S rRNA (MTRNR1) gen) (Dizi Analizi) (Postnatal)	Aminoglycoside-induced deafness (A1555G) (MTRNR1 gene) (Sequence analysis) (mtDNA 12S rRNA (MTRNR1) gene) (Dizi Analizi) (Postnatal)	mtDNA 12S rRNA (MTRNR1)	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AML - MDS Paneli (Del (5q) - t(15;17) (q22;q21) (PML/ RARA) - TP53 - t(8;21) (q22;q22) (ETO/ AML1) - Trizomi 8 - t(11;?) (q23;?) (MLL/ ?) - -7/ Del (7q) (Monozomi 7/ Delesyon 7q) - inv(16) (p13;q22) (CBFB/ MYH11) - Del (20q)) (FISH)	AML - MDS Panel (Del (5q) - t(15;17) (q22;q21) (PML/ RARA) - TP53 - t(8;21) (q22;q22) (ETO/ AML1) - Trisomy 8 - t(11;?) (q23;?) (MLL/ ?) - -7/ Del (7q) (Monosomy 7/ Deleasyon 7q) - inv(16) (p13;q22) (CBFB/ MYH11) - Del (20q)) (FISH) (PANEL)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Amniyon Sıvısından Kromozom Analizi (Prenatal)	Amniotic fluid- Chromosome analysis (Prenatal)	.	.	Kromozom analizi/ Karyotype analysis	Amniyon sıvısı Enjektör içinde (SİYAH PİSTONLU OLMAMALIDIR)	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid Injector (SHOULD NOT HAVE BLACK PISTON)
Amiloidoz, 3 Veya Daha Fazla Tip, 105200, Otozomal Dominant (Kalıtsal Amiloidoz-Primer Böbrek Tutulum) (APOA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, 3 or more types, 105200, Autosomal dominant (Hereditary amyloidosis with primary renal involement) (APOA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA1	APOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269|PubMed:1502149, ECO:0000269|PubMed:2123470, ECO:0000269|PubMed:3142462, ECO:0000269|PubMed:8208902}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amiloidoz, Ailesel Visseral (Kalıtsal Amiloidoz-Primer Böbrek Tutulumu) (APOA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	AMYLOIDOSIS, FAMILIAL VISCERAL (Hereditary amyloidosis with primary renal involement) (APOA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA1	APOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269|PubMed:1502149, ECO:0000269|PubMed:2123470, ECO:0000269|PubMed:3142462, ECO:0000269|PubMed:8208902}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amiloidoz, Ailesel Visseral, 105200, Otozomal Dominant (Kalıtsal Amiloidoz-Primer Böbrek Tutulum) (FGA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, familial visceral, 105200, Autosomal dominant (Hereditary amyloidosis with primary renal involement) (FGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269|PubMed:8097946}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Amyloidosis, Ailesel Visseral, 105200, Otozomal Dominant (Kalıtsal Amiloidoz-Primer Böbrek Tutulum) (B2M gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, familial visceral, 105200, Autosomal dominant (Hereditary amyloidosis with primary renal involement) (B2M gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B2M	B2M, IMD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amiloidoz, Fin Tipi, 105120, Otozomal Dominant (Agel Amiloidoz) (GSN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, Finnish type, 105120, Autosomal dominant (AGel amyloidosis) (GSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSN	GSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 5 (AMYL5) [MIM:105120]: A hereditary generalized amyloidosis due to gelsolin amyloid deposition. It is typically characterized by cranial neuropathy and lattice corneal dystrophy. Most patients have modest involvement of internal organs, but severe systemic disease can develop in some individuals causing peripheral polyneuropathy, amyloid cardiomyopathy, and nephrotic syndrome leading to renal failure. {ECO:0000269|PubMed:1338910, ECO:0000269|PubMed:2176481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Amiloidoz, Herediter, Transtiretin İle İlişkili, 105210, Otozomal Dominant (ATTRV30M Amiloidoz) (TTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, hereditary, transthyretin-related, 105210, Autosomal dominant (ATTRv30M amyloidosis) (TTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTR	TTR, PALB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis, transthyretin-related (AMYL-TTR) [MIM:105210]: A hereditary generalized amyloidosis due to transthyretin amyloid deposition. Protein fibrils can form in different tissues leading to amyloid polyneuropathies, amyloidotic cardiomyopathy, carpal tunnel syndrome, systemic senile amyloidosis. The disease includes leptomeningeal amyloidosis that is characterized by primary involvement of the central nervous system. Neuropathologic examination shows amyloid in the walls of leptomeningeal vessels, in pia arachnoid, and subpial deposits. Some patients also develop vitreous amyloid deposition that leads to visual impairment (oculoleptomeningeal amyloidosis). Clinical features include seizures, stroke-like episodes, dementia, psychomotor deterioration, variable amyloid deposition in the vitreous humor. {ECO:0000269|PubMed:10036587, ECO:0000269|PubMed:10071047, ECO:0000269|PubMed:10211412, ECO:0000269|PubMed:10436378, ECO:0000269|PubMed:10439117, ECO:0000269|PubMed:10611950, ECO:0000269|PubMed:10627135, ECO:0000269|PubMed:10694917, ECO:0000269|PubMed:10842705, ECO:0000269|PubMed:10842718, ECO:0000269|PubMed:10882995, ECO:0000269|PubMed:11445644, ECO:0000269|PubMed:11866053, ECO:0000269|PubMed:12050338, ECO:0000269|PubMed:12557757, ECO:0000269|PubMed:12771253, ECO:0000269|PubMed:1301926, ECO:0000269|PubMed:1351039,	EDTA Blood Sample
Amiloidoz, Primer Lokalize Kutanöz, 1, 105250, Otozomal Dominant; PLCA1 (Alesel Primer Lokalize Kutanöz Amiloidoz) (OSMR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, primary localized cutaneous, 1, 105250, Autosomal dominant; PLCA1 (Familial primary localized cutaneous amyloidosis) (OSMR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OSMR	OSMR, OSMRB, PLCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis, primary localized cutaneous, 1 (PLCA1) [MIM:105250]: A primary amyloidosis characterized by localized cutaneous amyloid deposition. This condition usually presents with itching (especially on the lower legs) and visible changes of skin hyperpigmentation and thickening that may be exacerbated by chronic scratching and rubbing. Primary localized cutaneous amyloidosis is often divided into macular and lichen subtypes although many affected individuals often show both variants coexisting. Lichen amyloidosis characteristically presents as a pruritic eruption of grouped hyperkeratotic papules with a predilection for the shins, calves, ankles and dorsa of feet and thighs. Papules may coalesce to form hyperkeratotic plaques that can resemble lichen planus, lichen simplex or nodular prurigo. Macular amyloidosis is characterized by small pigmented macules that may merge to produce macular hyperpigmentation, sometimes with a reticulate or rippled pattern. In macular and lichen amyloidosis, amyloid is deposited in the papillary dermis in association with grouped colloid bodies, thought to represent degenerate basal keratinocytes. The amyloid deposits probably reflect a combination of degenerate keratin filaments, serum amyloid P component, and deposition of immunoglobulins. {ECO:0000269|PubMed:18179886, ECO:0000269|PubMed:19690585}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amiloidoz, Primer Lokalize Kutanöz, 2, 613955, Otozomal Dominant; PLCA2 (Alesel Primer Lokalize Kutanöz Amiloidoz) (IL31RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, primary localized cutaneous, 2, 613955, Autosomal dominant; PLCA2 (Familial primary localized cutaneous amyloidosis) (IL31RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL31RA	IL31RA, GLMR, GPL, PLCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis, primary localized cutaneous, 2 (PLCA2) [MIM:613955]: A primary amyloidosis characterized by localized cutaneous amyloid deposition. This condition usually presents with itching (especially on the lower legs) and visible changes of skin hyperpigmentation and thickening that may be exacerbated by chronic scratching and rubbing. Primary localized cutaneous amyloidosis is often divided into macular and lichen subtypes although many affected individuals often show both variants coexisting. Lichen amyloidosis characteristically presents as a pruritic eruption of grouped hyperkeratotic papules with a predilection for the shins, calves, ankles and dorsa of feet and thighs. Papules may coalesce to form hyperkeratotic plaques that can resemble lichen planus, lichen simplex or nodular prurigo. Macular amyloidosis is characterized by small pigmented macules that may merge to produce macular hyperpigmentation, sometimes with a reticulate or rippled pattern. In macular and lichen amyloidosis, amyloid is deposited in the papillary dermis in association with grouped colloid bodies, thought to represent degenerate basal keratinocytes. The amyloid deposits probably reflect a combination of degenerate keratin filaments, serum amyloid P component, and deposition of immunoglobulins. {ECO:0000269	PubMed:19690585}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amiloidoz, Renal, 105200, Otozomal Dominant (Kalıtsal Amiloidoz-Primer Böbrek Tutulum) (LYZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, renal, 105200, Autosomal dominant (Hereditary amyloidosis with primary renal involement) (LYZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LYZ	LYZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amiloidoz, Sekonder, Yatkınlık (APCS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyloidosis, secondary, susceptibility to (APCS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APCS	APCS, SAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 1, 105400, Otozomal Resesif, Otozomal Dominant (Amyotrofik Lateral Skleroz) (SOD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 1, 105400, Autosomal recessive, Autosomal dominant (Amyotrophic lateral sclerosis) (SOD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOD1	SOD1, ALS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 1 (ALS1) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:1043043S, ECO:0000269|PubMed:10732812, ECO:0000269|PubMed:11369193, ECO:0000269|PubMed:11535232, ECO:0000269|PubMed:12402272, ECO:0000269|PubMed:14506039, ECO:0000269|PubMed:21220647, ECO:0000269|PubMed:21247266, ECO:0000269|PubMed:7496165, ECO:0000269|PubMed:7501156, ECO:0000269|PubMed:7647793, ECO:0000269|PubMed:7655468, ECO:0000269|PubMed:7655469, ECO:0000269|PubMed:7655471, ECO:0000269|PubMed:7700376, ECO:0000269|PubMed:7795609, ECO:0000269|PubMed:7836951, ECO:0000269|PubMed:7870076, ECO:0000269|PubMed:7881433, ECO:0000269|PubMed:7887412, ECO:0000269|PubMed:7951252, ECO:0000269|PubMed:7985016,	EDTA Blood Sample
Amyotrofik Lateral Skleroz 10, ± Frontotemporal Demans, 612069, Otozomal Dominant; ALS10 (Amyotrofik Lateral Skleroz) (TARDBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 10, with or without FTD, 612069, Autosomal dominant; ALS10 (Amyotrophic lateral sclerosis) (TARDBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TARDBP	TARDBP, TDP43, ALS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 10 (ALS10) [MIM:612069]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5- 10% of the cases. {ECO:0000269|PubMed:18288693, ECO:0000269|PubMed:18309045, ECO:0000269|PubMed:18372902, ECO:0000269|PubMed:18396105, ECO:0000269|PubMed:19439952, ECO:0000269|PubMed:19224587, ECO:0000269|PubMed:19695877, ECO:0000269|PubMed:21220647, ECO:0000269|PubMed:21418058, ECO:0000269|PubMed:22456481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amyotrofik Lateral Skleroz 11, 612577, Otozomal Dominant; ALS11 (Amyotrofik Lateral Skleroz) (FIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 11, 612577, Autosomal dominant; ALS11 (Amyotrophic lateral sclerosis) (FIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIG4	FIG4, KIAA0274, SAC3, ALS11, YVS, BTOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 12, 613435; ALS12 (Amyotrofik Lateral Skleroz) (OPTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 12, 613435; ALS12 (Amyotrophic lateral sclerosis) (OPTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPTN	OPTN, GLC1E, FIP2, HYPL, NRP, ALS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 14, ± Frontotemporal Demans, 613954; ALS14 (Amyotrofik Lateral Skleroz) (VCP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 14, with or without frontotemporal dementia, 613954; ALS14 (Amyotrophic lateral sclerosis) (VCP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCP	VCP, IBMPFD1, ALS14, CMT2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 15, ± Frontotemporal Demans, 300857, X'e Bağlı Dominant; ALS15 (Amyotrofik Lateral Skleroz) (UBQLN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia, 300857, X-linked dominant; ALS15 (Amyotrophic lateral sclerosis) (UBQLN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBQLN2	UBQLN2, PLIC2, CHAP1, ALS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 15, with or without frontotemporal dementia (ALS15) [MIM:300857]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. Patients with ALS15 may develop frontotemporal dementia. {ECO:0000269|PubMed:21857683, ECO:0000269|PubMed:22560112, ECO:0000269|PubMed:22717235, ECO:0000269|PubMed:22892309, ECO:0000269|PubMed:24215460, ECO:0000269|PubMed:25610951}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Amyotrofik Lateral Skleroz 16, Jüvenil, 614373, Otozomal Resesif (Juvenil Amyotrofik Lateral Sklerozl (SIGMAR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 16, juvenile, 614373, Autosomal recessive (Juvenile amyotrophic lateral sclerosis) (SIGMAR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIGMAR1	SIGMAR1, SRBP, ALS16, DSMA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 17, 614696, Otozomal Dominant; ALS17 (Amyotrofik Lateral Sklerozl (CHMP2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 17, 614696, Autosomal dominant; ALS17 (Amyotrophic lateral sclerosis) (CHMP2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHMP2B	CHMP2B, DMT1, VPS2B, ALS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Skleroz 18, 614808; ALS18 (Amyotrofik Lateral Sklerozl (PFN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 18, 614808; ALS18 (Amyotrophic lateral sclerosis) (PFN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PFN1	PFN1, ALS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 18 (ALS18) [MIM:614808]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5- 10% of the cases. {ECO:0000269	PubMed:22801503}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Skleroz 19, 615515, Otozomal Dominant; ALS19 (Amyotrofik Lateral Sklerozl (ERBB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 19, 615515, Autosomal dominant; ALS19 (Amyotrophic lateral sclerosis) (ERBB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERBB4	ERBB4, HER4, ALS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 19 (ALS19) [MIM:615515]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5- 10% of the cases. {ECO:0000269	PubMed:24119685}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 2, Jüvenil, 205100, Otozomal Resesif (Juvenil Amyotrofik Lateral Sklerozl (ALS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 2, juvenile, 205100, Autosomal recessive (Juvenile amyotrophic lateral sclerosis) (ALS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALS2	ALS2, ALSJ, PLSJ, IAHSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 2 (ALS2) [MIM:205100]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269	PubMed:11586298}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 20, 615426, Otozomal Dominant; ALS20 (Amyotrofik Lateral Sklerozl (HNRNPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 20, 615426, Autosomal dominant; ALS20 (Amyotrophic lateral sclerosis) (HNRNPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPA1	HNRNPA1, IBMPFD3, ALS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 21, 606070, Otozomal Dominant; ALS21 (Amyotrofik Lateral Sklerozl (MATR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 21, 606070, Autosomal dominant; ALS21 (Amyotrophic lateral sclerosis) (MATR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MATR3	MATR3, MPD2, ALS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 21 (ALS21) [MIM:606070]: A neurodegenerative disorder affecting upper and lower motor neurons, resulting in muscle weakness and respiratory failure. Some patients may develop myopathic features or dementia. {ECO:0000269|PubMed:19344878, ECO:0000269|PubMed:24686783, ECO:0000269|PubMed:25771394}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Skleroziis 22, ± Frontotemporal Demans, 616208, Otozomal Dominant; ALS22 (Amyotrofik Lateral Sklerozl (TUBA4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 22 with or without frontotemoral dementia, 616208, Autosomal dominant; ALS22 (Amyotrophic lateral sclerosis) (TUBA4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBA4A	TUBA4A, TUBA1, ALS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 22, with or without frontotemporal dementia (ALS22) [MIM:616208]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. Patients with ALS22 may develop frontotemporal dementia. {ECO:0000269	PubMed:25374358}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 4, Jüvenil, 602433, Otozomal Dominant (Amyotrofik Lateral Sklerozl Tip 4) (SETX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 4, juvenile, 602433, Autosomal dominant (Amyotrophic lateral sclerosis type 4) (SETX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETX	SETX, SCAR1, AOA2, ALS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 4 (ALS4) [MIM:602433]: A form of amyotrophic lateral sclerosis with childhood- or adolescent-onset, and characterized by slow disease progression and the sparing of bulbar and respiratory muscles. Amyotrophic lateral sclerosis is a neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:14770181, ECO:0000269|PubMed:15106121, ECO:0000269|PubMed:21190393, ECO:0000269|PubMed:24105744, ECO:0000269|PubMed:24244371}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 4, Jüvenil, 602433, Otozomal Dominant (Amyotrofik Lateral Sklerozl Tip 4) (MLPA)	Amyotrophic lateral sclerosis 4, juvenile, 602433, Autosomal dominant (Amyotrophic lateral sclerosis type 4) (MLPA)	SETX	SETX, SCAR1, AOA2, ALS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Amyotrophic lateral sclerosis 4 (ALS4) [MIM:602433]: A form of amyotrophic lateral sclerosis with childhood- or adolescent-onset, and characterized by slow disease progression and the sparing of bulbar and respiratory muscles. Amyotrophic lateral sclerosis is a neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:14770181, ECO:0000269|PubMed:15106121, ECO:0000269|PubMed:21190393, ECO:0000269|PubMed:24105744, ECO:0000269|PubMed:24244371}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 5, Jüvenil, 602099, Otozomal Resesif (Juvenil Amyotrofik Lateral Sklerozl (SPG11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 5, juvenile, 602099, Autosomal recessive (Juvenile amyotrophic lateral sclerosis) (SPG11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG11	SPG11, KIAA1840, FLJ21439, ALS5, CMT2X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Amyotrofik Lateral Skleroz 6, ± Frontotemporal Demans 608030; ALS6 (Amyotrofik Lateral Sklerozl (FUS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia, 608030; ALS6 (Amyotrophic lateral sclerosis) (FUS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUS	FUS, TLS, ALS6, ETM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia (ALS6) [MIM:608030]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:19251627, ECO:0000269|PubMed:19251628, ECO:0000269|PubMed:19861302, ECO:0000269|PubMed:20124201}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 8, 608627, Otozomal Dominant; ALS8 (Amyotrofik Lateral Sklerozl (VAPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 8, 608627, Autosomal dominant; ALS8 (Amyotrophic lateral sclerosis) (VAPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VAPB	VAPB, VAPC, ALS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotrofik Lateral Sklerozz 9, 611895; ALS9 (Amyotrofik Lateral Sklerozl (ANG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis 9, 611895; ALS9 (Amyotrophic lateral sclerosis) (ANG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANG	ANG, RNASE5, ALS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 9 (ALS9) [MIM:611895]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:15557516, ECO:0000269|PubMed:16501576, ECO:0000269|PubMed:17703939, ECO:0000269|PubMed:17886298, ECO:0000269|PubMed:18087731, ECO:0000269|PubMed:22292843, ECO:0000269|PubMed:25370231}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Amyotrofik Lateral Skleroz-Parkinsonizm / Demans Kompleksi, Yetkinlik, 105500, Otozomal Dominant (Amyotrofik Lateral Skleroz-Parkinsonizm-Demans Kompleksi) (TRPM7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis-parkinsonism/dementia complex, susceptibility to, 105500, Autosomal dominant (Amyotrophic lateral sclerosis-parkinsonism-dementia complex) (TRPM7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPM7	TRPM7, LTRPC7, CHAK, ALSPDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis-parkinsonism/dementia complex 1 (ALS-PDC1) [MIM:105500]: A neurodegenerative disorder characterized by chronic, progressive and uniformly fatal amyotrophic lateral sclerosis and parkinsonism-dementia. Both diseases are known to occur in the same kindred, the same sibship and even the same individual. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample

Amyotropik Lateral Skleroz, Yatkınlık, 105400, Otozomal Resesif, Otozomal Dominant (Amyotrofik Lateral Skleroz) (PRPH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis, susceptibility to, 105400, Autosomal recessive, Autosomal dominant (Amyotrophic lateral sclerosis) (PRPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH	PRPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Amyotropik Lateral Skleroz, Yatkınlık, 105400, Otozomal Resesif, Otozomal Dominant (Amyotrofik Lateral Skleroz) (DCTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis, susceptibility to, 105400, Autosomal recessive, Autosomal dominant (Amyotrophic lateral sclerosis) (DCTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCTN1	DCTN1, HMN7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis (ALS) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:15326253, ECO:0000269|PubMed:16240349}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Amyotrofik Lateral Skleroz, Yatkınlık, 105400, Otozomal Resesif, Otozomal Dominant; ALS1 (Amyotrofik Lateral Skleroz) (NEFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis, susceptibility to, 105400, Autosomal recessive, Autosomal dominant; ALS1 (Amyotrophic lateral sclerosis) (NEFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEFH	NEFH, CMT2CC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis (ALS) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:7849698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Amyotrofik Lateral Skleroz, Yatkınlık, 13, 183090, Otozomal Dominant (Spinoserebellar Ataksi Tip 2) (ATXN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophic lateral sclerosis, susceptibility to, 13, 183090, Autosomal dominant (Spinocerebellar ataxia type 2) (ATXN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN2	ATXN2, ATX2, SCA2, ASL13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 2 (SCA2) [MIM:183090]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA2 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA2 is characterized by hyporeflexia, myoclonus and action tremor and dopamine-responsive parkinsonism. In some patients, SCA2 presents as pure familial parkinsonism without cerebellar signs. {ECO:0000269|PubMed:8896555, ECO:0000269|PubMed:8896556, ECO:0000269|PubMed:8899537}. Note=The disease is caused by mutations affecting the gene represented in this entry. SCA2 is caused by expansion of a CAG repeat resulting in about 36 to 52 repeats in some patients. Longer expansions result in earlier the expansion, onset of the disease.	EDTA Blood Sample
Amyotrofi, Kalıtsal Nevraljik, 162100, Otozomal Dominant; HNA (Nöraljik Amyotrofi) (SEPT9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Amyotrophy, hereditary neuralgic, 162100, Autosomal dominant; HNA (Neuralgic amyotrophy) (SEPT9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEPT9	SEPT9, MSF, MSF1, NAP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Analbuminemi, 616000; ANALBA (Konjenital Analbuminemi) (ALB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Analbuminemia, 616000; ANALBA (Congenital analbuminemia) (ALB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALB	ALB, ANALBA, FDAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Analbuminemi, 616000; ANALBA (Konjenital Analbuminemi) (ALB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Analbuminemia, 616000; ANALBA (Congenital analbuminemia) (ALB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALB	ALB, ANALBA, FDAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anauksetik Displazi, 607095, Otozomal Resesif; ANXD1 (Anauksetik Displazi) (RMRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anauxetic dysplasia, 607095, Autosomal recessive; ANXD1 (Anauxetic dysplasia) (RMRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anauksetik Displazi, 607095, Otozomal Resesif; ANXD1 (Anauksetik Displazi) (RMRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anauxetic dysplasia, 607095, Autosomal recessive; ANXD1 (Anauxetic dysplasia) (RMRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Andersen Sendromu, 170390, Otozomal Dominant (Kardiodsitmik Potasyum Duyarlı Periyodik Paralizi (Andersen Sendromu)) (KCNJ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Andersen syndrome, 170390, Autosomal dominant (Cardiodysrhythmic potassium-sensitive periodic paralysis) (KCNJ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. {ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324964}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Andersen Sendromu, 170390, Otozomal Dominant (Kardiodsitmik Potasyum Duyarlı Periyodik Paralizi (Andersen Sendromu)) (MLPA)	Andersen syndrome, 170390, Autosomal dominant (Cardiodysrhythmic potassium-sensitive periodic paralysis) (MLPA)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. {ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324964}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Andersen Sendromu, 170390, Otozomal Dominant (Kardiodsitmik Potasyum Duyarlı Periyodik Paralizi (Andersen Sendromu)) (KCNJ2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Andersen syndrome, 170390, Autosomal dominant (Cardiodysrhythmic potassium-sensitive periodic paralysis) (KCNJ2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. {ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324964}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Andersen Sendromu, 170390, Otozomal Dominant (Kardiodsitmik Potasyum Duyarlı Periyodik Paralizi (Andersen Sendromu)) (Prenatal) (MLPA)	Andersen syndrome, 170390, Autosomal dominant (Cardiodysrhythmic potassium-sensitive periodic paralysis) (Prenatal) (MLPA)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. {ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324964}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Androjen Duyarsızlığı, 300068, X'e Bağlı Resesif; AIS (Komplet Androjen Duyarsızlık Sendromu) (AR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Androgen insensitivity, 300068, X-linked recessive; AIS (Complete androgen insensitivity syndrome) (AR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487245, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1608793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398,	EDTA Blood Sample

Androjen Duyarsızlığı, 300068, X'e Bağlı Resesif; AIS (Komplet Androjen Duyarsızlık Sendromu) (MLPA)	Androgen insensitivity, 300068, X-linked recessive; AIS (Complete androgen insensitivity syndrome) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398,	EDTA Blood Sample
Androjen Duyarsızlığı, 300068, X'e Bağlı Resesif; AIS (Komplet Androjen Duyarsızlık Sendromu) (AR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Androgen insensitivity, 300068, X-linked recessive; AIS (Complete androgen insensitivity syndrome) (AR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Androjen Duyarsızlığı, 300068, X'e Bağlı Resesif; AIS (Komplet Androjen Duyarsızlık Sendromu) (Prenatal) (MLPA)	Androgen insensitivity, 300068, X-linked recessive; AIS (Complete androgen insensitivity syndrome) (Prenatal) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Androjen Duyarsızlığı, Parsiyel, ± Meme Kanseri, 312300, X'e Bağlı Resesif; PAIS (Parsiyel Androjen Insensitivite Sendromu) (AR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Androgen insensitivity, partial, with or without breast cancer, 312300, X-linked recessive; PAIS (Partial androgen insensitivity syndrome) (AR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Androjen Duyarsızlığı, Parsiyel, ± Meme Kanseri, 312300, X'e Bağlı Resesif; PAIS (Parsiyel Androjen Insensitivite Sendromu) (MLPA)	Androgen insensitivity, partial, with or without breast cancer, 312300, X-linked recessive; PAIS (Partial androgen insensitivity syndrome) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Androjen Duyarsızlığı, Parsiyel, ± Meme Kanseri, 312300, X'e Bağlı Resesif; PAIS (Parsiyel Androjen Insensitivite Sendromu) (AR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Androgen insensitivity, partial, with or without breast cancer, 312300, X-linked recessive; PAIS (Partial androgen insensitivity syndrome) (AR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Androjen Duyarsızlığı, Parsiyel, ± Meme Kanseri, 312300, X'e Bağlı Resesif; PAIS (Parsiyel Androjen Insensitivite Sendromu) (Prenatal) (MLPA)	Androgen insensitivity, partial, with or without breast cancer, 312300, X-linked recessive; PAIS (Partial androgen insensitivity syndrome) (Prenatal) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Konjenital Diseritropoietik, Tip 2; Cdan2 (Konjenital Diseritropoetik Anemi Tip 2) (SEC23B geni) (Tüm Gen Dizi Analizi) (Postnatal)	ANEMIA, CONGENITAL DYSERYTHROPOIETIC, TYPE II; CDAN2 (Congenital dyserythropoietic anemia type II) (SEC23B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC23B	SEC23B, CDAN2, HEMPAS, CWS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, congenital dyserythropoietic, 2 (CDAN2) [MIM:224100]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, normocytic anemia, iron overload, jaundice, and variable splenomegaly. Ultrastructural features include bi- or multinucleated erythroblasts in bone marrow, karyorrhexis, and the presence of Gaucher-like bone marrow histiocytes. The main biochemical feature of the disease is defective glycosylation of some red blood cells membrane proteins. {ECO:0000269|PubMed:19561605, ECO:0000269|PubMed:19621418}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Anemi, Konjenital Diseritropoietik, Tip 2; Cdan2 (Konjenital Diseritropoetik Anemi Tip 2) (SEC23B geni) (Tüm Gen Dizi Analizi) (Prenatal)	ANEMIA, CONGENITAL DYSERYTHROPOIETIC, TYPE II; CDAN2 (Congenital dyserythropoietic anemia type II) (SEC23B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEC23B	SEC23B, CDAN2, HEMPAS, CWS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, congenital dyserythropoietic, 2 (CDAN2) [MIM:224100]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, normocytic anemia, iron overload, jaundice, and variable splenomegaly. Ultrastructural features include bi- or multinucleated erythroblasts in bone marrow, karyorrhexis, and the presence of Gaucher-like bone marrow histiocytes. The main biochemical feature of the disease is defective glycosylation of some red blood cells membrane proteins. {ECO:0000269|PubMed:19561605, ECO:0000269|PubMed:19621418}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Hemolitik, UMPH1 Eksikliğine Bağlı, 266120, Otozomal Resesif (Hemolitik Anemi, Primidine 5' Nükleotidaz Eksikliğine Bağlı) (NT5C3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, hemolytic, due to UMPH1 deficiency, 266120, Autosomal recessive (Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency) (NT5C3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NT5C3A	NT5C3A, NT5C3, UMPH1, PSN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: PSN deficiency (PSND) [MIM:266120]: Autosomal recessive condition causing hemolytic anemia characterized by marked basophilic stippling and the accumulation of high concentrations of pyrimidine nucleotides within the erythrocyte. It is implicated in the anemia of lead poisoning and is possibly associated with learning difficulties. {ECO:0000269|PubMed:11369620, ECO:0000269|PubMed:12930399, ECO:0000269|PubMed:15238149, ECO:0000269|PubMed:15604219, ECO:0000269|PubMed:15988458, ECO:0000269|PubMed:16461318, ECO:0000269|PubMed:18499901, ECO:0000269|PubMed:25153905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Anemi, Hemolitik Rh Null, Regülatör Tipi, 268150, Otozomal Resesif (Rh Eksikliği Sendromu) (RHAG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, hemolytic, Rh-null, regulator type, 268150, Autosomal recessive (Rh deficiency syndrome) (RHAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHAG	RHAG, RH50A, OHST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Anemi, Hipokrom Mikrositer, Aşırı Demir Yüklenmesi 1, 206100, Otozomal Resesif; AHMIO1 (Mikrositik Anemi-Karaciğer Demir Yüklenmesi) (SLC11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, hypochromic microcytic, with iron overload 1, 206100, Autosomal recessive; AHMIO1 (Microcytic anemia with liver iron overload) (SLC11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC11A2	SLC11A2, NRAMP2, DCT1, DMT1, AHMIO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, hypochromic microcytic, with iron overload 1 (AHMIO1) [MIM:206100]: A hematologic disease characterized by abnormal hemoglobin content in the erythrocytes which are reduced in size. The disorder is due to an error of iron metabolism that results in high serum iron, massive hepatic iron deposition, and absence of sideroblasts and stainable bone marrow iron store. Despite adequate transferrin-iron complex, delivery of iron to the erythroid bone marrow is apparently insufficient for the demands of hemoglobin synthesis. {ECO:0000269|PubMed:15459009, ECO:0000269|PubMed:16160008, ECO:0000269|PubMed:16439678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Anemi, Hipokrom Mikrositer, Aşın Demir Yükü 2, 615234, Otozomal Dominant; AHMIO2 (Ağır Konjenital Hipokrom Anemi-Halka Sekilinde Sideroblastlar) (STEAP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, hypochromic microcytic, with iron overload 2, 615234, Autosomal dominant; AHMIO2 (Severe congenital hypochromic anemia with ringed sideroblasts) (STEAP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STEAP3	STEAP3, TSAP6, AHMIO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, hypochromic microcytic, with iron overload 2 (AHMIO2) [MIM:615234]: A hematologic disease characterized by abnormal hemoglobin content in the erythrocytes which are reduced in size, severe anemia, erythropoietic hyperplasia of bone marrow, massive hepatic iron deposition, and hepatosplenomegaly. (ECO:0000269|PubMed:22031663). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Anemi, Neonatal Hemolitik, Fatal Veya Yüksek Riskli (Kalıtsal Sferositoz) (SPTB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, neonatal hemolytic, fatal and near-fatal (Hereditary spherocytosis) (SPTB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTB	SPTB, SPH2, EL3, HS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anemi, Neonatal Hemolitik, Fatal Veya Yüksek Riskli (Kalıtsal Sferositoz) (SPTB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, neonatal hemolytic, fatal and near-fatal (Hereditary spherocytosis) (SPTB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPTB	SPTB, SPH2, EL3, HS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Sideroblastik, 1, 300751, X'e Bağlı; SIDBA1 (X'e Bağlı Sideroblastik Anemi) (ALAS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, sideroblastic, 1, 300751, X-linked recessive; SIDBA1 (X-linked sideroblastic anemia) (ALAS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALAS2	ALAS2, ANH1, ASB, XLEPP, XLSA, ANH1, SIDBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anemi, Sideroblastik, 1, 300751, X'e Bağlı; SIDBA1 (X'e Bağlı Sideroblastik Anemi) (ALAS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, sideroblastic, 1, 300751, X-linked recessive; SIDBA1 (X-linked sideroblastic anemia) (ALAS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALAS2	ALAS2, ANH1, ASB, XLEPP, XLSA, ANH1, SIDBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Sideroblastik, 2, Piridoksin Dirençli, 205950, Otozomal Resesif; SIDBA2 (Otozomal Resesif Sideroblastik Anemi) (SLC25A38 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950, Autosomal recessive; SIDBA2 (Autosomal recessive sideroblastic anemia) (SLC25A38 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A38	SLC25A38, SIDBA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anemi, Sideroblastik, 2, Piridoksin Dirençli, 205950, Otozomal Resesif; SIDBA2 (Otozomal Resesif Sideroblastik Anemi) (SLC25A38 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, sideroblastic, 2, pyridoxine-refractory, 205950, Autosomal recessive; SIDBA2 (Autosomal recessive sideroblastic anemia) (SLC25A38 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A38	SLC25A38, SIDBA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Sideroblastik, 3, Piridoksin Dirençli, 616860, Otozomal Resesif; SIDBA3 (Yetişkin Başlangıçlı Otozomal Resesif Sideroblastik Anemi) (GLRX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860, Autosomal recessive; SIDBA3 (Adult-onset autosomal recessive sideroblastic anemia) (GLRX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLRX5	GLRX5, C14orf87, PRO1238, FLB4739, PRSA, SIDBA3, SPAH2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive (PRARSA) [MIM:205950]: A form of sideroblastic anemia not responsive to pyridoxine. Sideroblastic anemia is characterized by anemia of varying severity, hypochromic peripheral erythrocytes, systemic iron overload secondary to chronic ineffective erythropoiesis, and the presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-loaded mitochondria clustered around the nucleus. (ECO:0000269|PubMed:17485548, ECO:0000269|PubMed:20364084, ECO:0000269|PubMed:25342667, ECO:0000269|PubMed:26100117). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Anemi, Sideroblastik, 3, Piridoksin Dirençli, 616860, Otozomal Resesif; SIDBA3 (Yetişkin Başlangıçlı Otozomal Resesif Sideroblastik Anemi) (GLRX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, sideroblastic, 3, pyridoxine-refractory, 616860, Autosomal recessive; SIDBA3 (Adult-onset autosomal recessive sideroblastic anemia) (GLRX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLRX5	GLRX5, C14orf87, PRO1238, FLB4739, PRSA, SIDBA3, SPAH2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive (PRARSA) [MIM:205950]: A form of sideroblastic anemia not responsive to pyridoxine. Sideroblastic anemia is characterized by anemia of varying severity, hypochromic peripheral erythrocytes, systemic iron overload secondary to chronic ineffective erythropoiesis, and the presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-loaded mitochondria clustered around the nucleus. (ECO:0000269|PubMed:17485548, ECO:0000269|PubMed:20364084, ECO:0000269|PubMed:25342667, ECO:0000269|PubMed:26100117). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Sideroblastik, 4, 182170, Otozomal Dominant; SIDBA4 (Otozomal Resesif Sideroblastik Anemi) (HSPA9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, sideroblastic, 4, 182170, Autosomal dominant; SIDBA4 (Autosomal recessive sideroblastic anemia) (HSPA9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPA9	HSPA9, HSPA9B, MOT2, GRP75, EVPLS, SIDBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anemi, Sideroblastik, 4, 182170, Otozomal Dominant; SIDBA4 (Otozomal Resesif Sideroblastik Anemi) (HSPA9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, sideroblastic, 4, 182170, Autosomal dominant; SIDBA4 (Autosomal recessive sideroblastic anemia) (HSPA9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPA9	HSPA9, HSPA9B, MOT2, GRP75, EVPLS, SIDBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, Sideroblastik, Ataksi İle, 301310, X'e Bağlı (X'e Bağlı Sideroblastik Anemi ve Ataksi) (ABCB7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, sideroblastic, with ataxia, 301310, X-linked recessive (X-linked sideroblastic anemia and ataxia) (ABCB7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB7	ABCB7, ABC7, ASAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, sideroblastic, spinocerebellar ataxia (ASAT) [MIM:301310]: A X-linked recessive disorder characterized by an infantile to early childhood onset of non-progressive cerebellar ataxia and mild anemia, with hypochromia and microcytosis. (ECO:0000269|PubMed:10196363, ECO:0000269|PubMed:11050011, ECO:0000269|PubMed:11843825, ECO:0000269|PubMed:22398176). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Anemi, Sideroblastik, Ataksi İle, 301310, X'e Bağlı (X'e Bağlı Sideroblastik Anemi ve Ataksi) (ABCB7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, sideroblastic, with ataxia, 301310, X-linked recessive (X-linked sideroblastic anemia and ataxia) (ABCB7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCB7	ABCB7, ABC7, ASAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, sideroblastic, spinocerebellar ataxia (ASAT) [MIM:301310]: A X-linked recessive disorder characterized by an infantile to early childhood onset of non-progressive cerebellar ataxia and mild anemia, with hypochromia and microcytosis. (ECO:0000269|PubMed:10196363, ECO:0000269|PubMed:11050011, ECO:0000269|PubMed:11843825, ECO:0000269|PubMed:22398176). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anemi, X'e Bağlı, ± Nötropeni ve / Veya Trombosit Anormallikleri,300835, X'bağlı Resesif; XLANP (X'e Bağlı Diseritropoetik Anemi-Anormal Trombositler ve Nötropeni) (GATA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835, X-linked recessive; XLANP (X-linked dyserythropoietic anemia with abnormal platelets and neutropenia) (GATA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA1	GATA1, GF1, ERYF1, NFE1, XLTDA, XLTT, XLANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked dyserythropoietic anemia and thrombocytopenia (XDAT) [MIM:300367]: Disorder characterized by erythrocytes with abnormal size and shape, and paucity of platelets in peripheral blood. The bone marrow contains abundant and abnormally small megakaryocytes. (ECO:0000269|PubMed:10700180, ECO:0000269|PubMed:11418466, ECO:0000269|PubMed:11675338, ECO:0000269|PubMed:11809723). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Anemi, X'e Bağlı, ± Nötropeni ve / Veya Trombosit Anormallikleri,300835, X'bağlı Resesif; XLANP (X'e Bağlı Diseritropoetik Anemi-Anormal Trombositler ve Nötropeni) (GATA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anemia, X-linked, with/without neutropenia and/or platelet abnormalities, 300835, X-linked recessive; XLANP (X-linked dyserythropoietic anemia with abnormal platelets and neutropenia) (GATA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA1	GATA1, GF1, ERYF1, NFE1, XLTDA, XLTT, XLANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: X-linked dyserythropoietic anemia and thrombocytopenia (XDAT) [MIM:300367]: Disorder characterized by erythrocytes with abnormal size and shape, and paucity of platelets in peripheral blood. The bone marrow contains abundant and abnormally small megakaryocytes. (ECO:0000269|PubMed:10700180, ECO:0000269|PubMed:11418466, ECO:0000269|PubMed:11675338, ECO:0000269|PubMed:11809723). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Angelman sendromu (UBE3A, MTHFR, GABRB3) (MLPA)	Angelman syndrome (UBE3A, MTHFR, GABRB3) (MLPA)	UBE3A, MTHFR, GABRB3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Angelman sendromu (UBE3A, MTHFR, GABRB3) (MLPA) (Prenatal)	Angelman syndrome (UBE3A, MTHFR, GABRB3) (MLPA) (Prenatal)	UBE3A, MTHFR, GABRB3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Angelman Sendromu, 105830, Sporadik Vakalar; AS (Angelman Sendromu) (UBE3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angelman syndrome, 105830, Isolated cases; AS (Angelman syndrome) (UBE3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBE3A	UBE3A, ANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. (ECO:0000269|PubMed:25212744, ECO:0000269|PubMed:9585605). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Angelman Sendromu, 105830, Sporadik Vakalar; AS (Angelman Sendromu) (UBE3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Angelman syndrome, 105830, Isolated cases; AS (Angelman syndrome) (UBE3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBE3A	UBE3A, ANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. (ECO:0000269|PubMed:25212744, ECO:0000269|PubMed:9585605). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ACE İnhibitörleri İle İndüklenen Anjiyoödem, Yatkınlık, 300909; AEACEI (Akkz Anjiyoödem) (XPNPEP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angioedema induced by ACE inhibitors, susceptibility to, 300909; AEACEI (Acquired angioedema) (XPNPEP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XPNPEP2	XPNPEP2, AEACEI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angioedema induced by ACE inhibitors (AEACEI) [MIM:300909]: A potentially life-threatening side effect of ACE inhibitors that appears in a subset of patients taking these drugs for hypertension and cardiovascular disease treatment. AEACEI is characterized by swelling of the face, lips, tongue, and airway that can lead to suffocation and death if severe. (ECO:0000269|PubMed:16175507, ECO:0000269|PubMed:20625347, ECO:0000269|PubMed:21898657). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Anjiyoödem, Kalıtsal, Tip 3, 610618, Otozomal Dominant; HAE3 (Kalıtsal Anjiyoödemi) (F12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angioedema, hereditary, type III, 610618, Autosomal dominant; HAE3 (Hereditary angioedema) (F12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F12	F12, HAF, HAE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anjiyoödem, Kalıtsal, Tip 3, 610618, Otozomal Dominant; HAE3 (Kalıtsal Anjiyoödemi) (MLPA)	Angioedema, hereditary, type III, 610618, Autosomal dominant; HAE3 (Hereditary angioedema) (MLPA)	F12	F12, HAF, HAE3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Factor XII deficiency (FA12D) [MIM:234000]: An asymptomatic anomaly of in vitro blood coagulation. Its diagnosis is based on finding a low plasma activity of the factor in coagulating assays. It is usually only accidentally discovered through pre-operative blood tests. Factor XII deficiency is divided into two categories, a cross-reacting material (CRM)-negative group (negative F12 antigen detection) and a CRM-positive group (positive F12 antigen detection). (ECO:0000269|PubMed:10361128, ECO:0000269|PubMed:11726307, ECO:0000269|PubMed:15205584, ECO:0000269|PubMed:15617741, ECO:0000269|PubMed:2510163, ECO:0000269|PubMed:2882793, ECO:0000269|PubMed:8049433, ECO:0000269|PubMed:8528215, ECO:0000269|PubMed:9354665). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Anjiyoödem, Kalıtsal, Tip 1 ve 2, 106100, Otozomal Dominant, HAE1 (Kalıtsal Anjiyoödem) (SERPING1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angioedema, hereditary, types I and II, 106100, Autosomal dominant; HAE1 (Hereditary angioedema) (SERPING1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPING1	C1NH, HAE1, HAE2, SERPING1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary angioedema (HAE) [MIM:106100]: An autosomal dominant disorder characterized by episodic local swelling involving subcutaneous or submucous tissue of the upper respiratory and gastrointestinal tracts, face, extremities, and genitalia. Hereditary angioedema due to C1 esterase inhibitor deficiency is comprised of two clinically indistinguishable forms. In hereditary angioedema type 1, serum levels of C1 esterase inhibitor are decreased, while in type 2, the levels are normal or elevated, but the protein is non-functional. {ECO:0000269|PubMed:12773530, ECO:0000269|PubMed:1363816, ECO:0000269|PubMed:1451784, ECO:0000269|PubMed:14635117, ECO:0000269|PubMed:16409206, ECO:0000269|PubMed:2118657, ECO:0000269|PubMed:2296585, ECO:0000269|PubMed:22994404, ECO:0000269|PubMed:2365061, ECO:0000269|PubMed:24456027, ECO:0000269|PubMed:3178731, ECO:0000269|PubMed:7814636, ECO:0000269|PubMed:7883978, ECO:0000269|PubMed:8172583, ECO:0000269|PubMed:8529136, ECO:0000269|PubMed:8755917, ECO:0000269|Ref.41}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Anjiyoödem, Kalıtsal, Tip 1 ve 2, 106100, Otozomal Dominant, HAE1 (Kalıtsal Anjiyoödem) (MLPA)	Angioedema, hereditary, types I and II, 106100, Autosomal dominant; HAE1 (Hereditary angioedema) (MLPA)	SERPING1	C1NH, HAE1, HAE2, SERPING1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary angioedema (HAE) [MIM:106100]: An autosomal dominant disorder characterized by episodic local swelling involving subcutaneous or submucous tissue of the upper respiratory and gastrointestinal tracts, face, extremities, and genitalia. Hereditary angioedema due to C1 esterase inhibitor deficiency is comprised of two clinically indistinguishable forms. In hereditary angioedema type 1, serum levels of C1 esterase inhibitor are decreased, while in type 2, the levels are normal or elevated, but the protein is non-functional. {ECO:0000269|PubMed:12773530, ECO:0000269|PubMed:1363816, ECO:0000269|PubMed:1451784, ECO:0000269|PubMed:14635117, ECO:0000269|PubMed:16409206, ECO:0000269|PubMed:2118657, ECO:0000269|PubMed:2296585, ECO:0000269|PubMed:22994404, ECO:0000269|PubMed:2365061, ECO:0000269|PubMed:24456027, ECO:0000269|PubMed:3178731, ECO:0000269|PubMed:7814636, ECO:0000269|PubMed:7883978, ECO:0000269|PubMed:8172583, ECO:0000269|PubMed:8529136, ECO:0000269|PubMed:8755917, ECO:0000269|Ref.41}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Anjiyofibroma, Somatik (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angiofibroma, somatic (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Anjiyopati, Kalıtsal, Nefropati, Anevrizmalar ve Kas Krampları, 611773, Otozomal Dominant; HANAC (Otozomal Dominant Ailesel Hematüri-Retina Arterioler Tortiosite-Kontraktür Sendromu) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Angiopathy, hereditary, with nephropathy, aneurysms, and muscle cramps, 611773, Autosomal dominant; HANAC (Autosomal dominant familial hematuria-retinal arteriolar tortuosity-contractures syndrome) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porencephaly 1 (POREN1) [MIM:175780]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres, neurologic manifestations, facial paresis, and visual defects. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 1 is usually unilateral and results from focal destructive lesions such as fetal vascular occlusion or birth trauma. {ECO:0000269|PubMed:15905400, ECO:0000269|PubMed:16107487, ECO:0000269|PubMed:19194877}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Anhidroz, İzole, Normal Ter Bezleri İle, 106190, Otozomal Resesif; ANHD (İzole Jeneralize Anhidrozis-Normal Ter Bezleri) (ITPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anhidrosis, isolated, with normal sweat glands, 106190, Autosomal recessive; ANHD (Isolated generalized anhidrosis with normal sweat glands) (ITPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR2	ITPR2, ANHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anhidrosis, isolated, with normal sweat glands (ANHD) [MIM:106100]: An autosomal recessive disorder characterized by generalized, isolated anhidrosis, severe heat intolerance, and morphologically normal eccrine sweat glands. Body growth, teeth, hair, nails, and skin are normal. {ECO:0000269|PubMed:25329695}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aniridi 2, 617141, Otozomal Dominant; AN2 (İzole Aniridi) (ELP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aniridia 2, 617141, Autosomal dominant; AN2 (Isolated aniridia) (ELP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELP4	ELP4, PAX6NEB, AN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aniridi 3, 617142, Otozomal Dominant; AN3 (İzole Aniridi) (TRIM44 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aniridia 3, 617142, Autosomal dominant; AN3 (Isolated aniridia) (TRIM44 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIM44	TRIM44, AN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Aniridi, 106210, Otozomal Dominant (İzole Aniridi) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aniridia, 106210, Autosomal dominant (isolated aniridia) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9858761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aniridi, 106210, Otozomal Dominant (İzole Aniridi) (MLPA)	Aniridia, 106210, Autosomal dominant (isolated aniridia) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9858761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ankiloblefaron-Ektodermal Defektler-Yarık Dudak / Palat (Ankiloblefaron-Ektodermal Defekt-Yarık Dudak / Damak Sendromu (Hay-Wells Sendromu)) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ANKYLOBLEPHARON-ECTODERMAL DEFECTS-CLEFT LIP/PALATE (Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anonişia Konjenita, 206800, Otozomal Resesif; NDNC4 (İzole Konjenital Anonişi) (RSPO4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anonychia congenita, 206800, Autosomal recessive; NDNC4 (Isolated congenital anonychia) (RSPO4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPO4	RSPO4, CRISTIN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nail disorder, non-syndromic congenital, 4 (NDNC4) [MIM:206800]: A nail disorder characterized by congenital anonychia or its milder phenotypic variant hyponychia. Anonychia/hyponychia is the absence or severe hypoplasia of all fingernails and toenails without significant bone anomalies. {ECO:0000269|PubMed:17041604}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Anoreksiya Nervoza, Yatkınlık, 606788 (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anorexia nervosa, susceptibility to, 606788 (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Anoreksiya Nervosa, Yatkınlık, 610269 (BDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anorexia nervosa, susceptibility to, 610269 (BDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Segment Disgenezileri 8, 617319, Otozomal Resesif (Ailesel Okküler Ön Segment Mezenkimal Disgenezisi) (CPAMD8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 8, 617319, Autosomal recessive (Familial ocular anterior segment mesenchymal dysgenesis) (CPAMD8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPAMD8	CPAMD8, KIAA1283, ASGD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Segment Anomalileri, ± Katarakt, 602588, Otozomal Dominant (Brankio-Otik Sendrom) (EYA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment anomalies with or without cataract, 602588, Autosomal dominant (Branchio-otic syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Segment Anomalileri, ± Katarakt, 602588, Otozomal Dominant (Brankio-Otik Sendrom) (EYA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anterior segment anomalies with or without cataract, 602588, Autosomal dominant (Branchio-otic syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ön Segment Disgenezisi 1, Multipl Alt Tipler, 107250, Otozomal Dominant; ASGD1 (Ailesel Okküler Ön Segment Mezenkimal Disgenezisi) (PITX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 1, multiple subtypes, 107250, Autosomal dominant; ASGD1 (Familial ocular anterior segment mesenchymal dysgenesis) (PITX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX3	PITX3, CTPP4, CTRCT11, ASGD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Ön Segment Disgenezisi 2, Multipl Alt Tipler, 610256, Otozomal Resesif; ASGD2 (Konjenital Primer Atalci) (FOXE3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 2, multiple subtypes, 610256, Autosomal recessive; ASGD2 (Congenital primary aphakia) (FOXE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXE3	FOXE3, FKHL12, ASMD, CTRCT34, ASGD2, AATL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anterior segment mesenchymal dysgenesis (ASMD) [MIM:107250]: A range of developmental defects in structures at the front of the eye, resulting from abnormal migration or differentiation of the neural crest derived mesenchymal cells that give rise to the cornea, iris, and other components of the anterior chamber during eye development. Different mature anterior segment anomalies may exist alone or in combination, and are associated with an increased risk of glaucoma and corneal opacity. Conditions falling within the phenotypic spectrum of anterior segment anomalies include aniridia, posterior embryotoxon, Axenfeld anomaly, Reiger anomaly/syndrome, Peters anomaly, and iridogoniodysgenesis. {ECO:0000269|PubMed:11159941, ECO:0000269|PubMed:11980846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ön Segment Disgenezisi 3, Multipl Alt Tipler, 601631, Otozomal Dominant; ASGD3 (Rieger Anomalisi) (FOXC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 3, multiple subtypes, 601631, Autosomal dominant; ASGD3 (Rieger anomaly) (FOXC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iridogoniodysgenesis anomaly (IGDA) [MIM:601631]: Autosomal dominant phenotype characterized by iris hypoplasia, goniodysgenesis, and juvenile glaucoma. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ön Segment Disgenezisi 3, Multipl Alt Tipler, 601631, Otozomal Dominant; ASGD3 (Rieger Anomalisi) (MLPA)	Anterior segment dysgenesis 3, multiple subtypes, 601631, Autosomal dominant; ASGD3 (Rieger anomaly) (MLPA)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Iridogoniodysgenesis anomaly (IGDA) [MIM:601631]: Autosomal dominant phenotype characterized by iris hypoplasia, goniodysgenesis, and juvenile glaucoma. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ön Segment Disgenezisi 4, 137600, Otozomal Dominant; ASGD4 (Rieger Anomalisi) (PITX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 4, 137600, Autosomal dominant; ASGD4 (Rieger anomaly) (PITX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iridogoniodysgenesis 2 (IRID2) [MIM:137600]: Autosomal dominant inherited disease. {ECO:0000269|PubMed:9437321, ECO:0000269|PubMed:9618168}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ön Segment Disgenezisi 4, 137600, Otozomal Dominant; ASGD4 (Rieger Anomalisi) (MLPA)	Anterior segment dysgenesis 4, 137600, Autosomal dominant; ASGD4 (Rieger anomaly) (MLPA)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Iridogoniodysgenesis 2 (IRID2) [MIM:137600]: Autosomal dominant inherited disease. {ECO:0000269|PubMed:9437321, ECO:0000269|PubMed:9618168}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ön Segment Disgenezileri 5, Multipl Alt Tipler, 604229; ASGD5 (Peters Anomalisi) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 5, multiple subtypes, 604229; ASGD5 (Peters anomaly) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Segment Disgenezileri 5, Multipl Alt Tipler, 604229; ASGD5 (Peters Anomalisi) (MLPA)	Anterior segment dysgenesis 5, multiple subtypes, 604229; ASGD5 (Peters anomaly) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ön Segment Disgenezileri 5, Multipl Alt Tipler, 604229; ASGD5 (Peters Anomalisi) (PAX6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Anterior segment dysgenesis 5, multiple subtypes, 604229; ASGD5 (Peters anomaly) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ön Segment Disgenezileri 5, Multipl Alt Tipler, 604229; ASGD5 (Peters Anomalisi) (Prenatal) (MLPA)	Anterior segment dysgenesis 5, multiple subtypes, 604229; ASGD5 (Peters anomaly) (Prenatal) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ön Segment Disgenezisi 6, Çok Sayıda Alt-Tipleri, 617315; ASGD6 (Ailesel Oküler Ön Segment Mezenkimal Disgenezisi) (CYP1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 6, multiple subtypes, 617315; ASGD6 (Familial ocular anterior segment mesenchymal dysgenesis) (CYP1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP1B1	CYP1B1, GLC3A, ASGD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Segment Disgenezisi 6, Çok Sayıda Alt-Tipleri, 617315; ASGD6 (Ailesel Oküler Ön Segment Mezenkimal Disgenezisi) (MLPA)	Anterior segment dysgenesis 6, multiple subtypes, 617315; ASGD6 (Familial ocular anterior segment mesenchymal dysgenesis) (MLPA)	CYP1B1	CYP1B1, GLC3A, ASGD6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ön Segment Disgenezisi 7, Sklerokornea 269400, Otozomal Resesif; ASGD7 (Konjenital Katarakt Mikrokornea-Korneal Opasite) (PXDN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anterior segment dysgenesis 7, with sclerocornea, 269400, Autosomal recessive; ASGD7 (Congenital cataract microcornea with corneal opacity) (PXDN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PXDN	PXDN, D2S448E, KIAA0230, PRG2, PXN, COPOA, ASGD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal opacification with other ocular anomalies (COPOA) [MIM:269400]: An ocular disease characterized by sclerocornea associated with other ocular anomalies, such as cataract, microcornea, microphthalmia, and anterior segment dysgenesis. Sclerocornea is a primary anomaly in which sclerialization of a peripheral part of the cornea, or the entire corneal tissue, occurs. In the peripheral type of sclerocornea, the affected area is vascularized with regular arcades of superficial scleral vessels. In total sclerocornea, the entire cornea is opaque and vascularized. {ECO:0000269	PubMed:21907015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Antisosyal Davranış, 300615, X'e Bağlı Resesif (Monoamin Oksidaz A Eksikliği (Brunner Sendromu)) (MAOA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Antisocial behavior, 300615, X-linked recessive (Monoamine oxidase A deficiency) (MAOA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAOA	MAOA, BRNRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brunner syndrome (BRUNS) [MIM:300615]: A form of X-linked non-dysmorphic mild mental retardation. Male patients are affected by borderline mental retardation and exhibit abnormal behavior, including disturbed regulation of impulsive aggression. Obligate female carriers have normal intelligence and behavior. {ECO:0000269	PubMed:8211186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Antley-Bixler, Genital Anomaliler ve Dizensiz Steroidogenez İle Sendromu, 201750, Otozomal Resesif; ABS1 (Antley-Bixler Sendromu-Genital Anomali ve Steroidojenez Bozukluğu) (POR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750, Autosomal recessive; ABS1 (Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis) (POR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POR	POR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. {ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Antley-Bixler, Genital Anomaliler ve Dizensiz Steroidogenez İle Sendromu, 201750, Otozomal Resesif; ABS1 (Antley-Bixler Sendromu-Genital Anomali ve Steroidojenez Bozukluğu) (MLPA)	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750, Autosomal recessive; ABS1 (Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis) (MLPA)	POR	POR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. {ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Antley-Bixler, Genital Anomaliler ve Dizensiz Steroidogenez İle Sendromu, 201750, Otozomal Resesif; ABS1 (Antley-Bixler Sendromu-Genital Anomali ve Steroidojenez Bozukluğu) (POR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750, Autosomal recessive; ABS1 (Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis) (POR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POR	POR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. {ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Antley-Bixler, Genital Anomaliler ve Dizensiz Steroidogenez İle Sendromu, 201750, Otozomal Resesif; ABS1 (Antley-Bixler Sendromu-Genital Anomali ve Steroidojenez Bozukluğu) (Prenatal) (MLPA)	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750, Autosomal recessive; ABS1 (Antley-Bixler syndrome with genital anomaly and disorder of steroidogenesis) (Prenatal) (MLPA)	POR	POR	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. {ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Antley-Bixler Sendromu, Genital Anomali Veya Dizensiz Steroidogenez Olmadan, 207410, Otozomal Resesif; ABS2 (Antley-Bixler Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410, Autosomal recessive; ABS2 (Antley-Bixler syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Antley-Bixler Sendromu, Genital Anomali Veya Dizensiz Steroidogenez Olmadan, 207410, Otozomal Resesif; ABS2 (Antley-Bixler Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410, Autosomal recessive; ABS2 (Antley-Bixler syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Anksiyete İle İlgili Kişilik Özellikleri, 607834 (SLC6A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Anxiety-related personality traits, 607834 (SLC6A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A4	SLC6A4, HTT, OCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 10, 617168, Otozomal Dominant (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (LOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 10, 617168, Autosomal dominant (Familial thoracic aortic aneurysm and aortic dissection) (LOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LOX	LOX, AAT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 11, Yatkınlık, 617349, Otozomal Dominant (Konjenital Primer Atalci) (FOXE3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 11, susceptibility to, 617349, Autosomal dominant (Congenital primary aphakia) (FOXE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXE3	FOXE3, FKHL12, ASMD, CTRCT34, ASGD2, AATL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 4, 132900, Otozomal Dominant; AAT4 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (MYH11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 4, 132900, Autosomal dominant; AAT4 (Familial thoracic aortic aneurysm and aortic dissection) (MYH11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH11	MYH11, AAT4, FAA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 4 (AAT4) [MIM:132900]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269	PubMed:16444274}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 6, 611788, Otozomal Dominant; AAT6 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (ACTA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 6, 611788, Autosomal dominant; AAT6 (Familial thoracic aortic aneurysm and aortic dissection) (ACTA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA2	ACTA2, ACTSA, AAT6, MYMY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 6 (AAT6) [MIM:611788]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269|PubMed:17904018, ECO:0000269|PubMed:19409525, ECO:0000269|PubMed:19639654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Aort Anevrizması, Ailesel Torakal 7, 613780, Otozomal Dominant; AAT7 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (MYLK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 7, 613780, Autosomal dominant; AAT7 (Familial thoracic aortic aneurysm and aortic dissection) (MYLK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYLK	MYLK, MLCK, AAT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 7 (AAT7) [MIM:613780]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269	PubMed:21055718}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 8, 615436, Otozomal Dominant (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (PRKG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 8, 615436, Autosomal dominant; AAT8 (Familial thoracic aortic aneurysm and aortic dissection) (PRKG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKG1	PRKG1, PRKG1B, PRKGR1B, AAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 8 (AAT8) [MIM:615436]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269	PubMed:23910461}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Aort Anevrizması, Ailesel Torakal 9, 616166, Otozomal Dominant; AAT9 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (MFAP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic aneurysm, familial thoracic 9, 616166, Autosomal dominant; AAT9 (Familial thoracic aortic aneurysm and aortic dissection) (MFAP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFAP5	MFAP5, MAGP2, AAT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 9 (AAT9) [MIM:616166]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269	PubMed:25434006}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Aort Kapak Hastalığı 1, 109730, Otozomal Dominant; AOVD1 (Ailesel Bikispit Aort Kapağı) (NOTCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic valve disease 1, 109730, Autosomal dominant; AOVD1 (Familial bicuspid aortic valve) (NOTCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH1	NOTCH1, TAN1, AOSS, AOVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic valve disease 1 (AOVD1) [MIM:109730]: A common defect in the aortic valve in which two rather than three leaflets are present. It is often associated with aortic valve calcification, stenosis and insufficiency. In extreme cases, the blood flow may be so restricted that the left ventricle fails to grow, resulting in hypoplastic left heart syndrome. {ECO:0000269	PubMed:16025100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aort Kapak Hastalığı 2, 614823, Otozomal Dominant; AOVD2 (Ailesel Bikispit Aort Kapağı) (SMAD6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aortic valve disease 2, 614823, Autosomal dominant; AOVD2 (Familial bicuspid aortic valve) (SMAD6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD6	SMAD6, MADH6, AOVD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic valve disease 2 (AOVD2) [MIM:614823]: A common defect in the aortic valve in which two rather than three leaflets are present. It is often associated with aortic valve calcification, stenosis and insufficiency. In extreme cases, the blood flow may be so restricted that the left ventricle fails to grow, resulting in hypoplastic left heart syndrome. {ECO:0000269|PubMed:22275001}. Note=The disease is caused by mutations affecting the gene represented in this entry. SMAD6 variants may contribute to increased risk of congenital cardiovascular malformations (CVM). CVM is a major cause of mortality and morbidity in childhood. In most sporadic cases that cannot be attributed to particular malformation syndromes or teratogenic exposures, there remains a substantial excess familial risk, indicating a significant genetic contribution to disease susceptibility (PubMed:22275001). {ECO:0000269|PubMed:22275001};.	EDTA Blood Sample
Apert Sendromu, 101200, Otozomal Dominant (Apert Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Apert syndrome, 101200, Autosomal dominant (Apert syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781877, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Apert Sendromu, 101200, Otozomal Dominant (Apert Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Apert syndrome, 101200, Autosomal dominant (Apert syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781877, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atalarlı, Parsiyel-Sindaktili ve Metatars Duplikasyonu 4 (Atalarlı-Sindaktili-Mikrosefali Sendromu) (2200)	APHALANGIA, PARTIAL, WITH SYNDACTYLY AND DUPLICATION OF METATARSAL IV (Aphalangy-syndactyly-microcephaly syndrome) (440)	.		Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.		EDTA Blood Sample and Heparinized blood sample
Atalarlı, Parsiyel-Sindaktili ve Metatars Duplikasyonu 4 (Atalarlı-Sindaktili-Mikrosefali Sendromu) (Prenatal)	APHALANGIA, PARTIAL, WITH SYNDACTYLY AND DUPLICATION OF METATARSAL IV (Aphalangy-syndactyly-microcephaly syndrome) (Prenatal)	.		Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Afazi, Primer Progresif, 607485, Otozomal Dominant (Progresif Akıcı Olmayan Afazi) (GRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aphasia, primary progressive, 607485, Autosomal dominant (Progressive non-fluent aphasia) (GRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRN	GRN, CLN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ubiquitin-positive frontotemporal dementia (UP-FTD) [MIM:607485]: Frontotemporal dementia (FTD) is the second most common cause of dementia in people under the age of 65 years. It is an autosomal dominant neurodegenerative disease. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Afazi, Primer Progresif, 607485, Otozomal Dominant (Progresif Akıcı Olmayan Afazi) (GRN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aphasia, primary progressive, 607485, Autosomal dominant (Progressive non-fluent aphasia) (GRN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRN	GRN, CLN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ubiquitin-positive frontotemporal dementia (UP-FTD) [MIM:607485]: Frontotemporal dementia (FTD) is the second most common cause of dementia in people under the age of 65 years. It is an autosomal dominant neurodegenerative disease. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplasia Cutis Konjenita, Sendromik Olmayan, 107600, Otozomal Resesif, Otozomal Dominant; ACC (Aplazia Kutis Konjenita) (BMS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplasia cutis congenita, nonsyndromic, 107600, Autosomal recessive, Autosomal dominant; ACC (Aplasia cutis congenita) (BMS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMS1	BMS1, BMS1L, KIAA0187, ACC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aplasia cutis congenita, non-syndromic (ACC) [MIM:107600]: A disorder characterized by congenital absence of a portion of skin in a localized or widespread area of the body. The lesions are most commonly localized on the scalp, however aplasia cutis congenita can affect any part of the body. {ECO:0000269	PubMed:23785305}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Aplasia Cutis Konjenita, Sendromik Olmayan, 107600, Otozomal Resesif, Otozomal Dominant; ACC (Aplazia Kutis Konjenita) (BMS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplasia cutis congenita, nonsyndromic, 107600, Autosomal recessive, Autosomal dominant; ACC (Aplasia cutis congenita) (BMS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMS1	BMS1, BMS1L, KIAA0187, ACC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aplasia cutis congenita, non-syndromic (ACC) [MIM:107600]: A disorder characterized by congenital absence of a portion of skin in a localized or widespread area of the body. The lesions are most commonly localized on the scalp, however aplasia cutis congenita can affect any part of the body. {ECO:0000269	PubMed:23785305}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lakrimal ve Tükürük Bezlerinin Aplazisi, 180920, Otozomal Dominant; ALSG (Gözyaşı ve Tükürük Bezi Aplazisi) (FGF10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplasia of lacrimal and salivary glands, 180920, Autosomal dominant; ALSG (Aplasia of lacrimal and salivary glands) (FGF10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF10	FGF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal			EDTA Blood Sample
Lakrimal ve Tükürük Bezlerinin Aplazisi, 180920, Otozomal Dominant; ALSG (Gözyaşı ve Tükürük Bezi Aplazisi) (MLPA)	Aplasia of lacrimal and salivary glands, 180920, Autosomal dominant; ALSG (Aplasia of lacrimal and salivary glands) (MLPA)	FGF10	FGF10	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.		EDTA Blood Sample
Aplastik Anemi (İdiopatik Aplastik Anemi) (SBDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	APLASTIC ANEMIA (idiopathic aplastic anemia) (SBDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.		EDTA Blood Sample
Aplastik Anemi (İdiopatik Aplastik Anemi) (SBDS geni) (Tüm Gen Dizi Analizi) (Prenatal)	APLASTIC ANEMIA (idiopathic aplastic anemia) (SBDS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (PRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (PRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRF1	PRF1, HPLH2, FLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.		EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (NBN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (NBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBN	NBN, NBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.		EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (IFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (IFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aplastic anemia (AA) [MIM:609135]: A form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. It is characterized by peripheral pancytopenia and marrow hypoplasia. {ECO:0000269	PubMed:15327519}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry..	EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (MLPA)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (MLPA)	PRF1	PRF1, HPLH2, FLH2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.		EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (PRF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (PRF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRF1	PRF1, HPLH2, FLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (NBN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (NBN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NBN	NBN, NBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (IFNG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (IFNG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aplastic anemia (AA) [MIM:609135]: A form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. It is characterized by peripheral pancytopenia and marrow hypoplasia. {ECO:0000269	PubMed:15327519}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplastik Anemi, 609135 (İdiopatik Aplastik Anemi) (MLPA)	Aplastic anemia, 609135 (Idiopathic aplastic anemia) (Prenatal) (MLPA)	PRF1	PRF1, HPLH2, FLH2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aplastik Anemi, 614743, Otozomal Dominant (İdiopatik Aplastik Anemi) (TERC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplastic anemia, 614743, Autosomal dominant (Idiopathic aplastic anemia) (TERC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.		EDTA Blood Sample
Aplastik Anemi, 614743, Otozomal Dominant (İdiopatik Aplastik Anemi) (TERC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplastic anemia, 614743, Autosomal dominant (Idiopathic aplastic anemia) (TERC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Aplastik Anemi, Yatkınlık, 609135 (İdiopatik Aplastik Anemi) (SBDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aplastic anemia, susceptibility to, 609135 (Idiopathic aplastic anemia) (SBDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aplastik Anemi, Yatkınlık, 609135 (İdiopatik Aplastik Anemi) (SBDS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aplastic anemia, susceptibility to, 609135 (Idiopathic aplastic anemia) (SBDS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Apne, Postanestezik (BCHE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Apnea, postanesthetic (BCHE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCHE	BCHE, CHE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Butyrylcholinesterase deficiency (BChE deficiency) [MIM:177400]: A metabolic disorder characterized by prolonged apnea after the use of certain anesthetic drugs, including the muscle relaxants succinylcholine or mivacurium and other ester local anesthetics. The duration of the prolonged apnea varies significantly depending on the extent of the enzyme deficiency. {ECO:0000269|PubMed:10404729, ECO:0000269|PubMed:11928765, ECO:0000269|PubMed:12881446, ECO:0000269|PubMed:1306123, ECO:0000269|PubMed:1340196, ECO:0000269|PubMed:1415224, ECO:0000269|PubMed:1556385, ECO:0000269|PubMed:15781196, ECO:0000269|PubMed:1611188, ECO:0000269|PubMed:16788378, ECO:0000269|PubMed:17700357, ECO:0000269|PubMed:18075469, ECO:0000269|PubMed:18300943, ECO:0000269|PubMed:25054547, ECO:0000269|PubMed:2526279, ECO:0000269|PubMed:2915989, ECO:0000269|PubMed:7634491, ECO:0000269|PubMed:8554068, ECO:0000269|PubMed:8608411, ECO:0000269|PubMed:9110359, ECO:0000269|PubMed:9191541, ECO:0000269|PubMed:9388484, ECO:0000269|PubMed:9543549, ECO:0000269|PubMed:9694584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Apoa-I ve Apoc-III Eksikliği, Kombine (Apolipoprotein A-I Eksikliği) (APOA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ApoA-I and apoC-III deficiency, combined (Apolipoprotein A-I deficiency) (APOA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA1	APOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Apolipoprotein A-2 Eksikliği (Homozigot Ailesel Hiperkolesterolemi) (APOA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Apolipoprotein A-II deficiency (homozygous familial hypercholesterolemia) (APOA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA2	APOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Apolipoprotein C-3 Eksikliği, 614028 (Kolesterol Ester Transfer Proteini Eksikliği) (APOC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Apolipoprotein C-III deficiency, 614028 (Cholesterol-ester transfer protein deficiency) (APOC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOC3	APOC3, HALP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Belirgin Mineralokortikoid Fazlalığı, 218030, Otozomal Resesif; AME (Belirgin Mineralokortikoid Artışı) (HSD11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Apparent mineralocorticoid excess, 218030, Autosomal recessive; AME (Apparent mineralocorticoid excess) (HSD11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD11B2	HSD11B2, HSD11K, AME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Belirgin Mineralokortikoid Fazlalığı, 218030, Otozomal Resesif; AME (Belirgin Mineralokortikoid Artışı) (HSD11B2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Apparent mineralocorticoid excess, 218030, Autosomal recessive; AME (Apparent mineralocorticoid excess) (HSD11B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD11B2	HSD11B2, HSD11K, AME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Argininemia, 207800, Otozomal Resesif (Arginineml) (ARG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Argininemia, 207800, Autosomal recessive (Argininemia) (ARG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARG1	ARG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Argininemia (ARGIN) [MIM:207800]: A rare autosomal recessive disorder of the urea cycle. Arginine is elevated in the blood and cerebrospinal fluid, and periodic hyperammonemia occurs. Clinical manifestations include developmental delay, seizures, mental retardation, hypotonia, ataxia and progressive spastic quadriplegia. {ECO:0000269|PubMed:1463019, ECO:0000269|PubMed:22959135, ECO:0000269|PubMed:23659858, ECO:0000269|PubMed:7649538}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Argininemia, 207800, Otozomal Resesif (Arginineml) (ARG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Argininemia, 207800, Autosomal recessive (Argininemia) (ARG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARG1	ARG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Argininemia (ARGIN) [MIM:207800]: A rare autosomal recessive disorder of the urea cycle. Arginine is elevated in the blood and cerebrospinal fluid, and periodic hyperammonemia occurs. Clinical manifestations include developmental delay, seizures, mental retardation, hypotonia, ataxia and progressive spastic quadriplegia. {ECO:0000269|PubMed:1463019, ECO:0000269|PubMed:22959135, ECO:0000269|PubMed:23659858, ECO:0000269|PubMed:7649538}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arjininosüksinik Asidüri, 207900, Otozomal Resesif (Arjininosüksinik Asidüri) (ASL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Argininosuccinic aciduria, 207900, Autosomal recessive (Argininosuccinic aciduria) (ASL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASL	ASL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Argininosuccinic aciduria (ARGINSA) [MIM:207900]: An autosomal recessive disorder of the urea cycle. The disease is characterized by mental and physical retardation, liver enlargement, skin lesions, dry and brittle hair showing trichorrhexis nodosa microscopically and fluorescing red, convulsions, and episodic unconsciousness. {ECO:0000269|PubMed:12408190, ECO:0000269|PubMed:1705937, ECO:0000269|PubMed:17326097, ECO:0000269|PubMed:2263616, ECO:0000269|PubMed:24168529}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Arjininosüksinik Asidüri, 207900, Otozomal Resesif (Arjininosüksinik Asidüri) (ASL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Argininosuccinic aciduria, 207900, Autosomal recessive (Argininosuccinic aciduria) (ASL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASL	ASL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Argininosuccinic aciduria (ARGINSA) [MIM:207900]: An autosomal recessive disorder of the urea cycle. The disease is characterized by mental and physical retardation, liver enlargement, skin lesions, dry and brittle hair showing trichorrhexis nodosa microscopically and fluorescing red, convulsions, and episodic unconsciousness. {ECO:0000269|PubMed:12408190, ECO:0000269|PubMed:1705937, ECO:0000269|PubMed:17326097, ECO:0000269|PubMed:2263616, ECO:0000269|PubMed:24168529}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aromataz Eksikliği, 613546 (Aromataz Eksikliği) (CYP19A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aromatase deficiency, 613546 (Aromatase deficiency) (CYP19A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP19A1	CYP19A1, CYP19, ARO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aromatase deficiency (AROD) [MIM:613546]: A rare disease in which fetal androgens are not converted into estrogens due to placental aromatase deficiency. Thus, pregnant women exhibit a hirsutism, which spontaneously resolves after post-partum. At birth, female babies present with pseudohermaphroditism due to virilization of extern genital organs. In adult females, manifestations include delay of puberty, breast hypoplasia and primary amenorrhoea with multicystic ovaries. {ECO:0000269|PubMed:24705274, ECO:0000269|PubMed:8265607, ECO:0000269|PubMed:8530621, ECO:0000269|PubMed:9211678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aromataz Eksikliği, 613546 (Aromataz Eksikliği) (CYP19A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aromatase deficiency, 613546 (Aromatase deficiency) (CYP19A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP19A1	CYP19A1, CYP19, ARO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aromatase deficiency (AROD) [MIM:613546]: A rare disease in which fetal androgens are not converted into estrogens due to placental aromatase deficiency. Thus, pregnant women exhibit a hirsutism, which spontaneously resolves after post-partum. At birth, female babies present with pseudohermaphroditism due to virilization of extern genital organs. In adult females, manifestations include delay of puberty, breast hypoplasia and primary amenorrhoea with multicystic ovaries. {ECO:0000269|PubMed:24705274, ECO:0000269|PubMed:8265607, ECO:0000269|PubMed:8530621, ECO:0000269|PubMed:9211678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aromataz Fazlalığı Sendromu, 139300, Otozomal Dominant; AEXS (Aromataz Fazlalığı Sendromu) (CYP19A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aromatase excess syndrome, 139300, Autosomal dominant; AEXS (Aromatase excess syndrome) (CYP19A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP19A1	CYP19A1, CYP19, ARO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aromatase deficiency (AROD) [MIM:613546]: A rare disease in which fetal androgens are not converted into estrogens due to placental aromatase deficiency. Thus, pregnant women exhibit a hirsutism, which spontaneously resolves after post-partum. At birth, female babies present with pseudohermaphroditism due to virilization of extern genital organs. In adult females, manifestations include delay of puberty, breast hypoplasia and primary amenorrhoea with multicystic ovaries. {ECO:0000269|PubMed:24705274, ECO:0000269|PubMed:8265607, ECO:0000269|PubMed:8530621, ECO:0000269|PubMed:9211678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aromataz Fazlalığı Sendromu, 139300, Otozomal Dominant; AEXS (Aromataz Fazlalığı Sendromu) (CYP19A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aromatase excess syndrome, 139300, Autosomal dominant; AEXS (Aromatase excess syndrome) (CYP19A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP19A1	CYP19A1, CYP19, ARO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aromatase deficiency (AROD) [MIM:613546]: A rare disease in which fetal androgens are not converted into estrogens due to placental aromatase deficiency. Thus, pregnant women exhibit a hirsutism, which spontaneously resolves after post-partum. At birth, female babies present with pseudohermaphroditism due to virilization of extern genital organs. In adult females, manifestations include delay of puberty, breast hypoplasia and primary amenorrhoea with multicystic ovaries. {ECO:0000269|PubMed:24705274, ECO:0000269|PubMed:8265607, ECO:0000269|PubMed:8530621, ECO:0000269|PubMed:9211678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aromatik L-Amino Asit Dekarboksilaz Eksikliği, 608643, Otozomal Resesif (Aromatik L-Amino Asit Dekarboksilaz Eksikliği) (DDC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aromatic L-amino acid decarboxylase deficiency, 608643, Autosomal recessive (Aromatic L-amino acid decarboxylase deficiency) (DDC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDC	DDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aromatic L-amino-acid decarboxylase deficiency (AADCD) [MIM:608643]: An inborn error in neurotransmitter metabolism that leads to combined serotonin and catecholamine deficiency. It causes developmental and psychomotor delay, poor feeding, lethargy, ptosis, intermittent hypothermia, gastrointestinal disturbances. The onset is early in infancy and inheritance is autosomal recessive. {ECO:0000269|PubMed:14991824, ECO:0000269|PubMed:15079002, ECO:0000269|Ref.12}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Aromatik L-Amino Asit Dekarboksilaz Eksikliği, 608643, Otozomal Resesif (Aromatik L-Amino Asit Dekarboksilaz Eksikliği) (DDC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aromatic L-amino acid decarboxylase deficiency, 608643, Autosomal recessive (Aromatic L-amino acid decarboxylase deficiency) (DDC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDC	DDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aromatic L-amino-acid decarboxylase deficiency (AADCD) [MIM:608643]: An inborn error in neurotransmitter metabolism that leads to combined serotonin and catecholamine deficiency. It causes developmental and psychomotor delay, poor feeding, lethargy, ptosis, intermittent hypothermia, gastrointestinal disturbances. The onset is early in infancy and inheritance is autosomal recessive. (ECO:0000269|PubMed:14991824, ECO:0000269|PubMed:15079002, ECO:0000269|Ref.12). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Array - CGH	Array-CGH	.	.	Array-CGH	EDTA'lı Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Aritmojenik Sağ Ventrikül Displazisi 1, 107970, Otozomal Dominant; ARVD1 (Un Anomalisi) (TGFB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 1, 107970, Autosomal dominant; ARVD1 (Uhl anomaly) (TGFB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFB3	TGFB3, ARVD1, RNHF, LDS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 1 (ARVD1) [MIM:107970]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. (ECO:0000269	PubMed:15639475). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 10, 610193, Otozomal Dominant (Alesel İzole Aritmojenik Sağ Ventrikül Displazisi) (DSG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 10, 610193, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (DSG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG2	DSG2, ARVD10, ARVC10, CMD1BB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 10 (ARVD10) [MIM:610193]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. (ECO:0000269|PubMed:16773573, ECO:0000269|PubMed:19863551, ECO:0000269|PubMed:20031617, ECO:0000269|PubMed:21062920). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi-Hafif Palmoplantar Keratoderma ve Yünsü Saçlı, 610476, Otozomal Resesif, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (DSC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476, Autosomal recessive, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (DSC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSC2	DSC2, DSC3, ARVD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 11 (ARVD11) [MIM:610476]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. (ECO:0000269|PubMed:17033975, ECO:0000269|PubMed:19863551, ECO:0000269|PubMed:21062920). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 11, 610476, Otozomal Resesif, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (DSC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 11, 610476, Autosomal recessive, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (DSC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSC2	DSC2, DSC3, ARVD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 11 (ARVD11) [MIM:610476]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. (ECO:0000269|PubMed:17033975, ECO:0000269|PubMed:19863551, ECO:0000269|PubMed:21062920). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aritmojenik Sağ Ventrikül 12, 611528, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (JUP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 12, 611528, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (JUP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JUP	JUP, DP3, PDGB, ARVD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 2, 600996, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (RYR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 2, 600996, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (RYR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR2	RYR2, VTSIP, ARVD2, ARVC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 2 (ARVD2) [MIM:600996]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. (ECO:0000269	PubMed:11159836). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 5, 604400, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (TMEM43 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 5, 604400, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (TMEM43 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM43	TMEM43, ARVD5, ARVC5, EDMD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 8, 607450, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (DSP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 8, 607450, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (DSP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi 9, 609040, Otozomal Dominant (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (PKP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia 9, 609040, Autosomal dominant (Familial isolated arrhythmogenic right ventricular dysplasia) (PKP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKP2	PKP2, ARVD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aritmojenik Sağ Ventrikül Displazisi, Aleseal, 13, 615816, Otozomal Dominant; ARVD13 (Aleseal İzole Aritmojenik Sağ Ventrikül Displazisi) (CTNNA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arrhythmogenic right ventricular dysplasia, familial, 13, 615816, Autosomal dominant; ARVD13 (Familial isolated arrhythmogenic right ventricular dysplasia) (CTNNA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNA3	CTNNA3, ARVD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 1, 208000, Otozomal Resesif; GAC1 (İnfantil Jeneralize Arteryal Kalsifikasyon) (ENPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arterial calcification, generalized, of infancy, 1, 208000, Autosomal recessive; GAC1 (Generalized arterial calcification of infancy) (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENPP1	ENPP1, PDNP1, NPPS, MES1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 1, 208000, Otozomal Resesif; GAC1 (İnfantil Jeneralize Arteryal Kalsifikasyon) (ENPP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arterial calcification, generalized, of infancy, 1, 208000, Autosomal recessive; GAC1 (Generalized arterial calcification of infancy) (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ENPP1	ENPP1, PDNP1, NPPS, MES1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 2, 614473, Otozomal Resesif; GAC2 (İnfantil Jeneralize Arteryal Kalsifikasyon) (ABCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arterial calcification, generalized, of infancy, 2, 614473, Autosomal recessive; GAC2 (Generalized arterial calcification of infancy) (ABCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 2, 614473, Otozomal Resesif; GAC2 (İnfantil Jeneralize Arteryal Kalsifikasyon) (MLPA)	Arterial calcification, generalized, of infancy, 2, 614473, Autosomal recessive; GAC2 (Generalized arterial calcification of infancy) (MLPA)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GAC2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 2, 614473, Otozomal Resesif; GAC2 (İnfantil Jeneralize Arteryal Kalsifikasyon) (ABCC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arterial calcification, generalized, of infancy, 2, 614473, Autosomal recessive; GAC2 (Generalized arterial calcification of infancy) (ABCC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arteriyel Kalsifikasyon, Jeneralize, İnfantil, 2, 614473, Otozomal Resesif; GAC2 (İnfantil Jeneralize Arteryal Kalsifikasyon) (Prenatal) (MLPA)	Arterial calcification, generalized, of infancy, 2, 614473, Autosomal recessive; GAC2 (Generalized arterial calcification of infancy) (Prenatal) (MLPA)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GAC2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arteryal Tortioziste Sendromu, 208050, Otozomal Resesif; ATS (Arteriyel Tortiosite Sendromu) (SLC2A10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arterial tortuosity syndrome, 208050, Autosomal recessive; ATS (Arterial tortuosity syndrome) (SLC2A10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A10	SLC2A10, GLUT10, ATS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arteryal Tortiosite Sendromu, 208050, Otozomal Resesif; ATS (Arteriyel Tortiosite Sendromu) (SLC2A10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arterial tortuosity syndrome, 208050, Autosomal recessive; ATS (Arterial tortuosity syndrome) (SLC2A10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC2A10	SLC2A10, GLUT10, ATS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arteriyovenöz Malformasyonlar, Beyinde (Serebral Arteriovenöz Malformasyon) (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ARTERIOVENOUS MALFORMATIONS OF THE BRAIN (Cerebral arteriovenous malformation) (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arteriyovenöz Malformasyonlar, Beyinde (Serebral Arteriovenöz Malformasyon) (IL6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ARTERIOVENOUS MALFORMATIONS OF THE BRAIN (Cerebral arteriovenous malformation) (IL6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis Multipleks Konjenita, Distal, Tip 1, 108120, Otozomal Dominant; DA1A (Dilfotalar Dismorfizmi) (TPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis multiplex congenita, distal, type 1, 108120, Autosomal dominant; DA1A (Digitotalar dysmorphism) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 1A (DA1A) [MIM:108120]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposes type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. (ECO:0000269|PubMed:12592607, ECO:0000269|PubMed:24692096). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Artrogrifozis Multipleks Konjenita, Distal, Tip 1, 108120, Otozomal Dominant; DA1A (Dilfotalar Dismorfizmi) (TPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis multiplex congenita, distal, type 1, 108120, Autosomal dominant; DA1A (Digitotalar dysmorphism) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 1A (DA1A) [MIM:108120]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposes type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. (ECO:0000269|PubMed:12592607, ECO:0000269|PubMed:24692096). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis Multipleks Konjenita, Distal 2B, 601680, Otozomal Dominant; DA2B (Sheldon-Hall Sendromu) (TNNI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis multiplex congenita, distal, type 2B, 601680, Autosomal dominant; DA2B (Sheldon-Hall syndrome) (TNNI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI2	TNNI2, AMCD2B, DA2B, FSSV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 2B (DA2B) [MIM:601680]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA2B is characterized by contractures of the hands and feet, and a distinctive face characterized by prominent nasolabial folds, small mouth and downslanting palpebral fissures. (ECO:0000269	PubMed:12592607). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Artrogrifozis Multipleks Konjenita, Distal 2B, 601680, Otozomal Dominant; DA2B (Sheldon-Hall Sendromu) (TNNI2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis multiplex congenita, distal, type 2B, 601680, Autosomal dominant; DA2B (Sheldon-Hall syndrome) (TNNI2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNNI2	TNNI2, AMCD2B, DA2B, FSSV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 2B (DA2B) [MIM:601680]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA2B is characterized by contractures of the hands and feet, and a distinctive face characterized by prominent nasolabial folds, small mouth and downslanting palpebral fissures. (ECO:0000269	PubMed:12592607). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Artrogrifozis, Distal Tip 1B, 614335; DA1B (Dijitotalar Dismorfizmi (MYBPC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 1B, 614335; DA1B (Digitotalar dysmorphism) (MYBPC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYBPC1	MYBPC1, LCCS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 1B (DA1B) [MIM:614335]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269|PubMed:20045868}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Artrogrifozis, Distal Tip 1B, 614335; DA1B (Dijitotalar Dismorfizmi (MYBPC1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 1B, 614335; DA1B (Digitotalar dysmorphism) (MYBPC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYBPC1	MYBPC1, LCCS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 1B (DA1B) [MIM:614335]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269|PubMed:20045868}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 2A, 193700, Otozomal Dominant; DA2A(Freeman-Sheldon Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 2A, 193700, Autosomal dominant; DA2A (Freeman-Sheldon syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 8 (DA8) [MIM:178110]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. {ECO:0000269|PubMed:25957469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Artrogrifozis, Distal, Tip 2A, 193700, Otozomal Dominant; DA2A (Freeman-Sheldon Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 2A, 193700, Autosomal dominant; DA2A (Freeman-Sheldon syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 8 (DA8) [MIM:178110]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. {ECO:0000269|PubMed:25957469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (TPM2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (TPM2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 3, 114300, Otozomal Dominant; DA3 (Gordon Sendromu) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 3, 114300, Autosomal dominant; DA3 (Gordon syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Distal, Tip 3, 114300, Otozomal Dominant; DA3 (Gordon Sendromu) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 3, 114300, Autosomal dominant; DA3 (Gordon syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 5, 108145, Otozomal Dominant; DA5 (Artrogrifozis-Okulomotor Sınırlılık ve Elektroretinal Anomalili) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 5, 108145, Autosomal dominant; DA5 (Arthrogryposis with oculomotor limitation and electroretinal anomalies) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 5 (DA5) [MIM:108145]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA5 features include ocular abnormalities, typically ptosis, ophthalmoplegia and/or strabismus, in addition to contractures of the skeletal muscles. Some patients have pulmonary hypertension as a result of restrictive lung disease. {ECO:0000269|PubMed:23407782}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Artrogrifozis, Distal, Tip 5, 108145, Otozomal Dominant; DA5 (Artrogrifozis-Okulomotor Sınırlılık ve Elektroretinal Anomalili) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 5, 108145, Autosomal dominant; DA5 (Arthrogryposis with oculomotor limitation and electroretinal anomalies) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 5 (DA5) [MIM:108145]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA5 features include ocular abnormalities, typically ptosis, ophthalmoplegia and/or strabismus, in addition to contractures of the skeletal muscles. Some patients have pulmonary hypertension as a result of restrictive lung disease. {ECO:0000269|PubMed:23407782}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip SD, 615065, Otozomal Resesif; DASD (Distal Artrogrifozis Tip 5D) (ECEL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type SD, 615065, Autosomal recessive; DASD (Distal arthrogryposis type 5D) (ECEL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ECEL1	ECEL1, XCE, DASD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, SD (DASD) [MIM:615065]: An autosomal recessive form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DASD is characterized by severe camptodactyly of the hands, mild camptodactyly of the toes, clubfoot and/or a calcaneovalgus deformity, extension contractures of the knee, unilateral ptosis or ptosis that is more severe on one side, a round-shaped face, arched eyebrows, a bulbous upturned nose, and micrognathia. Patients do not have ophthalmoplegia. {ECO:0000269|PubMed:23236030, ECO:0000269|PubMed:23261301, ECO:0000269|PubMed:23808592, ECO:0000269|PubMed:23829171}. Note=The disease is caused by mutations affecting the gene represented in this entry. ECEL1 mutations have also been found in patients with arthrogryposis, significant ophthalmoplegia, and refractive errors (PubMed:23808592). {ECO:0000269|PubMed:23808592}.;	EDTA Blood Sample
Artrogrifozis, Distal, Tip SD, 615065, Otozomal Resesif; DASD (Distal Artrogrifozis Tip 5D) (ECEL1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type SD, 615065, Autosomal recessive; DASD (Distal arthrogryposis type 5D) (ECEL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ECEL1	ECEL1, XCE, DASD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, SD (DASD) [MIM:615065]: An autosomal recessive form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DASD is characterized by severe camptodactyly of the hands, mild camptodactyly of the toes, clubfoot and/or a calcaneovalgus deformity, extension contractures of the knee, unilateral ptosis or ptosis that is more severe on one side, a round-shaped face, arched eyebrows, a bulbous upturned nose, and micrognathia. Patients do not have ophthalmoplegia. {ECO:0000269|PubMed:23236030, ECO:0000269|PubMed:23261301, ECO:0000269|PubMed:23808592, ECO:0000269|PubMed:23829171}. Note=The disease is caused by mutations affecting the gene represented in this entry. ECEL1 mutations have also been found in patients with arthrogryposis, significant ophthalmoplegia, and refractive errors (PubMed:23808592). {ECO:0000269|PubMed:23808592}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 7; Da7 (Trismus-Psödokamptodaktili Sendromu) (MYH8 gen) (Tüm Gen Dizi Analizi) (Postnatal)	ARTHROGRYPOSIS, DISTAL, TYPE 7; DA7 (Trismus-pseudocamptodactyly syndrome) (MYH8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH8	MYH8, DA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 7 (DA7) [MIM:158300]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA7 is characterized by an inability to open the mouth fully (trismus) and pseudocamptodactyly in which wrist dorsiflexion, but not volarflexion, produces involuntary flexion contracture of distal and proximal interphalangeal joints. Additional features include shortened hamstring muscles and short stature. {ECO:0000269|PubMed:15282353, ECO:0000269|PubMed:20949528}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Artrogrifozis, Distal, Tip 7; Da7 (Trismus-Psödokamptodaktili Sendromu) (MYH8 gen) (Tüm Gen Dizi Analizi) (Prenatal)	ARTHROGRYPOSIS, DISTAL, TYPE 7; DA7 (Trismus-pseudocamptodactyly syndrome) (MYH8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH8	MYH8, DA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 7 (DA7) [MIM:158300]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA7 is characterized by an inability to open the mouth fully (trismus) and pseudocamptodactyly in which wrist dorsiflexion, but not volarflexion, produces involuntary flexion contracture of distal and proximal interphalangeal joints. Additional features include shortened hamstring muscles and short stature. {ECO:0000269|PubMed:15282353, ECO:0000269|PubMed:20949528}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 8, 178110, Otozomal Dominant; DA8 (Otozomal Dominant Multipl Pterijum Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 8, 178110, Autosomal dominant; DA8 (Autosomal dominant multiple pterygium syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 8 (DA8) [MIM:178110]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. {ECO:0000269|PubMed:25957469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Artrogrifozis, Distal, Tip 8, 178110, Otozomal Dominant; DA8 (Otozomal Dominant Multipl Pterijum Sendromu) (MYH3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 8, 178110, Autosomal dominant; DA8 (Autosomal dominant multiple pterygium syndrome) (MYH3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH3	MYH3, DA2A, DA2B, DA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 8 (DA8) [MIM:178110]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. {ECO:0000269|PubMed:25957469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogripozis, Distal, Propriyosepsiyon ve Dokunma Bozukluğu, 617146, Otozomal Resesif (Marden-Walker Sendromu) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, with impaired proprioception and touch, 617146, Autosomal recessive (Marden-Walker syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogripozis, Distal, Propriyosepsiyon ve Dokunma Bozukluğu, 617146, Otozomal Resesif (Marden-Walker Sendromu) (PIEZO2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, with impaired proprioception and touch, 617146, Autosomal recessive (Marden-Walker syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Artrogrifozis, Letal, Ön Boynuz Hücre Hastalığı, 611890; LAAHD (Letal Artrogrifozis-Ön Boynuz Hücre Hastalığı Sendromu) (GLE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, lethal, with anterior horn cell disease, 611890; LAAHD (Lethal arthrogryposis-anterior horn cell disease syndrome) (GLE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLE1	GLE1, GLE1L, LCCS, LCCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Letal, Ön Boynuz Hücre Hastalığı, 611890; LAAHD (Letal Artrogrifozis-Ön Boynuz Hücre Hastalığı Sendromu) (GLE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, lethal, with anterior horn cell disease, 611890; LAAHD (Lethal arthrogryposis-anterior horn cell disease syndrome) (GLE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLE1	GLE1, GLE1L, LCCS, LCCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Mental Retardasyon ve Nöbet 615553, Otozomal Resesif; AMRS (Otizm Spektrum Bozukluğu-Epilepsi-Artrogrifozis Sendromu) (SLC35A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, mental retardation, and seizures, 615553, Autosomal recessive; AMRS (Autism spectrum disorder-epilepsy-arthrogryposis syndrome) (SLC35A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC35A3	SLC35A3, AMRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, mental retardation, and seizures (AMRS) [MIM:615553]: A disease characterized by arthrogryposis, mental retardation, autism spectrum disorder, and epilepsy. Additional features include limb malformations, distal joint involvement, microcephaly, retromicrognathia, and general muscle hypotonia. {ECO:0000269	PubMed:24031089}. Note=The disease is caused by mutations affecting the gene represented in this entry. In Golgi vesicles isolated from patient fibroblasts the transport of the respective nucleotide sugar is significantly reduced causing a massive decrease in the content of cell surface expressed highly branched N-glycans and a concomitant sharp increase of lower branched glycoforms.;	EDTA Blood Sample
Artrogrifozis, Mental Retardasyon ve Nöbet 615553, Otozomal Resesif; AMRS (Otizm Spektrum Bozukluğu-Epilepsi-Artrogrifozis Sendromu) (SLC35A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, mental retardation, and seizures, 615553, Autosomal recessive; AMRS (Autism spectrum disorder-epilepsy-arthrogryposis syndrome) (SLC35A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC35A3	SLC35A3, AMRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, mental retardation, and seizures (AMRS) [MIM:615553]: A disease characterized by arthrogryposis, mental retardation, autism spectrum disorder, and epilepsy. Additional features include limb malformations, distal joint involvement, microcephaly, retromicrognathia, and general muscle hypotonia. {ECO:0000269	PubMed:24031089}. Note=The disease is caused by mutations affecting the gene represented in this entry. In Golgi vesicles isolated from patient fibroblasts the transport of the respective nucleotide sugar is significantly reduced causing a massive decrease in the content of cell surface expressed highly branched N-glycans and a concomitant sharp increase of lower branched glycoforms.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Perthes Hastalığı ve Yukarı Bakış Paralizisi, 614262, Otozomal Resesi; APUG (NEK9 İle İlişkili Letal İskelet Displazisi) (NEK9 geni) (Tüm Gen Dizi Analizi (Postnatal)	Arthrogryposis, Perthes disease, and upward gaze palsy, 614262, Autosomal recessive; APUG (NEK9-related lethal skeletal dysplasia) (NEK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK9	NEK9, NERCC1, LCCS10, APUG, NC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Perthes Hastalığı ve Yukarı Bakış Paralizisi, 614262, Otozomal Resesi; APUG (NEK9 İle İlişkili Letal İskelet Displazisi) (NEK9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, Perthes disease, and upward gaze palsy, 614262, Autosomal recessive; APUG (NEK9-related lethal skeletal dysplasia) (NEK9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEK9	NEK9, NERCC1, LCCS10, APUG, NC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Böbrek Fonksiyon Bozukluğu ve Kolestaz 1, 208085, Otozomal Resesif; ARCS1 (Artrogrifozis-Böbrek İşlev Bozukluğu-Kolestaz Sendromu) (VPS33B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085, Autosomal recessive; ARCS1 (Arthrogryposis-renal dysfunction-cholestasis syndrome) (VPS33B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS33B	VPS33B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, renal dysfunction and cholestasis syndrome 1 (ARCS1) [MIM:208085]: A multisystem disorder, characterized by neurogenic arthrogryposis multiplex congenita, renal tubular dysfunction and neonatal cholestasis with bile duct hypoplasia and low gamma glutamyl transpeptidase activity. Platelet dysfunction is common. {ECO:0000269	PubMed:15052268, ECO:0000269	PubMed:18853461}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Artrogrifozis, Böbrek Fonksiyon Bozukluğu ve Kolestaz 1, 208085, Otozomal Resesif; ARCS1 (Artrogrifozis-Böbrek İşlev Bozukluğu-Kolestaz Sendromu) (VPS33B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, renal dysfunction, and cholestasis 1, 208085, Autosomal recessive; ARCS1 (Arthrogryposis-renal dysfunction-cholestasis syndrome) (VPS33B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS33B	VPS33B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, renal dysfunction and cholestasis syndrome 1 (ARCS1) [MIM:208085]: A multisystem disorder, characterized by neurogenic arthrogryposis multiplex congenita, renal tubular dysfunction and neonatal cholestasis with bile duct hypoplasia and low gamma glutamyl transpeptidase activity. Platelet dysfunction is common. {ECO:0000269	PubMed:15052268, ECO:0000269	PubMed:18853461}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Böbrek Fonksiyon Bozukluğu ve Kolestaz 2, 613404, Otozomal Resesif; ARCS2 (Artrogrifozis-Böbrek İşlev Bozukluğu-Kolestaz Sendromu) (VIPAS39 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404, Autosomal recessive; ARCS2 (Arthrogryposis-renal dysfunction-cholestasis syndrome) (VIPAS39 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VIPAS39	VIPAS39, VIPAR, SPE39, C14orf133	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, renal dysfunction and cholestasis syndrome 2 (ARCS2) [MIM:613404]: A multisystem disorder, characterized by neurogenic arthrogryposis multiplex congenita, renal tubular dysfunction and neonatal cholestasis with bile duct hypoplasia and low gamma glutamyl transpeptidase activity. Platelet dysfunction is common. Note=The disease is caused by mutations affecting the gene represented in this entry. In liver, CEACAM5 and ABCB11 are mislocalized and E-cadherin expression is decreased.;	EDTA Blood Sample
Artrogrifozis, Böbrek Fonksiyon Bozukluğu ve Kolestaz 2, 613404, Otozomal Resesif; ARCS2 (Artrogrifozis-Böbrek İşlev Bozukluğu-Kolestaz Sendromu) (VIPAS39 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, renal dysfunction, and cholestasis 2, 613404, Autosomal recessive; ARCS2 (Arthrogryposis-renal dysfunction-cholestasis syndrome) (VIPAS39 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VIPAS39	VIPAS39, VIPAR, SPE39, C14orf133	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, renal dysfunction and cholestasis syndrome 2 (ARCS2) [MIM:613404]: A multisystem disorder, characterized by neurogenic arthrogryposis multiplex congenita, renal tubular dysfunction and neonatal cholestasis with bile duct hypoplasia and low gamma glutamyl transpeptidase activity. Platelet dysfunction is common. Note=The disease is caused by mutations affecting the gene represented in this entry. In liver, CEACAM5 and ABCB11 are mislocalized and E-cadherin expression is decreased.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artropati, Progresif Psödoromatoid, Çocukluk Çağı, 208230, Otozomal Resesif; PPAC (Çocukluk Çağı Progresif Psödoromatoid Artropatisi (Progresif Artropati İle Spondiloepifyal Displazi Tarda)) (WISP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthropathy, progressive pseudorheumatoid, of childhood, 208230, Autosomal recessive; PPAC (Progressive pseudorheumatoid arthropathy of childhood) (WISP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WISP3	WISP3, PPAC, PPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artropati, Progresif Psödoromatoid, Çocukluk Çağı, 208230, Otozomal Resesif; PPAC (Çocukluk Çağı Progresif Psödoromatoid Artropatisi (Progresif Artropati İle Spondiloepifyal Displazi Tarda)) (WISP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthropathy, progressive pseudorheumatoid, of childhood, 208230, Autosomal recessive; PPAC (Progressive pseudorheumatoid arthropathy of childhood) (WISP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WISP3	WISP3, PPAC, PPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (TNNT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (TNNT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT3	TNNT3, AMCD2B, DA2B, FSSV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Artrogrifozis, Distal, Tip 2B, 601680, Otozomal Dominant (Sheldon-Hall Sendromu) (TNNT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arthrogryposis, distal, type 2B, 601680, Autosomal dominant (Sheldon-Hall syndrome) (TNNT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNNT3	TNNT3, AMCD2B, DA2B, FSSV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Arts Sendromu, 301835, X'e Bağlı Resesif; ARTS (Letal Ataksi-Sağırlık ve Optik Atrofi (Arts Sendromu)) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Arts syndrome, 301835, X-linked recessive; ARTS (Lethal ataxia with deafness and optic atrophy) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Arts Sendromu, 301835, X'e Bağlı Resesif; ARTS (Letal Ataksi-Sağırlık ve Optik Atrofi (Arts Sendromu)) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Arts syndrome, 301835, X-linked recessive; ARTS (Lethal ataxia with deafness and optic atrophy) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Asparagin Sentetaz Eksikliği, 615574, Otozomal Resesif (Konjenital Mikrosefali-Ağır Ensefalopati-Progresif Serebral Atrofi Sendromu) (ASNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asparagine synthetase deficiency, 615574, Autosomal recessive (Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome) (ASNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASNS	ASNS, ASNSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asparagine synthetase deficiency (ASNSD) [MIM:615574]: An inborn error of asparagine biosynthesis that results in a severe neurologic disorder characterized by microcephaly, severely delayed psychomotor development, progressive encephalopathy, cortical atrophy, and seizure or hyperekplexic activity. {ECO:0000269	PubMed:24139043}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Asparagin Sentetaz Eksikliği, 615574, Otozomal Resesif (Konjenital Mikrosefali-Ağır Ensefalopati-Progresif Serebral Atrofi Sendromu) (ASNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Asparagine synthetase deficiency, 615574, Autosomal recessive (Congenital microcephaly-severe encephalopathy-progressive cerebral atrophy syndrome) (ASNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASNS	ASNS, ASNSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Asparagine synthetase deficiency (ASNSD) [MIM:615574]: An inborn error of asparagine biosynthesis that results in a severe neurologic disorder characterized by microcephaly, severely delayed psychomotor development, progressive encephalopathy, cortical atrophy, and seizure or hyperekplexic activity. {ECO:0000269	PubMed:24139043}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aspartat Aminotransferaz, QTL1, Serum Düzeyi, 614419 (GOT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aspartate aminotransferase, serum level of, QTL1, 614419 (GOT1 gene) (Sequence Analysis-All Codno Exonsi (Postnatal)	GOT1	GOT1, ASTQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aspartat Aminotransferaz, QTL1, Serum Düzeyi, 614419 (GOT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aspartate aminotransferase, serum level of, QTL1, 614419 (GOT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GOT1	GOT1, ASTQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aspartilglikozaminüri, 208400, Otozomal Resesif; AGU (Aspartilglukosaminüri) (AGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aspartylglucosaminuria, 208400, Autosomal recessive; AGU (Aspartylglucosaminuria) (AGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGA	AGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aspartylglucosaminuria (AGU) [MIM:208400]: An inborn lysosomal storage disease causing excess accumulation of glycoasparagine in the body tissues and its increased excretion in urine. Clinical features include mild to severe mental retardation manifesting from the age of two, coarse facial features and mild connective tissue abnormalities. {ECO:0000269|PubMed:11309371, ECO:0000269|PubMed:1703489, ECO:0000269|PubMed:1904874, ECO:0000269|PubMed:2011603, ECO:0000269|PubMed:8776587, ECO:0000269|PubMed:9137882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aspartilglikozaminüri, 208400, Otozomal Resesif; AGU (Aspartilglukosaminüri) (AGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aspartylglucosaminuria, 208400, Autosomal recessive; AGU (Aspartylglucosaminuria) (AGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGA	AGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aspartylglucosaminuria (AGU) [MIM:208400]: An inborn lysosomal storage disease causing excess accumulation of glycoasparagine in the body tissues and its increased excretion in urine. Clinical features include mild to severe mental retardation manifesting from the age of two, coarse facial features and mild connective tissue abnormalities. {ECO:0000269|PubMed:11309371, ECO:0000269|PubMed:1703489, ECO:0000269|PubMed:1904874, ECO:0000269|PubMed:2011603, ECO:0000269|PubMed:8776587, ECO:0000269|PubMed:9137882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Asperger Sendromu Yatkınlık, X'e Bağlı 1, 300494, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (NLGN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asperger syndrome susceptibility, X-linked 1, 300494, Isolated cases, X-linked, Multifactorial (NLGN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLGN3	NLGN3, ASPGX1, AUTSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asperger syndrome, X-linked, 1 (ASPGX1) [MIM:300494]: A syndrome with features similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:12669065}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample

Asperger Sendromu Yatkınlık, X'e Bağlı 1, 300494, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (NLGN3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Asperger syndrome susceptibility, X-linked 1, 300494, Isolated cases, X-linked, Multifactorial (NLGN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLGN3	NLGN3, ASPGX1, AUTSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Asperger syndrome, X-linked, 1 (ASPGX1) [MIM:300494]: A syndrome with features similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:12669065}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Asperger Sendromu Yatkınlık, X'e Bağlı 2, 300497, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (NLGN4X gene) (Tüm Gen Dizi Analizi) (Postnatal)	Asperger syndrome susceptibility, X-linked 2, 300497, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asperger syndrome, X-linked, 2 (ASPGX2) [MIM:300497]: A syndrome with features similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:14963808}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Asperger Sendromu Yatkınlık, X'e Bağlı 2, 300497, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (NLGN4X gene) (Tüm Gen Dizi Analizi) (Prenatal)	Asperger syndrome susceptibility, X-linked 2, 300497, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Asperger syndrome, X-linked, 2 (ASPGX2) [MIM:300497]: A syndrome with features similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:14963808}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aspergilloz, Yatkınlık, 614079 (Aspergillozis) (CLEC7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aspergillosis, susceptibility to, 614079 (Aspergillosis) (CLEC7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLEC7A	CLEC7A, CLECSF12, DECTIN1, CANDF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aspergilloz, Yatkınlık, 614079 (Aspergillozis) (CLEC7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Aspergillosis, susceptibility to, 614079 (Aspergillosis) (CLEC7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLEC7A	CLEC7A, CLECSF12, DECTIN1, CANDF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aspleni, İzole Konjenital, 271400, Otozomal Dominant; ICAS (Ailesel İzole Konjenital Aspleni) (RPSA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asplenia, isolated congenital, 271400, Autosomal dominant; ICAS (Familial isolated congenital asplenia) (RPSA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPSA	RPSA, LAMR1, LAMBR, ICAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asplenia, isolated congenital (ICAS) [MIM:271400]: A rare primary immunodeficiency and life-threatening condition, often presenting with pneumococcal sepsis. Most affected individuals die of severe bacterial infections in early childhood. Isolated asplenia is distinct from asplenia associated with other complex visceral defects, notably heterotaxy syndromes such as Ivemark syndrome. {ECO:0000269	PubMed:23579497}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Aspleni, İzole Konjenital, 271400, Otozomal Dominant; ICAS (Ailesel İzole Konjenital Aspleni) (RPSA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Asplenia, isolated congenital, 271400, Autosomal dominant; ICAS (Familial isolated congenital asplenia) (RPSA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPSA	RPSA, LAMR1, LAMBR, ICAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Asplenia, isolated congenital (ICAS) [MIM:271400]: A rare primary immunodeficiency and life-threatening condition, often presenting with pneumococcal sepsis. Most affected individuals die of severe bacterial infections in early childhood. Isolated asplenia is distinct from asplenia associated with other complex visceral defects, notably heterotaxy syndromes such as Ivemark syndrome. {ECO:0000269	PubMed:23579497}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Astım ve Nazal Polipler, 208550, Otozomal Resesif (TBX21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma and nasal polyps, 208550, Autosomal recessive (TBX21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX21	TBX21, TBET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım Yatkınlık 5, 611064 (IRAK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma susceptibility 5, 611064 (IRAK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRAK3	IRAK3, IRAKM, ASRT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astımla İlgili Özellikler, Yatkınlık, 7, 611960 (CHI3L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma-related traits, susceptibility to, 7, 611960 (CHI3L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHI3L1	CHI3L1, GP39, YKL40, ASRT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asthma-related traits 7 (ASRT7) [MIM:611960]: Asthma- related traits include clinical symptoms of asthma, such as coughing, wheezing, dyspnea, bronchial hyperresponsiveness as assessed by methacholine challenge test, serum IgE levels, atopy and atopic dermatitis. {ECO:0000269	PubMed:18403759}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Astım, 600807, Otozomal Dominant (PHF11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, 600807, Autosomal dominant (PHF11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHF11	PHF11, NYREN34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Aspirine Bağlı, Yatkınlık, 208550, Otozomal Resesif (TBX21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, aspirin-induced, susceptibility to, 208550, Autosomal recessive (TBX21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX21	TBX21, TBET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Aspirine Bağlı, Yatkınlık, 208550, Otozomal Resesif (PTGER2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, aspirin-induced, susceptibility to, 208550, Autosomal recessive (PTGER2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTGER2	PTGER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Antileukotrien Tedavisine Azalmış Yanıt, 600807, Otozomal Dominant (ALOX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, diminished response to antileukotriene treatment in, 600807, Autosomal dominant (ALOX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALOX5	ALOX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Noktural, Yatkınlık, 600807, Otozomal Dominant (ADRB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, nocturnal, susceptibility to, 600807, Autosomal dominant (ADRB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB2	ADRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Karşı Koruma, 600807, Otozomal Dominant (MUC7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, protection against, 600807, Autosomal dominant (MUC7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUC7	MUC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asthma (ASTHMA) [MIM:600807]: The most common chronic disease affecting children and young adults. It is a complex genetic disorder with a heterogeneous phenotype, largely attributed to the interactions among many genes and between these genes and the environment. It is characterized by recurrent attacks of paroxysmal dyspnea, with wheezing due to spasmodic contraction of the bronchi. {ECO:0000269	PubMed:11378823}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Astım, Yatkınlık, 1, 607277 (PTGDR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 1, 607277 (PTGDR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTGDR	PTGDR, AS1, ASRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 2, 608584 (NPSR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 2, 608584 (NPSR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPSR1	NPSR1, GPR154, GPRA, VRR1, PGR14, ASRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (TNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (TNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNF	TNF, TNFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (SCGB3A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (SCGB3A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCGB3A2	SCGB3A2, UGRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (PLA2G7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (PLA2G7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G7	PLA2G7, PAFAH, PAFAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atopic hypersensitivity (ATOPY) [MIM:147050]: A condition characterized by predisposition to develop hypersensitivity reactions. Atopic individuals can develop eczema, allergic rhinitis and allergic asthma. {ECO:0000269	PubMed:10733466}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (IL13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (IL13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL13	IL13, ALRH, BHR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (HNMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (HNMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNMT	HNMT, MRTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (HLA-G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (HLA-G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-G	HLA-G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Astım, Yatkınlık, 600807, Otozomal Dominant (CCL11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Asthma, susceptibility to, 600807, Autosomal dominant (CCL11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL11	CCL11, SCYA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İzole Vitamin E Eksikliği Ataksisi, 277460, Otozomal Resesif; VED (Vitamin E Eksikliğine Bağlı Ataksi) (TTPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia with isolated vitamin E deficiency, 277460, Autosomal recessive; VED (Ataxia with vitamin E deficiency) (TTPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTPA	TTPA, TTP1, AVED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia with isolated vitamin E deficiency (AVED) [MIM:277460]: An autosomal recessive disease characterized by undetectable or markedly reduced plasma levels of vitamin E, spinocerebellar degeneration, ataxia, areflexia and proprioception loss. {ECO:0000269|PubMed:15304640, ECO:0000269|PubMed:7566022, ECO:0000269|PubMed:8602747, ECO:0000269|PubMed:9463307}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Ataksi Okülomotor Apraksi 3, 615217, Otozomal Resesif; AOA3 (Spinoserebellar ataksi with Aksiki-Aksonal Nöropati Tip 3) (PIK3R5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-oculomotor apraxia 3, 615217, Autosomal recessive; AOA3 (Spinocerebellar ataxia with axonal neuropathy type 2) (PIK3R5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3R5	PIK3R5, p101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia-oculomotor apraxia 3 (AOA3) [MIM:615217]: An autosomal recessive disease characterized by cerebellar ataxia, oculomotor apraxia, areflexia and peripheral neuropathy. {ECO:0000269	PubMed:22065524}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Ataksi Okülomotor Apraksi 4, 616267, Otozomal Resesif (Ataksi Okülomotor Apraksi Tip 4) (PNKP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-oculomotor apraxia 4, 616267, Autosomal recessive (Ataxia-oculomotor apraxia type 4) (PNKP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNKP	PNKP, PNK, MCSZ, EIEE10, MCSZ, AOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ataksi Parsitoperi Sendromu, 159550, Otozomal Dominant; ATXPC (Ataksi Parsitoperi Sendromu) (SAMD9L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-pancytopenia syndrome, 159550, Autosomal dominant; ATXPC (Ataxia-pancytopenia syndrome) (SAMD9L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAMD9L	SAMD9L, ATXPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ataksi Parsitoperi Sendromu, 159550, Otozomal Dominant; ATXPC (Ataksi Parsitoperi Sendromu) (SAMD9L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ataxia-pancytopenia syndrome, 159550, Autosomal dominant; ATXPC (Ataxia-pancytopenia syndrome) (SAMD9L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SAMD9L	SAMD9L, ATXPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Ataksi-Telenjiektazi Benzeri Hastalık, 604391, Otozomal Resesif (Ataksi-Telenjiektazi Benzeri Bozukluk) (MRE11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-telangiectasia-like disorder, 604391, Autosomal recessive (Ataxia-telangiectasia-like disorder) (MRE11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRE11	MRE11A, MRE11, ATLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ataksi-Telenjiektazi Benzeri Hastalık, 604391, Otozomal Resesif (Ataksi-Telenjiektazi Benzeri Bozukluk) (MRE11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ataxia-telangiectasia-like disorder, 604391, Autosomal recessive (Ataxia-telangiectasia-like disorder) (MRE11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MRE11	MRE11A, MRE11, ATLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ataksi Telenjiektazi Benzeri Hastalık, 615919, Otozomal Resesif; ATLD2 (Nörodejeneratif Fotosensitivite Sendromu, PCNA İle İlişkili (Ataksi-Telenjiektazi Benzeri Bozukluk)) (PCNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-telangiectasia-like disorder, 615919, Autosomal recessive; ATLD2 (PCNA-related progressive neurodegenerative photosensitivity syndrome) (PCNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCNA	PCNA, ATLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia-telangiectasia-like disorder 2 (ATLD2) [MIM:615919]: A neurodegenerative disorder due to defects in DNA excision repair. ATLD2 is characterized by developmental delay, ataxia, sensorineural hearing loss, short stature, cutaneous and ocular telangiectasia, and photosensitivity. {ECO:0000269	PubMed:24911150}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ataksi Telenjiektazi Benzeri Hastalık, 615919, Otozomal Resesif; ATLD2 (Nörodejeneratif Fotosensitivite Sendromu, PCNA İle İlişkili (Ataksi-Telenjiektazi Benzeri Bozukluk)) (PCNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ataxia-telangiectasia-like disorder, 615919, Autosomal recessive; ATLD2 (PCNA-related progressive neurodegenerative photosensitivity syndrome) (PCNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCNA	PCNA, ATLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ataxia-telangiectasia-like disorder 2 (ATLD2) [MIM:615919]: A neurodegenerative disorder due to defects in DNA excision repair. ATLD2 is characterized by developmental delay, ataxia, sensorineural hearing loss, short stature, cutaneous and ocular telangiectasia, and photosensitivity. {ECO:0000269	PubMed:24911150}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ataksi Telenjiektazi, 208900, Otozomal Resesif; AT (Ataksi-Telenjiektazi) (ATM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia-telangiectasia, 208900, Autosomal recessive; AT (Ataxia-telangiectasia) (ATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333}. Note=The disease is caused	EDTA Blood Sample
Ataksi Telenjiektazi, 208900, Otozomal Resesif; AT (Ataksi-Telenjiektazi) (MLPA)	Ataxia-telangiectasia, 208900, Autosomal recessive; AT (Ataxia-telangiectasia) (MLPA)	ATM	ATM, ATA, AT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333}. Note=The disease is caused	EDTA Blood Sample
Ataksi Telenjiektazi, 208900, Otozomal Resesif; AT (Ataksi-Telenjiektazi) (ATM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ataxia-telangiectasia, 208900, Autosomal recessive; AT (Ataxia-telangiectasia) (ATM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333}. Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ataksi Telenjiektazi, 208900, Otozomal Resesif; AT (Ataksi-Telenjiektazi) (Prenatal) (MLPA)	Ataxia-telangiectasia, 208900, Autosomal recessive; AT (Ataxia-telangiectasia) (Prenatal) (MLPA)	ATM	ATM, ATA, AT1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333}. Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ataksi, Serebellar, Cayman Tipi, 601238, Otozomal Resesif; ATCAY (Serebellar Ataksi, Cayman Tipi) (ATCAY geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia, cerebellar, Cayman type, 601238, Autosomal recessive; ATCAY (Cerebellar ataxia, Cayman type) (ATCAY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATCAY	ATCAY, CLAC, KIAA1872	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, cayman type (ATCAY) [MIM:601238]: Found in a population isolate on Grand Cayman Island and causes a marked psychomotor retardation and prominent nonprogressive cerebellar dysfunction including nystagmus, intention tremor, dysarthria, and wide-based ataxic gait. Hypotonia is present from early childhood. {ECO:0000269|PubMed:14556008}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ataksi, Kombine Serebeller ve Periferal, İşitme Kaybı ve Diyabetes Mellitus, 616192, Otozomal Resesif; ACPHD (Juvenil Başlangıçlı Diyabet-Santral ve Periferal Nörodejenerasyon Sendromu) (DNAJC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus, 616192, Autosomal recessive; ACPHD (Juvenile-onset diabetes mellitus-central and peripheral neurodegeneration syndrome) (DNAJC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC3	DNAJC3, PRKRI, PS8, ACPHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia, combined cerebellar and peripheral, with hearing loss and diabetes mellitus (ACPHD) [MIM:616192]: A disease characterized by juvenile-onset diabetes and neurodegeneration, resulting in ataxia, upper-motor-neuron damage, peripheral neuropathy, hearing loss, and cerebral atrophy. {ECO:0000269|PubMed:25468070}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ataksi, Erken Başlangıçlı, Okülomotor Apraksi ve Hipoalbuminemi, 208920, Otozomal Resesif; EAOH (Ataksi-Okulomotor Apraksi Tip 1) (APTX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920, Autosomal recessive; EAOH (Ataxia-oculomotor apraxia type 1) (APTX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APTX	APTX, AOA, AOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia-oculomotor apraxia syndrome (AOA) [MIM:208920]: An autosomal recessive syndrome characterized by early-onset cerebellar ataxia, oculomotor apraxia, early areflexia and late peripheral neuropathy. {ECO:0000269|PubMed:11586299, ECO:0000269|PubMed:11586300, ECO:0000269|PubMed:12196655, ECO:0000269|PubMed:12629250, ECO:0000269|PubMed:14506070, ECO:0000269|PubMed:15699391, ECO:0000269|PubMed:15852392}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Ataksi, Erken Başlangıçlı, Okülomotor Apraksi ve Hipoalbüminemi, 208920, Otozomal Resesif; EAOH (Ataksi-Okülomotor Apraksi Tip 1) (MLPA)	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920, Autosomal recessive; EAOH (Ataxia-oculomotor apraxia type 1) (MLPA)	APTX	APTX, AOA, AOA1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ataxia-oculomotor apraxia syndrome (AOA) [MIM:208920]: An autosomal recessive syndrome characterized by early-onset cerebellar ataxia, oculomotor apraxia, early areflexia and late peripheral neuropathy. {ECO:0000269|PubMed:11586299, ECO:0000269|PubMed:11598300, ECO:0000269|PubMed:12196655, ECO:0000269|PubMed:12629250, ECO:0000269|PubMed:14506070, ECO:0000269|PubMed:15699391, ECO:0000269|PubMed:15852392}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ataksi, Posterior Kolon, Retinitis Pigmentoza, 609033, Otozomal Resesif; AXPC1 (Arka Kolon Ataksi-Retinitis Pigmentoza Sendromu) (FLVCR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia, posterior column, with retinitis pigmentosa, 609033, Autosomal recessive; AXPC1 (Posterior column ataxia-retinitis pigmentosa syndrome) (FLVCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLVCR1	FLVCR1, AXPC1, PCARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Posterior column ataxia with retinitis pigmentosa (PCARP) [MIM:609033]: A neurodegenerative syndrome beginning in infancy with areflexia and retinitis pigmentosa. Nyctalopia (night blindness) and peripheral visual field loss are usually evident during late childhood or teenage years, with subsequent progressive constriction of the visual fields and loss of central retinal function over time. A sensory ataxia caused by degeneration of the posterior columns of the spinal cord results in a loss of proprioceptive sensation that is clinically evident in the second decade of life and gradually progresses. Scoliosis, camptodactyly, achalasia, gastrointestinal dysmotility, and a sensory peripheral neuropathy are variable features of the disease. Affected individuals have no clinical or radiological evidence of cerebral or cerebellar involvement. {ECO:0000269|PubMed:21070897, ECO:0000269|PubMed:21267618}. Note=The disease is caused by mutations affecting the gene represented in this entry. Defective neuronal heme transmembrane export due to FLVCR1 mutations may abrogate the neuroprotective effects of neuroglobin and initiate an apoptotic cascade that results in the selective degeneration of photoreceptors in the neurosensory retina and sensory neurons in the posterior spinal cord.;	EDTA Blood Sample
Ataksi, Duyusal, 1, Otozomal Dominant, 608984, Otozomal Dominant (RNF170 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ataxia, sensory, 1, autosomal dominant, 608984, Autosomal dominant (RNF170 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF170	RNF170, SNAX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atelosteogenezis 2 256050, Otozomal Resesif; AO2 (Atelosteogenesis Tip 2) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atelosteogenesis II, 256050, Autosomal recessive; AO2 (Atelosteogenesis type II) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atelosteogenezis 2 256050, Otozomal Resesif; AO2 (Atelosteogenesis Tip 2) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Atelosteogenesis II, 256050, Autosomal recessive; AO2 (Atelosteogenesis type II) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atelosteogenezis, Tip 1, 108720, Otozomal Dominant; AO1 (Atelosteogenezis Tip 1) (FLNB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atelosteogenesis, type I, 108720, Autosomal dominant; AO1 (Atelosteogenesis type I) (FLNB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269	PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Atelosteogenezis, Tip 1, 108720, Otozomal Dominant; AO1 (Atelosteogenezis Tip 1) (FLNB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Atelosteogenesis, type I, 108720, Autosomal dominant; AO1 (Atelosteogenesis type I) (FLNB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269	PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atelosteogenezis, Tip 3, 108721, Otozomal Dominant; AO3 (Atelosteogenesis Tip 3) (FLNB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atelosteogenesis, type III, 108721, Autosomal dominant; AO3 (Atelosteogenesis type III) (FLNB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atelosteogenezis, Tip 3, 108721, Otozomal Dominant; AO3 (Atelosteogenesis Tip 3) (FLNB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Atelosteogenesis, type III, 108721, Autosomal dominant; AO3 (Atelosteogenesis type III) (FLNB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Athabaskan Beyin Disgenezileri Sendromu, 601536; ABDS (Boskey Salih-Akrainy Sendromu (Atabaşkan Beyin Disgenezisi Sendromu)) (HOXA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Athabaskan brainstem dysgenesis syndrome, 601536; ABDS (Boskey-Salih-Akrainy syndrome) (HOXA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA1	HOXA1, HOX1F, BSAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Athabaskan brainstem dysgenesis syndrome (ABDS) [MIM:601536]: Characterized by horizontal gaze palsy, sensorineural deafness, central hypoventilation, and developmental delay. Some patients had swallowing dysfunction, vocal cord paralysis, facial paresis, seizures, and cardiac outflow tract anomalies. {ECO:0000269	PubMed:16155570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Athabaskan Beyin Disgenezileri Sendromu, 601536; ABDS (Boskey Salih-Akrainy Sendromu (Atabaşkan Beyin Disgenezisi Sendromu)) (HOXA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Athabaskan brainstem dysgenesis syndrome, 601536; ABDS (Boskey-Salih-Akrainy syndrome) (HOXA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXA1	HOXA1, HOX1F, BSAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Athabaskan brainstem dysgenesis syndrome (ABDS) [MIM:601536]: Characterized by horizontal gaze palsy, sensorineural deafness, central hypoventilation, and developmental delay. Some patients had swallowing dysfunction, vocal cord paralysis, facial paresis, seizures, and cardiac outflow tract anomalies. {ECO:0000269	PubMed:16155570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ateroskleroz, Yatkınlık (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atherosclerosis, susceptibility to (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ateroskleroz, Yatkınlık (ALOX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atherosclerosis, susceptibility to (ALOX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALOX5	ALOX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atopi, 147050, Otozomal Dominant (SPINK5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, 147050, Autosomal dominant (SPINK5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINK5	SPINK5, LEKTI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atopi, Direnç, 147050, Otozomal Dominant (HAVCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, resistance to, 147050, Autosomal dominant (HAVCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HAVCR1	HAVCR1, HAVCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atopi, Yatkınlık, 147050, Otozomal Dominant (SELP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, susceptibility to, 147050, Autosomal dominant (SELP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SELP	SELP, GRMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atopi, Yatkınlık, 147050, Otozomal Dominant (PLA2G7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, susceptibility to, 147050, Autosomal dominant (PLA2G7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G7	PLA2G7, PAFAH, PAFAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atopic hypersensitivity (ATOPY) [MIM:147050]: A condition characterized by predisposition to develop hypersensitivity reactions. Atopic individuals can develop eczema, allergic rhinitis and allergic asthma. {ECO:0000269	PubMed:10733468}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Atopi, Yatkınlık, 147050, Otozomal Dominant (MS4A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, susceptibility to, 147050, Autosomal dominant (MS4A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MS4A2	MS4A2, FCER1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atopi, Yatkınlık, 147050, Otozomal Dominant (IL4R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atopy, susceptibility to, 147050, Autosomal dominant (IL4R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL4R	IL4R, IL4RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atp Sentaz 6; Mtatp6 (MT-ATP6 geni) (Dizi Analizi) (Postnatal)	ATP SYNTHASE 6; MTATP6 (MT-ATP6 gene) (Dizi Analizi) (Postnatal)	MT-ATP6		Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atp Sentaz 8; Mtatp8 (MT-ATP8 geni) (Dizi Analizi) (Postnatal)	ATP SYNTHASE 8; MTATP8 (MT-ATP8 gene) (Dizi Analizi) (Postnatal)	MT-ATP8		Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atransferinemi 209300, Otozomal Resesif (Konjenital Atransferinemi) (TF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atransferrinemia, 209300, Autosomal recessive (Congenital atransferrinemia) (TF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TF	TF, TFQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atransferrinemia (ATRAF) [MIM:209300]: A rare autosomal recessive disorder characterized by abnormal synthesis of transferrin leading to iron overload and microcytic hypochromic anemia. {ECO:0000269|PubMed:11110675, ECO:0000269|PubMed:15466165}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Atransferinemi 209300, Otozomal Resesif (Konjenital Atransferinemi) (TF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Atransferrinemia, 209300, Autosomal recessive (Congenital atransferrinemia) (TF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TF	TF, TFQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Atransferrinemia (ATRAF) [MIM:209300]: A rare autosomal recessive disorder characterized by abnormal synthesis of transferrin leading to iron overload and microcytic hypochromic anemia. {ECO:0000269|PubMed:11110675, ECO:0000269|PubMed:15466165}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atriyal Fibrilasyon 15, 615770, Otozomal Resesif; ATFB15 (Ailesel Atriyal Fibrilasyon) (NUP155 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation 15, 615770, Autosomal recessive; ATFB15 (Familial atrial fibrillation) (NUP155 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP155	NUP155, KIAA0791, ATFB15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 15 (ATFB15) [MIM:615770]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. {ECO:0000269	PubMed:19070573}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 10, 614022, Otozomal Dominant; ATFB10 (Ailesel Atriyal Fibrilasyon) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 10, 614022, Autosomal dominant; ATFB10 (Familial atrial fibrillation) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 11, 614049, Otozomal Dominant; ATFB11 (Ailesel Atriyal Fibrilasyon) (GJA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 11, 614049, Autosomal dominant; ATFB11 (Familial atrial fibrillation) (GJA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA5	GJA5, CX40, ATFB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 12, 614050, Otozomal Dominant; ATFB12 (Ailesel Atriyal Fibrilasyon) (ABCC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 12, 614050, Autosomal dominant; ATFB12 (Familial atrial fibrillation) (ABCC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC9	ABCC9, SUR2, CMD1O, ATFB12, CANTU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 13, 615377, Otozomal Dominant; ATFB13 (Ailesel Atriyal Fibrilasyon) (SCN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 13, 615377, Autosomal dominant; ATFB13 (Familial atrial fibrillation) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEES2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 14, 615378, Otozomal Dominant; ATFB14 (Ailesel Atriyal Fibrilasyon) (SCN2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 14, 615378, Autosomal dominant; ATFB14 (Familial atrial fibrillation) (SCN2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN2B	SCN2B, ATFB14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 16, 613120, Otozomal Dominant (Brugada Sendromu) (SCN3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 16, 613120, Autosomal dominant (Brugada syndrome) (SCN3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN3B	SCN3B, SCNB3, BRGDA7, ATFB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 16 (ATFB16) [MIM:613120]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. {ECO:0000269|PubMed:20558140, ECO:0000269|PubMed:21054419}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Atriyal Fibrilasyon, Ailesel, 17, 611819, Otozomal Dominant (Ailesel Uzun QT Sendromu) (SCN4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 17, 611819, Autosomal dominant (Familial long QT syndrome) (SCN4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4B	SCN4B, LQT10, ATFB17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 18, 617280, Otozomal Dominant (Ailesel Atriyal Fibrilasyon) (MYL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 18, 617280, Autosomal dominant (Familial atrial fibrillation) (MYL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYL4	MYL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 3, 607554, Otozomal Dominant; ATFB3 (Ailesel Atriyal Fibrilasyon) (KCNQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 3, 607554, Autosomal dominant; ATFB3 (Familial atrial fibrillation) (KCNQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB3, SQT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 4, 611493; ATFB4 (Ailesel Atriyal Fibrilasyon) (KCNE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 4, 611493; ATFB4 (Familial atrial fibrillation) (KCNE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNE2	KCNE2, MiRP1, LQT6, ATFB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 4 (ATFB4) [MIM:611493]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. {ECO:0000269	PubMed:15368194}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 6, 612201, Otozomal Dominant; ATFB6 (Ailesel Atriyal Fibrilasyon) (NPPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 6, 612201, Autosomal dominant; ATFB6 (Familial atrial fibrillation) (NPPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPPA	NPPA, PND, ANP, ATFB6, ATRST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 6 (ATFB6) [MIM:612201]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. {ECO:0000269	PubMed:18614783}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 7, 612240, Otozomal Dominant; ATFB7 (Ailesel Atriyal Fibrilasyon) (KCNA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 7, 612240, Autosomal dominant; ATFB7 (Familial atrial fibrillation) (KCNA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNA5	KCNA5, ATFB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 7 (ATFB7) [MIM:612240]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. {ECO:0000269	PubMed:16772329}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 9, 613980, Otozomal Dominant; ATFB9 (Ailesel Atriyal Fibrilasyon) (KCNJ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial fibrillation, familial, 9, 613980, Autosomal dominant; ATFB9 (Familial atrial fibrillation) (KCNJ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Fibrilasyon, Ailesel, 9, 613980, Otozomal Dominant; ATFB9 (Ailesel Atriyal Fibrilasyon) (MLPA)	Atrial fibrillation, familial, 9, 613980, Autosomal dominant; ATFB9 (Familial atrial fibrillation) (MLPA)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 2, 607941, Otozomal Dominant; ASD2 (Interatriyal Kominikasyon) (GATA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 2, 607941, Autosomal dominant; ASD2 (Interatrial communication) (GATA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 2, 607941, Otozomal Dominant; ASD2 (Interatriyal Kominikasyon) (MLPA)	Atrial septal defect 2, 607941, Autosomal dominant; ASD2 (Interatrial communication) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 3, 614089; ASD3 (İnteratriyal Kominikasyon) (MYH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 3, 614089; ASD3 (Interatrial communication) (MYH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH6	MYH6, ASD3, MYHCA, CMD1EE, CMH14, SSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 4, 611363; ASD4 (İnteratriyal Kominikasyon) (TBX20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 4, 611363; ASD4 (Interatrial communication) (TBX20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX20	TBX20, ASD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial septal defect 4 (ASD4) [MIM:611363]: A congenital heart malformation characterized by incomplete closure of the wall between the atria resulting in blood flow from the left to the right atria. Patients show other heart abnormalities including defects in septation, chamber growth and valvulogenesis. The disease is not associated with defects in the cardiac conduction system or with non-cardiac abnormalities. {ECO:0000269	PubMed:17668378, ECO:0000269	PubMed:19762328}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Atriyal Septal Defekt 5, 612794, Otozomal Dominant; ASD5 (İnteratriyal Kominikasyon) (ACTC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 5, 612794, Autosomal dominant; ASD5 (Interatrial communication) (ACTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTC1	ACTC1, CMD1R, CMH11, ASD5, LVNC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 6, 613087, Otozomal Dominant; ASD6 (İnteratriyal Kominikasyon) (TLL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 6, 613087, Autosomal dominant; ASD6 (Interatrial communication) (TLL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLL1	TLL1, TLL, ASD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial septal defect 6 (ASD6) [MIM:613087]: A congenital heart malformation characterized by incomplete closure of the wall between the atria resulting in blood flow from the left to the right atria. {ECO:0000269	PubMed:18830233}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Atriyal Septal Defekt 7, ± AV İletim Defekti, 108900, Otozomal Dominant; ASD7 (Atriyal Septal Defekt-Atriyoventriküler İletim Defekti Sendromu) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 7, with or without AV conduction defects, 108900, Autosomal dominant; ASD7 (Atrial septal defect-atrioventricular conduction defects syndrome) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 8, 614433, Otozomal Dominant; ASD8 (İnteratriyal Kominikasyon) (CITED2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 8, 614433, Autosomal dominant; ASD8 (Interatrial communication) (CITED2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CITED2	CITED2, MRG1, P35SRJ, VSD2, ASD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 9, 614475, Otozomal Dominant (İnteratriyal Kominikasyon) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial septal defect 9, 614475, Autosomal dominant (Interatrial communication) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Septal Defekt 9, Aut9 (İnteratriyal Kominikasyon) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ATRIAL SEPTAL DEFECT 9; ASD9 (Interatrial communication) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Standstill 2, 615745, Otozomal Resesif; ATRST2 (Atriyal Durma) (NPPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial standstill 2, 615745, Autosomal recessive; ATRST2 (Atrial standstill) (NPPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPPA	NPPA, PND, ANP, ATFB6, ATRST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyal Standstill, Dijenik (GJA5 / SCN5A), 108770, Otozomal Dominant; ATRST1 (Atriyal Duraklaması) (GJA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrial standstill, digenic (GJA5/SCN5A), 108770, Autosomal dominant; ATRST1 (Atrial standstill) (GJA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA5	GJA5, CX40, ATFB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial standstill 1 (ATRST1) [MIM:108770]: A rare arrhythmia characterized by the absence of electrical and mechanical activity in the atria. Electrocardiographically, it is characterized by bradycardia, the absence of P waves, and a junctional narrow complex escape rhythm. {ECO:0000269	PubMed:16790700}. Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry. A rare GJA5 genotype has been detected in combination with a mutation in SCN5A in a large family with atrial standstill.	EDTA Blood Sample
Papüler Lezyonlar-Atrişi, 209500, Otozomal Resesif; APL (Atrişi-Papüler Lezyonlar) (HR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrichia with papular lesions, 209500, Autosomal recessive; APL (Atrichia with papular lesions) (HR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HR	HR, AU, MUHH1, HYPT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 3, 600309, Otozomal Dominant (Atriyoventriküler Kanal Defekti) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrioventricular septal defect 3, 600309, Autosomal dominant (Atrioventricular canal defect) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 3; Avsd3 (Atriyoventriküler Kanal Defekti) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ATRIOVENTRICULAR SEPTAL DEFECT 3; AVSD3 (Atrioventricular canal defect) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 4, 614430, Otozomal Dominant; AVSD4 (Atriyoventriküler Kanal Defekti) (GATA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrioventricular septal defect 4, 614430, Autosomal dominant; AVSD4 (Atrioventricular canal defect) (GATA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 4, 614430, Otozomal Dominant; AVSD4 (Atriyoventriküler Kanal Defekti) (MLPA)	Atrioventricular septal defect 4, 614430, Autosomal dominant; AVSD4 (Atrioventricular canal defect) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 5, 614474, Otozomal Dominant (Atriyoventriküler Kanal Defekti) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrioventricular septal defect 5, 614474, Autosomal dominant (Atrioventricular canal defect) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt 5; Avsd5 (Atriyoventriküler Kanal Defekti) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ATRIOVENTRICULAR SEPTAL DEFECT 5; AVSD5 (Atrioventricular canal defect) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt, Parsiyel, Heterotaksi Sendromu, 606217, Otozomal Dominant; AVSD2 (Atriyoventriküler Kanal Defekti) (CRELD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217, Autosomal dominant; AVSD2 (Atrioventricular canal defect) (CRELD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRELD1	CRELD1, AVSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atriyoventriküler Septal Defekt, Yatkınlık, 2, 606217, Otozomal Dominant; AVSD2 (Atriyoventriküler Kanal Defekti) (CRELD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Atrioventricular septal defect, susceptibility to, 2, 606217, Autosomal dominant; AVSD2 (Atrioventricular canal defect) (CRELD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRELD1	CRELD1, AVSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrioventricular septal defect 2 (AVSD2) [MIM:606217]: A congenital heart malformation characterized by a common atrioventricular junction coexisting with deficient atrioventricular septation. The complete form involves underdevelopment of the lower part of the atrial septum and the upper part of the ventricular septum; the valve itself is also shared. A less severe form, known as ostium primum atrial septal defect, is characterized by separate atrioventricular valvar orifices despite a common junction. {ECO:0000269	PubMed:12632326, ECO:0000269	PubMed:15857420}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.,	EDTA Blood Sample
Dikkat Eksikliği-Hiperaktivite Bozukluğu, 143465, Otozomal Dominant (DRD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Attention deficit-hyperactivity disorder, 143465, Autosomal dominant (DRD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD4	DRD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dikkat Eksikliği-Hiperaktivite Bozukluğu, Yatkınlık, 143465, Otozomal Dominant (DRD5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Attention deficit-hyperactivity disorder, susceptibility to, 143465, Autosomal dominant (DRD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD5	DRD5, DRD1B, DRD1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dikkat Eksikliği-Hiperaktivite Bozukluğu, Yatkınlık, 7, 613003 (TPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Attention deficit-hyperactivity disorder, susceptibility to, 7, 613003 (TPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPH2	TPH2, NTPH, ADHD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Atipik hemolitik-üremik sendrom paneli (CFH, CFHR3, CFHR1, CFHR2, 1q23) (MLPA)	Atypical hemolytic-uremic syndrome panel (CFH, CFHR3, CFHR1, CFHR2, 1q23) (MLPA)	CFH, CFHR3, CFHR1, CFHR2, 1q23	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Atipik hemolitik-üremik sendrom paneli (CFH, CFHR3, CFHR1, CFHR2, 1q23) (MLPA) (Prenatal)	Atypical hemolytic-uremic syndrome panel (CFH, CFHR3, CFHR1, CFHR2, 1q23) (MLPA) (Prenatal)	CFH, CFHR3, CFHR1, CFHR2, 1q23	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atipik Rett sendromu paneli (CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3) (MLPA)	Atypical Rett syndrome panel (CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3) (MLPA)	CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Atipik Rett sendromu paneli (CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3) (MLPA) (Prenatal)	Atypical Rett syndrome panel (CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3) (MLPA) (Prenatal)	CDKL5 Xp22, NTNG1 1p13.3, ARX Xp21.3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atipik Rett sendromu paneli (MEF2C Sq14.3, FOXG1 14q12) (MLPA)	Atypical Rett syndrome panel (MEF2C Sq14.3, FOXG1 14q12) (MLPA)	MEF2C Sq14.3, FOXG1 14q12	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Atipik Rett sendromu paneli (MEF2C 5q14.3, FOXG1 14q12) (MLPA) (Prenatal)	Atypical Rett syndrome panel (MEF2C 5q14.3, FOXG1 14q12) (MLPA) (Prenatal)	MEF2C 5q14.3, FOXG1 14q12	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Au-Kline Sendromu, 616580, Otozomal Dominant; AUKS (Nöro-Gelişimsel Bozukluk-Kraniofasiyal Dismorfizm-Kalp Defekti-Kalça Displazisi Sendromu) (HNRNPK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Au-Kline syndrome, 616580, Autosomal dominant; AUKS (Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome) (HNRNPK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPK	HNRNPK, HNRPK, AUKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Au-Kline Sendromu, 616580, Otozomal Dominant; AUKS (Nöro-Gelişimsel Bozukluk-Kraniofasiyal Dismorfizm-Kalp Defekti-Kalça Displazisi Sendromu) (HNRNPK gen) (Tüm Gen Dizi Analizi) (Prenatal)	Au-Kline syndrome, 616580, Autosomal dominant; AUKS (Neurodevelopmental disorder-craniofacial dysmorphism-cardiac defect-hip dysplasia syndrome) (HNRNPK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HNRNPK	HNRNPK, HNRPK, AUKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İşitsel Nöropati, Otozomal Dominant, 1, 609129, Otozomal Dominant; AUNA1 (Otozomal Dominant Sendromik Olmayan Sensörinöral İşitme Kaybı Tipi DFNA) (DIAPH6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Auditory neuropathy, autosomal dominant, 1, 609129, Autosomal dominant; AUNA1 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (DIAPH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIAPH3	DIAPH3, DIA2, DRF3, AUNA1, NSDAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Auditory neuropathy, autosomal dominant, 1 (AUNA1) [MIM:609129]: A form of sensorineural hearing loss with absent or severely abnormal auditory brainstem response, in the presence of normal cochlear outer hair cell function and normal otoacoustic emissions. Auditory neuropathies result from a lesion in the area including the inner hair cells, connections between the inner hair cells and the cochlear branch of the auditory nerve, the auditory nerve itself and auditory pathways of the brainstem. {ECO:0000269|PubMed:20624953}. Note=The disease is caused by mutations affecting the gene represented in this entry. A disease- causing mutation in the conserved 5'-UTR leads to increased protein expression (PubMed:20624953). {ECO:0000269|PubMed:20624953}.;	EDTA Blood Sample
İşitsel Nöropati, Otozomal Resesif, 1, 601071, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensörinöral İşitme Kaybı Tipi DFNB) (OTOF gen) (Tüm Gen Dizi Analizi) (Postnatal)	Auditory neuropathy, autosomal recessive, 1, 601071, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (OTOF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTOF	OTOF, DFNB9, NSRD9, AUNB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 9 (DFNB9) [MIM:601071]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:10192385, ECO:0000269|PubMed:12127154, ECO:0000269|PubMed:16097006, ECO:0000269|PubMed:16283880, ECO:0000269|PubMed:16371502}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İşitsel Ateşli, Konjenital, 607842, Otozomal Dominant; CAA (Dış Kulak Yolu Aplazisi / Hipoplazi) (TSHZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Aural atresia, congenital, 607842, Autosomal dominant; CAA (External auditory canal aplasia/hypoplasia) (TSHZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHZ1	TSHZ1, TSH1, CAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aurikulospondiler Sendrom 1, 602483, Otozomal Dominant; ARCND1 (Aurikulokondiler Sendromi) (GNAI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Auriculocondylar syndrome 1, 602483, Autosomal dominant; ARCND1 (Auriculocondylar syndrome) (GNAI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAI3	GNAI3, ARCND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Auriculocondylar syndrome 1 (ARCND1) [MIM:602483]: An autosomal dominant craniofacial malformation syndrome characterized by variable mandibular anomalies, including mild to severe micrognathia, temporomandibular joint ankylosis, cleft palate, and a characteristic ear malformation that consists of separation of the lobule from the external ear, giving the appearance of a question mark (question-mark ear). Other frequently described features include prominent cheeks, cupped and posteriorly rotated ears, preauricular tags, and microstomia. {ECO:0000269|PubMed:22560091}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Aurikülokondiler Sendrom 2, 614669, Otozomal Resesif, Otozomal Dominant; ARCND2 (Aurikulokondiler Sendromi) (PLCB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Auriculocondylar syndrome 2, 614669, Autosomal recessive, Autosomal dominant; ARCND2 (Auriculocondylar syndrome) (PLCB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCB4	PLCB4, ARCND2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aurikulokondiler Sendromu 3, 615706, Otozomal Resesif; ARCND3 (Aurikulokondiler Sendromi) (EDN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Auriculocondylar syndrome 3, 615706, Autosomal recessive; ARCND3 (Auriculocondylar syndrome) (EDN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN1	EDN1, ARCND3, QME, HDLCQ7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Auriculocondylar syndrome 3 (ARCND3) [MIM:615706]: A craniofacial malformation syndrome characterized by variable mandibular anomalies, including mild to severe micrognathia, temporomandibular joint ankylosis, cleft palate, and a characteristic ear malformation that consists of separation of the lobule from the external ear, giving the appearance of a question mark (question-mark ear). Other frequently described features include prominent cheeks, cupped and posteriorly rotated ears, preauricular tags, and microstomia. {ECO:0000269|PubMed:24268655}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otizm paneli (15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13) (MLPA)	Autism panel (15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13) (MLPA)	15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal		.	EDTA Blood Sample
Otizm paneli (15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13) (MLPA) (Prenatal)	Autism panel (15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13) (MLPA) (Prenatal)	15q11-13: UBE3A, GABRB3; 16p11; SHANK3 22q13	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık 14A, 611913 (Proksimal 16p11.2 Mikrodelesyon Sendromu) (2200	Autism susceptibility 14A, 611913 (Proximal 16p11.2 microdeletion syndrome) (440	.	DEL16p11.2, C16DEL p11.2, AUTS14A	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Otizm Yatkınlık 14A, 611913 (Proksimal 16p11.2 Mikrodelesyon Sendromu) (Prenatal)	Autism susceptibility 14A, 611913 (Proximal 16p11.2 microdeletion syndrome) (Prenatal)	.	DEL16p11.2, C16DEL p11.2, AUTS14A	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık 15, 612100 (Kortikal Displazi-Fokal Epilepsi Sendromu) (CNTNAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility 15, 612100 (Cortical dysplasia-focal epilepsy syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 15 (AUTS15) [MIM:612100]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Otizm Yatkınlık 15, 612100 (Kortikal Displazi-Fokal Epilepsi Sendromu) (CNTNAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility 15, 612100 (Cortical dysplasia-focal epilepsy syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 15 (AUTS15) [MIM:612100]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık 16, 613410 (SLC9A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility 16, 613410 (SLC9A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC9A9	SLC9A9, AUTS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 16 (AUTS16) [MIM:613410]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. AUTS16 can be associated with epilepsy. {ECO:0000269|PubMed:18621663}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Otizm Yatkınlık 16, 613410 (SLC9A9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility 16, 613410 (SLC9A9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC9A9	SLC9A9, AUTS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 16 (AUTS16) [MIM:613410]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. AUTS16 can be associated with epilepsy. {ECO:0000269|PubMed:18621663}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık 17, 613436 (SHANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility 17, 613436 (SHANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHANK2	SHANK2, CORTBP1, AUTS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 17 (AUTS17) [MIM:613436]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:20473310}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Otizm Yatkınlık 17, 613436 (SHANK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility 17, 613436 (SHANK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHANK2	SHANK2, CORTBP1, AUTS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 17 (AUTS17) [MIM:613436]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:20473310}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık 4, 608636, Otozomal Dominant (15q11q13 Mikroduplikasyon Sendromu) (2200)	Autism susceptibility 4, 608636, Autosomal dominant (15q11q13 microduplication syndrome) (440)	.	AUTS4	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Otizm Yatkınlık 4, 608636, Otozomal Dominant (15q11q13 Mikroduplikasyon Sendromu) (Prenatal)	Autism susceptibility 4, 608636, Autosomal dominant (15q11q13 microduplication syndrome) (Prenatal)	.	AUTS4	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 1, 300425, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (NLGN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility, X-linked 1, 300425, Isolated cases, X-linked, Multifactorial (NLGN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLGN3	NLGN3, ASPGX1, AUTSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 1, 300425, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (NLGN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility, X-linked 1, 300425, Isolated cases, X-linked, Multifactorial (NLGN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLGN3	NLGN3, ASPGX1, AUTSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 2, 300495, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (NLGN4X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility, X-linked 2, 300495, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 2, 300495, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (NLGN4X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility, X-linked 2, 300495, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 3, 300496, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (Rett Sendromu) (MECP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism susceptibility, X-linked 3, 300496, Isolated cases, X-linked, Multifactorial (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 3, 300496, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (Rett Sendromu) (MLPA)	Autism susceptibility, X-linked 3, 300496, Isolated cases, X-linked, Multifactorial (Rett syndrome) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 3, 300496, Sporadik Vakalar, X'e Bağlı, Multifaktöryel (Rett Sendromu) (MECP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism susceptibility, X-linked 3, 300496, Isolated cases, X-linked, Multifactorial (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Otizm Yatkınlık, X'e Bağlı 3, 300496, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (Rett Sendromu) (Prenatal) (MLPA)	Autism susceptibility, X-linked 3, 300496, Isolated cases, X-linked, Multifactorial (Rett syndrome) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRX513, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm, Yatkınlık, 14B, 614671 (Proksimal 16p11.2 Mikroduplikasyon Sendromu) (2200)	Autism, susceptibility to, 14B, 614671 (Proximal 16p11.2 microduplication syndrome) (440)	.	DUP16p11.2, C16DUPp11.2, AUTS14B	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Otizm, Yatkınlık, 14B, 614671 (Proksimal 16p11.2 Mikroduplikasyon Sendromu) (Prenatal)	Autism, susceptibility to, 14B, 614671 (Proximal 16p11.2 microduplication syndrome) (Prenatal)	.	DUP16p11.2, C16DUPp11.2, AUTS14B	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm, Yatkınlık, 18, 615032, Otozomal Dominant (CHD8 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Autism, susceptibility to, 18, 615032, Autosomal dominant (CHD8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHD8	CHD8, DUPLIN, KIAA1564, AUTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 18 (AUTS18) [MIM:615032]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269	PubMed:23160955}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Otizm, Yatkınlık, 18, 615032, Otozomal Dominant (CHD8 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Autism, susceptibility to, 18, 615032, Autosomal dominant (CHD8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHD8	CHD8, DUPLIN, KIAA1564, AUTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 18 (AUTS18) [MIM:615032]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269	PubMed:23160955}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm, Yatkınlık, 19, 615091 (EIF4E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism, susceptibility to, 19, 615091 (EIF4E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF4E	EIF4E, EIF4EL1, AUTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 19 (AUTS19) [MIM:615091]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:19556253}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A heterozygous single-nucleotide insertion has been found in families affected by autism. The variant results in increased promoter activity and is involved in disease pathogenesis through EIF4E deregulation (PubMed:19556253). {ECO:0000269|PubMed:19556253}.	EDTA Blood Sample
Otizm, Yatkınlık, 19, 615091 (EIF4E geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism, susceptibility to, 19, 615091 (EIF4E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF4E	EIF4E, EIF4EL1, AUTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 19 (AUTS19) [MIM:615091]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:19556253}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A heterozygous single-nucleotide insertion has been found in families affected by autism. The variant results in increased promoter activity and is involved in disease pathogenesis through EIF4E deregulation (PubMed:19556253). {ECO:0000269|PubMed:19556253}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm, Yatkınlık, X'e Bağlı 4, 300830, X'e Bağlı Resesif (PTCHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism, susceptibility to, X-linked 4, 300830, X-linked recessive (PTCHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCHD1	PTCHD1, AUTSX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otizm, Yatkınlık, X'e Bağlı 4, 300830, X'e Bağlı Resesif (PTCHD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism, susceptibility to, X-linked 4, 300830, X-linked recessive (PTCHD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTCHD1	PTCHD1, AUTSX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm, Yatkınlık, X'e Bağlı 5, 300847 (RPL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism, susceptibility to, X-linked 5, 300847 (RPL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL10	RPL10, DXS648, QM, AUTSX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism, X-linked 5 (AUTSX5) [MIM:300847]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:16940977, ECO:0000269|PubMed:21567917}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. RPL10 is involved in autism only in rare cases. Two hypomorphic variants affecting the translation process have been found in families with autism spectrum disorders, suggesting that aberrant translation may play a role in disease mechanisms.;	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 5, 300847 (RPL10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism, susceptibility to, X-linked 5, 300847 (RPL10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL10	RPL10, DXS648, QM, AUTSX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism, X-linked 5 (AUTSX5) [MIM:300847]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. {ECO:0000269|PubMed:16940977, ECO:0000269|PubMed:21567917}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. RPL10 is involved in autism only in rare cases. Two hypomorphic variants affecting the translation process have been found in families with autism spectrum disorders, suggesting that aberrant translation may play a role in disease mechanisms.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 6, 300872, X'e Bağlı Resesif (TMLHE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autism, susceptibility to, X-linked 6, 300872, X-linked recessive (TMLHE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMLHE	TMLHE, BBOX2, TMLH, TMLHED, AUTSX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otizm Yatkınlık, X'e Bağlı 6, 300872, X'e Bağlı Resesif (TMLHE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autism, susceptibility to, X-linked 6, 300872, X-linked recessive (TMLHE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMLHE	TMLHE, BBOX2, TMLH, TMLHED, AUTSX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Hastalık, Multisistem, İnfantil Başlangıç, 1, 615952, Otozomal Dominant (Erken Başlangıçlı Multisistem Otoimmün Hastalık, STAT3 İle İlişkili) (STAT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune disease, multisystem, infantile-onset, 1, 615952, Autosomal dominant (STAT3-related early-onset multisystem autoimmune disease) (STAT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT3	STAT3, APRF, HIES, ADMIO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otoimmün Hastalığı, Multisistem, İnfantil Başlangıçlı, 2, 617006, Otozomal Resesif (Kombine İmmün Yetmezlik, ZAP70 Eksikliğine Bağlı) (ZAP70 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune disease, multisystem, infantile-onset, 2, 617006, Autosomal recessive (Combined immunodeficiency due to ZAP70 deficiency) (ZAP70 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZAP70	ZAP70, SRK, ADMIO2, IMD48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otoimmün Hastalık, Multisistem, Fasiyal Dismorfizm, 613385, Otozomal Resesif (Sendromik Multisistem Otoimmün Hastalık, Itch Eksikliğine Bağlı) (ITCH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune disease, multisystem, with facial dysmorphism, 613385, Autosomal recessive (Syndromic multisystem autoimmune disease due to Itch deficiency) (ITCH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITCH	ITCH, AIP4, NAPP1, ADMFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune disease, multisystem, with facial dysmorphism (ADMFD) [MIM:613385]: A disorder characterized by organomegaly, failure to thrive, developmental delay, dysmorphic features and autoimmune inflammatory cell infiltration of the lungs, liver and gut. {ECO:0000269	PubMed:20170897}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün Hastalık, Yatkınlık, 1, 607836, Otozomal Dominant (FOXD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune disease, susceptibility to, 1, 607836, Autosomal dominant (FOXD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXD3	FOXD3, AIS1, VAMAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otoimmün Hastalık, Yatkınlık, 6, 613551 (SIAE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune disease, susceptibility to, 6, 613551 (SIAE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIAE	SIAE, AIS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otoimmün İnterstisyel Akciğer, Eklem ve Böbrek Hastalığı, 616414, Otozomal Dominant; AILJK (Otoimmün İnterstisyel Akciğer Hastalığı-Artritis Sendromu) (COPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune interstitial lung, joint, and kidney disease, 616414, Autosomal dominant; AILJK (Autoimmune interstitial lung disease-arthritis syndrome) (COPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COPA	COPA, AILJK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune interstitial lung, joint, and kidney disease (AILJK) [MIM:616414]: An autoimmune disease characterized by inflammatory arthritis, interstitial lung disease, and immune complex-mediated renal disease. {ECO:0000269	PubMed:25894502}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün İnterstisyel Akciğer, Eklem ve Böbrek Hastalığı, 616414, Otozomal Dominant; AILJK (Otoimmün İnterstisyel Akciğer Hastalığı-Artritis Sendromu) (COPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune interstitial lung, joint, and kidney disease, 616414, Autosomal dominant; AILJK (Autoimmune interstitial lung disease-arthritis syndrome) (COPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COPA	COPA, AILJK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune interstitial lung, joint, and kidney disease (AILJK) [MIM:616414]: An autoimmune disease characterized by inflammatory arthritis, interstitial lung disease, and immune complex-mediated renal disease. {ECO:0000269	PubMed:25894502}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, 601859, Otozomal Dominant; ALPS (Otoimmün Lenfoproliferatif Sendromi) (FAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, 601859, Autosomal dominant; ALPS (Autoimmune lymphoproliferative syndrome) (FAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAS	FAS, TNFRSF6, APT1, CD95, ALPS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 1A (ALPS1A) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:10090885, ECO:0000269|PubMed:10340403, ECO:0000269|PubMed:10515860, ECO:0000269|PubMed:11418480, ECO:0000269|PubMed:17336828, ECO:0000269|PubMed:7540117, ECO:0000269|PubMed:8929361, ECO:0000269|PubMed:9028321, ECO:0000269|PubMed:9028957, ECO:0000269|PubMed:9322534, ECO:0000269|PubMed:9821419, ECO:0000269|PubMed:9927496}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, 601859, Otozomal Dominant; ALPS (Otoimmün Lenfoproliferatif Sendromi) (FAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, 601859, Autosomal dominant; ALPS (Autoimmune lymphoproliferative syndrome) (FAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAS	FAS, TNFRSF6, APT1, CD95, ALPS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune lymphoproliferative syndrome 1A (ALPS1A) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:10090885, ECO:0000269|PubMed:10340403, ECO:0000269|PubMed:10515860, ECO:0000269|PubMed:11418480, ECO:0000269|PubMed:17336828, ECO:0000269|PubMed:7540117, ECO:0000269|PubMed:8929361, ECO:0000269|PubMed:9028321, ECO:0000269|PubMed:9028957, ECO:0000269|PubMed:9322534, ECO:0000269|PubMed:9821419, ECO:0000269|PubMed:9927496}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Otoimmün Lenfoproliferatif Sendrom, Tip 1B, 601859, Otozomal Dominant (Otoimmün Lenfoproliferatif Sendromi) (FASLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, type IB, 601859, Autosomal dominant (Autoimmune lymphoproliferative syndrome) (FASLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FASLG	FASLG, TNFSF6, APT1LG1, FASL, ALPS1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 1B (ALPS1B) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:8787672}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 1B, 601859, Otozomal Dominant (Otoimmün Lenfoproliferatif Sendromi) (FASLG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, type IB, 601859, Autosomal dominant (Autoimmune lymphoproliferative syndrome) (FASLG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FASLG	FASLG, TNFSF6, APT1LG1, FASL, ALPS1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune lymphoproliferative syndrome 1B (ALPS1B) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:8787672}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 2, 603909, Otozomal Dominant; ALPS2A (Otoimmün Lenfoproliferatif Sendromi) (CASP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, type II, 603909, Autosomal dominant; ALPS2A (Autoimmune lymphoproliferative syndrome) (CASP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP10	CASP10, MCH4, ALPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 2A (ALPS2A) [MIM:603909]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:10412980, ECO:0000269|PubMed:16446975}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 2, 603909, Otozomal Dominant; ALPS2A (Otoimmün Lenfoproliferatif Sendromi) (CASP10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, type II, 603909, Autosomal dominant; ALPS2A (Autoimmune lymphoproliferative syndrome) (CASP10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASP10	CASP10, MCH4, ALPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune lymphoproliferative syndrome 2A (ALPS2A) [MIM:603909]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:10412980, ECO:0000269|PubMed:16446975}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 2B, 607271, Otozomal Resesif (Otoimmün Lenfoproliferatif Sendrom-Tekrarlayan Viral Enfeksiyonlar) (CASP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, type IIB, 607271, Autosomal recessive (Autoimmune lymphoproliferative syndrome with recurrent viral infections) (CASP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP8	CASP8, MCH5, ALPS2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caspase-8 deficiency (CASP8D) [MIM:607271]: Disorder resembling autoimmune lymphoproliferative syndrome (ALPS). It is characterized by lymphadenopathy, splenomegaly, and defective CD95-induced apoptosis of peripheral blood lymphocytes (PBLs). It leads to defects in activation of T-lymphocytes, B-lymphocytes, and natural killer cells leading to immunodeficiency characterized by recurrent sinopulmonary and herpes simplex virus infections and poor responses to immunization. {ECO:0000269|PubMed:12353035}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 2B, 607271, Otozomal Resesif (Otoimmün Lenfoproliferatif Sendrom-Tekrarlayan Viral Enfeksiyonlar) (CASP8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, type IIB, 607271, Autosomal recessive (Autoimmune lymphoproliferative syndrome with recurrent viral infections) (CASP8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASP8	CASP8, MCH5, ALPS2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caspase-8 deficiency (CASP8D) [MIM:607271]: Disorder resembling autoimmune lymphoproliferative syndrome (ALPS). It is characterized by lymphadenopathy, splenomegaly, and defective CD95-induced apoptosis of peripheral blood lymphocytes (PBLs). It leads to defects in activation of T-lymphocytes, B-lymphocytes, and natural killer cells leading to immunodeficiency characterized by recurrent sinopulmonary and herpes simplex virus infections and poor responses to immunization. {ECO:0000269|PubMed:12353035}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 3, 615559, Otozomal Resesif; ALPS3 (Otoimmün Lenfoproliferatif Sendromi) (PRKCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, type III, 615559, Autosomal recessive; ALPS3 (Autoimmune lymphoproliferative syndrome) (PRKCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCD	PRKCD, CVID9, ALPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 3 (ALPS3) [MIM:615559]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. CVID9 patients have B-cell deficiency and severe autoimmunity. {ECO:0000269|PubMed:23319571}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 3, 615559, Otozomal Resesif; ALPS3 (Otoimmün Lenfoproliferatif Sendromi) (PRKCD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, type III, 615559, Autosomal recessive; ALPS3 (Autoimmune lymphoproliferative syndrome) (PRKCD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKCD	PRKCD, CVID9, ALPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune lymphoproliferative syndrome 3 (ALPS3) [MIM:615559]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. CVID9 patients have B-cell deficiency and severe autoimmunity. {ECO:0000269|PubMed:23319571}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 5, 616100, Otozomal Dominant; ALPS5 (Otoimmün Lenfoproliferatif Sendrom, CTLA4 Haploinsuffiency Bağlı) (CTLA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune lymphoproliferative syndrome, type V, 616100, Autosomal dominant; ALPS5 (Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency) (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Otoimmün Lenfoproliferatif Sendrom, Tip 5, 616100, Otozomal Dominant; ALPS5 (Otoimmün Lenfoproliferatif Sendrom, CTLA4 Haploinsuffiency Bağlı) (CTLA4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune lymphoproliferative syndrome, type V, 616100, Autosomal dominant; ALPS5 (Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency) (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Poliendokrinopati Sendromu, Tip 1, ± Otozomal Dominant, Geri Dönüşümlü Metaflzyal Displazi, 240300, Otozomal Resesif; APS1 (Otoimmün Poliendokrinopati Tip 1) (AIRE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia, 240300, Autosomal recessive, Autosomal dominant; APS1 (Autoimmune polyendocrinopathy type 1) (AIRE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIRE	AIRE, APECED, APS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune polyendocrine syndrome 1, with or without reversible metaphyseal dysplasia (APS1) [MIM:240300]: A rare disease characterized by the combination of chronic mucocutaneous candidiasis, hypoparathyroidism and Addison disease. Symptoms of mucocutaneous candidiasis manifest first, followed by hypotension or fatigue occurring as a result of Addison disease. APS1 is associated with other autoimmune disorders including diabetes mellitus, vitiligo, alopecia, hepatitis, pernicious anemia and primary hypothyroidism. {ECO:0000269|PubMed:10677297, ECO:0000269|PubMed:11275943, ECO:0000269|PubMed:11524731, ECO:0000269|PubMed:11524733, ECO:0000269|PubMed:11600535, ECO:0000269|PubMed:11836330, ECO:0000269|PubMed:12050215, ECO:0000269|PubMed:12173802, ECO:0000269|PubMed:12625412, ECO:0000269|PubMed:15712268, ECO:0000269|PubMed:9398839, ECO:0000269|PubMed:9888391}. Note=The disease is caused by mutations affecting the gene represented in this entry. Most of the mutations alter the nucleus-cytoplasm distribution of AIRE and disturb its association with nuclear dots and cytoplasmic filaments. Most of the mutations also decrease transactivation of the protein. The HSR domain is responsible for the homomultimerization activity of AIRE. All the missense mutations of the HSR and the SAND domains decrease this activity, but those in other domains do not. The AIRE protein is present in soluble high-molecular-weight complexes. Mutations in the HSR domain and deletion of PHD zinc fingers disturb the formation of these complexes. {PubMed:14974083}.	EDTA Blood Sample
Otoimmün Poliendokrinopati Sendromu, Tip 1, ± Otozomal Dominant, Geri Dönüşümlü Metaflzyal Displazi, 240300, Otozomal Resesif; APS1 (Otoimmün Poliendokrinopati Tip 1) (AIRE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoimmune polyendocrinopathy syndrome, type I, with or without reversible metaphyseal dysplasia, 240300, Autosomal recessive, Autosomal dominant; APS1 (Autoimmune polyendocrinopathy type 1) (AIRE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AIRE	AIRE, APECED, APS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autoimmune polyendocrine syndrome 1, with or without reversible metaphyseal dysplasia (APS1) [MIM:240300]: A rare disease characterized by the combination of chronic mucocutaneous candidiasis, hypoparathyroidism and Addison disease. Symptoms of mucocutaneous candidiasis manifest first, followed by hypotension or fatigue occurring as a result of Addison disease. APS1 is associated with other autoimmune disorders including diabetes mellitus, vitiligo, alopecia, hepatitis, pernicious anemia and primary hypothyroidism. {ECO:0000269|PubMed:10677297, ECO:0000269|PubMed:11275943, ECO:0000269|PubMed:11524731, ECO:0000269|PubMed:11524733, ECO:0000269|PubMed:11600535, ECO:0000269|PubMed:11836330, ECO:0000269|PubMed:12050215, ECO:0000269|PubMed:12173802, ECO:0000269|PubMed:12625412, ECO:0000269|PubMed:15712268, ECO:0000269|PubMed:9398839, ECO:0000269|PubMed:9888391}. Note=The disease is caused by mutations affecting the gene represented in this entry. Most of the mutations alter the nucleus-cytoplasm distribution of AIRE and disturb its association with nuclear dots and cytoplasmic filaments. Most of the mutations also decrease transactivation of the protein. The HSR domain is responsible for the homomultimerization activity of AIRE. All the missense mutations of the HSR and the SAND domains decrease this activity, but those in other domains do not. The AIRE protein is present in soluble high-molecular-weight complexes. Mutations in the HSR domain and deletion of PHD zinc fingers disturb the formation of these complexes. {PubMed:14974083}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoimmün Tiroid Hastalığı, Yatkınlık, 3, 608175 (ZFAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune thyroid disease, susceptibility to, 3, 608175 (ZFAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFAT	ZFAT1, ZNF406, AITD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Otoimmün Tiroid Hastalığı, Yatkınlık, 3, 608175 (Ailesel Tiroid Dishormonogenezis) (TG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoimmune thyroid disease, susceptibility to, 3, 608175 (Familial thyroid dyshormonogenesis) (TG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TG	TG, AITD3, TDH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroid dyshormonogenesis 3 (TDH3) [MIM:274700]: A disorder due to thyroid dyshormonogenesis, causing large goiters of elastic and soft consistency in the majority of patients. Although the degree of thyroid dysfunction varies considerably among patients with defective thyroglobulin synthesis, patients usually have a relatively high serum free triiodothyronine (T3) concentration with disproportionately low free tetraiodothyronine (T4) level. The maintenance of relatively high free T3 levels prevents profound tissue hypothyroidism except in brain and pituitary, which are dependent on T4 supply, resulting in neurologic and intellectual defects in some cases. {ECO:0000269|PubMed:10199792, ECO:0000269|PubMed:16477365, ECO:0000269|PubMed:17244789, ECO:0000269|PubMed:17532758, ECO:0000269|PubMed:19509106}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Otoinflamasyon, Artrit ve Diseratozis, 617388, Otozomal Resesif, Otozomal Dominant; AIADK (Kornea İntraepiteliyal Diskeratozisi-Palmoplantar Hiperkeratoz-Laringeal Diskeratozis Sendromu) (NLRP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammation with arthritis and dyskeratosis, 617388, Autosomal recessive, Autosomal dominant; AIADK (Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome) (NLRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP1	NLRP1, NALP1, KIAA0926, DEFCAP, CARD7, SLEV1, VAMAS1, MSPC, AIADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitiligo-associated multiple autoimmune disease 1 (VAMAS1) [MIM:606579]: A disorder characterized by the association of vitiligo with several autoimmune and autoinflammatory diseases including autoimmune thyroid disease, rheumatoid arthritis and systemic lupus erythematosus. [ECO:0000269	PubMed:17377159]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Otoinflamasyon, Artrit ve Diseratozis, 617388, Otozomal Resesif, Otozomal Dominant; AIADK (Kornea İntraepiteliyal Diskeratozisi-Palmoplantar Hiperkeratoz-Laringeal Diskeratozis Sendromu) (NLRP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoinflammation with arthritis and dyskeratosis, 617388, Autosomal recessive, Autosomal dominant; AIADK (Corneal intraepithelial dyskeratosis-palmoplantar hyperkeratosis-laryngeal dyskeratosis syndrome) (NLRP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLRP1	NLRP1, NALP1, KIAA0926, DEFCAP, CARD7, SLEV1, VAMAS1, MSPC, AIADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Vitiligo-associated multiple autoimmune disease 1 (VAMAS1) [MIM:606579]: A disorder characterized by the association of vitiligo with several autoimmune and autoinflammatory diseases including autoimmune thyroid disease, rheumatoid arthritis and systemic lupus erythematosus. [ECO:0000269	PubMed:17377159]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoinflamasyon, İnfantil Enterokolit 616050, Otozomal Dominant; AFEC (Periyodik Ateş-İnfantil Enterokolit-Otoinflamatuar Sendrom) (NLRC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammation with infantile enterocolitis, 616050, Autosomal dominant; AIFEC (Periodic fever-infantile enterocolitis-autoinflammatory syndrome) (NLRC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRC4	NLRC4, CARD12, CLAN, IPAF, AIFEC, FCAS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoinflammation with infantile enterocolitis (AIFEC) [MIM:616050]: An autosomal dominant disorder characterized by neonatal-onset enterocolitis, periodic fever, and fatal or near-fatal episodes of autoinflammation. Affected individuals tend to have poor overall growth and gastrointestinal symptoms in infancy, recurrent febrile episodes with splenomegaly, and sometimes hematologic disturbances, arthralgias, or myalgias. [ECO:0000269	PubMed:25217959, ECO:0000269	PubMed:25217960]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otoinflamasyon, Antikor Eksikliği ve İmmün Disregülasyon Sendromu, 614878, Otozomal Dominant (Otoinflamasyon-PLCG2 İlişkili Antikor Eksikliği-İmmün Disregülasyon) (PLCG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878, Autosomal dominant (Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation) (PLCG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCG2	PLCG2, FCAS3, APLAID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial cold autoinflammatory syndrome 3 (FCAS3) [MIM:614468]: An autosomal dominant immune disorder characterized by the development of cutaneous urticaria, erythema, and pruritis in response to cold exposure. Affected individuals have variable additional immunologic defects, including antibody deficiency, decreased numbers of B-cells, defective B-cells, increased susceptibility to infection, and increased risk of autoimmune disorders. [ECO:0000269	PubMed:22236196]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otoinflamasyon, Antikor Eksikliği ve İmmün Disregülasyon Sendromu, 614878, Otozomal Dominant (Otoinflamasyon-PLCG2 İlişkili Antikor Eksikliği-İmmün Disregülasyon) (PLCG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878, Autosomal dominant (Autoinflammation-PLCG2-associated antibody deficiency-immune dysregulation) (PLCG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLCG2	PLCG2, FCAS3, APLAID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial cold autoinflammatory syndrome 3 (FCAS3) [MIM:614468]: An autosomal dominant immune disorder characterized by the development of cutaneous urticaria, erythema, and pruritis in response to cold exposure. Affected individuals have variable additional immunologic defects, including antibody deficiency, decreased numbers of B-cells, defective B-cells, increased susceptibility to infection, and increased risk of autoimmune disorders. [ECO:0000269	PubMed:22236196]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otoinflamasyon, Lipodistrofi ve Dermatoz Sendromu, 256040, Otozomal Resesif; ALDD (Nakajo-Nishimura Sendromu) (PSMB8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammation, lipodystrophy, and dermatosis syndrome, 256040, Autosomal recessive; ALDD (Nakajo-Nishimura syndrome) (PSMB8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSMB8	PSMB8, LMP7, RING10, JMP, NKJO, ALDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nakajo syndrome (NKJO) [MIM:256040]: An autosomal recessive autoinflammatory disorder characterized by early childhood onset of recurrent fever, joint stiffness and severe contractures of the hands and feet, and erythematous skin lesions with subsequent development of lipodystrophy and laboratory evidence of immune dysregulation. Accompanying features may include muscle weakness and atrophy, hepatosplenomegaly, and microcytic anemia. [ECO:0000269	PubMed:21129723, ECO:0000269	PubMed:21852578, ECO:0000269	PubMed:21881205]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otoinflamasyon, Panikülit, ve Dermatoz Sendromu, 617099, Otozomal Resesif; AIPDS (OTULIN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammation, panniculitis, and dermatosis syndrome, 617099, Autosomal recessive; AIPDS (OTULIN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTULIN	OTULIN, FAM105B, GUIM, AIPDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otoinflamatuvar Sendrom, Ailesel, Behcet, Benzeri, 616744, Otozomal Dominant; AISBL (Kalıtsal Pediatrik Behçet Benzeri Hastalık) (TNFAIP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autoinflammatory syndrome, familial, Behcet-like, 616744, Autosomal dominant; AISBL (Hereditary pediatric Behçet-like disease) (TNFAIP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFAIP3	TNFAIP3, A20, OTUD7C, AISBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Otonom Sinir Sistemi İşlev Bozukluğu (DRD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Autonomic nervous system dysfunction (DRD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD4	DRD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Femur Başı Avasküler Nekrozu, Primer, 2, 617383, Otozomal Dominant; ANFH2 (Ailesel Femur Başı Avasküler Nekrozu) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Avascular necrosis of femoral head, primary, 2, 617383, Autosomal dominant; ANFH2 (Familial avascular necrosis of femoral head) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Femur Başının Avasküler Nekrozu, 608805, Otozomal Dominant; ANFH1 (Ailesel Femur Başı Avasküler Nekrozu) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Avascular necrosis of the femoral head, 608805, Autosomal dominant; ANFH1 (Familial avascular necrosis of femoral head) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Femur Başının Avasküler Nekrozu, 608805, Otozomal Dominant; ANFH1 (Ailesel Femur Başı Avasküler Nekrozu) (MLPA)	Avascular necrosis of the femoral head, 608805, Autosomal dominant; ANFH1 (Familial avascular necrosis of femoral head) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 1, 180500, Otozomal Dominant; RIEG1 (Axenfeld-Rieger Sendromu) (PITX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Axenfeld-Rieger syndrome, type 1, 180500, Autosomal dominant; RIEG1 (Axenfeld-Rieger syndrome) (PITX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 1, 180500, Otozomal Dominant; RIEG1 (Axenfeld-Rieger Sendromu) (MLPA)	Axenfeld-Rieger syndrome, type 1, 180500, Autosomal dominant; RIEG1 (Axenfeld-Rieger syndrome) (MLPA)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 1, 180500, Otozomal Dominant; RIEG1 (Axenfeld-Rieger Sendromu) (PITX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Axenfeld-Rieger syndrome, type 1, 180500, Autosomal dominant; RIEG1 (Axenfeld-Rieger syndrome) (PITX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 1, 180500, Otozomal Dominant; RIEG1 (Axenfeld-Rieger Sendromu) (Prenatal) (MLPA)	Axenfeld-Rieger syndrome, type 1, 180500, Autosomal dominant; RIEG1 (Axenfeld-Rieger syndrome) (Prenatal) (MLPA)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 3, 602482, Otozomal Dominant; RIEG3 (Axenfeld-Rieger Sendromu) (FOXC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Axenfeld-Rieger syndrome, type 3, 602482, Autosomal dominant; RIEG3 (Axenfeld-Rieger syndrome) (FOXC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 3, 602482, Otozomal Dominant; RIEG3 (Axenfeld-Rieger Sendromu) (MLPA)	Axenfeld-Rieger syndrome, type 3, 602482, Autosomal dominant; RIEG3 (Axenfeld-Rieger syndrome) (MLPA)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 3, 602482, Otozomal Dominant; RIEG3 (Axenfeld-Rieger Sendromu) (FOXC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Axenfeld-Rieger syndrome, type 3, 602482, Autosomal dominant; RIEG3 (Axenfeld-Rieger syndrome) (FOXC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Axenfeld-Rieger Sendromu, Tip 3, 602482, Otozomal Dominant; RIEG3 (Axenfeld-Rieger Sendromu) (Prenatal) (MLPA)	Axenfeld-Rieger syndrome, type 3, 602482, Autosomal dominant; RIEG3 (Axenfeld-Rieger syndrome) (Prenatal) (MLPA)	FOXC1	FOXC1, FKHL7, FREAC3, IRID1, RIEG3, ASGD3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ayme-Gripp Sendromu, 601088, Otozomal Dominant; AYGRP (Ayme-Gripp Sendromu) (MAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ayme-Gripp syndrome, 601088, Autosomal dominant; AYGRP (Aymé-Gripp syndrome) (MAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAF	MAF, CCA4, CTRCT21, AYGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ayme-Gripp syndrome (AYGRP) [MIM:601088]: A multisystem disorder characterized by congenital cataracts, sensorineural deafness, intellectual disability, seizures, brachycephaly, distinctive flat facial appearance, skeletal anomalies, mammary gland hypoplasia, and reduced growth. [ECO:0000269	PubMed:25865493]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Ayme-Gripp Sendromu, 601088, Otozomal Dominant; AYGRP (Ayme-Gripp Sendromu) (MAF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ayme-Gripp syndrome, 601088, Autosomal dominant; AYGRP (Aymé-Gripp syndrome) (MAF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAF	MAF, CCA4, CTRCT21, AYGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ayme-Gripp syndrome (AYGRP) [MIM:601088]: A multisystem disorder characterized by congenital cataracts, sensorineural deafness, intellectual disability, seizures, brachycephaly, distinctive flat facial appearance, skeletal anomalies, mammary gland hypoplasia, and reduced growth. [ECO:0000269	PubMed:25865493]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
B-Hücre Artışı, NF-KB ve T-Hücreli Anerjisi 616452, Otozomal Dominant; BENTA (BENTA Hastalığı) (CARD11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	B-cell expansion with NFKB and T-cell anergy, 616452, Autosomal dominant; BENTA (BENTA disease) (CARD11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARD11	CARD11, CARMA1, BIMP3, PPBL, BENTA, IMD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B Hücreli Non-Hodgkin Lenfoma, Yüksek Gradeli (BCL7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	B-cell non-Hodgkin lymphoma, high-grade (BCL7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL7A	BCL7A, BCL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Chromosomal aberrations involving BCL7A may be a cause of B-cell non-Hodgkin lymphoma. Three-way translocation t(8;14;12)(q24.1;q32.3;p24.1) with MYC and with immunoglobulin gene regions. [ECO:0000269	PubMed:8605326]..	EDTA Blood Sample
Bakteriyemi, Karşı Koruma, 614382 (Tüberküloz) (TIRAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bacteremia, protection against, 614382 (Tuberculosis) (TIRAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIRAP	TIRAP, BACTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bakteriyemi, Yatkınlık, 614383 (CISH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bacteremia, susceptibility to, 614383 (CISH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CISH	CISH, BACTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bainbridge-Ropers Sendromu, 615485 (Ağır Beslenme Zorlukları-Gelişme Gerilliği-Mikrosefali,ASXL3 Eksikliği Sendromuna Bağlı) (ASXL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bainbridge-Ropers syndrome, 615485 (Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome) (ASXL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASXL3	ASXL3, KIAA1713, BRPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bainbridge-Ropers Sendromu, 615485 (Ağır Beslenme Zorlukları-Gelişme Gerilliği-Mikrosefali,ASXL3 Eksikliği Sendromuna Bağlı) (ASXL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bainbridge-Ropers syndrome, 615485 (Severe feeding difficulties-failure to thrive-microcephaly due to ASXL3 deficiency syndrome) (ASXL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASXL3	ASXL3, KIAA1713, BRPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Baller-Gerold Sendromu, 218600, Otozomal Resesif; BGS (Baller-Gerold Sendromu) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Baller-Gerold syndrome, 218600, Autosomal recessive; BGS (Baller-Gerold syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. [ECO:0000269	PubMed:15964893]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Baller-Gerold Sendromu, 218600, Otozomal Resesif; BGS (Baller-Gerold Sendromu) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Baller-Gerold syndrome, 218600, Autosomal recessive; BGS (Baller-Gerold syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. [ECO:0000269	PubMed:15964893]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Bamforth-Lazarus Sendromu, 241850, Otozomal Resesif (Bamforth-Lazarus Sendromu) (FOXE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bamforth-Lazarus syndrome, 241850, Autosomal recessive (Bamforth-Lazarus syndrome) (FOXE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. (ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Bamforth-Lazarus Sendromu, 241850, Otozomal Resesif (Bamforth-Lazarus Sendromu) (MLPA)	Bamforth-Lazarus syndrome, 241850, Autosomal recessive (Bamforth-Lazarus syndrome) (MLPA)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. (ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Bamforth-Lazarus Sendromu, 241850, Otozomal Resesif (Bamforth-Lazarus Sendromu) (FOXE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bamforth-Lazarus syndrome, 241850, Autosomal recessive (Bamforth-Lazarus syndrome) (FOXE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. (ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bamforth-Lazarus Sendromu, 241850, Otozomal Resesif (Bamforth-Lazarus Sendromu) (Prenatal) (MLPA)	Bamforth-Lazarus syndrome, 241850, Autosomal recessive (Bamforth-Lazarus syndrome) (Prenatal) (MLPA)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. (ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bant Heterotopi, 600348, Otozomal Resesif; BH (Subkortikal Bant Heterotopisi) (EML1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Band heterotopia, 600348, Autosomal recessive; BH (Subcortical band heterotopia) (EML1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EML1	EML1, EMAPL, EMAP, BH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Mutations in this gene are associated with atypical heterotopia, epilepsy and mental retardation. Patients present giant bilateral periventricular and ribbon-like subcortical heterotopia with polymicrogyria and agenesis of the corpus callosum.	EDTA Blood Sample
Bant Heterotopi, 600348, Otozomal Resesif; BH (Subkortikal Bant Heterotopisi) (EML1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Band heterotopia, 600348, Autosomal recessive; BH (Subcortical band heterotopia) (EML1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EML1	EML1, EMAPL, EMAP, BH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Mutations in this gene are associated with atypical heterotopia, epilepsy and mental retardation. Patients present giant bilateral periventricular and ribbon-like subcortical heterotopia with polymicrogyria and agenesis of the corpus callosum.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bant Benzeri kalsifikasyon, Azalmış Girasyon ve Polimikrogiri, 251290, Otozomal Resesif (Konjenital Intrauterin Enfeksiyon Benzeri Sendrom (Psödo-TORCH Sendromu)) (OCLN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Band-like calcification with simplified gyration and polymicrogyria, 251290, Autosomal recessive (Congenital intrauterine infection-like syndrome) (OCLN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCLN	OCLN, BLCPMG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Band-like calcification with simplified gyration and polymicrogyria (BLCPMG) [MIM:251290]: A neurologic disorder with characteristic clinical and neuroradiologic features that mimic intrauterine TORCH infection in the absence of evidence of infection. Affected individuals have congenital microcephaly, intracranial calcifications, and severe developmental delay. (ECO:0000269	PubMed:20727516). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Bant Benzeri kalsifikasyon, Azalmış Girasyon ve Polimikrogiri, 251290, Otozomal Resesif (Konjenital Intrauterin Enfeksiyon Benzeri Sendrom (Psödo-TORCH Sendromu)) (OCLN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Band-like calcification with simplified gyration and polymicrogyria, 251290, Autosomal recessive (Congenital intrauterine infection-like syndrome) (OCLN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OCLN	OCLN, BLCPMG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Band-like calcification with simplified gyration and polymicrogyria (BLCPMG) [MIM:251290]: A neurologic disorder with characteristic clinical and neuroradiologic features that mimic intrauterine TORCH infection in the absence of evidence of infection. Affected individuals have congenital microcephaly, intracranial calcifications, and severe developmental delay. (ECO:0000269	PubMed:20727516). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bannayan-Riley-Ruvalcaba Sendromu, 153480, Otozomal Dominant; BRRS (Bannayan-Riley-Ruvalcaba Sendromu) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bannayan-Riley-Ruvalcaba syndrome, 153480, Autosomal dominant; BRRS (Bannayan-Riley-Ruvalcaba syndrome) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. (ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bannayan-Riley-Ruvalcaba Sendromu, 153480, Otozomal Dominant; BRRS (Bannayan-Riley-Ruvalcaba Sendromu) (MLPA)	Bannayan-Riley-Ruvalcaba syndrome, 153480, Autosomal dominant; BRRS (Bannayan-Riley-Ruvalcaba syndrome) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. (ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bannayan-Riley-Ruvalcaba Sendromu, 153480, Otozomal Dominant; BRRS (Bannayan-Riley-Ruvalcaba Sendromu) (PTEN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bannayan-Riley-Ruvalcaba syndrome, 153480, Autosomal dominant; BRRS (Bannayan-Riley-Ruvalcaba syndrome) (PTEN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. (ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bannayan-Riley-Ruvalcaba Sendromu, 153480, Otozomal Dominant; BRRS (Bannayan-Riley-Ruvalcaba Sendromu) (Prenatal) (MLPA)	Bannayan-Riley-Ruvalcaba syndrome, 153480, Autosomal dominant; BRRS (Bannayan-Riley-Ruvalcaba syndrome) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. (ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Baraitser-Winter Sendromu 1, 243310, Otozomal Dominant; BRWS1 (Gelişimsel Malformasyonlar-Sağırlık-Distoni Sendromu) (ACTB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Baraitser-Winter syndrome 1, 243310, Autosomal dominant; BRWS1 (Developmental malformations-deafness-dystonia syndrome) (ACTB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTB	ACTB, BRWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Baraitser-Winter syndrome 1 (BRWS1) [MIM:243310]: A rare developmental disorder characterized by the combination of congenital ptosis, high-arched eyebrows, hypertelorism, ocular colobomata, and a brain malformation consisting of anterior- predominant lissencephaly. Other typical features include postnatal short stature and microcephaly, intellectual disability, seizures, and hearing loss. (ECO:0000269	PubMed:22366783). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Baraitser-Winter Sendromu 1, 243310, Otozomal Dominant; BRWS1 (Gelişimsel Malformasyonlar-Sağırlık-Distoni Sendromu) (ACTB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Baraitser-Winter syndrome 1, 243310, Autosomal dominant; BRWS1 (Developmental malformations-deafness-dystonia syndrome) (ACTB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTB	ACTB, BRWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Baraitser-Winter syndrome 1 (BRWS1) [MIM:243310]: A rare developmental disorder characterized by the combination of congenital ptosis, high-arched eyebrows, hypertelorism, ocular colobomata, and a brain malformation consisting of anterior- predominant lissencephaly. Other typical features include postnatal short stature and microcephaly, intellectual disability, seizures, and hearing loss. (ECO:0000269	PubMed:22366783). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Baraitser-Winter Sendromu 2, 614583, Otozomal Dominant; BRWS2 (Baraitser-Winter Sendromu) (ACTG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Baraitser-Winter syndrome 2, 614583, Autosomal dominant; BRWS2(Baraitser-Winter syndrome) (ACTG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTG1	ACTG1, DFNA20, DFNA26, BRWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Baraitser-Winter Sendromu 2, 614583, Otozomal Dominant; BRWS2 (Baraitser-Winter Sendromu) (ACTG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Baraitser-Winter syndrome 2, 614583, Autosomal dominant; BRWS2(Baraitser-Winter syndrome) (ACTG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTG1	ACTG1, DFNA20, DFNA26, BRWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Barber-Say Sendromu, 209885, Otozomal Dominant; BBRSAY (Barber-Say Sendromu) (TWIST2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Barber-Say syndrome, 209885, Autosomal dominant; BBRSAY (Barber-Say syndrome) (TWIST2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST2	TWIST2, DERMO1, SETLS5, FFDD3, BBRSAY, AMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Barber-Say Sendromu, 209885, Otozomal Dominant; BBRSAY (Barber-Say Sendromu) (TWIST2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Barber-Say syndrome, 209885, Autosomal dominant; BBRSAY (Barber-Say syndrome) (TWIST2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST2	TWIST2, DERMO1, SETLS5, FFDD3, BBRSAY, AMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu 1, 209900, Otozomal Resesif, Digenik Resesif, BBS1 (Bardet-Biedl Sendromu) (BBS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 1, 209900, Autosomal recessive, Digenic recessive, BBS1 (Bardet-Biedl syndrome) (BBS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS1	BBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 1 (BBS1) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:12118255, ECO:0000269|PubMed:12524598, ECO:0000269|PubMed:12567324, ECO:0000269|PubMed:12677556, ECO:0000269|PubMed:12920096, ECO:0000269|PubMed:15770229, ECO:0000269|PubMed:21052717, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu 1, 209900, Otozomal Resesif, Digenik Resesif, BBS1 (Bardet-Biedl Sendromu) (Prenatal) (BBS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 1, 209900, Autosomal recessive, Digenic recessive, BBS1 (Bardet-Biedl syndrome) (BBS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS1	BBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 1 (BBS1) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:12118255, ECO:0000269|PubMed:12524598, ECO:0000269|PubMed:12567324, ECO:0000269|PubMed:12677556, ECO:0000269|PubMed:12920096, ECO:0000269|PubMed:15770229, ECO:0000269|PubMed:21052717, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Bardet-Biedl Sendromu 1, Modifiye Edici, 209900, Otozomal Resesif, Digenik Resesif; BBS1 (Bardet-Biedl Sendromu) (CCDC28B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 1, modifier of, 209900, Autosomal recessive, Digenic recessive; BBS1 (Bardet-Biedl syndrome) (CCDC28B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC28B	CCDC28B, MGC1203	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:16327777). Note=The gene represented in this entry acts as a disease modifier.	EDTA Blood Sample
Bardet-Biedl Sendromu 1, Modifiye Edici, 209900, Otozomal Resesif, Digenik Resesif; BBS1 (Bardet-Biedl Sendromu) (ARL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 1, modifier of, 209900, Autosomal recessive, Digenic recessive; BBS1 (Bardet-Biedl syndrome) (ARL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL6	ARL6, BBS3, RP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 3 (BBS3) [MIM:600151]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:15258860, ECO:0000269|PubMed:15314642, ECO:0000269|PubMed:23219996). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bardet-Biedl Sendromu 1, Modifiye Edici, 209900, Otozomal Resesif, Digenik Resesif; BBS1 (Bardet-Biedl Sendromu) (CCDC28B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 1, modifier of, 209900, Autosomal recessive, Digenic recessive; BBS1 (Bardet-Biedl syndrome) (CCDC28B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC28B	CCDC28B, MGC1203	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:16327777). Note=The gene represented in this entry acts as a disease modifier.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu 1, Modifiye Edici, 209900, Otozomal Resesif, Digenik Resesif; BBS1 (Bardet-Biedl Sendromu) (ARL6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 1, modifier of, 209900, Autosomal recessive, Digenic recessive; BBS1 (Bardet-Biedl syndrome) (ARL6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARL6	ARL6, BBS3, RP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 10, 615987, Otozomal Resesif; BBS10 (Bardet-Biedl Sendromu) (BBS10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 10, 615987, Autosomal recessive; BBS10 (Bardet-Biedl syndrome) (BBS10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS10	BBS10, C12orf58, FLJ23560	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 10 (BBS10) [MIM:615987]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:16582908, ECO:0000269|PubMed:20120035, ECO:0000269|PubMed:21344540, ECO:0000269|PubMed:23219996). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 10, 615987, Otozomal Resesif; BBS10 (Bardet-Biedl Sendromu) (BBS10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 10, 615987, Autosomal recessive; BBS10 (Bardet-Biedl syndrome) (BBS10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS10	BBS10, C12orf58, FLJ23560	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 10 (BBS10) [MIM:615987]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:16582908, ECO:0000269|PubMed:20120035, ECO:0000269|PubMed:21344540, ECO:0000269|PubMed:23219996). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 11, 615988, Otozomal Resesif; BBS11 (Bardet-Biedl Sendromu) (TRIM32 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 11, 615988, Autosomal recessive; BBS11 (Bardet-Biedl syndrome) (TRIM32 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIM32	TRIM32, HT2A, LGMD2H, BBS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 11, 615988, Otozomal Resesif; BBS11 (Bardet-Biedl Sendromu) (TRIM32 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 11, 615988, Autosomal recessive; BBS11 (Bardet-Biedl syndrome) (TRIM32 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIM32	TRIM32, HT2A, LGMD2H, BBS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 12, 615989, Otozomal Resesif; BBS12 (Bardet-Biedl Sendromu) (BBS12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 12, 615989, Autosomal recessive; BBS12 (Bardet-Biedl syndrome) (BBS12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS12	BBS12, FLJ35630, C4orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 12 (BBS12) [MIM:615989]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:17160889, ECO:0000269|PubMed:20120035, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 12, 615989, Otozomal Resesif; BBS12 (Bardet-Biedl Sendromu) (BBS12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 12, 615989, Autosomal recessive; BBS12 (Bardet-Biedl syndrome) (BBS12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS12	BBS12, FLJ35630, C4orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 12 (BBS12) [MIM:615989]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:17160889, ECO:0000269|PubMed:20120035, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 13, 615990, Otozomal Resesif; BBS13 (Bardet-Biedl Sendromu) (MKS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 13, 615990, Autosomal recessive; BBS13 (Bardet-Biedl syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 13, 615990, Otozomal Resesif; BBS13 (Bardet-Biedl Sendromu) (MKS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 13, 615990, Autosomal recessive; BBS13 (Bardet-Biedl syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 14, 615991, Otozomal Resesif (Bardet-Biedl Sendromu) (CEP290 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 14, 615991, Autosomal recessive (Bardet-Biedl syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 14, 615991, Otozomal Resesif (Bardet-Biedl Sendromu) (CEP290 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 14, 615991, Autosomal recessive (Bardet-Biedl syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 14, Modifiye Edici, 615991, Otozomal Resesif; BBS14 (Bardet-Biedl Sendromu) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 14, modifier of, 615991, Autosomal recessive; BBS14 (Bardet-Biedl syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:18327255). Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	EDTA Blood Sample
Bardet-Biedl Sendromu, 14, Modifiye Edici, 615991, Otozomal Resesif; BBS14 (Bardet-Biedl Sendromu) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 14, modifier of, 615991, Autosomal recessive; BBS14 (Bardet-Biedl syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:18327255). Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 15, 615992, Otozomal Resesif; BBS15 (Bardet-Biedl Sendromu) (WDPCP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 15, 615992, Autosomal recessive; BBS15 (Bardet-Biedl syndrome) (WDPCP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDPCP	WDPCP, C2orf86, BBS15, CHDTHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 15, 615992, Otozomal Resesif; BBS15 (Bardet-Biedl Sendromu) (WDPCP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 15, 615992, Autosomal recessive; BBS15 (Bardet-Biedl syndrome) (WDPCP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDPCP	WDPCP, C2orf86, BBS15, CHDTHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 16, 615993, Otozomal Resesif (Bardet-Biedl Sendromu) (SDCCAG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 16, 615993, Autosomal recessive; BBS16 (Bardet-Biedl syndrome) (SDCCAG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDCCAG8	SDCCAG8, CCCAP, SLSN7, BBS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 16 (BBS16) [MIM:615993]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:20835237, ECO:0000269|PubMed:22626039). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Bardet-Biedl Sendromu, 16, 615993, Otozomal Resesif (Bardet-Biedl Sendromu) (SDCCAG8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 16, 615993, Autosomal recessive; BBS16 (Bardet-Biedl syndrome) (SDCCAG8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SDCCAG8	SDCCAG8, CCCAP, SLSN7, BBS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 16 (BBS16) [MIM:615993]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:20835237, ECO:0000269|PubMed:22626039). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu 17, 615994, Otozomal Resesif; BBS17 (Bardet-Biedl Sendromu) (LZTFL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 17, 615994, Autosomal recessive; BBS17 (Bardet-Biedl syndrome) (LZTFL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LZTFL1	LZTFL1, BBS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 17 (BBS17) [MIM:615994]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:22510444, ECO:0000269|PubMed:23692385). Note=The disease is caused by mutations affecting the gene represented in this entry. Patients carrying LZTFL1 mutations manifest mesoaxial polydactyly, a clinical feature very uncommon for Bardet-Biedl syndrome (PubMed:22510444 and PubMed:23692385). Some patients manifest situs inversus (PubMed:22510444). (ECO:0000269|PubMed:22510444).;	EDTA Blood Sample
Bardet-Biedl Sendromu 17, 615994, Otozomal Resesif; BBS17 (Bardet-Biedl Sendromu) (LZTFL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 17, 615994, Autosomal recessive; BBS17 (Bardet-Biedl syndrome) (LZTFL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LZTFL1	LZTFL1, BBS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 17 (BBS17) [MIM:615994]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:22510444, ECO:0000269|PubMed:23692385). Note=The disease is caused by mutations affecting the gene represented in this entry. Patients carrying LZTFL1 mutations manifest mesoaxial polydactyly, a clinical feature very uncommon for Bardet-Biedl syndrome (PubMed:22510444 and PubMed:23692385). Some patients manifest situs inversus (PubMed:22510444). (ECO:0000269|PubMed:22510444).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 15, 615992, Otozomal Resesif; BBS18 (Bardet-Biedl Sendromu) (BBIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 18, 615995, Autosomal recessive; BBS18 (Bardet-Biedl syndrome) (BBIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBIP1	BBIP1, NCRNA00081, BBIP10, BBS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 18 (BBS18) [MIM:615995]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:24026985). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 15, 615992, Otozomal Resesif; BBS18 (Bardet-Biedl Sendromu) (BBIP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 18, 615995, Autosomal recessive; BBS18 (Bardet-Biedl syndrome) (BBIP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBIP1	BBIP1, NCRNA00081, BBIP10, BBS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 18 (BBS18) [MIM:615995]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:24026985). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 19, 615996, Otozomal Resesif; BBS19 (Bardet-Biedl Sendromu) (IFT27 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 19, 615996, Autosomal recessive; BBS19 (Bardet-Biedl syndrome) (IFT27 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT27	IFT27, RABL4, BBS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 19 (BBS19) [MIM:615996]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:24688770). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 19, 615996, Otozomal Resesif; BBS19 (Bardet-Biedl Sendromu) (IFT27 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 19, 615996, Autosomal recessive; BBS19 (Bardet-Biedl syndrome) (IFT27 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT27	IFT27, RABL4, BBS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 19 (BBS19) [MIM:615996]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:24688770). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu 2, 615981, Otozomal Resesif; BBS2 (Bardet-Biedl Sendromu) (BBS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 2, 615981, Autosomal recessive; BBS2 (Bardet-Biedl syndrome) (BBS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS2	BBS2, RP74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu 2, 615981, Otozomal Resesif; BBS2 (Bardet-Biedl Sendromu) (BBS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 2, 615981, Autosomal recessive; BBS2 (Bardet-Biedl syndrome) (BBS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS2	BBS2, RP74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 20, 617119, Otozomal Resesif (Bardet-Biedl Sendromu) (IFT74 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 20, 617119, Autosomal recessive (Bardet-Biedl syndrome) (IFT74 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT74	IFT74, CCDC2, CMG1, BBS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 20, 617119, Otozomal Resesif (Bardet-Biedl Sendromu) (IFT74 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 20, 617119, Autosomal recessive (Bardet-Biedl syndrome) (IFT74 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT74	IFT74, CCDC2, CMG1, BBS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 3, 600151, Otozomal Resesif; BBS3 (Bardet-Biedl Sendromu) (ARL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 3, 600151, Autosomal recessive; BBS3 (Bardet-Biedl syndrome) (ARL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL6	ARL6, BBS3, RP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 3 (BBS3) [MIM:600151]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:15258860, ECO:0000269|PubMed:15314642, ECO:0000269|PubMed:23219996). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bardet-Biedl Sendromu, 3, 600151, Otozomal Resesif; BBS3 (Bardet-Biedl Sendromu) (ARL6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 3, 600151, Autosomal recessive; BBS3 (Bardet-Biedl syndrome) (ARL6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARL6	ARL6, BBS3, RP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 3 (BBS3) [MIM:600151]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:15258860, ECO:0000269|PubMed:15314642, ECO:0000269|PubMed:23219996). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu 4, 615982, Otozomal Resesif; BBS4 (Bardet-Biedl Sendromu) (BBS4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 4, 615982, Autosomal recessive; BBS4 (Bardet-Biedl syndrome) (BBS4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS4	BBS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bardet-Biedl Sendromu, 4, 615982, Otozomal Resesif; BBS4 (Bardet-Biedl Sendromu) (BBS4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 4, 615982, Autosomal recessive; BBS4 (Bardet-Biedl syndrome) (BBS4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS4	BBS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 5, 615983, Otozomal Resesif; BBS5 (Bardet-Biedl Sendromu) (BBS5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 5, 615983, Autosomal recessive; BBS5 (Bardet-Biedl syndrome) (BBS5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS5	BBS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 5 (BBS5) [MIM:615983]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:15137946, ECO:0000269|PubMed:18203199, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 5, 615983, Otozomal Resesif; BBS5 (Bardet-Biedl Sendromu) (BBS5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 5, 615983, Autosomal recessive; BBS5 (Bardet-Biedl syndrome) (BBS5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS5	BBS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 5 (BBS5) [MIM:615983]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269|PubMed:15137946, ECO:0000269|PubMed:18203199, ECO:0000269|PubMed:21344540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 6, 605231, Otozomal Resesif; BBS6 (Bardet-Biedl Sendromu) (MKKS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 6, 605231, Autosomal recessive; BBS6 (Bardet-Biedl syndrome) (MKKS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKKS	MKKS, HMCS, KMS, MKS, BBS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: McKusick-Kaufman syndrome (MKKS) [MIM:236700]: Autosomal recessive developmental disorder. It is characterized by hydrometrocolpos, postaxial polydactyly and congenital heart defects. (ECO:0000269	PubMed:10802661). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Bardet-Biedl Sendromu, 6, 605231, Otozomal Resesif; BBS6 (Bardet-Biedl Sendromu) (MKKS gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 6, 605231, Autosomal recessive; BBS6 (Bardet-Biedl syndrome) (MKKS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MKKS	MKKS, HMCS, KMS, MKS, BBS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: McKusick-Kaufman syndrome (MKKS) [MIM:236700]: Autosomal recessive developmental disorder. It is characterized by hydrometrocolpos, postaxial polydactyly and congenital heart defects. {ECO:0000269	PubMed:10802661}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 7, 615984, Otozomal Resesif; BBS7 (Bardet-Biedl Sendromu) (BBS7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 7, 615984, Autosomal recessive; BBS7 (Bardet-Biedl syndrome) (BBS7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS7	BBS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 7 (BBS7) [MIM:615984]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:12567324, ECO:0000269|PubMed:12677556, ECO:0000269|PubMed:15770229, ECO:0000269|PubMed:21344540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 7, 615984, Otozomal Resesif; BBS7 (Bardet-Biedl Sendromu) (BBS7 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 7, 615984, Autosomal recessive; BBS7 (Bardet-Biedl syndrome) (BBS7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS7	BBS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 7 (BBS7) [MIM:615984]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:12567324, ECO:0000269|PubMed:12677556, ECO:0000269|PubMed:15770229, ECO:0000269|PubMed:21344540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 8, 615985, Otozomal Resesif; BBS8 (Bardet-Biedl Sendromu) (TTC8 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 8, 615985, Autosomal recessive; BBS8 (Bardet-Biedl syndrome) (TTC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC8	TTC8, BBS8, RP51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 8 (BBS8) [MIM:615985]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:14520415, ECO:0000269|PubMed:16308660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 8, 615985, Otozomal Resesif; BBS8 (Bardet-Biedl Sendromu) (TTC8 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 8, 615985, Autosomal recessive; BBS8 (Bardet-Biedl syndrome) (TTC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTC8	TTC8, BBS8, RP51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 8 (BBS8) [MIM:615985]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:14520415, ECO:0000269|PubMed:16308660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bardet-Biedl Sendromu, 9, 615986, Otozomal Resesif; BBS9 (Bardet-Biedl Sendromu) (BBS9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bardet-Biedl syndrome 9, 615986, Autosomal recessive; BBS9 (Bardet-Biedl syndrome) (BBS9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS9	PTHB1, BBS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 9 (BBS9) [MIM:615986]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:16380913}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bardet-Biedl Sendromu, 9, 615986, Otozomal Resesif; BBS9 (Bardet-Biedl Sendromu) (BBS9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bardet-Biedl syndrome 9, 615986, Autosomal recessive; BBS9 (Bardet-Biedl syndrome) (BBS9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BBS9	PTHB1, BBS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 9 (BBS9) [MIM:615986]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:16380913}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1 (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	BARE LYMPHOCYTE SYNDROME, TYPE I (Immunodeficiency by defective expression of HLA class 1) (TAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAP1	TAP1, ABCB2, TAP1, RING4, PSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:10074494}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1 (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	BARE LYMPHOCYTE SYNDROME, TYPE I (Immunodeficiency by defective expression of HLA class 1) (TAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAP1	TAP1, ABCB2, TAP1, RING4, PSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:10074494}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAPBP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type I, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAPBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAPBP	TAPBP, TPSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type I, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAP1	TAP1, ABCB2, TAP1, RING4, PSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:10074494}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAPBP gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type I, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAPBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAPBP	TAPBP, TPSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type I, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAP1	TAP1, ABCB2, TAP1, RING4, PSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:10074494}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, TAP2 Eksikliğine Bağlı, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAP2	TAP2, ABCB3, PSF2, RING11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:7517574}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 1, TAP2 Eksikliğine Bağlı, 604571, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf I Defektif Ekspresyonuna Bağlı) (TAP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571, Autosomal recessive (Immunodeficiency by defective expression of HLA class 1) (TAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAP2	TAP2, ABCB3, PSF2, RING11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 1 (BLS1) [MIM:604571]: A HLA class I deficiency. Contrary to bare lymphocyte syndromes type 2 and type 3, which are characterized by early-onset severe combined immunodeficiency, class I antigen deficiencies are not accompanied by particular pathologic manifestations during the first years of life. Systemic infections have not been described. Chronic bacterial infections, often beginning in the first decade of life, are restricted to the respiratory tract. {ECO:0000269	PubMed:7517574}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 2, Komplementasyon Grubu A, 209920, Otozomal Resesif (İmmun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (CIITA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type II, complementation group A, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (CIITA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIITA	CIITA, MHC2TA, C2TA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. {ECO:0000269|PubMed:10501838, ECO:0000269|PubMed:11466404, ECO:0000269|PubMed:11862382, ECO:0000269|PubMed:7749984, ECO:0000269|PubMed:8402893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Çıplak Lenfosit Sendromu, Tip 2, Komplementasyon Grubu A, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (CIITA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type II, complementation group A, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (CIITA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CIITA	CIITA, MHC2TA, C2TA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. (ECO:0000269|PubMed:10501838, ECO:0000269|PubMed:11466404, ECO:0000269|PubMed:11862382, ECO:0000269|PubMed:7749984, ECO:0000269|PubMed:8402693). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 2, Komplementasyon Grubu C, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (RFX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type II, complementation group C, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RFX5	RFX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. (ECO:0000269	PubMed:10825209). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 2, Komplementasyon Grubu C, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (RFX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type II, complementation group C, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFX5	RFX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. (ECO:0000269	PubMed:10825209). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 2,Komplementasyon Grubu D, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (RFXAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bare lymphocyte syndrome, type II, complementation group D, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFXAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RFXAP	RFXAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. (ECO:0000269|PubMed:10072068, ECO:0000269|PubMed:9118643). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Çıplak Lenfosit Sendromu, Tip 2,Komplementasyon Grubu D, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı (Komplementasyon Grup A)) (RFXAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bare lymphocyte syndrome, type II, complementation group D, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFXAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFXAP	RFXAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. (ECO:0000269|PubMed:10072068, ECO:0000269|PubMed:9118643). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Barret Özofagus / Özofageal Adenokarsinoma, 614266 (Özofageal Adenokarsinom) (MSR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Barrett esophagus/esophageal adenocarcinoma, 614266 (Adenocarcinoma of esophagus) (MSR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSR1	MSR1, SCARA1, SRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Barrett esophagus (BE) [MIM:614266]: A condition characterized by a metaplastic change in which normal esophageal squamous epithelium is replaced by a columnar and intestinal-type epithelium. Patients with Barrett esophagus have an increased risk of esophageal adenocarcinoma. The main cause of Barrett esophagus is gastroesophageal reflux. The retrograde movement of acid and bile salts from the stomach into the esophagus causes prolonged injury to the esophageal epithelium and induces chronic esophagitis, which in turn is believed to trigger the pathologic changes. (ECO:0000269	PubMed:21791690). Note=The disease may be caused by mutations affecting the gene represented in this entry. Genetic variants in MSR1 have been found in individuals with Barrett esophagus and are thought to contribute to disease susceptibility.;	EDTA Blood Sample
Barret Özofagus / Özofageal Adenokarsinoma, 614266 (Özofageal Adenokarsinom) (CTHRC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Barrett esophagus/esophageal adenocarcinoma, 614266 (Adenocarcinoma of esophagus) (CTHRC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTHRC1	CTHRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Barrett esophagus (BE) [MIM:614266]: A condition characterized by a metaplastic change in which normal esophageal squamous epithelium is replaced by a columnar and intestinal-type epithelium. Patients with Barrett esophagus have an increased risk of esophageal adenocarcinoma. The main cause of Barrett esophagus is gastroesophageal reflux. The retrograde movement of acid and bile salts from the stomach into the esophagus causes prolonged injury to the esophageal epithelium and induces chronic esophagitis, which in turn is believed to trigger the pathologic changes. (ECO:0000269	PubMed:21791690). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Barret Özofagus / Özofageal Adenokarsinoma, 614266 (Özofageal Adenokarsinom) (ASCC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Barrett esophagus/esophageal adenocarcinoma, 614266 (Adenocarcinoma of esophagus) (ASCC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASCC1	ASCC1, p50, SMABF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bart-Pumfrey Sendromu, 149200, Otozomal Dominant (Parmak Yastıkları-Lökonişi-Sensorinöral İşitme Kaybı-Palmoplantar Hiperkeratoz Sendromu) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bart-Pumphrey syndrome, 149200, Autosomal dominant (Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bart-Pumfrey Sendromu, 149200, Otozomal Dominant (Parmak Yastıkları-Lökonişi-Sensorinöral İşitme Kaybı-Palmoplantar Hiperkeratoz Sendromu) (GJB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bart-Pumphrey syndrome, 149200, Autosomal dominant (Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Barth Sendromu, 302060, X'e Bağlı Resesif (Barth Sendromu) (TAZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Barth syndrome, 302060, X-linked recessive; BTHS (Barth syndrome) (TAZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAZ	TAZ, EFE2, BTHS, CMD3A, LVNCX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Barth syndrome (BTHS) [MIM:302060]: An X-linked disease characterized by dilated cardiomyopathy with endocardial fibroelastosis, a predominantly proximal skeletal myopathy, growth retardation, neutropenia, and organic aciduria, particularly excess of 3-methylglutaconic acid. Additional features include hypertrophic cardiomyopathy, isolated left ventricular non-compaction, ventricular arrhythmia, motor delay, poor appetite, fatigue and exercise intolerance, hypoglycemia, lactic acidosis, hyperammonemia, and dramatic late catch-up growth after growth delay throughout childhood. (ECO:0000269|PubMed:11238270, ECO:0000269|PubMed:12032589, ECO:0000269|PubMed:9382096, ECO:0000269|PubMed:9382097). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Barth Sendromu, 302060, X'e Bağlı Resesif (Barth Sendromu) (TAZ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Barth syndrome, 302060, X-linked recessive; BTHS (Barth syndrome) (TAZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAZ	TAZ, EFE2, BTHS, CMD3A, LVNCX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Barth syndrome (BTHS) [MIM:302060]: An X-linked disease characterized by dilated cardiomyopathy with endocardial fibroelastosis, a predominantly proximal skeletal myopathy, growth retardation, neutropenia, and organic aciduria, particularly excess of 3-methylglutaconic acid. Additional features include hypertrophic cardiomyopathy, isolated left ventricular non-compaction, ventricular arrhythmia, motor delay, poor appetite, fatigue and exercise intolerance, hypoglycemia, lactic acidosis, hyperammonemia, and dramatic late catch-up growth after growth delay throughout childhood. (ECO:0000269|PubMed:11238270, ECO:0000269|PubMed:12032589, ECO:0000269|PubMed:9382096, ECO:0000269|PubMed:9382097). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 1, 601678, Otozomal Resesif, BART S1 (Bartter Sendromu) (SLC12A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 1, 601678, Autosomal recessive, BARTS1 (Bartter syndrome) (SLC12A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC12A1	SLC12A1, NKCC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bartter Sendromu, Tip 1, 601678, Otozomal Resesif, BART S1 (Bartter Sendromu) (SLC12A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 1, 601678, Autosomal recessive, BARTS1 (Bartter syndrome) (SLC12A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC12A1	SLC12A1, NKCC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 2, 241200, Otozomal Resesif, BART S2 (Bartter Sendromu) (KCNJ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 2, 241200, Autosomal recessive; BARTS2 (Bartter syndrome) (KCNJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ1	KCNJ1, ROMK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bartter syndrome 2 (BS2) [MIM:241200]: An autosomal recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 2 is a life-threatening condition beginning in utero, with marked fetal polyuria that leads to polyhydramnios and premature delivery. Another hallmark is a marked hypercalciuria and, as a secondary consequence, the development of nephrocalcinosis and osteopenia. (ECO:0000269|PubMed:8841184, ECO:0000269|PubMed:9002665, ECO:0000269|PubMed:9727001). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bartter Sendromu, Tip 2, 241200, Otozomal Resesif, BART S2 (Bartter Sendromu) (KCNJ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 2, 241200, Autosomal recessive; BARTS2 (Bartter syndrome) (KCNJ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNJ1	KCNJ1, ROMK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bartter syndrome 2 (BS2) [MIM:241200]: An autosomal recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 2 is a life-threatening condition beginning in utero, with marked fetal polyuria that leads to polyhydramnios and premature delivery. Another hallmark is a marked hypercalciuria and, as a secondary consequence, the development of nephrocalcinosis and osteopenia. (ECO:0000269|PubMed:8841184, ECO:0000269|PubMed:9002665, ECO:0000269|PubMed:9727001). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 3, 607364, Otozomal Resesif, BART S3 (Bartter Sendromu) (CLCNKB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 3, 607364, Autosomal recessive; BARTS3 (Bartter syndrome) (CLCNKB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCNKB	CLCNKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bartter Sendromu, Tip 3, 607364, Otozomal Resesif, BART S3 (Bartter Sendromu) (MLPA)	Bartter syndrome, type 3, 607364, Autosomal recessive; BARTS3 (Bartter syndrome) (MLPA)	CLCNKB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample

Bartter Sendromu, Tip 3, 607364, Otozomal Resesif, BARTS3 (Bartter Sendromu) (CLCNKB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 3, 607364, Autosomal recessive; BARTS3 (Bartter syndrome) (CLCNKB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCNKB	CLCNKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 3, 607364, Otozomal Resesif, BARTS3 (Bartter Sendromu) (Prenatal) (MLPA)	Bartter syndrome, type 3, 607364, Autosomal recessive; BARTS3 (Bartter syndrome) (Prenatal) (MLPA)	CLCNKB	CLCNKB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 4a, 602522, Otozomal Resesif, BARTS4A (Bartter Sendromu) (BSND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 4a, 602522, Autosomal recessive; BARTS4A (Bartter syndrome) (BSND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSND	BSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bartter syndrome 4A (BS4A) [MIM:602522]: An autosomal recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4 is associated with sensorineural deafness. (ECO:0000269|PubMed:11687798, ECO:0000269|PubMed:12574213). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bartter Sendromu, Tip 4a, 602522, Otozomal Resesif, BARTS4A (Bartter Sendromu) (BSND geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 4a, 602522, Autosomal recessive; BARTS4A (Bartter syndrome) (BSND gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BSND	BSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bartter syndrome 4A (BS4A) [MIM:602522]: An autosomal recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4 is associated with sensorineural deafness. (ECO:0000269|PubMed:11687798, ECO:0000269|PubMed:12574213). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif (Bartter Sendromu) (CLCNKB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive (Bartter syndrome) (CLCNKB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCNKB	CLCNKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bartter syndrome 4B (BS4B) [MIM:613090]: A digenic, recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4B is associated with sensorineural deafness. (ECO:0000269|PubMed:15044642, ECO:0000269|PubMed:18310267). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Loss-of-function of both CLCNKA and CLCNKB results in the disease phenotype (PubMed:18310267).	EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif (Bartter Sendromu) (MLPA)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive (Bartter syndrome) (MLPA)	CLCNKB	CLCNKB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bartter syndrome 4B (BS4B) [MIM:613090]: A digenic, recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4B is associated with sensorineural deafness. (ECO:0000269|PubMed:15044642, ECO:0000269|PubMed:18310267). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Loss-of-function of both CLCNKA and CLCNKB results in the disease phenotype (PubMed:18310267).	EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif (Bartter Sendromu) (CLCNKB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive (Bartter syndrome) (CLCNKB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCNKB	CLCNKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bartter syndrome 4B (BS4B) [MIM:613090]: A digenic, recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4B is associated with sensorineural deafness. (ECO:0000269|PubMed:15044642, ECO:0000269|PubMed:18310267). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Loss-of-function of both CLCNKA and CLCNKB results in the disease phenotype (PubMed:18310267).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif (Bartter Sendromu) (Prenatal) (MLPA)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive (Bartter syndrome) (Prenatal) (MLPA)	CLCNKB	CLCNKB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bartter syndrome 4B (BS4B) [MIM:613090]: A digenic, recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4B is associated with sensorineural deafness. (ECO:0000269|PubMed:15044642, ECO:0000269|PubMed:18310267). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Loss-of-function of both CLCNKA and CLCNKB results in the disease phenotype (PubMed:18310267).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif, BARTS4B (Bartter Sendromu) (CLCNKA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive; BARTS4B (Bartter syndrome) (CLCNKA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCNKA	CLCNKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Bartter Sendromu, Tip 4b, Dijenik, 613090, Dijenik Resesif, BARTS4B (Bartter Sendromu) (CLCNKA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 4b, digenic, 613090, Digenic recessive; BARTS4B (Bartter syndrome) (CLCNKA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCNKA	CLCNKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bartter Sendromu, Tip 5, Doğum Öncesi, Geçici, 300971, X'e Bağlı Resesif, BARTS5 (Bartter Sendromu) (MAGED2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bartter syndrome, type 5, antenatal, transient, 300971, X-linked recessive; BARTS5 (Bartter syndrome) (MAGED2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAGED2	MAGED2, MAGED, BARTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bartter Sendromu, Tip 5, Doğum Öncesi, Geçici, 300971, X'e Bağlı Resesif, BARTS5 (Bartter Sendromu) (MAGED2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bartter syndrome, type 5, antenatal, transient, 300971, X-linked recessive; BARTS5 (Bartter syndrome) (MAGED2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAGED2	MAGED2, MAGED, BARTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Hücre Karsinomu 7, 614740 (TP53 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell carcinoma 7, 614740 (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bazal Hücre Karsinomu 7, 614740 (MLPA)	Basal cell carcinoma 7, 614740 (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Bazal Hücreli Karsinom, Somatik, 605462 (RASA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell carcinoma, somatic, 605462 (RASA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RASA1	RASA1, GAP, CMAVM, PKWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Bazal Hücreli Karsinom, Somatik, 605462 (SMO gen) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell carcinoma, somatic, 605462 (SMO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMO	SMOH, SMO, CRJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Bazal Hücreli Karsinom, Somatik, 605462 (Gorlin Sendromu) (PTCH2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell carcinoma, somatic, 605462 (Gorlin syndrome) (PTCH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH2	PTCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Bazal Hücreli Karsinom, Somatik, 605462 (PTCH1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell carcinoma, somatic, 605462 (PTCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH1	PTCH1, NBCCS, BCNS, HPE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Basal cell nevus syndrome (BCNS) [MIM:109400]: An autosomal dominant disease characterized by nevoid basal cell carcinomas and developmental abnormalities such as rib and craniofacial alterations, polydactyly, syndactyly, and spina bifida. In addition, the patients suffer from a multitude of tumors like basal cell carcinomas, fibromas of the ovaries and heart, cysts of the skin, jaws and mesentery, as well as medulloblastomas and meningiomas. (ECO:0000269|PubMed:11231326, ECO:0000269|PubMed:15459969, ECO:0000269|PubMed:8840969, ECO:0000269|PubMed:8981943, ECO:0000269|PubMed:9620294). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant (Gorlin Sendromu) (SUFU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell nevus syndrome, 109400, Autosomal dominant (Gorlin syndrome) (SUFU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUFU	SUFU, SUFUXL, SUFUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant (Gorlin Sendromu) (PTCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell nevus syndrome, 109400, Autosomal dominant (Gorlin syndrome) (PTCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH1	PTCH1, NBCCS, BCNS, HPE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Basal cell nevus syndrome (BCNS) [MIM:109400]: An autosomal dominant disease characterized by nevoid basal cell carcinomas and developmental abnormalities such as rib and craniofacial alterations, polydactyly, syndactyly, and spina bifida. In addition, the patients suffer from a multitude of tumors like basal cell carcinomas, fibromas of the ovaries and heart, cysts of the skin, jaws and mesentery, as well as medulloblastomas and meningiomas. (ECO:0000269|PubMed:11231326, ECO:0000269|PubMed:15459969, ECO:0000269|PubMed:8840969, ECO:0000269|PubMed:8981943, ECO:0000269|PubMed:9620294). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant (Gorlin Sendromu) (SUFU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal cell nevus syndrome, 109400, Autosomal dominant (Gorlin syndrome) (SUFU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUFU	SUFU, SUFUXL, SUFUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant (Gorlin Sendromu) (PTCH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal cell nevus syndrome, 109400, Autosomal dominant (Gorlin syndrome) (PTCH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTCH1	PTCH1, NBCCS, BCNS, HPE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Basal cell nevus syndrome (BCNS) [MIM:109400]: An autosomal dominant disease characterized by nevoid basal cell carcinomas and developmental abnormalities such as rib and craniofacial alterations, polydactyly, syndactyly, and spina bifida. In addition, the patients suffer from a multitude of tumors like basal cell carcinomas, fibromas of the ovaries and heart, cysts of the skin, jaws and mesentery, as well as medulloblastomas and meningiomas. (ECO:0000269|PubMed:11231326, ECO:0000269|PubMed:15459969, ECO:0000269|PubMed:8840969, ECO:0000269|PubMed:8981943, ECO:0000269|PubMed:9620294). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant, BCNS (Gorlin Sendromu) (PTCH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal cell nevus syndrome, 109400, Autosomal dominant; BCNS (Gorlin syndrome) (PTCH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH2	PTCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bazal Hücre Nevus Sendromu, 109400, Otozomal Dominant, BCNS (Gorlin Sendromu) (PTCH2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Basal cell nevus syndrome, 109400, Autosomal dominant; BCNS (Gorlin syndrome) (PTCH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTCH2	PTCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Bazal Ganglıyon Kalsifikasyonu, İdyopatik, 1, 213600, Otozomal Dominant; IBGC1 (Bilateral Striopallidodentat Kalsinozis) (SLC20A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal ganglia calcification, idiopathic, 1, 213600, Autosomal dominant; IBGC1 (Bilateral striopallidodentate calcinosis) (SLC20A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC20A2	SLC20A2, MLVAR, GLVR2, IBGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Basal ganglia calcification, idiopathic, 1 (IBGC1) [MIM:213600]: A form of basal ganglia calcification, an autosomal dominant condition characterized by symmetric calcification in the basal ganglia and other brain regions. Affected individuals can either be asymptomatic or show a wide spectrum of neuropsychiatric symptoms, including parkinsonism, dystonia, tremor, ataxia, dementia, psychosis, seizures, and chronic headache. Serum levels of calcium, phosphate, alkaline phosphatase and parathyroid hormone are normal. The neuropathological hallmark of the disease is vascular and pericapillary calcification, mainly of calcium phosphate, in the affected brain areas. {ECO:0000269|PubMed:22327515, ECO:0000269|PubMed:23334463, ECO:0000269|PubMed:23406454, ECO:0000269|PubMed:23939468, ECO:0000269|PubMed:24463626, ECO:0000269|PubMed:25284758}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bazal Ganglıyon Kalsifikasyonu, İdyopatik, 1, 213600, Otozomal Dominant; IBGC1 (Bilateral Striopallidodentat Kalsinozis) (SLC20A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal ganglia calcification, idiopathic, 1, 213600, Autosomal dominant; IBGC1 (Bilateral striopallidodentate calcinosis) (SLC20A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC20A2	SLC20A2, MLVAR, GLVR2, IBGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Basal ganglia calcification, idiopathic, 1 (IBGC1) [MIM:213600]: A form of basal ganglia calcification, an autosomal dominant condition characterized by symmetric calcification in the basal ganglia and other brain regions. Affected individuals can either be asymptomatic or show a wide spectrum of neuropsychiatric symptoms, including parkinsonism, dystonia, tremor, ataxia, dementia, psychosis, seizures, and chronic headache. Serum levels of calcium, phosphate, alkaline phosphatase and parathyroid hormone are normal. The neuropathological hallmark of the disease is vascular and pericapillary calcification, mainly of calcium phosphate, in the affected brain areas. {ECO:0000269|PubMed:22327515, ECO:0000269|PubMed:23334463, ECO:0000269|PubMed:23406454, ECO:0000269|PubMed:23939468, ECO:0000269|PubMed:24463626, ECO:0000269|PubMed:25284758}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 4, 615007, Otozomal Dominant; IBGC4 (Bilateral Striopallidodentat Kalsinozis) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal ganglia calcification, idiopathic, 4, 615007, Autosomal dominant; IBGC4 (Bilateral striopallidodentate calcinosis) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 4, 615007, Otozomal Dominant; IBGC4 (Bilateral Striopallidodentat Kalsinozis) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal ganglia calcification, idiopathic, 4, 615007, Autosomal dominant; IBGC4 (Bilateral striopallidodentate calcinosis) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 5, 615483, Otozomal Dominant; IBGC5 (Bilateral Striopallidodentat Kalsinozis) (PDGFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal ganglia calcification, idiopathic, 5, 615483, Autosomal dominant; IBGC5 (Bilateral striopallidodentate calcinosis) (PDGFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFB	PDGFB, SIS, IBGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 5, 615483, Otozomal Dominant; IBGC5 (Bilateral Striopallidodentat Kalsinozis) (PDGFB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal ganglia calcification, idiopathic, 5, 615483, Autosomal dominant; IBGC5 (Bilateral striopallidodentate calcinosis) (PDGFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDGFB	PDGFB, SIS, IBGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 6, 616413, Otozomal Dominant; IBGC6 (Bilateral Striopallidodentat Kalsinozis) (XPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal ganglia calcification, idiopathic, 6, 616413, Autosomal dominant; IBGC6 (Bilateral striopallidodentate calcinosis) (XPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XPR1	XPR1, SYG1, IBGC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Basal ganglia calcification, idiopathic, 6 (IBGC6) [MIM:616413]: A form of basal ganglia calcification, an autosomal dominant condition characterized by symmetric calcification in the basal ganglia and other brain regions. Affected individuals can either be asymptomatic or show a wide spectrum of neuropsychiatric symptoms, including parkinsonism, dystonia, tremor, ataxia, dementia, psychosis, seizures, and chronic headache. Serum levels of calcium, phosphate, alkaline phosphatase and parathyroid hormone are normal. The neuropathological hallmark of the disease is vascular and pericapillary calcification, mainly of calcium phosphate, in the affected brain areas. {ECO:0000269	PubMed:25938945}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bazal Ganglıon Kalsifikasyonu, İdyopatik, 6, 616413, Otozomal Dominant; IBGC6 (Bilateral Striopallidodentat Kalsinozis) (XPR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basal ganglia calcification, idiopathic, 6, 616413, Autosomal dominant; IBGC6 (Bilateral striopallidodentate calcinosis) (XPR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XPR1	XPR1, SYG1, IBGC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Basal ganglia calcification, idiopathic, 6 (IBGC6) [MIM:616413]: A form of basal ganglia calcification, an autosomal dominant condition characterized by symmetric calcification in the basal ganglia and other brain regions. Affected individuals can either be asymptomatic or show a wide spectrum of neuropsychiatric symptoms, including parkinsonism, dystonia, tremor, ataxia, dementia, psychosis, seizures, and chronic headache. Serum levels of calcium, phosphate, alkaline phosphatase and parathyroid hormone are normal. The neuropathological hallmark of the disease is vascular and pericapillary calcification, mainly of calcium phosphate, in the affected brain areas. {ECO:0000269	PubMed:25938945}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bazal Laminer Druzen, 126700, Otozomal Dominant (Aleksel Druzen) (CFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basal laminar drusen, 126700, Autosomal dominant (Familial drusen) (CFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bazal Laminer Druzen, 126700, Otozomal Dominant (Aleksel Druzen) (MLPA)	Basal laminar drusen, 126700, Autosomal dominant (Familial drusen) (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Basan Sendromu, 129200, Otozomal Dominant (Ektodermal Displazi-Dermatoglifik Yokluğu Sendromu) (SMARCAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basan syndrome, 129200, Autosomal dominant (Ectodermal dysplasia-absent dermatoglyphs syndrome) (SMARCAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCAD1	SMARCAD1, KIAA1122, ETL1, HEL1, ADERM, BASNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Basan Sendromu, 129200, Otozomal Dominant (Ektodermal Displazi-Dermatoglifik Yokluğu Sendromu) (SMARCAD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basan syndrome, 129200, Autosomal dominant (Ectodermal dysplasia-absent dermatoglyphs syndrome) (SMARCAD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCAD1	SMARCAD1, KIAA1122, ETL1, HEL1, ADERM, BASNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Basel Vanagait-Smirin-Yosef'e Sendromu, 616449, Otozomal Resesif; BVSYS (Konjenital Kataralt-Mikrosefali-Nevus Flammeus Simpleks-Şiddetli Mental Retardasyon Sendromu) (MED25 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Basel-Vanagait-Smirin-Yosef syndrome, 616449, Autosomal recessive; BVSYS (Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome) (MED25 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED25	MED25, PTOV2, ARC92, CMT2B2, BVSYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Basel-Vanagaite-Smirin-Yosef syndrome (BVSYS) [MIM:616449]: An autosomal recessive syndrome characterized by eye, brain, cardiac and palatal abnormalities as well as growth retardation, microcephaly and severe intellectual disability. {ECO:0000269	PubMed:25792360}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Basel Vanagait-Smirin-Yosef'e Sendromu, 616449, Otozomal Resesif; BVSYS (Konjenital Kataralt-Mikrosefali-Nevus Flammeus Simpleks-Şiddetli Mental Retardasyon Sendromu) (MED25 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Basel-Vanagait-Smirin-Yosef syndrome, 616449, Autosomal recessive; BVSYS (Congenital cataract-microcephaly-nevus flammeus simplex-severe intellectual disability syndrome) (MED25 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED25	MED25, PTOV2, ARC92, CMT2B2, BVSYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Basel-Vanagaite-Smirin-Yosef syndrome (BVSYS) [MIM:616449]: An autosomal recessive syndrome characterized by eye, brain, cardiac and palatal abnormalities as well as growth retardation, microcephaly and severe intellectual disability. {ECO:0000269	PubMed:25792360}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
BCL2 Breakapart (FISH)	BCL2 Breakapart (FISH)	18q21.33	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Beare-Stevenson Kutis Girata Sendromu, 123790, Otozomal Dominant; BSTVS (Kutis Girata-Akantozis Nigricans-Kraniyosinostozis Sendromu (Beare-Stevenson Sendromu)) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beare-Stevenson cutis gyrata syndrome, 123790, Autosomal dominant; BSTVS (Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beare-Stevenson Kutis Girata Sendromu, 123790, Otozomal Dominant; BSTVS (Kutis Girata-Akantozis Nigricans-Kraniyosinostozis Sendromu (Beare-Stevenson Sendromu)) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beare-Stevenson cutis gyrata syndrome, 123790, Autosomal dominant; BSTVS (Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beaulieu-Boykot-Innes Sendromu, 613680, Otozomal Resesif; BBIS (Gelişimsel Gecikme-Mikrosefali-Fasiyal Dismorfizm Sendromu, Hutterite Tipi) (THOC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beaulieu-Boycott-Innes syndrome, 613680, Autosomal recessive; BBIS (Developmental delay-microcephaly-facial dysmorphism syndrome, Hutterite type) (THOC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THOC6	THOC6, FSAP35, BBIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beaulieu-Boycott-Innes syndrome (BBIS) [MIM:613680]: An autosomal recessive neurodevelopmental disorder characterized by delayed development, moderate intellectual disability, and dysmorphic facial features. Other developmental anomalies, such as cardiac and renal defects, may also occur. {ECO:0000269	PubMed:23621916}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Beaulieu-Boykot-Innes Sendromu, 613680, Otozomal Resesif; BBIS (Gelişimsel Gecikme-Mikrosefali-Fasiyal Dismorfizm Sendromu, Hutterite Tipi) (THOC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beaulieu-Boycott-Innes syndrome, 613680, Autosomal recessive; BBIS (Developmental delay-microcephaly-facial dysmorphism syndrome, Hutterite type) (THOC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	THOC6	THOC6, FSAP35, BBIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Beaulieu-Boycott-Innes syndrome (BBIS) [MIM:613680]: An autosomal recessive neurodevelopmental disorder characterized by delayed development, moderate intellectual disability, and dysmorphic facial features. Other developmental anomalies, such as cardiac and renal defects, may also occur. {ECO:0000269	PubMed:23621916}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Becker Müsküler Distrofi, 300376, X'e Bağlı (Becker Müsküler Distrofi) (DMD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Becker muscular dystrophy, 300376, X-linked recessive (Becker muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Becker Müsküler Distrofi, 300376, X'e Bağlı (Becker Müsküler Distrofi) (MLPA)	Becker muscular dystrophy, 300376, X-linked recessive (Becker muscular dystrophy) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Becker Müsküler Distrofi, 300376, X'e Bağlı (Becker Müsküler Distrofi) (DMD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Becker muscular dystrophy, 300376, X-linked recessive (Becker muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Becker Müsküler Distrofi, 300376, X'e Bağlı (Becker Müsküler Distrofi) (Prenatal) (MLPA)	Becker muscular dystrophy, 300376, X-linked recessive (Becker muscular dystrophy) (Prenatal) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. [ECO:0000269]PubMed:10573008). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith- Wiedemann Sendromu (BWS), Russell-Silver Sendromu (RSS) (Delesyon + Metilasyon) (11p15, H19, IGF2, CDKN1C, KCNQ1) (MLPA)	Beckwith-Wiedemann Syndrome (BWS), Russell-Silver Syndrome (RSS) (Deletion + Methylation) (11p15, H19, IGF2, CDKN1C, KCNQ1) (MLPA)	11p15, H19, IGF2, CDKN1C, KCNQ1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Beckwith- Wiedemann Sendromu (BWS), Russell-Silver Sendromu (RSS) (Delesyon + Metilasyon) (11p15, H19, IGF2, CDKN1C, KCNQ1) (MLPA) (Prenatal)	Beckwith-Wiedemann Syndrome (BWS), Russell-Silver Syndrome (RSS) (Deletion + Methylation) (11p15, H19, IGF2, CDKN1C, KCNQ1) (MLPA) (Prenatal)	11p15, H19, IGF2, CDKN1C, KCNQ1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (KCNQ1OT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (KCNQ1OT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1OT1	KCNQ1OT1, LIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (H19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (H19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (CDKN1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (CDKN1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN1C	CDKN1C, KIP2, BWS, IMAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:10424811). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA'lı Kan (2-4 ml)
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (MLPA)	H19	H19, D11S813E, ASM1, BWS, WT2	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (MLPA)	CDKN1C	CDKN1C, KIP2, BWS, IMAGE	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:10424811). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (KCNQ1OT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (KCNQ1OT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNQ1OT1	KCNQ1OT1, LIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (H19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (H19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (Prenatal) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (Prenatal) (MLPA)	H19	H19, D11S813E, ASM1, BWS, WT2	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant (Beckwith-Wiedemann Sendromu) (Prenatal) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant (Beckwith-Wiedemann syndrome) (Prenatal) (MLPA)	CDKN1C	CDKN1C, KIP2, BWS, IMAGE	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:10424811). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant; BWS (Beckwith-Wiedemann Sendromu) (NSD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant; BWS (Beckwith-Wiedemann syndrome) (NSD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSD1	NSD1, ARA267, STO, SOTOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:14997421). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant; BWS (Beckwith-Wiedemann Sendromu) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant; BWS (Beckwith-Wiedemann syndrome) (MLPA)	NSD1	NSD1, ARA267, STO, SOTOS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:14997421). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant; BWS (Beckwith-Wiedemann Sendromu) (NSD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant; BWS (Beckwith-Wiedemann syndrome) (NSD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSD1	NSD1, ARA267, STO, SOTOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:14997421). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beckwith-Wiedemann Sendromu, 130650, Otozomal Dominant; BWS (Beckwith-Wiedemann Sendromu) (Prenatal) (MLPA)	Beckwith-Wiedemann syndrome, 130650, Autosomal dominant; BWS (Beckwith-Wiedemann syndrome) (Prenatal) (MLPA)	NSD1	NSD1, ARA267, STO, SOTOS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. [ECO:0000269]PubMed:14997421). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Behr Sendromu, 210000, Otozomal Resesif; BEHRS (Behr Sendromu) (OPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Behr syndrome, 210000, Autosomal recessive; BEHRS (Behr syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Behr Sendromu, 210000, Otozomal Resesif; BEHRS (Behr Sendromu) (OPA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Behr syndrome, 210000, Autosomal recessive; BEHRS (Behr syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kafatasnmş Kemik Displazisi Sendromu, 614592, Otozomal Dominant; BBDS (FGFR2 İlişkili Kafatasnmş Kemik Displazisi) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bent bone dysplasia syndrome, 614592, Autosomal dominant; BBDS (FGFR2-related bent bone dysplasia) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kafatasnmş Kemik Displazisi Sendromu, 614592, Otozomal Dominant; BBDS (FGFR2 İlişkili Kafatasnmş Kemik Displazisi) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bent bone dysplasia syndrome, 614592, Autosomal dominant; BBDS (FGFR2-related bent bone dysplasia) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Benzen Toksisitesi, Yatkınlık (NQO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Benzene toxicity, susceptibility to (NQO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NQO1	NQO1, DIA4, NMOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bernard-Soulier Sendromu, Tip A1 (Resesif), 231200, Otozomal Resesif (Bernard-Soulier Sendromu) (GP1BA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bernard-Soulier syndrome, type A1 (recessive), 231200, Autosomal recessive (Bernard-Soulier) syndrome) (GP1BA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BA	GP1BA, BSS, BDPLT1, VWDP, BDPLT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bernard-Soulier Sendromu, Tip A2 (Dominant), 153670, Otozomal Dominant; BSSA2 (Bernard-Soulier Sendromu) (GP1BA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bernard-Soulier syndrome, type A2 (dominant), 153670, Autosomal dominant; BSSA2 (Bernard-Soulier syndrome) (GP1BA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BA	GP1BA, BSS, BDPLT1, VWDP, BDPLT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bernard-Soulier Sendromu, Tip B, 231200, Otozomal Resesif (Bernard-Soulier Sendromu) (GP1BB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bernard-Soulier syndrome, type B, 231200, Autosomal recessive (Bernard-Soulier syndrome) (GP1BB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BB	GP1BB, BS, BDPLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bernard-Soulier Sendromu, C, 231200, Otozomal Resesif; BSS (Bernard-Soulier Sendromu) (GP9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bernard-Soulier syndrome, type C, 231200, Autosomal recessive; BSS (Bernard-Soulier syndrome) (GP9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP9	GP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Berilyum Hastalığı, Kronik, Yatkınlık (HLA-DPB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beryllium disease, chronic, susceptibility to (HLA-DPB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DPB1	HLA-DPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bestrofinopati, Otozomal Resesif, 611809; ARB (Otozomal Resesif Bestrofinopati) (BEST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bestrophinopathy, autosomal recessive, 611809; ARB (Autosomal recessive bestrophinopathy) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beta-2-Adrenoreseptör Agonisti, Azalmış Yanıt (ADRB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beta-2-adrenoreceptor agonist, reduced response to (ADRB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB2	ADRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beta-Talasemi (Beta-Talasemi) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	BETA-THALASSEMIA (Beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beta-Talasemi (Beta-Talasemi) (MLPA)	BETA-THALASSEMIA (Beta-thalassemia) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Beta-Talasemi (Beta-Talasemi) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	BETA-THALASSEMIA (Beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beta-Talasemi (Beta-Talasemi) (Prenatal) (MLPA)	BETA-THALASSEMIA (Beta-thalassemia) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beta Talasemi (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beta-thalassemia (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beta Talasemi (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beta-thalassemia (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beta-Üreidopropionaz Eksikliği, 613161, Otozomal Resesif; UPB1D (Beta-Üreidopropionaz Eksikliği) (UPB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Beta-ureidopropionase deficiency, 613161, Autosomal recessive; UPB1D (Beta-ureidopropionase deficiency) (UPB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UPB1	UPB1, BUP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beta-ureidopropionase deficiency (BUPD) [MIM:613161]: An inborn error of metabolism due to a defect in pyrimidine degradation. It is characterized by muscular hypotonia, dystonic movements, scoliosis, microcephaly and severe developmental delay. Patients show strongly elevated levels of N-carbamyl-beta-alanine and N-carbamyl-beta-aminoisobutyric acid in plasma, cerebrospinal fluid and urine. [ECO:0000269]PubMed:15385443). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Beta-Üreidopropionaz Eksikliği, 613161, Otozomal Resesif. UPB1D (Beta-Ureidopropionaz Eksikliği) (UPB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Beta-ureidopropionase deficiency, 613161, Autosomal recessive; UPB1D (Beta-ureidopropionase deficiency) (UPB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UPB1	UPB1, BUP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beta-ureidopropionase deficiency (BUPD) [MIM:613161]: An inborn error of metabolism due to a defect in pyrimidine degradation. It is characterized by muscular hypotonia, dystonic movements, scoliosis, microcephaly and severe developmental delay. Patients show strongly elevated levels of N-carbamyl-beta-alanine and N-carbamyl-beta-aminoisobutyric acid in plasma, cerebrospinal fluid and urine. (ECO:0000269	PubMed:15385443). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bethlem Miyopati 1, 158810, Otozomal Resesif, Otozomal Dominant (Bethlem Miyopati) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bethlem myopathy 1, 158810, Autosomal recessive, Autosomal dominant (Bethlem myopathy) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bethlem Miyopati 1, 158810, Otozomal Resesif, Otozomal Dominant (Bethlem Miyopati) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bethlem myopathy 1, 158810, Autosomal recessive, Autosomal dominant (Bethlem myopathy) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bethlem Miyopati 1, 158810, Otozomal Resesif, Otozomal Dominant; BTHLM1 (Bethlem Miyopati) (COL6A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bethlem myopathy 1, 158810, Autosomal recessive, Autosomal dominant; BTHLM1 (Bethlem myopathy) (COL6A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A3	COL6A3, DYT27, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:10399756, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:9536084). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bethlem Miyopati 1, 158810, Otozomal Resesif, Otozomal Dominant; BTHLM1 (Bethlem Miyopati) (COL6A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bethlem myopathy 1, 158810, Autosomal recessive, Autosomal dominant; BTHLM1 (Bethlem myopathy) (COL6A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL6A3	COL6A3, DYT27, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:10399756, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:9536084). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bethlem Miyopati 2, 616471, BTHLM2 (Bethlem Miyopati) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bethlem myopathy 2, 616471; BTHLM2 (Bethlem myopathy) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Bethlem Miyopati 2, 616471, BTHLM2 (Bethlem Miyopati) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bethlem myopathy 2, 616471; BTHLM2 (Bethlem myopathy) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bietti Kristalin Comeoretina Distrofisi, 210370, Otozomal Resesif. BCD (Bietti Kristalin Distrofisi) (CYP4V2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bietti crystalline corneoretinal dystrophy, 210370, Autosomal recessive; BCD (Bietti crystalline dystrophy) (CYP4V2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP4V2	CYP4V2, BCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bifid Burun, ± Anorektal ve Böbrek Anomalileri, 608980 (BNAR Sendromu) (FREM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bifid nose with or without anorectal and renal anomalies, 608980 (BNAR syndrome) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bifid Burun, ± Anorektal ve Böbrek Anomalileri, 608980 (BNAR Sendromu) (FREM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bifid nose with or without anorectal and renal anomalies, 608980 (BNAR syndrome) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Emilim Bozukluğu, Primer, 613291, Otozomal Resesif (SLC10A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid malabsorption, primary, 613291, Autosomal recessive (SLC10A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC10A2	SLC10A2, NTCP2, PBAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary bile acid malabsorption (PBAM) [MIM:613291]: An intestinal disorder associated with chronic watery diarrhea, excess fecal bile acids, steatorrhea and interruption of the enterohepatic circulation of bile acids. (ECO:0000269	PubMed:9109432). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 1, 607765, Otozomal Resesif; CBAS1 (Konjenital Safra Asidi Sentezi Defekti Tip 1) (HSD3B7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 1, 607765, Autosomal recessive; CBAS1 (Congenital bile acid synthesis defect type 1) (HSD3B7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD3B7	HSD3B7, CBAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 1 (CBAS1) [MIM:607765]: A primary defect in bile synthesis leading to progressive liver disease. Clinical features include neonatal jaundice, severe intrahepatic cholestasis, cirrhosis. (ECO:0000269|PubMed:11067870, ECO:0000269|PubMed:12679481). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 1, 607765, Otozomal Resesif; CBAS1 (Konjenital Safra Asidi Sentezi Defekti Tip 1) (HSD3B7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 1, 607765, Autosomal recessive; CBAS1 (Congenital bile acid synthesis defect type 1) (HSD3B7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD3B7	HSD3B7, CBAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital bile acid synthesis defect 1 (CBAS1) [MIM:607765]: A primary defect in bile synthesis leading to progressive liver disease. Clinical features include neonatal jaundice, severe intrahepatic cholestasis, cirrhosis. (ECO:0000269|PubMed:11067870, ECO:0000269|PubMed:12679481). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 2, 235555, Otozomal Resesif; CBAS2 (Konjenital Safra Asidi Sentezi Defekti Tip 2) (AKR1D1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 2, 235555, Autosomal recessive; CBAS2 (Congenital bile acid synthesis defect type 2) (AKR1D1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKR1D1	AKR1D1, SRD5B1, CBAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 2, 235555, Otozomal Resesif; CBAS2 (Konjenital Safra Asidi Sentezi Defekti Tip 2) (AKR1D1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 2, 235555, Autosomal recessive; CBAS2 (Congenital bile acid synthesis defect type 2) (AKR1D1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AKR1D1	AKR1D1, SRD5B1, CBAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 3, 613812, Otozomal Resesif; CBAS3 (Konjenital Safra Asidi Sentez Defekti Tip 3) (CYP7B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 3, 613812, Autosomal recessive; CBAS3 (Congenital bile acid synthesis defect type 3) (CYP7B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP7B1	CYP7B1, CBAS3, SPG5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 3 (CBAS3) [MIM:613812]: A disorder resulting in severe cholestasis, cirrhosis and liver synthetic failure. Hepatic microsomal oxysterol 7-alpha-hydroxylase activity is undetectable. (ECO:0000269	PubMed:9802883). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 3, 613812, Otozomal Resesif; CBAS3 (Konjenital Safra Asidi Sentez Defekti Tip 3) (CYP7B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 3, 613812, Autosomal recessive; CBAS3 (Congenital bile acid synthesis defect type 3) (CYP7B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP7B1	CYP7B1, CBAS3, SPG5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital bile acid synthesis defect 3 (CBAS3) [MIM:613812]: A disorder resulting in severe cholestasis, cirrhosis and liver synthetic failure. Hepatic microsomal oxysterol 7-alpha-hydroxylase activity is undetectable. (ECO:0000269	PubMed:9802883). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 4, 214950, Otozomal Resesif; CBAS4 (Konjenital Safra Asidi Sentezi Defekti, Tip 4) (AMACR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 4, 214950, Autosomal recessive; CBAS4 (Congenital bile acid synthesis defect type 4) (AMACR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMACR	AMACR, CBAS4, AMACRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 4 (CBAS4) [MIM:214950]: A disorder characterized by the presence of trihydroxycoprostanic acid in the bile and absence of cholic acid. Patients manifest neonatal jaundice, intrahepatic cholestasis and bile duct deficiency. (ECO:0000269|PubMed:10655068, ECO:0000269|PubMed:12512044). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 4, 214950, Otozomal Resesif; CBAS4 (Konjenital Safra Asidi Sentezi Defekti, Tip 4) (AMACR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 4, 214950, Autosomal recessive; CBAS4 (Congenital bile acid synthesis defect type 4) (AMACR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMACR	AMACR, CBAS4, AMACRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital bile acid synthesis defect 4 (CBAS4) [MIM:214950]: A disorder characterized by the presence of trihydroxycoprostanic acid in the bile and absence of cholic acid. Patients manifest neonatal jaundice, intrahepatic cholestasis and bile duct deficiency. (ECO:0000269|PubMed:10655068, ECO:0000269|PubMed:12512044). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 5, 616278, Otozomal Resesif; CBAS5 (ABCD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 5, 616278, Autosomal recessive; CBAS5 (ABCD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCD3	ABCD3, PXMP1, PMP70, CBAS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 5 (CBAS5) [MIM:616278]: An autosomal recessive disorder characterized by hepatosplenomegaly, hepatic fibrosis, progressive liver failure, and accumulation of peroxisomal C27-bile acid intermediates in plasma. (ECO:0000269	PubMed:25168382). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 5, 616278, Otozomal Resesif; CBAS5 (ABCD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 5, 616278, Autosomal recessive; CBAS5 (ABCD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCD3	ABCD3, PXMP1, PMP70, CBAS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital bile acid synthesis defect 5 (CBAS5) [MIM:616278]: An autosomal recessive disorder characterized by hepatosplenomegaly, hepatic fibrosis, progressive liver failure, and accumulation of peroxisomal C27-bile acid intermediates in plasma. (ECO:0000269	PubMed:25168382). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 6, 617308, Otozomal Resesif (ACOX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bile acid synthesis defect, congenital, 6, 617308, Autosomal recessive (ACOX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACOX2	ACOX2, BRCACOX, CBAS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Safra Asidi Sentez Defekti, Konjenital, 6, 617308, Otozomal Resesif (ACOX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bile acid synthesis defect, congenital, 6, 617308, Autosomal recessive (ACOX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACOX2	ACOX2, BRCACOX, CBAS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Biotinidaz Eksikliği, 253260, Otozomal Resesif (Biotinidaz Eksikliği) (BTD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Biotinidase deficiency, 253260, Autosomal recessive (Biotinidase deficiency) (BTD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BTD	BTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Biotinidase deficiency (BTD deficiency) [MIM:253260]: A juvenile form of multiple carboxylase deficiency, an autosomal recessive disorder of biotin metabolism, characterized by ketoacidosis, hyperammonemia, excretion of abnormal organic acid metabolites, and dermatitis. Biotinidase deficiency is characterized by seizures, hypotonia, skin rash, alopecia, ataxia, hearing loss, and optic atrophy. If untreated, symptoms usually become progressively worse, and coma and death may occur. (ECO:0000269|PubMed:10206677, ECO:0000269|PubMed:9099842, ECO:0000269|PubMed:9654207). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Biotinidaz Eksikliği, 253260, Otozomal Resesif (Biotinidaz Eksikliği) (BTD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Biotinidase deficiency, 253260, Autosomal recessive (Biotinidase deficiency) (BTD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BTD	BTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Biotinidase deficiency (BTD deficiency) [MIM:253260]: A juvenile form of multiple carboxylase deficiency, an autosomal recessive disorder of biotin metabolism, characterized by ketoacidosis, hyperammonemia, excretion of abnormal organic acid metabolites, and dermatitis. Biotinidase deficiency is characterized by seizures, hypotonia, skin rash, alopecia, ataxia, hearing loss, and optic atrophy. If untreated, symptoms usually become progressively worse, and coma and death may occur. (ECO:0000269|PubMed:10206677, ECO:0000269|PubMed:9099842, ECO:0000269|PubMed:9654207). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Birk-Barel Mental Retardasyon Dismorfizm Sendromu, 612292 (Mental Retardasyon, Birk-Barel Tipi) (KCNK9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Birk-Barel mental retardation dysmorphism syndrome, 612292 (Intellectual disability, Birk-Barel type) (KCNK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNK9	KCNK9, TASK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Birk-Barel Mental Retardasyon Dismorfizm Sendromu, 612292 (Mental Retardasyon, Birk-Barel Tipi) (KCNK9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Birk-Barel mental retardation dysmorphism syndrome, 612292 (Intellectual disability, Birk-Barel type) (KCNK9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNK9	KCNK9, TASK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Birt-Hogg-Dubé sendromu (FLCN 17p11, Smith-Magenis sendromu bölgesi) (MLPA)	Birt-Hogg-Dubé syndrome (FLCN 17p11, Smith-Magenis syndrome region) (MLPA)	FLCN 17p11, Smith-Magenis sendromu bölgesi	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Birt-Hogg-Dube Sendromu, 135150, Otozomal Dominant; BHD (Birt-Hogg-Dubé Sendromu) (FLCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Birt-Hogg-Dube syndrome, 135150, Autosomal dominant; BHD (Birt-Hogg-Dubé syndrome) (FLCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLCN	FLCN, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Birt-Hogg-Dube syndrome (BHD) [MIM:135150]: A rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of Birt- Hogg-Dube syndrome skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. Birt-Hogg-Dube syndrome is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in Birt-Hogg- Dube syndrome patients. {ECO:0000269|PubMed:12204536, ECO:0000269|PubMed:15852235, ECO:0000269|PubMed:18234728, ECO:0000269|PubMed:19785621}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Birt-Hogg-Dube Sendromu, 135150, Otozomal Dominant; BHD (Birt-Hogg-Dubé Sendromu) (MLPA)	Birt-Hogg-Dube syndrome, 135150, Autosomal dominant; BHD (Birt-Hogg-Dubé syndrome) (MLPA)	FLCN	FLCN, BHD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Birt-Hogg-Dube syndrome (BHD) [MIM:135150]: A rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of Birt- Hogg-Dube syndrome skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. Birt-Hogg-Dube syndrome is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in Birt-Hogg- Dube syndrome patients. {ECO:0000269|PubMed:12204536, ECO:0000269|PubMed:15852235, ECO:0000269|PubMed:18234728, ECO:0000269|PubMed:19785621}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Birt-Hogg-Dube Sendromu, 135150, Otozomal Dominant; BHD (Birt-Hogg-Dubé Sendromu) (FLCN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Birt-Hogg-Dube syndrome, 135150, Autosomal dominant; BHD (Birt-Hogg-Dubé syndrome) (FLCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLCN	FLCN, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Birt-Hogg-Dube syndrome (BHD) [MIM:135150]: A rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of Birt- Hogg-Dube syndrome skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. Birt-Hogg-Dube syndrome is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in Birt-Hogg- Dube syndrome patients. {ECO:0000269|PubMed:12204536, ECO:0000269|PubMed:15852235, ECO:0000269|PubMed:18234728, ECO:0000269|PubMed:19785621}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bjornstad Sendromu, 262000, Otozomal Resesif; BJS (Björnstad Sendromu) (BCS1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bjornstad syndrome, 262000, Autosomal recessive; BJS (Björnstad syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bjornstad Sendromu, 262000, Otozomal Resesif; BJS (Björnstad Sendromu) (BCS1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bjornstad syndrome, 262000, Autosomal recessive; BJS (Björnstad syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mesane Kanseri, Somatik, 109800 (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bladder cancer, somatic, 109800 (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mesane Kanseri, Somatik, 109800 (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bladder cancer, somatic, 109800 (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Bladder cancer (BLC) [MIM:109800]: A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas that begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. {ECO:0000269|PubMed:6298635, ECO:0000269|PubMed:6844927}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mesane Kanseri, Somatik, 109800 (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bladder cancer, somatic, 109800 (KRAS gene) (Sequence Analysis) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mesane Kanseri, Somatik, 109800 (RB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bladder cancer, somatic, 109800 (RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1	RB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Bladder cancer (BLC) [MIM:109800]: A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas that begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Blau Sendromu, 186580, Otozomal Dominant; BLAUS (Blau Sendromu) (NOD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blau syndrome, 186580, Autosomal dominant; BLAUS (Blau syndrome) (NOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOD2	NOD2, CARD15, IBD1, CD, YAOS, BLAUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blau syndrome (BLAUS) [MIM:186580]: A rare autosomal dominant disorder characterized by early-onset granulomatous arthritis, uveitis and skin rash. {ECO:0000269|PubMed:11526384, ECO:0000269|PubMed:15812565, ECO:0000269|PubMed:19116920, ECO:0000269|PubMed:19169908, ECO:0000269|PubMed:19479837, ECO:0000269|PubMed:20199415, ECO:0000269|PubMed:24960071, ECO:0000269|PubMed:25093298, ECO:0000269|PubMed:25692065, ECO:0000269|PubMed:25724124}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kanama Bozukluğu, P2RX1 Defekti, Somatik, 609821, BDPLT8 (P2Y12 Defekti) (P2RX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder due to P2RX1 defect, somatic, 609821, BDPLT8 (P2Y12 defect) (P2RX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P2RX1	P2RX1, P2X1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 11, 614201, Otozomal Resesif; BDPLT11 (Kanama Diyatezi, Kolajen Reseptör Defektine Bağlı) (GP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 11, 614201, Autosomal recessive; BDPLT11 (Bleeding diathesis due to a collagen receptor defect) (GP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP6	GP6, GPIV, BDPLT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 11 (BDPLT11) [MIM:614201]: A mild to moderate bleeding disorder caused by defective platelet activation and aggregation in response to collagen. {ECO:0000269|PubMed:19549989, ECO:0000269|PubMed:19553682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 13, Yatkınlık, 614009, Otozomal Dominant; GHDD (Kanama Diyatezi, Tromboksan Sentez Eksikliğine Bağlı) (TBXA2R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 13, susceptibility to, 614009, Autosomal dominant; GHDD (Bleeding diathesis due to thromboxane synthesis deficiency) (TBXA2R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBXA2R	TBXA2R, BDPLT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 13 (BDPLT13) [MIM:614009]: A disorder characterized by reduced platelet aggregation and a tendency to mild mucocutaneous bleeding. {ECO:0000269|PubMed:7929844}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 15, 615193, Otozomal Dominant; BDPLT15 (Otozomal Dominant Makrotrombositopeni) (ACTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 15, 615193, Autosomal dominant; BDPLT15 (Autosomal dominant macrothrombocytopenia) (ACTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN1	ACTN1, BDPLT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 15 (BDPLT15) [MIM:615193]: An autosomal dominant form of macrothrombocytopenia. Affected individuals usually have no or only mild bleeding tendency, such as epistaxis. Laboratory studies show decreased numbers of large platelets and anisocytosis, but the platelets show no in vitro functional abnormalities. {ECO:0000269|PubMed:23434115, ECO:0000269|PubMed:24069336}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 16, Otozomal Dominant, 187800, Otozomal Dominant (Otozomal Dominant Makrotrombositopeni) (ITGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800, Autosomal dominant (Autosomal dominant macrothrombocytopenia) (ITGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB3	ITGB3, GP3A, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (BDPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. {ECO:0000269|PubMed:18065693}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 16, Otozomal Dominant, 187800, Otozomal Dominant; BDPLT16 (Otozomal Dominant Makrotrombositopeni) (ITGA2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 16, autosomal dominant, 187800, Autosomal dominant; BDPLT16 (Autosomal dominant macrothrombocytopenia) (ITGA2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA2B	ITGA2B, GP2B, CD41B, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (BDPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. {ECO:0000269|PubMed:21454453, ECO:0000269|PubMed:9834222}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kanama Bozukluğu, Trombosit Tipi, 17, 187900, Otozomal Dominant; BDPLT17 (Gri Trombosit Sendromu) (GF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 17, 187900, Autosomal dominant; BDPLT17 (Gray platelet syndrome) (GF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GF1B	GF1B, BDPLT17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 17 (BDPLT17) [MIM:187900]: An autosomal dominant disorder characterized by increased bleeding tendency due to platelet dysfunction, and associated with macrothrombocytopenia and red cell anisopoikilocytosis. Platelets appear abnormal on light microscopy, while electron microscopy shows a heterogeneous decrease of alpha granules within platelets. Bone marrow biopsy shows increased numbers of abnormal megakaryocytes, suggesting a defect in megakaryopoiesis and platelet production. The severity of bleeding is variable with some affected individuals experiencing spontaneous bleeding while other exhibit only abnormal bleeding with surgery. (ECO:0000269|PubMed:23927492, ECO:0000269|PubMed:24325358). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 18, 615888, Otozomal Resesif; BDPLT18 (CalDAG-GEFI Eksikliğine Kanama Bozukluğu Diyatezi) (RASGRP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 18, 615888, Autosomal recessive; BDPLT18 (Bleeding disorder due to CalDAG-GEFI deficiency) (RASGRP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RASGRP2	RASGRP2, CDC25L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 18 (BDPLT18) [MIM:615888]: A disorder characterized by increased bleeding tendency due to platelet dysfunction. Clinical features include epistaxis, hematomas, bleeding after tooth extraction, and menorrhagia. (ECO:0000269	PubMed:24958846). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 19, 616176, Otozomal Resesif; BDPLT19 (Ağır Otozomal Resesif Makrotrombositopeni) (PRKACG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 19, 616176, Autosomal recessive; BDPLT19 (Severe autosomal recessive macrothrombocytopenia) (PRKACG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKACG	PRKACG, BDPLT19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 20, 616913, Otozomal Dominant; BDPLT20 (Otozomal Dominant Trombositopeni-Trombosit Salgılama Defekti) (SLFN14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 20, 616913, Autosomal dominant; BDPLT20 (Autosomal dominant thrombocytopenia with platelet secretion defect) (SLFN14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLFN14	SLFN14, BDPLT20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kanama Bozukluğu, Trombosit Tipi, 8, 609821, Otozomal Resesif (P2Y12 Defekti) (P2RY12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bleeding disorder, platelet-type, 8, 609821, Autosomal recessive (P2Y12 defect) (P2RY12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P2RY12	P2RY12, P2Y12, BDPLT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 8 (BDPLT8) [MIM:609821]: A condition characterized by mild to moderate mucocutaneous bleeding, and excessive bleeding after surgery or trauma. The defect is due to severe impairment of platelet response to ADP resulting in defective platelet aggregation. (ECO:0000269|PubMed:11196645, ECO:0000269|PubMed:12578987, ECO:0000269|PubMed:25428217). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Blefarofimozis, Epikantus İnversus ve Ptozis, Tip 1, 110100, Otozomal Dominant; BPES (Blefarofimozis-Epikantus İnversus-Ptoz Sendromu) (FOXL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100, Autosomal dominant; BPES (Blepharophimosis-epicanthus inversus-ptosis syndrome) (FOXL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. (ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2). Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Blefarofimozis, Epikantus İnversus ve Ptozis, Tip 1, 110100, Otozomal Dominant; BPES (Blefarofimozis-Epikantus İnversus-Ptoz Sendromu) (MLPA)	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100, Autosomal dominant; BPES (Blepharophimosis-epicanthus inversus-ptosis syndrome) (MLPA)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. (ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2). Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Blefarofimozis, Epikantus İnversus ve Ptozis, Tip 2, 110100, Otozomal Dominant; BPES (Blefarofimozis-Epikantus İnversus-Ptoz Sendromu) (FOXL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100, Autosomal dominant; BPES (Blepharophimosis-epicanthus inversus-ptosis syndrome) (FOXL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. (ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2). Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Blefarofimozis, Epikantus İnversus ve Ptozis, Tip 2, 110100, Otozomal Dominant; BPES (Blefarofimozis-Epikantus İnversus-Ptoz Sendromu) (MLPA)	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100, Autosomal dominant; BPES (Blepharophimosis-epicanthus inversus-ptosis syndrome) (MLPA)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. (ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2). Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Blefarospazm, Primer Benign, 606798, Sporadik Vakalar (DRD5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blepharospasm, primary benign, 606798, Isolated cases (DRD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD5	DRD5, DRD1B, DRD1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kan Grubu - Lutheran İnhibitörü, 111150 (KLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blood group--Lutheran inhibitor, 111150 (KLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Bloom Sendromu, 210900, Otozomal Resesif; BLM (Bloom Sendromu) (BLM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bloom syndrome, 210900, Autosomal recessive; BLM (Bloom syndrome) (BLM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLM	RECQL3, RECQ2, BLM, BS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bloom syndrome (BLM) [MIM:210900]: An autosomal recessive disorder. It is characterized by proportionate pre- and postnatal growth deficiency, sun-sensitive telangiectatic hypo- and hyperpigmented skin, predisposition to malignancy, and chromosomal instability. (ECO:0000269|PubMed:10862105, ECO:0000269|PubMed:7585968, ECO:0000269|PubMed:9285778). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bloom Sendromu, 210900, Otozomal Resesif; BLM (Bloom Sendromu) (BLM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bloom syndrome, 210900, Autosomal recessive; BLM (Bloom syndrome) (BLM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BLM	RECQL3, RECQ2, BLM, BS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bloom syndrome (BLM) [MIM:210900]: An autosomal recessive disorder. It is characterized by proportionate pre- and postnatal growth deficiency, sun-sensitive telangiectatic hypo- and hyperpigmented skin, predisposition to malignancy, and chromosomal instability. (ECO:0000269|PubMed:10862105, ECO:0000269|PubMed:7585968, ECO:0000269|PubMed:9285778). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mavi Kon Monokromazi, 303700, X'e Bağlı (Mavi Kon Monokromatizm) (OPN1MW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blue cone monochromacy, 303700, X-linked recessive (Blue cone monochromatism) (OPN1MW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPN1MW	OPN1MW, GCP, CBD, CBBM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blue cone monochromacy (BCM) [MIM:303700]: A rare X- linked congenital stationary cone dysfunction syndrome characterized by the absence of functional long wavelength-sensitive and medium wavelength-sensitive cones in the retina. Color discrimination is severely impaired from birth, and vision is derived from the remaining preserved blue (S) cones and rod photoreceptors. BCM typically presents with reduced visual acuity, pendular nystagmus, and photophobia. Patients often have myopia. (ECO:0000269	PubMed:8966378). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mavi Kon Monokromazi, 303700, X'e Bağlı Resesif; BCM (Mavi Kon Monokromatizmi) (OPN1LW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Blue cone monochromacy, 303700, X-linked recessive; BCM (Blue cone monochromatism) (OPN1LW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPN1LW	OPN1LW, RCP, CBP, CBBM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blue cone monochromacy (BCM) [MIM:303700]: A rare X- linked congenital stationary cone dysfunction syndrome characterized by the absence of functional long wavelength-sensitive and medium wavelength-sensitive cones in the retina. Color discrimination is severely impaired from birth, and vision is derived from the remaining preserved blue (S) cones and rod (photoreceptors. BCM typically presents with reduced visual acuity, pendular nystagmus, and photophobia. Patients often have myopia. {ECO:0000269|PubMed:8213841, ECO:0000269|PubMed:8666378}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bohring Opitz Sendromu, 605039, Otozomal Dominant; BOPS (Bohring-Opitz Sendromu) (ASXL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bohring-Opitz syndrome, 605039, Autosomal dominant; BOPS (Bohring-Opitz syndrome) (ASXL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASXL1	ASXL1, KIAA0978, BOPS, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bohring-Opitz syndrome (BOPS) [MIM:605039]: A syndrome characterized by severe intrauterine growth retardation, poor feeding, profound mental retardation, trigonocephaly, prominent metopic suture, exophthalmos, nevus flammeus of the face, upslanting palpebral fissures, hirsutism, and flexion of the elbows and wrists with deviation of the wrists and metacarpophalangeal joints. {ECO:0000269|PubMed:21706002}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bohring Opitz Sendromu, 605039, Otozomal Dominant; BOPS (Bohring-Opitz Sendromu) (ASXL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bohring-Opitz syndrome, 605039, Autosomal dominant; BOPS (Bohring-Opitz syndrome) (ASXL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASXL1	ASXL1, KIAA0978, BOPS, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bohring-Opitz syndrome (BOPS) [MIM:605039]: A syndrome characterized by severe intrauterine growth retardation, poor feeding, profound mental retardation, trigonocephaly, prominent metopic suture, exophthalmos, nevus flammeus of the face, upslanting palpebral fissures, hirsutism, and flexion of the elbows and wrists with deviation of the wrists and metacarpophalangeal joints. {ECO:0000269|PubMed:21706002}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bombay Fenotipi - Para Bombay Fenotipi , H Antijeni (FUT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	BOMBAY PHENOTYPE - PARA-BOMBAY PHENOTYPE (FUT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT1	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 1, 614675, Otozomal Dominant; BMFS1 (Otozomal Dominant Aplazisi ve Myelodisplazi (SRP72 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone marrow failure syndrome 1, 614675, Autosomal dominant; BMFS1 (Autosomal dominant aplasia and myelodysplasia) (SRP72 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRP72	SRP72, BMFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bone marrow failure syndrome 1 (BMFS1) [MIM:614675]: An autosomal dominant disease characterized by aplastic anemia and myelodysplasia resulting from bone marrow failure. Aplastic anemia is a form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. Myelodysplasia is a clonal hematopoietic stem cell disorder in which immature cells in the bone marrow become malformed and dysfunctional. {ECO:0000269|PubMed:22541560}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 1, 614675, Otozomal Dominant; BMFS1 (Otozomal Dominant Aplazisi ve Myelodisplazi) (SRP72 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bone marrow failure syndrome 1, 614675, Autosomal dominant; BMFS1 (Autosomal dominant aplasia and myelodysplasia) (SRP72 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRP72	SRP72, BMFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bone marrow failure syndrome 1 (BMFS1) [MIM:614675]: An autosomal dominant disease characterized by aplastic anemia and myelodysplasia resulting from bone marrow failure. Aplastic anemia is a form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. Myelodysplasia is a clonal hematopoietic stem cell disorder in which immature cells in the bone marrow become malformed and dysfunctional. {ECO:0000269|PubMed:22541560}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 2, 615715, Otozomal Resesif; BMFS2 (Pansitopeni-Gelişimsel Gecikme Sendromu) (ERCC6L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone marrow failure syndrome 2, 615715, Autosomal recessive; BMFS2 (Pancytopenia-developmental delay syndrome) (ERCC6L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6L2	ERCC6L2, RAD26L, BMFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bone marrow failure syndrome 2 (BMFS2) [MIM:615715]: An autosomal recessive disorder characterized by trilineage bone marrow failure, bone marrow hypocellularity, learning difficulties, and microcephaly. Insufficient hematopoiesis results in peripheral blood cytopenias, affecting myeloid, erythroid and megakaryocyte lines. Cutaneous features and increased chromosome breakage are not features. {ECO:0000269|PubMed:24507776}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 2, 615715, Otozomal Resesif; BMFS2 (Pansitopeni-Gelişimsel Gecikme Sendromu) (ERCC6L2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bone marrow failure syndrome 2, 615715, Autosomal recessive; BMFS2 (Pancytopenia-developmental delay syndrome) (ERCC6L2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC6L2	ERCC6L2, RAD26L, BMFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bone marrow failure syndrome 2 (BMFS2) [MIM:615715]: An autosomal recessive disorder characterized by trilineage bone marrow failure, bone marrow hypocellularity, learning difficulties, and microcephaly. Insufficient hematopoiesis results in peripheral blood cytopenias, affecting myeloid, erythroid and megakaryocyte lines. Cutaneous features and increased chromosome breakage are not features. {ECO:0000269|PubMed:24507776}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 3, 617052, Otozomal Resesif; BMFS3 (DNAJC21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone marrow failure syndrome 3, 617052, Autosomal recessive; BMFS3 (DNAJC21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC21	DNAJC21, DNAJA5, BMFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kemik İliği Yetmezliği Sendromu 3, 617052, Otozomal Resesif; BMFS3 (DNAJC21 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bone marrow failure syndrome 3, 617052, Autosomal recessive; BMFS3 (DNAJC21 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNAJC21	DNAJC21, DNAJA5, BMFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kemik İliği Kromozom Analizi	Bone Marrow- Chromosome analysis			Kromozom analizi/ Karyotype analysis	Heparinli Kemik İliği (2-3 ml)	*	15	45			21	Postnatal			Heparinized bone marrow
Kemik Mineral Yoğunluğu QTL 12, Osteoporoz, 612560 (UGT2B17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone mineral density QTL 12, osteoporosis, 612560 (UGT2B17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT2B17	UGT2B17, BMND12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kemik Mineral Yoğunluğu QTL18, Osteoporoz, 300910, X'e Bağlı Dominant (X'e Bağlı Osteoporoz-Fraktürler) (PLS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone mineral density QTL18, osteoporosis, 300910, X-linked dominant (X-linked osteoporosis with fractures) (PLS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLS3	PLS3, BMND18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. {ECO:0000269|PubMed:24088043}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kemik Mineral Yoğunluğu Kantitatif Karakter Loküsü 16; Bmnd16 (Idiopatik Jüvenil Osteoporoz) (WNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	BONE MINERAL DENSITY QUANTITATIVE TRAIT LOCUS 16; BMND16 (Idiopathic juvenile osteoporosis) (WNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT1	WNT1, INT1, OI15, BMND16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. {ECO:0000269|PubMed:23499309, ECO:0000269|PubMed:23656646}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kemik Mineral Yoğunluğu Değişimi QTL, Osteoporoz, 166710, Otozomal Dominant (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone mineral density variation QTL, osteoporosis, 166710, Autosomal dominant (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kemik Mineral Yoğunluğu Değişimi QTL, Osteoporoz, 166710, Otozomal Dominant (MLPA)	Bone mineral density variation QTL, osteoporosis, 166710, Autosomal dominant (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kemik Mineral Yoğunluğu, Düşük, Yatkınlık, 615311 (LGR4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bone mineral density, low, susceptibility to, 615311 (LGR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LGR4	LGR4, GPR48, BNMD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. {ECO:0000269|PubMed:23644456}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. A LGR4 nonsense mutation creating a stop codon after position 126 (c.376C>T) is strongly associated with low bone mineral density and osteoporotic fractures (PubMed:23644456). This mutation probably causes degradation of the transcript by nonsense-mediated decay (NMD). The c.376C>T mutation is also associated with electrolyte imbalance, late onset of menarche and reduced testosterone levels, as well as an increased risk of squamous cell carcinoma of the skin and biliary tract cancer (PubMed:23644456). {ECO:0000269|PubMed:23644456}.;	EDTA Blood Sample
Boomerang Displazi, 112310, Otozomal Dominant (Boomerang Displazi) (FLNB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Boomerang dysplasia, 112310, Autosomal dominant (Boomerang dysplasia) (FLNB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Boomerang Displazi, 112310, Otozomal Dominant (Boomerang Displazi) (FLNB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Boomerang dysplasia, 112310, Autosomal dominant (Boomerang dysplasia) (FLNB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bor-Duane Hidrosefali Komşu Gen Sendromu, 600257, Otozomal Dominant (2200)	Bor-Duane hydrocephalus contiguous gene syndrome, 600257, Autosomal dominant (440)	.	DEL8q12q21, C8DELq12q21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Bor-Duane Hidrosefali Komşu Gen Sendromu, 600257, Otozomal Dominant (Prenatal)	Bor-Duane hydrocephalus contiguous gene syndrome, 600257, Autosomal dominant (Prenatal)	.	DEL8q12q21, C8DELq12q21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Borjeson-Forssman-Lehmann Sendromu, 301900, X'e Bağlı Resesif; BFLS (Borjeson-Forssman-Lehmann Sendromu) (PHF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Borjeson-Forssman-Lehmann syndrome, 301900, X-linked recessive; BFLS (Borjeson-Forssman-Lehmann syndrome) (PHF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHF6	PHF6, BFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Borjeson-Forssman-Lehmann syndrome (BFLS) [MIM:301900]: A X-linked recessive disorder characterized by moderate to severe mental retardation, epilepsy, hypogonadism, hypometabolism, obesity with marked gynecomastia, swelling of subcutaneous tissue of the face, narrow palpebral fissure and large but not deformed ears. {ECO:0000269|PubMed:12415272}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Borjeson-Forssman-Lehmann Sendromu, 301900, X'e Bağlı Resesif; BFLS (Borjeson-Forssman-Lehmann Sendromu) (PHF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Borjeson-Forssman-Lehmann syndrome, 301900, X-linked recessive; BFLS (Borjeson-Forssman-Lehmann syndrome) (PHF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHF6	PHF6, BFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Borjeson-Forssman-Lehmann syndrome (BFLS) [MIM:301900]: A X-linked recessive disorder characterized by moderate to severe mental retardation, epilepsy, hypogonadism, hypometabolism, obesity with marked gynecomastia, swelling of subcutaneous tissue of the face, narrow palpebral fissure and large but not deformed ears. {ECO:0000269|PubMed:12415272}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bosch Boonstra-Schaaf Optik Atrofi Sendromu, 615722, Otozomal Dominant; BBSOAS (Optik Atrofi-Mental Retardasyon Sendromu) (NR2F1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722, Autosomal dominant; BBSOAS (Optic atrophy-intellectual disability syndrome) (NR2F1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR2F1	NR2F1, TFCOUP1, ERBAL3, EAR3, BBSOAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS) [MIM:615722]: An autosomal dominant disorder characterized by optic atrophy associated with developmental delay and intellectual disability. Most patients also have evidence of cerebral visual impairment. {ECO:0000269|PubMed:24462372}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Bosch Boonstra-Schaaf Optik Atrofi Sendromu, 615722, Otozomal Dominant, BBSOAS (Optik Atrofi-Mental Retardasyon Sendromu) (NR2F1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bosch-Boonstra-Schaaf optic atrophy syndrome, 615722, Autosomal dominant; BBSOAS (Optic atrophy-intellectual disability syndrome) (NR2F1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR2F1	NR2F1, TFCOUP1, ERBAL3, EAR3, BBSOAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bosch-Boonstra-Schaaf optic atrophy syndrome (BBSOAS) [MIM:615722]: An autosomal dominant disorder characterized by optic atrophy associated with developmental delay and intellectual disability. Most patients also have evidence of cerebral visual impairment. {ECO:0000269	PubMed:24462372}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Boskey Salih-Alorainy Sendromu, 601536 (Boskey Salih-Alorainy Sendromu (Atabaşkan Beyin Dişgenezisi Sendromu)) (HOXA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Boskey-Salih-Alorainy syndrome, 601536 (Boskey-Salih-Alorainy syndrome) (HOXA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA1	HOXA1, HOX1F, BSAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Boskey Salih-Alorainy Sendromu, 601536 (Boskey Salih-Alorainy Sendromu (Atabaşkan Beyin Dişgenezisi Sendromu)) (HOXA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Boskey-Salih-Alorainy syndrome, 601536 (Boskey-Salih-Alorainy syndrome) (HOXA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXA1	HOXA1, HOX1F, BSAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bosma Arini Mikroftalmi Sendromu, 603457, Otozomal Dominant; BAMS (Arini-Koanal Atrezi-Mikroftalmi Sendromu) (SMCHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bosma arhinia microphthalmia syndrome, 603457, Autosomal dominant; BAMS (Arrhinia-choanal atresia-microphthalmia syndrome) (SMCHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMCHD1	SMCHD1, KIAA0650, BAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bosma Arini Mikroftalmi Sendromu, 603457, Otozomal Dominant; BAMS (Arini-Koanal Atrezi-Mikroftalmi Sendromu) (SMCHD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bosma arhinia microphthalmia syndrome, 603457, Autosomal dominant; BAMS (Arrhinia-choanal atresia-microphthalmia syndrome) (SMCHD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMCHD1	SMCHD1, KIAA0650, BAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bothnia Retina Distrofisi, 607475, Otozomal Resesif (Bothnia Retina Distrofi (RLBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bothnia retinal dystrophy, 607475, Autosomal recessive (Bothnia retinal dystrophy) (RLBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RLBP1	RLBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Boucher-Neuhauser Sendromu, 215470, Otozomal Resesif; BHHS (Ataksi-Hipogonadizm-Koroid Distrofi Sendromu (Boucher-Neuhauser Sendromu)) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Boucher-Neuhauser syndrome, 215470, Autosomal recessive; BHHS (Ataxia-hypogonadism-choroidal dystrophy syndrome) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA6	PNPLA6, NTE_SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. {ECO:0000269|PubMed:24357708, ECO:0000269|PubMed:25033069}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bowen-Conradi Sendromu, 211180, Otozomal Resesif; BWCNS (Bowen-Conradi Sendromu) (EMG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bowen-Conradi syndrome, 211180, Autosomal recessive; BWCNS (Bowen-Conradi syndrome) (EMG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EMG1	EMG1, NEP1, C2F, BWCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bowen-Conradi syndrome (BWCNS) [MIM:211180]: A combination of malformations characterized in newborns by low birth weight, microcephaly, mild joint restriction, a prominent nose, micrognathia, fifth finger clinodactyly, and 'rocker-bottom' feet. The syndrome is transmitted as an autosomal recessive trait. The prognosis is poor, with all infants dying within the first few months of life. {ECO:0000269	PubMed:19463982}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bowen-Conradi Sendromu, 211180, Otozomal Resesif; BWCNS (Bowen-Conradi Sendromu) (EMG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Bowen-Conradi syndrome, 211180, Autosomal recessive; BWCNS (Bowen-Conradi syndrome) (EMG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EMG1	EMG1, NEP1, C2F, BWCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bowen-Conradi syndrome (BWCNS) [MIM:211180]: A combination of malformations characterized in newborns by low birth weight, microcephaly, mild joint restriction, a prominent nose, micrognathia, fifth finger clinodactyly, and 'rocker-bottom' feet. The syndrome is transmitted as an autosomal recessive trait. The prognosis is poor, with all infants dying within the first few months of life. {ECO:0000269	PubMed:19463982}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brakidaktili-Sindaktili Sendromu, 610713; BDSD (Brakidaktili-Sindaktili, Zhao Tipi) (HOXD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly-syndactyly syndrome, 610713; BDSD (Brachydactyly-syndactyly, Zhao type) (HOXD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili-Sindaktili Sendromu, 610713; BDSD (Brakidaktili-Sindaktili, Zhao Tipi) (MLPA)	Brachydactyly-syndactyly syndrome, 610713; BDSD (Brachydactyly-syndactyly, Zhao type) (MLPA)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip A1, 112500, Otozomal Dominant; BDA1 (Brakidaktili Tip A1) (IHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A1, 112500, Autosomal dominant; BDA1 (Brachydactyly type A1) (IHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IHH	IHH, BDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A1 (BDA1) [MIM:112500]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type A1 is characterized by middle phalanges of all the digits rudimentary or fused with the terminal phalanges. The proximal phalanges of the thumbs and big toes are short. {ECO:0000269|PubMed:11455389, ECO:0000269|PubMed:12384778}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip A1, C, 615072, Otozomal Resesif, Otozomal Dominant; BDA1C (Brakidaktili Tip A1) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A1, C, 615072, Autosomal recessive, Autosomal dominant; BDA1C (Brachydactyly type A1) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:16127465, ECO:0000269|PubMed:18203755, ECO:0000269|PubMed:21976273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip A1, D, 616849, Otozomal Dominant; BDA1D (Brakidaktili Tip A1) (BMPR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A1, D, 616849, Autosomal dominant; BDA1D (Brachydactyly type A1) (BMPR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1B	BMPR1B, ALK6, AMDD, BDA2, BDA1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:14523231, ECO:0000269|PubMed:16957682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brakidaktili, Tip A2, 112600, Otozomal Dominant (Brakidaktili Tip A2) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A2, 112600, Autosomal dominant (Brachydactyly type A2) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:16127465, ECO:0000269|PubMed:18203755, ECO:0000269|PubMed:21976273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip A2, 112600, Otozomal Dominant (Brakidaktili Tip A2) (BMP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A2, 112600, Autosomal dominant (Brachydactyly type A2) (BMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP2	BMP2, BMP2A, BDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brakidaktili, Tip A2, 112600, Otozomal Dominant; BDA2 (Brakidaktili Tip A2) (BMPR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type A2, 112600, Autosomal dominant; BDA2 (Brachydactyly type A2) (BMPR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1B	BMPR1B, ALK6, AMDD, BDA2, BDA1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:14523231, ECO:0000269|PubMed:16957682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brakidaktili, B1, 113000, Otozomal Dominant; BDB1 (Brakidaktili Tip B) (ROR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type B1, 113000, Autosomal dominant; BDB1 (Brachydactyly type B) (ROR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROR2	ROR2, BDB1, BDB, NTRKR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, B1, 113000, Otozomal Dominant; BDB1 (Brakidaktili Tip B) (MLPA)	Brachydactyly, type B1, 113000, Autosomal dominant; BDB1 (Brachydactyly type B) (MLPA)	ROR2	ROR2, BDB1, BDB, NTRKR2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip B2, 611377, Otozomal Dominant; BDB2 (Brakidaktili Tip B2) (NOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type B2, 611377, Autosomal dominant; BDB2 (Brachydactyly type B2) (NOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOG	NOG, SYM1, SYNS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brakidaktili, Tip C, 113100, Otozomal Dominant; BDC (Brakidaktili Tip C) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type C, 113100, Autosomal dominant; BDC (Brachydactyly type C) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Brakidaktili, Tip D, 113200, Otozomal Dominant (HOXD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type D, 113200, Autosomal dominant (HOXD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip D, 113200, Otozomal Dominant (MLPA)	Brachydactyly, type D, 113200, Autosomal dominant (MLPA)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brakidaktili, Tip E, 113300, Otozomal Dominant; BDE1 (Brakidaktili Tip E) (HOXD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type E, 113300, Autosomal dominant; BDE1 (Brachydactyly type E) (HOXD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brakidaktili, Tip E, 113300, Otozomal Dominant; BDE1 (Brakidaktili Tip E) (MLPA)	Brachydactyly, type E , 113300, Autosomal dominant; BDE1 (Brachydactyly type E) (MLPA)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Brakidaktili, Tip E2, 613382, Otozomal Dominant; BDE2 (Brakidaktili Tip E) (PTHLH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachydactyly, type E2, 613382, Autosomal dominant; BDE2 (Brachydactyly type E) (PTHLH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTHLH	PTHLH, BDE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly E2 (BDE2) [MIM:613382]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type E is characterized by shortening of the fingers mainly in the metacarpals and metatarsals. Wide variability in the number of digits affected occurs from person to person, even in the same family. Some individuals are moderately short of stature. In brachydactyly type E2 variable combinations of metacarpals are involved, with shortening also of the first and third distal and the second and fifth middle phalanges. {ECO:0000269	PubMed:20170896}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brakiolmi 4, Hafif Epifizyal ve Metafizyal Değişiklikler, 612847, Otozomal Resesif; BCYM4 (Spondiloepimetafizyal Displazi, PAPSS2 Tipi) (PAPSS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachyolmia 4 with mild epiphyseal and metaphyseal changes, 612847, Autosomal recessive; BCYM4 (Spondyloepimetaphyseal dysplasia, PAPSS2 type) (PAPSS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAPSS2	PAPSS2, ATPSK2, BCYM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brakiolmi Tip 3, 113500, Otozomal Dominant; BCYM3 (Otozomal Dominant Brakiolmi) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brachyolmia type 3, 113500, Autosomal dominant; BCYM3 (Autosomal dominant brachyolmia) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269	PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Bradiposi, 608415 (Bradyopsi) (RGS9BP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bradyopsia, 608415 (Bradyopsia) (RGS9BP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RGS9BP	RGS9BP, RSAP, RGS9, PERRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bradiopsi, 608415 (Bradiyopsi) (RGS9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bradyopsia, 608415 (Bradyopsia) (RGS9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RGS9	RGS9, PERRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beyin Küçük Damar Hastalığı ± Oküler Anomaliler, 607595, Otozomal Dominant; BSVD (Ailesel Vasküler Lökoensefalopati) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brain small vessel disease with or without ocular anomalies, 607595, Autosomal dominant; BSVD (Familial vascular leukoencephalopathy) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beyin Küçük Damar Hastalığı ± Oküler Anomaliler, 607595, Otozomal Dominant; BSVD (Ailesel Vasküler Lökoensefalopati) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Brain small vessel disease with or without ocular anomalies, 607595, Autosomal dominant; BSVD (Familial vascular leukoencephalopathy) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beyin Tümörü-Polipozis Sendromu 2, 175100, Otozomal Dominant (Turcot Sendromu-Polipozis) (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brain tumor-polyposis syndrome 2, 175100, Autosomal dominant (Turcot syndrome with polyposis) (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beyin Tümörü-Polipozis Sendromu 2, 175100, Otozomal Dominant (Turcot Sendromu-Polipozis) (MLPA)	Brain tumor-polyposis syndrome 2, 175100, Autosomal dominant (Turcot syndrome with polyposis) (MLPA)	APC	APC, GS, FPC, BTPS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Dallı Zincirli Ketoasit Dehidrojenaz Kinaz Eksikliği, 614923; BCKDKD (Otizm Epilepsi Sendromu, Dallı Zincirli Ketoasit Dehidrojenaz Kinaz Eksikliğine Bağlı) (BCKDK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branched-chain ketoacid dehydrogenase kinase deficiency, 614923; BCKDKD (Autism-epilepsy syndrome due to branched chain ketoacid dehydrogenase kinase deficiency) (BCKDK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCKDK	BCKDK, BDK, BCKDKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branched-chain ketoacid dehydrogenase kinase deficiency (BCKDKD) [MIM:614923]: A metabolic disorder characterized by autism, epilepsy, intellectual disability, and reduced branched- chain amino acids. {ECO:0000269|PubMed:22956686, ECO:0000269|PubMed:24449431}. Note=The disease is caused by mutations affecting the gene represented in this entry. A diet enriched in branched amino acids (BCAAs) allows to normalize plasma BCAA levels. This suggests that it may be possible to treat patients with mutations in BCKDK with BCAA supplementation .	EDTA Blood Sample
Brankiookülofasiyal Sendrom, 113620, Otozomal Dominant; BOFS (Brankio-Okülo-Fasiyal Sendrom (BOR Sendromu)) (TFAP2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branchiooculofacial syndrome, 113620, Autosomal dominant; BOFS (Branchio-oculo-facial syndrome) (TFAP2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFAP2A	TFAP2A, AP2TF, BOFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branchiooculofacial syndrome (BOFS) [MIM:113620]: A syndrome characterized by growth retardation, bilateral branchial sinus defects with hemangiomatous, scarred skin, cleft lip with or without cleft palate, pseudocleft of the upper lip, nasolacrimal duct obstruction, low set ears with posterior rotation, a malformed, asymmetrical nose with a broad bridge and flattened tip, conductive or sensorineural deafness, ocular and renal anomalies. {ECO:0000269	PubMed:18423521}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brankiookülofasiyal Sendrom, 113620, Otozomal Dominant; BOFS (Brankio-Okülo-Fasiyal Sendrom (BOR Sendromu)) (TFAP2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Branchiooculofacial syndrome, 113620, Autosomal dominant; BOFS (Branchio-oculo-facial syndrome) (TFAP2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TFAP2A	TFAP2A, AP2TF, BOFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Branchiooculofacial syndrome (BOFS) [MIM:113620]: A syndrome characterized by growth retardation, bilateral branchial sinus defects with hemangiomatous, scarred skin, cleft lip with or without cleft palate, pseudocleft of the upper lip, nasolacrimal duct obstruction, low set ears with posterior rotation, a malformed, asymmetrical nose with a broad bridge and flattened tip, conductive or sensorineural deafness, ocular and renal anomalies. {ECO:0000269	PubMed:18423521}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brankiootik Sendrom 1, 602588, Otozomal Dominant; BOS1 (Brankio-Otik Sendrom) (EYA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branchiootic syndrome 1, 602588, Autosomal dominant; BOS1 (Branchio-otic syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. {ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brankiootik Sendrom 1, 602588, Otozomal Dominant; BOS1 (Brankio-Otik Sendrom) (EYA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Branchiootic syndrome 1, 602588, Autosomal dominant; BOS1 (Branchio-otic syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. {ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brankiootik Sendrom 3, 608389, Otozomal Dominant; BOS3 (Brankio-Otik Sendrom) (SIX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branchiootic syndrome 3, 608389, Autosomal dominant; BOS3 (Branchio-otic syndrome) (SIX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX1	SIX1, BOS3, DFNA23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brankiootik Sendrom 3, 608389, Otozomal Dominant; BOS3 (Brankio-Otik Sendrom) (SIX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Branchiootic syndrome 3, 608389, Autosomal dominant; BOS3 (Branchio-otic syndrome) (SIX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIX1	SIX1, BOS3, DFNA23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brankiootorenal Sendrom 1, ± Katarakt, 113650, Otozomal Dominant; BOR1 (BOR Sendromu) (EYA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branchiootorenal syndrome 1, with or without cataracts, 113650, Autosomal dominant; BOR1 (BOR syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. {ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brankiootorenal Sendrom 1, ± Katarakt, 113650, Otozomal Dominant; BOR1 (BOR Sendromu) (EYA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Branchiootorenal syndrome 1, with or without cataracts, 113650, Autosomal dominant; BOR1 (BOR syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. {ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brankiootorenal Sendrom 2, 610896; BOR2 (BOR Sendromu) (SIX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Branchiootorenal syndrome 2, 610896; BOR2 (BOR syndrome) (SIX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX5	SIX5, DMAHP, BOR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branchiootorenal syndrome 2 (BOR2) [MIM:610896]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. {ECO:0000269	PubMed:17357085}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Brankiootorenal Sendrom 2, 610896; BOR2 (BOR Sendromu) (SIX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Branchiootorenal syntrome 2, 610896; BOR2 (BOR syntrome) (SIX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIX5	SIX5, DMAHP, BOR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (IKİSİ BIRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Branchiootorenal syndrome 2 (BOR2) [MIM:610896]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. (ECO:0000269|PubMed:17357085). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meme ve Kolorektal Kanser, Yatkınlık (CHEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast and colorectal cancer, susceptibility to (CHEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. (ECO:0000269|PubMed:12094328).;	EDTA Blood Sample
Meme ve Kolorektal Kanser, Yatkınlık (MLPA)	Breast and colorectal cancer, susceptibility to (MLPA)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. (ECO:0000269|PubMed:12094328).;	EDTA Blood Sample
Meme kanseri (BRCA1 bölgesi Delesyon/ Duplikasyon paneli) (BRCA1 region) (MLPA)	Breast cancer (BRCA1 region Deletion / Duplication panel) (BRCA1 region) (MLPA)	BRCA1 region	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Kanseri (ERBB2 (HER2-NEU) 17q21.1) (MLPA)	Breast Cancer (ERBB2 (HER2-NEU) 17q21.1) (MLPA)	ERBB2 (HER2-NEU) 17q21.1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Kanseri Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (NQO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer susceptibility, 114480, Autosomal dominant (Hereditary breast cancer) (NQO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NQO2	NQO2, NMOR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, 114480, Autosomal dominant (Hereditary breast cancer) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (PPM1D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, 114480, Autosomal dominant (Hereditary breast cancer) (PPM1D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPM1D	PPM1D, WIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, 114480, Autosomal dominant (Hereditary breast cancer) (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (MLPA)	Breast cancer, 114480, Autosomal dominant (Hereditary breast cancer) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Erken Başlangıç, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (BRIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, early-onset, 114480, Autosomal dominant (Hereditary breast cancer) (BRIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRIP1	BRIP1, BACH1, FANCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:11301018). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Kanseri, İnvaziv Dutkal, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (RAD54L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, invasive ductal, 114480, Autosomal dominant (Hereditary breast cancer) (RAD54L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD54L	RAD54L, HR54, HRAD54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Lobüler, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (CDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, lobular, 114480, Autosomal dominant (Hereditary breast cancer) (CDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH1	CDH1, UVO, LCAM, ECAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Erkek, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, male, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Kanseri, Erkek, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (MLPA)	Breast cancer, male, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Kanseri, Kemoterapi Sonrası Kötü Sağkalım (NQO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, poor survival after chemotherapy for (NQO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NQO1	NQO1, DIA4, NMOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Karşı Koruma, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (CASP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, protection against, 114480, Autosomal dominant (Hereditary breast cancer) (CASP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP8	CASP8, MCH5, ALPS2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Somatik, 114480 (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:16353168). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Kanseri, Somatik, 114480 (TSG101 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (TSG101 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSG101	TSG101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Kanseri, Somatik, 114480 (SLC22A18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (SLC22A18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A18	SLC22A1L, BWSCR1A, IMPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Kanseri, Somatik, 114480 (RB1CC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (RB1CC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1CC1	RB1CC1, CC1, KIAA0203	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Kanseri, Somatik, 114480 (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Kanseri, Somatik, 114480 (Ekzon 4) (AKT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, somatic, 114480 (Exon 4) (AKT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269|PubMed:17611497). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (CHEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (CHEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25618629}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (MLPA)	Breast cancer, susceptibility to, 114480, Autosomal dominant (MLPA)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25618629}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (XRCC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (XRCC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XRCC3	XRCC3, CMM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:12023962}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (RAD51 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (RAD51 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51	RAD51A, RECA, MRMV2, FANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:10807537}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (PHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (PHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHB	PHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (PALB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (PALB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PALB2	PALB2, FANCN, PNCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:17287723, ECO:0000269|PubMed:22241545}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Breast cancer susceptibility is strongly associated with PALB2 truncating mutations. Conversely, rare missense mutations do not strongly influence breast cancer risk (PubMed:22241545). {ECO:0000269|PubMed:22241545}.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (HMMR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (HMMR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMMR	HMMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (BARD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (BARD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BARD1	BARD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (ATM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (ATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Yatkınlık, 114480, Otozomal Dominant (Kalıtsal Meme Kanseri) (MLPA)	Breast cancer, susceptibility to, 114480, Autosomal dominant (Hereditary breast cancer) (MLPA)	ATM	ATM, ATA, AT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Ailesel, 1, 604370, Otozomal Dominant, Multifaktöriyel; BROVCA1 (Ailesel Meme-Over Kanseri Sendromu) (BRCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast-ovarian cancer, familial, 1, 604370, Autosomal dominant, Multifactorial, BROVCA1 (Hereditary breast and ovarian cancer syndrome) (BRCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA1	BRCA1, PSCP, BROVCA1, PNCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme Kanseri, Ailesel, 1, 604370, Otozomal Dominant, Multifaktöriyel; BROVCA1 (Ailesel Meme-Over Kanseri Sendromu) (MLPA)	Breast-ovarian cancer, familial, 1, 604370, Autosomal dominant, Multifactorial, BROVCA1 (Hereditary breast and ovarian cancer syndrome) (MLPA)	BRCA1	BRCA1, PSCP, BROVCA1, PNCA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Over Kanseri, Ailesel, 2, 612555, Otozomal Dominant; BROVCA2 (Ailesel Meme-Over Kanseri Sendromu) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast-ovarian cancer, familial, 2, 612555, Autosomal dominant; BROVCA2 (Hereditary breast and ovarian cancer syndrome) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ailesel Meme ve Over Kanseri Paneli - EDTA'lı Kan (BRCA1, BRCA2) (2 gen)	Breast-ovarian cancer, familial, 1- 2 - EDTA Blood sample (BRCA1, BRCA2 genes)	BRCA1, BRCA2	BRCA1, PSCP, BROVCA1, PNCA4, BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	60			60	Postnatal		EDTA Blood Sample
Ailesel Meme ve Over Kanseri Paneli - Parafin Blok (BRCA1, BRCA2) (2 gen)	Breast-ovarian cancer, familial, 1- 2 - Parafinized tissue sample (BRCA1, BRCA2 genes)	BRCA1, BRCA2	BRCA1, PSCP, BROVCA1, PNCA4, BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu	*	30	60			60	Postnatal		Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meme Over Kanseri, Ailesel, 2, 612555, Otozomal Dominant; BROVCA2 (Ailesel Meme-Over Kanseri Sendromu) (MLPA)	Breast-ovarian cancer, familial, 2, 612555, Autosomal dominant; BROVCA2 (Hereditary breast and ovarian cancer syndrome) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Meme Over Kanseri, Kalıtsal, Yatkınlık, 3, 613399; BROVCA3 (Ailesel Meme-Over Kanseri Sendromu) (RAD51C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast-ovarian cancer, familial, susceptibility to, 3, 613399; BROVCA3 (Hereditary breast and ovarian cancer syndrome) (RAD51C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51C	RAD51C, FANCO, BROVCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group O (FANCO) [MIM:613390]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000268	PubMed:20400963}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meme Over Kanseri, Kalıtsal, Yatkınlık, 4, 614291; BROVCA4 (Ailesel Meme-Over Kanseri Sendromu) (RAD51D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breast-ovarian cancer, familial, susceptibility to, 4, 614291; BROVCA4 (Hereditary breast and ovarian cancer syndrome) (RAD51D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51D	RAD51D, RAD51L3, BROVCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meme ve / Veya Areola, Aplazi Veya Hipoplazi, 2, 616001, Otozomal Resesif; BNAH2 (Izole Konjenital Meme Hipoplazisi / Aplazisi) (PTPRF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001, Autosomal recessive; BNAH2 (Isolated congenital breast hypoplasia/aplasia) (PTPRF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRF	PTPRF, LAR, BNAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aplasia or hypoplasia of the breasts and/or nipples 2 (BNAH2) [MIM:616001]: A group of congenital deformities encompassing total absence of breasts and nipple (amastia), absence of the nipple (athelia), and absence of the mammary gland (amazia). {ECO:0000269	PubMed:24781087}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brevican; BCAN (BCAN geni) (Tüm Gen Dizi Analizi) (Postnatal)	BREVICAN; BCAN (BCAN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAN	BCAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frajil Kornea Sendromu 1, 229200, Otozomal Resesif; BCS1 (Kırılgan Kornea Sendromu) (ZNF469 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brittle cornea syndrome 1, 229200, Autosomal recessive; BCS1 (Brittle cornea syndrome) (ZNF469 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF469	ZNF469, KIAA1858, BCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frajil Kornea Sendromu 2, 614170, Otozomal Resesif; BCS2 (Kırılgan Kornea Sendromu) (PRDM5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brittle cornea syndrome 2, 614170, Autosomal recessive; BCS2 (Brittle cornea syndrome) (PRDM5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM5	PRDM5, BCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brody Miyopati, 601003, Otozomal Resesif (Brody Miyopati) (ATP2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brody myopathy, 601003, Autosomal recessive (Brody myopathy) (ATP2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2A1	ATP2A1, SERCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brody myopathy (BRM) [MIM:601003]: A disorder of muscle function that is characterized by painless muscle cramping and exercise-induced impairment of muscle relaxation. {ECO:0000269	PubMed:10914677}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brody Miyopati, 601003, Otozomal Resesif (Brody Miyopati) (ATP2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Brody myopathy, 601003, Autosomal recessive (Brody myopathy) (ATP2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP2A1	ATP2A1, SERCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE AİNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Brody myopathy (BRM) [MIM:601003]: A disorder of muscle function that is characterized by painless muscle cramping and exercise-induced impairment of muscle relaxation. {ECO:0000269	PubMed:10914677}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bronşektazi, z Ter Klorüründe Artış 1, 211400, Otozomal Dominant (İdiopatik Bronşektazi) (SCNN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bronchiectasis with or without elevated sweat chloride 1, 211400, Autosomal dominant (Idiopathic bronchiectasis) (SCNN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1B	SCNN1B, BESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bronşektazi, z Ter Klorür Artışı 1, Modifiye Edici, 211400, Otozomal Dominant; BESC1 (İdiopatik Bronşektazi) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bronchiectasis with or without elevated sweat chloride 1, modifier of, 211400, Autosomal dominant; BESC1 (Idiopathic bronchiectasis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bronşektazi, z Ter Klorür Artışı 1, Modifiye Edici, 211400, Otozomal Dominant; BESC1 (İdiopatik Bronşektazi) (MLPA)	Bronchiectasis with or without elevated sweat chloride 1, modifier of, 211400, Autosomal dominant; BESC1 (Idiopathic bronchiectasis) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Bronşektazi, z Yüksek Ter Klorürü 2, 613021, Otozomal Dominant; BESC2 (İdiopatik Bronşektazi) (SCNN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bronchiectasis with or without elevated sweat chloride 2, 613021, Autosomal dominant, BESC2 (Idiopathic bronchiectasis) (SCNN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1A	SCNN1A, BESC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bronşektazi, z Ter Klorüründe Artış 1, 211400, Otozomal Dominant; BESC3 (İdiopatik Bronşektazi) (SCNN1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Bronchiectasis with or without elevated sweat chloride 3, 613071, Autosomal dominant, BESC3 (Idiopathic bronchiectasis) (SCNN1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1G	SCNN1G, PHA1, BESC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brooke-Spiegler Sendromu, 605041, Otozomal Dominant; BRSS (Brooke-Spiegler Sendromu) (CYLD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Brooke-Spiegler syndrome, 605041, Autosomal dominant; BRSS (Brooke-Spiegler syndrome) (CYLD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYLD	CYLD, CDMT, EAC, MFT1, KIAA0849, BRSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Brooke-Spiegler Sendromu, 605041, Otozomal Dominant; BRSS (Brooke-Spiegler Sendromu) (CYLD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Brooke-Spiegler syndrome, 605041, Autosomal dominant; BRSS (Brooke-Spiegler syndrome) (CYLD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYLD	CYLD, CDMT, EAC, MFT1, KIAA0849, BRSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brown-Vialetto Van Laere Sendromu 1, 211530, Otozomal Resesif; BVVLS1 (Riboflavin Transporter Eksikliği (Brown-Vialetto-Van Laere Sendromu Tip 2)) (SLC52A3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brown-Vialetto-Van Laere syndrome 1, 211530, Autosomal recessive; BVVLS1 (Riboflavin transporter deficiency) (SLC52A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC52A3	SLC52A3, C20orf54, RFT2, BVVLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brown-Vialetto Van Laere Sendromu 1, 211530, Otozomal Resesif; BVVLS1 (Riboflavin Transporter Eksikliği (Brown-Vialetto-Van Laere Sendromu Tip 2)) (SLC52A3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Brown-Vialetto-Van Laere syndrome 1, 211530, Autosomal recessive; BVVLS1 (Riboflavin transporter deficiency) (SLC52A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC52A3	SLC52A3, C20orf54, RFT2, BVVLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brown-Vialetto Van Laere Sendromu 2, 614707, Otozomal Resesif; BVVLS2 (Riboflavin Transporter Eksikliği (Brown-Vialetto-Van Laere Sendromu Tip 2)) (SLC52A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brown-Vialetto-Van Laere syndrome 2, 614707, Autosomal recessive; BVVLS2 (Riboflavin transporter deficiency) (SLC52A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC52A2	SLC52A2, GPR172A, GPCR41, PAR1, FLJ11856, BVVLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brown-Vialetto Van Laere Sendromu 2, 614707, Otozomal Resesif; BVVLS2 (Riboflavin Transporter Eksikliği (Brown-Vialetto-Van Laere Sendromu Tip 2)) (SLC52A2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Brown-Vialetto-Van Laere syndrome 2, 614707, Autosomal recessive; BVVLS2 (Riboflavin transporter deficiency) (SLC52A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC52A2	SLC52A2, GPR172A, GPCR41, PAR1, FLJ11856, BVVLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bruck Sendromu 1, 259450, Otozomal Resesif; BRKS1 (Bruck Sendromu) (FKBP10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bruck syndrome 1, 259450, Autosomal recessive; BRKS1 (Bruck syndrome) (FKBP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKBP10	FKBP10, FKBP65, OI11, BRKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bruck syndrome 1 (BRKS1) [MIM:259450]: A disease characterized by generalized osteopenia, congenital joint contractures, fragile bones with onset of fractures in infancy or early childhood, short stature, severe limb deformity, progressive scoliosis, and pterygia. {ECO:0000269|PubMed:20839288, ECO:0000269|PubMed:22949511}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Bruck Sendromu 1, 259450, Otozomal Resesif; BRKS1 (Bruck Sendromu) (FKBP10 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bruck syndrome 1, 259450, Autosomal recessive; BRKS1 (Bruck syndrome) (FKBP10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKBP10	FKBP10, FKBP65, OI11, BRKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bruck syndrome 1 (BRKS1) [MIM:259450]: A disease characterized by generalized osteopenia, congenital joint contractures, fragile bones with onset of fractures in infancy or early childhood, short stature, severe limb deformity, progressive scoliosis, and pterygia. {ECO:0000269|PubMed:20839288, ECO:0000269|PubMed:22949511}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bruck Sendromu 2, 609220, Otozomal Resesif; BRKS2 (Bruck Sendromu) (PLOD2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bruck syndrome 2, 609220, Autosomal recessive; BRKS2 (Bruck syndrome) (PLOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLOD2	PLOD2, LH2, TLH, BRKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bruck Sendromu 2, 609220, Otozomal Resesif; BRKS2 (Bruck Sendromu) (PLOD2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Bruck syndrome 2, 609220, Autosomal recessive; BRKS2 (Bruck syndrome) (PLOD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLOD2	PLOD2, LH2, TLH, BRKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Brugada Sendromu 1, 601144, Otozomal Dominant; BRGDA1 (Brugada Sendromu) (SCN5A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 1, 601144, Autosomal dominant; BRGDA1 (Brugada syndrome) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 2, 611777; BRGDA2 (Brugada Sendromu) (GPD1L gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 2, 611777; BRGDA2 (Brugada syndrome) (GPD1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPD1L	GPD1L, KIAA0089	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 3, 611875; BRGDA3 (Brugada Sendromu) (CACNA1C gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 3, 611875; BRGDA3 (Brugada syndrome) (CACNA1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1C	CACNA1C, CACNL1A1, CCHL1A1, TS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 4, 611876; BRGDA4 (Brugada Sendromu) (CACNB2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 4, 611876; BRGDA4 (Brugada syndrome) (CACNB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNB2	CACNB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brugada syndrome 4 (BRGDA4) [MIM:611876]: A heart disease characterized by the association of Brugada syndrome with shortened QT intervals. Brugada syndrome is a tachyarrhythmia characterized by right bundle branch block and ST segment elevation on an electrocardiogram (ECG). It can cause the ventricles to beat so fast that the blood is prevented from circulating efficiently in the body. When this situation occurs, the individual will faint and may die in a few minutes if the heart is not reset. {ECO:0000269	PubMed:17224476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brugada Sendromu 5, 612838; BRGDA5 (Ailesel Progresif Kardiyak İletim Defekti) (SCN1B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 5, 612838; BRGDA5 (Familial progressive cardiac conduction defect) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEE52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brugada syndrome 5 (BRGDA5) [MIM:612838]: A tachyarrhythmia characterized by right bundle branch block and ST segment elevation on an electrocardiogram (ECG). It can cause the ventricles to beat so fast that the blood is prevented from circulating efficiently in the body. When this situation occurs, the individual will faint and may die in a few minutes if the heart is not reset. {ECO:0000269	PubMed:18464934}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Brugada Sendromu 6, 613119; BRGDA6 (Brugada Sendromu) (KCNE3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 6, 613119; BRGDA6 (Brugada syndrome) (KCNE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNE3	KCNE3, HOKPP, HYPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 7, 613120, Otozomal Dominant; BRGDA7 (Brugada Sendromu) (SCN3B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 7, 613120, Autosomal dominant; BRGDA7 (Brugada syndrome) (SCN3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN3B	SCN3B, SCN83, BRGDA7, AITFB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 8, 613123; BRGDA8 (Brugada Sendromu) (HCN4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 8, 613123; BRGDA8 (Brugada syndrome) (HCN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCN4	HCN4, SSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brugada Sendromu 9, 616399, Otozomal Dominant; BRGDA9 (Brugada Sendromu) (KCND3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brugada syndrome 9, 616399, Autosomal dominant; BRGDA9 (Brugada syndrome) (KCND3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCND3	KCND3, KCND3S, KCND3L, SCA19, SCA22, BRGDA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brunner Sendromu, 300615, X'e Bağlı Resesif; BRNRS (Monoamin Oksidaz A Eksikliği (Brunner Sendromu)) (MAOA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Brunner syndrome, 300615, X-linked recessive; BRNRS (Monoamine oxidase A deficiency) (MAOA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAOA	MAOA, BRNRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brunner syndrome (BRUNS) [MIM:300615]: A form of X-linked non-dysmorphic mild mental retardation. Male patients are affected by borderline mental retardation and exhibit abnormal behavior, including disturbed regulation of impulsive aggression. Obligate female carriers have normal intelligence and behavior. {ECO:0000269	PubMed:8211186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Brunner Sendromu, 300615, X'e Bağlı Resesif; BRNRS (Monoamin Oksidaz A Eksikliği (Brunner Sendromu)) (MAOA gen) (Tüm Gen Dizi Analizi) (Prenatal)	Brunner syndrome, 300615, X-linked recessive; BRNRS (Monoamine oxidase A deficiency) (MAOA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAOA	MAOA, BRNRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brunner syndrome (BRUNS) [MIM:300615]: A form of X-linked non-dysmorphic mild mental retardation. Male patients are affected by borderline mental retardation and exhibit abnormal behavior, including disturbed regulation of impulsive aggression. Obligate female carriers have normal intelligence and behavior. {ECO:0000269	PubMed:8211186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Budd-Chiari Sendromu, 600880, Otozomal Resesif; BDCHS (Budd-Chiari Sendromu) (F5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Budd-Chiari syndrome, 600880, Autosomal recessive; BDCHS (Budd-Chiari syndrome) (F5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Budd-Chiari Sendromu, 600880, Otozomal Resesif; BDCHS (Budd-Chiari Sendromu) (F5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Budd-Chiari syndrome, 600880, Autosomal recessive; BDCHS (Budd-Chiari syndrome) (F5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Budd-Chiari Sendromu, Somatik, 600800 (JAK2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Budd-Chiari syndrome, somatic, 600800 (JAK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAK2	JAK2, THCYT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Bulimia Nervoza, Kilo Kaybının Başlama Yaşı, 607499, Multifaktöriyel (BDNF gen) (Tüm Gen Dizi Analizi) (Postnatal)	Bulimia nervosa, age of onset of weight loss in, 607499, Multifactorial (BDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Burkitt Lenfoma, 113970, Sporadik Vakalar; BL (Burkitt Lenfoma) (MYC gen) (Tüm Gen Dizi Analizi) (Postnatal)	Burkitt lymphoma, 113970, Isolated cases; BL (Burkitt lymphoma) (MYC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYC	MYC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Burkitt lymphoma (BL) [MIM:113970]: A form of undifferentiated malignant lymphoma commonly manifested as a large osteolytic lesion in the jaw or as an abdominal mass. {ECO:0000269|PubMed:2186998, ECO:0000269|PubMed:8220424}. Note=The gene represented in this entry is involved in disease pathogenesis. Chromosomal aberrations involving MYC are usually found in Burkitt lymphoma. Translocations t(8;14), t(8;22) or t(2;8) which juxtapose MYC to one of the heavy or light chain immunoglobulin gene loci.;	EDTA Blood Sample
Burn Mckeown Sendromu, 608572, Otozomal Resesif; BMKS (Koanal Atrezi-İşitme Kaybı-Kalp Defekti-Kraniyofasiyal Dismorfizm Sendromu (Burn-Mckeown Sendromu)) (TXNL4A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Burn-McKeown syndrome, 608572, Autosomal recessive; BMKS (Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome) (TXNL4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TXNL4A	TXNL4A, DIM1, BMKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Burn-McKeown syndrome (BMKS) [MIM:608572]: A disease characterized by choanal atresia, sensorineural deafness, cardiac defects, and typical craniofacial dysmorphism consisting of narrow palpebral fissures, coloboma of the lower eyelids, prominent nose with high nasal bridge, short philtrum, cleft lip and/or palate, and large and protruding ears. Intellectual development is normal. {ECO:0000269	PubMed:25434003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Burn Mckeown Sendromu, 608572, Otozomal Resesif; BMKS (Koanal Atrezi-İşitme Kaybı-Kalp Defekti-Kraniyofasiyal Dismorfizm Sendromu (Burn-Mckeown Sendromu)) (TXNL4A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Burn-McKeown syndrome, 608572, Autosomal recessive; BMKS (Choanal atresia-hearing loss-cardiac defects-craniofacial dysmorphism syndrome) (TXNL4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TXNL4A	TXNL4A, DIM1, BMKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Burn-McKeown syndrome (BMKS) [MIM:608572]: A disease characterized by choanal atresia, sensorineural deafness, cardiac defects, and typical craniofacial dysmorphism consisting of narrow palpebral fissures, coloboma of the lower eyelids, prominent nose with high nasal bridge, short philtrum, cleft lip and/or palate, and large and protruding ears. Intellectual development is normal. {ECO:0000269	PubMed:25434003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Burul Ülser, Yatkınlık, 610446 (SLC11A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Burril ulcer, susceptibility to, 610446 (SLC11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC11A1	NRAMP1, NRAMP, SLC11A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Buschke-Ollendorff Sendromu, 166700, Otozomal Dominant; BOS (İzole Osteopoikiloz) (LEMD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Buschke-Ollendorff syndrome, 166700, Autosomal dominant; BOS (isolated osteopoikilosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Buschke-Ollendorff Sendromu, 166700, Otozomal Dominant; BOS (İzole Osteopoikiloz) (LEMD3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Buschke-Ollendorff syndrome, 166700, Autosomal dominant; BOS (isolated osteopoikilosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

C Sendromu, 211750, Otozomal Resesif (C Sendromu) (CD96 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C syndrome, 211750, Autosomal recessive (C syndrome) (CD96 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD96	CD96, TACTILE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: C syndrome (CSYN) [MIM:211750]: A syndrome characterized by trigonocephaly, severe mental retardation, hypotonia, variable cardiac defects, redundant skin, and dysmorphic facial features, including upslanted palpebral fissures, epicanthal folds, depressed nasal bridge, and low-set, posteriorly rotated ears. {ECO:0000269	PubMed:17847009}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving CD96 has been found in a patient with C syndrome. Translocation t(3;18)(q13.13;q12.1). CD96 gene was located at the 3q13.13 breakpoint. Precise structural analysis around the breakpoint showed that the gene was disrupted by the translocation in exon 5, probably leading to premature termination or loss of expression of CD96 protein. No gene was detected at the chromosome 18 breakpoint.;	EDTA Blood Sample
C Sendromu, 211750, Otozomal Resesif (C Sendromu) (CD96 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C syndrome, 211750, Autosomal recessive (C syndrome) (CD96 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD96	CD96, TACTILE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: C syndrome (CSYN) [MIM:211750]: A syndrome characterized by trigonocephaly, severe mental retardation, hypotonia, variable cardiac defects, redundant skin, and dysmorphic facial features, including upslanted palpebral fissures, epicanthal folds, depressed nasal bridge, and low-set, posteriorly rotated ears. {ECO:0000269	PubMed:17847009}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving CD96 has been found in a patient with C syndrome. Translocation t(3;18)(q13.13;q12.1). CD96 gene was located at the 3q13.13 breakpoint. Precise structural analysis around the breakpoint showed that the gene was disrupted by the translocation in exon 5, probably leading to premature termination or loss of expression of CD96 protein. No gene was detected at the chromosome 18 breakpoint.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QB geni) (Tüm Gen Dizi Analizi) (Postnatal)	C1q deficiency, 613652, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C1QB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1QB	C1QB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:9476130}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QA geni) (Tüm Gen Dizi Analizi) (Postnatal)	C1q deficiency, 613652, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C1QA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1QA	C1QA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QB geni) (Tüm Gen Dizi Analizi) (Prenatal)	C1q deficiency, 613652, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C1QB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C1QB	C1QB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:9476130}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QA geni) (Tüm Gen Dizi Analizi) (Prenatal)	C1q deficiency, 613652, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C1QA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C1QA	C1QA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif; C1QD (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QC geni) (Tüm Gen Dizi Analizi) (Postnatal)	C1q deficiency, 613652, Autosomal recessive; C1QD (Immunodeficiency due to a classical component pathway complement deficiency) (C1QC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1QC	C1QC, C1QG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:8630118}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
C1q Eksikliği, 613652, Otozomal Resesif; C1QD (İmmün Yetmezlik, Klasik Komponent Yolağı Komplemen Eksikliğine Bağlı) (C1QC geni) (Tüm Gen Dizi Analizi) (Prenatal)	C1q deficiency, 613652, Autosomal recessive; C1QD (Immunodeficiency due to a classical component pathway complement deficiency) (C1QC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C1QC	C1QC, C1QG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component C1q deficiency (C1QD) [MIM:613652]: A disorder caused by impaired activation of the complement classical pathway. It generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:8630118}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C1s Eksikliği, 613783 (İmmün Yetmezlik, Klasik Komponent Yolağı Kompleman Eksikliğine Bağlı) (C1S geni) (Tüm Gen Dizi Analizi) (Postnatal)	C1s deficiency, 613783 (Immunodeficiency due to a classical component pathway complement deficiency) (C1S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1S	C1S, EDSPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component C1s deficiency (C1SD) [MIM:613783]: A rare defect resulting in C1 deficiency and impaired activation of the complement classical pathway. C1 deficiency generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:11390518}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
C1s Eksikliği, 613783 (İmmün Yetmezlik, Klasik Komponent Yolağı Kompleman Eksikliğine Bağlı) (C1S geni) (Tüm Gen Dizi Analizi) (Prenatal)	C1s deficiency, 613783 (Immunodeficiency due to a classical component pathway complement deficiency) (C1S gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C1S	C1S, EDSPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component C1s deficiency (C1SD) [MIM:613783]: A rare defect resulting in C1 deficiency and impaired activation of the complement classical pathway. C1 deficiency generally leads to severe immune complex disease with features of systemic lupus erythematosus and glomerulonephritis. {ECO:0000269	PubMed:11390518}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C2-Eksikliği, 217000, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Kompleman Eksikliğine Bağlı) (C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C2 deficiency, 217000, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C2	C2, ARMD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 2 deficiency (C2D) [MIM:217000]: A rare defect of the complement classical pathway associated with the development of autoimmune disorders, mainly systemic lupus erythematosus. Skin and joint manifestations are common and renal disease is relatively rare. Patients with complement component 2 deficiency are also reported to have recurrent invasive infections. {ECO:0000269|PubMed:8621452, ECO:0000269|PubMed:9670930}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
C2-Eksikliği, 217000, Otozomal Resesif (İmmün Yetmezlik, Klasik Komponent Yolağı Kompleman Eksikliğine Bağlı) (C2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C2 deficiency, 217000, Autosomal recessive (Immunodeficiency due to a classical component pathway complement deficiency) (C2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C2	C2, ARMD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 2 deficiency (C2D) [MIM:217000]: A rare defect of the complement classical pathway associated with the development of autoimmune disorders, mainly systemic lupus erythematosus. Skin and joint manifestations are common and renal disease is relatively rare. Patients with complement component 2 deficiency are also reported to have recurrent invasive infections. {ECO:0000269|PubMed:8621452, ECO:0000269|PubMed:9670930}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C3 Eksikliği, 613779, Otozomal Resesif (Komplemen Komponent 3 Eksikliği) (C3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C3 deficiency, 613779, Autosomal recessive (Complement component 3 deficiency) (C3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C3	C3, ARMD9, AHUS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
C3 Eksikliği, 613779, Otozomal Resesif (Kompleman Komponent 3 Eksikliği) (C3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C3 deficiency, 613779, Autosomal recessive (Complement component 3 deficiency) (C3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C3	C3, ARMD9, AHUS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C5 Eksikliği, 609536 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C5 deficiency, 609536 (Immunodeficiency due to a late component of complement deficiency) (C5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C5	C5, C5D, ECLZB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 5 deficiency (C5D) [MIM:609536]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
C5 Eksikliği, 609536 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C5 deficiency, 609536 (Immunodeficiency due to a late component of complement deficiency) (C5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C5	C5, C5D, ECLZB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 5 deficiency (C5D) [MIM:609536]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C6 Eksikliği, 612446 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C6 deficiency, 612446 (Immunodeficiency due to a late component of complement deficiency) (C6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C6	C6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 6 deficiency (C6D) [MIM:612446]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269	PubMed:15565285}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
C6 Eksikliği, 612446 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C6 deficiency, 612446 (Immunodeficiency due to a late component of complement deficiency) (C6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C6	C6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 6 deficiency (C6D) [MIM:612446]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269	PubMed:15565285}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C7 Eksikliği, 610102 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	C7 deficiency, 610102 (Immunodeficiency due to a late component of complement deficiency) (C7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C7	C7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 7 deficiency (C7D) [MIM:610102]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269|PubMed:8871666, ECO:0000269|PubMed:9218625, ECO:0000269|PubMed:9856499}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
C7 Eksikliği, 610102 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	C7 deficiency, 610102 (Immunodeficiency due to a late component of complement deficiency) (C7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C7	C7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 7 deficiency (C7D) [MIM:610102]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269|PubMed:8871666, ECO:0000269|PubMed:9218625, ECO:0000269|PubMed:9856499}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C8 Eksikliği, Tip 1, 613790, Otozomal Resesif (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	C8 deficiency, type I, 613790, Autosomal recessive (Immunodeficiency due to a late component of complement deficiency) (C8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C8A	C8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 8 deficiency, 1 (C8D1) [MIM:613790]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
C8 Eksikliği, Tip 1, 613790, Otozomal Resesif (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	C8 deficiency, type I, 613790, Autosomal recessive (Immunodeficiency due to a late component of complement deficiency) (C8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C8A	C8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 8 deficiency, 1 (C8D1) [MIM:613790]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

C8 Eksikliği, Tip 2, 613789, Otozomal Resesif İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C8B gen) (Tüm Gen Dizi Analizi) (Postnatal)	C8 deficiency, type II, 613789, Autosomal recessive (Immunodeficiency due to a late component of complement deficiency) (Immunodeficiency due to a late component of complement deficiency) (C8B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C8B	C8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 8 deficiency, 2 (C8D2) [MIM:613789]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
C8 Eksikliği, Tip 2, 613789, Otozomal Resesif İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C8B gen) (Tüm Gen Dizi Analizi) (Prenatal)	C8 deficiency, type II, 613789, Autosomal recessive (Immunodeficiency due to a late component of complement deficiency) (Immunodeficiency due to a late component of complement deficiency) (C8B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C8B	C8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 8 deficiency, 2 (C8D2) [MIM:613789]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
C9 Eksikliği, 613825 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	C9 deficiency, 613825 (Immunodeficiency due to a late component of complement deficiency) (C9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C9	C9, C9D, ARMD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 9 deficiency (C9D) [MIM:613825]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Some patients may develop dermatomyositis. {ECO:0000269|PubMed:9634479}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
C9 Eksikliği, 613825 (İmmun Yetmelik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	C9 deficiency, 613825 (Immunodeficiency due to a late component of complement deficiency) (C9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C9	C9, C9D, ARMD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 9 deficiency (C9D) [MIM:613825]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Some patients may develop dermatomyositis. {ECO:0000269|PubMed:9634479}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Caffey Hastalığı, 114000, Otozomal Dominant (Caffey Hastalığı) (COL1A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Caffey disease, 114000, Autosomal dominant (Caffey disease) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Caffey Hastalığı, 114000, Otozomal Dominant (Caffey Hastalığı) (MLPA)	Caffey disease, 114000, Autosomal dominant (Caffey disease) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Eklem ve Arterlerin Kalsifikasyonu, 211800, Otozomal Resesif (Kalitsal Arter ve Eklem Multipl Kalsifikasyon Sendromu) (NT5E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Calcification of joints and arteries, 211800, Autosomal recessive (Hereditary arterial and articular multiple calcification syndrome) (NT5E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NT5E	NT5E, NT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Calcification of joints and arteries (CALJA) [MIM:211800]: A condition characterized by adult-onset calcification of the lower extremity arteries, including the iliac, femoral and tibial arteries, and hand and foot capsule joints. Age of onset has been reported as early as the second decade of life, usually involving intense joint pain or calcification in the hands. {ECO:0000269|PubMed:21288095}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kalsiyum, Serum Seviyesi (Allesel Hipokalsürik Hiperkalsemi) (CASR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Calcium, serum level of (Familial hypocalciuric hypercalcemia) (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11782699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kalsiyum, Serum Seviyesi (Allesel Hipokalsürik Hiperkalsemi) (MLPA)	Calcium, serum level of (Familial hypocalciuric hypercalcemia) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11782699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kampomelik Displazi-Otozomal Cinsiyet Uyumsuzluğu, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Campomelic dysplasia with autosomal sex reversal, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kampomelik Displazi-Otozomal Cinsiyet Uyumsuzluğu, 114290, Otozomal Dominant (Kampomelik Displazi) (MLPA)	Campomelic dysplasia with autosomal sex reversal, 114290, Autosomal dominant (Campomelic dysplasia) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kampomelik Displazi-Otozomal Cinsiyet Uyumsuzluğu, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Campomelic dysplasia with autosomal sex reversal, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kampomelik Displazi-Otozomal Cinsiyet Uyumsuzluğu, 114290, Otozomal Dominant (Kampomelik Displazi) (Prenatal) (MLPA)	Campomelic dysplasia with autosomal sex reversal, 114290, Autosomal dominant (Campomelic dysplasia) (Prenatal) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (MLPA)	Campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (SOX9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (SOX9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX9	SOX9, CMD1, SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kampomelik Displazi, 114290, Otozomal Dominant (Kampomelik Displazi) (Prenatal) (MLPA)	Campomelic dysplasia, 114290, Autosomal dominant (Campomelic dysplasia) (Prenatal) (MLPA)	SOX9	SOX9, CMD1, SRA1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Campomelic dysplasia (CMD1) [MIM:114290]: A rare, often lethal, osteochondrodysplasia characterized by congenital bowing and angulation of long bones. Other skeletal defects include unusually small scapula, deformed pelvis and spine, and a missing pair of ribs. Craniofacial and ear defects are common. Most patients die soon after birth due to respiratory distress which has been attributed to hypoplasia of the tracheobronchial cartilage and small thoracic cage. Up to two-thirds of affected XY individuals have genital defects or may develop as phenotypic females. {ECO:0000269|PubMed:10446171, ECO:0000269|PubMed:10951468, ECO:0000269|PubMed:11323423, ECO:0000269|PubMed:11754051, ECO:0000269|PubMed:12783851, ECO:0000269|PubMed:19921652, ECO:0000269|PubMed:20513132, ECO:0000269|PubMed:7485151, ECO:0000269|PubMed:9002675, ECO:0000269|PubMed:9452059}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kamptodaktili-Artropati-Koksa Vara-Perikardit Sendromu, 208250, Otozomal Resesif; CACP (Kamptodaktili-Artropati-Koksa Vara-Perikardit Sendromu) (PRG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250, Autosomal recessive; CACP (Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome) (PRG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRG4	PRG4, CACP, MSF, SZP, HAPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Camptodactyly-arthropathy-coxa vara-pericarditis syndrome (CACP) [MIM:208250]: An autosomal recessive disorder characterized by the association of congenital or early-onset camptodactyly and non-inflammatory arthropathy with synovial hyperplasia. Individuals with CACP have normal appearing joints at birth but with advancing age develop joint failure, non-inflammatory synoviocyte hyperplasia and subintimal fibrosis of the synovial capsule. Some patients also manifest progressive coxa vara deformity and/or non-inflammatory pericardial or pleural effusions. {ECO:0000269|PubMed:10545050}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kamptodaktili-Artropati-Koksa Vara-Perikardit Sendromu, 208250, Otozomal Resesif; CACP (Kamptodaktili-Artropati-Koksa Vara-Perikardit Sendromu) (PRG4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250, Autosomal recessive; CACP (Camptodactyly-arthropathy-coxa-vara-pericarditis syndrome) (PRG4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRG4	PRG4, CACP, MSF, SZP, HAPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Camptodactyly-arthropathy-coxa vara-pericarditis syndrome (CACP) [MIM:208250]: An autosomal recessive disorder characterized by the association of congenital or early-onset camptodactyly and non-inflammatory arthropathy with synovial hyperplasia. Individuals with CACP have normal appearing joints at birth but with advancing age develop joint failure, non-inflammatory synoviocyte hyperplasia and subintimal fibrosis of the synovial capsule. Some patients also manifest progressive coxa vara deformity and/or non-inflammatory pericardial or pleural effusions. {ECO:0000269|PubMed:10545050}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kamptopolisindaktili, Kompleks, 607539, Otozomal Resesif; CCSPD (BHLHA9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Camptosynpolydactyly, complex, 607539, Autosomal recessive; CCSPD (BHLHA9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BHLHA9	BHLHA9, BHLHF42, MSSD, CCSPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kamptopolisindaktili, Kompleks, 607539, Otozomal Resesif; CCSPD (BHLHA9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Camptosynpolydactyly, complex, 607539, Autosomal recessive; CCSPD (BHLHA9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BHLHA9	BHLHA9, BHLHF42, MSSD, CCSPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Camurati-Engelmann Hastalığı, 131300, Otozomal Dominant; CAEND (Camurati-Engelmann Hastalığı) (TGFB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Camurati-Engelmann disease, 131300, Autosomal dominant; CAEND (Camurati-Engelmann disease) (TGFB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFB1	TGFB1, DPD1, CED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Camurati-Engelmann disease (CAEND) [MIM:131300]: An autosomal dominant disorder characterized by hyperostosis and sclerosis of the diaphyses of long bones. The disease typically presents in early childhood with pain, muscular weakness and waddling gait, and in some cases other features such as exophthalmos, facial paralysis, hearing difficulties and loss of vision. {ECO:0000269|PubMed:10973241, ECO:0000269|PubMed:11062463, ECO:0000269|PubMed:15103729}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Camurati-Engelmann Hastalığı, 131300, Otozomal Dominant; CAEND (Camurati-Engelmann Hastalığı) (TGFB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Camurati-Engelmann disease, 131300, Autosomal dominant; CAEND (Camurati-Engelmann disease) (TGFB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGFB1	TGFB1, DPD1, CED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Camurati-Engelmann disease (CAEND) [MIM:131300]: An autosomal dominant disorder characterized by hyperostosis and sclerosis of the diaphyses of long bones. The disease typically presents in early childhood with pain, muscular weakness and waddling gait, and in some cases other features such as exophthalmos, facial paralysis, hearing difficulties and loss of vision. {ECO:0000269|PubMed:10973241, ECO:0000269|PubMed:11062463, ECO:0000269|PubMed:15103729}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Canavan Hastalığı, 271900, Otozomal Resesif (Canavan Hastalığı) (ASPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Canavan disease, 271900, Autosomal recessive (Canavan disease) (ASPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPA	ASPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Canavan Hastalığı, 271900, Otozomal Resesif (Canavan Hastalığı) (MLPA)	Canavan disease, 271900, Autosomal recessive (Canavan disease) (MLPA)	ASPA	ASPA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Canavan Hastalığı, 271900, Otozomal Resesif (Canavan Hastalığı) (ASPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Canavan disease, 271900, Autosomal recessive (Canavan disease) (ASPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASPA	ASPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Canavan Hastalığı, 271900, Otozomal Resesif (Canavan Hastalığı) (Prenatal) (MLPA)	Canavan disease, 271900, Autosomal recessive (Canavan disease) (Prenatal) (MLPA)	ASPA	ASPA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kanser Progresyonu/ Metastaz (FGFR4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cancer progression/metastasis (FGFR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR4	FGFR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kandidiyazis, Ailesel, 2, Otozomal Resesif, 212050, Otozomal Resesif; CANDF2 (İnvazif Mantar Hastalığına Yatkınlık,CARD9 Eksikliğine Bağlı) (CARD9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Candidiasis, familial, 2, autosomal recessive, 212050, Autosomal recessive; CANDF2 (Predisposition to invasive fungal disease due to CARD9 deficiency) (CARD9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARD9	CARD9, CANDF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 2 (CANDF2) [MIM:212050]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269|PubMed:19864672, ECO:0000269|PubMed:23335372, ECO:0000269|PubMed:24131138). Note=The disease is caused by mutations affecting the gene represented in this entry. Defects induce reduced numbers of CD4(+) Th17 lymphocytes as well as a lack of monocyte-derived cytokines in response to Candida strains. Neutrophils show a selective Candida albicans killing defect with abnormal ultrastructural phagolysosomes and outgrowth of hyphae (PubMed:23335372). (ECO:0000269|PubMed:23335372).	EDTA Blood Sample
Kandidiyazis, Ailesel, 2, Otozomal Resesif, 212050, Otozomal Resesif; CANDF2 (İnvazif Mantar Hastalığına Yatkınlık,CARD9 Eksikliğine Bağlı) (Prenatal) (CARD9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Candidiasis, familial, 2, autosomal recessive, 212050, Autosomal recessive; CANDF2 (Predisposition to invasive fungal disease due to CARD9 deficiency) (CARD9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CARD9	CARD9, CANDF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Candidiasis, familial, 2 (CANDF2) [MIM:212050]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269|PubMed:19864672, ECO:0000269|PubMed:23335372, ECO:0000269|PubMed:24131138). Note=The disease is caused by mutations affecting the gene represented in this entry. Defects induce reduced numbers of CD4(+) Th17 lymphocytes as well as a lack of monocyte-derived cytokines in response to Candida strains. Neutrophils show a selective Candida albicans killing defect with abnormal ultrastructural phagolysosomes and outgrowth of hyphae (PubMed:23335372). (ECO:0000269|PubMed:23335372).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kandidiyazis, Ailesel, 4, Otozomal Resesif, 613108, Otozomal Resesif; CANDF4 (Kronik Mukokutanöz Kandidiyazi) (CLEC7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Candidiasis, familial, 4, autosomal recessive, 613108, Autosomal recessive; CANDF4 (Chronic mucocutaneous candidiasis) (CLEC7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLEC7A	CLEC7A, CLECSF12, DECTIN1, CANDF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 4 (CANDF4) [MIM:613108]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:19864674). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kandidiyazis, Ailesel, 4, Otozomal Resesif, 613108, Otozomal Resesif; CANDF4 (Kronik Mukokutanöz Kandidiyazi) (Prenatal) (CLEC7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Candidiasis, familial, 4, autosomal recessive, 613108, Autosomal recessive; CANDF4 (Chronic mucocutaneous candidiasis) (CLEC7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLEC7A	CLEC7A, CLECSF12, DECTIN1, CANDF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Candidiasis, familial, 4 (CANDF4) [MIM:613108]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:19864674). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kandidiasis, Ailesel, 6, Otozomal Dominant, 613956; CANDF6 (Kronik Mukokutanöz Kandidiyazi (IL17F geni) (Tüm Gen Dizi Analizi) (Postnatal)	Candidiasis, familial, 6, autosomal dominant, 613956; CANDF6 (Chronic mucocutaneous candidiasis) (IL17F gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL17F	IL17F, ML1, CANDF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kandidiasis, Ailesel, 6, Otozomal Dominant, 613956; CANDF6 (Kronik Mukokutanöz Kandidiyazi (Prenatal) (IL17F geni) (Tüm Gen Dizi Analizi) (Prenatal)	Candidiasis, familial, 6, autosomal dominant, 613956; CANDF6 (Chronic mucocutaneous candidiasis) (IL17F gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL17F	IL17F, ML1, CANDF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kandidiasis, Ailesel, 8, 615527, Otozomal Resesif; CANDF8 (Kronik Mukokutanöz Kandidiyazı (TRAF3IP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Candidiasis, familial, 8, 615527, Autosomal recessive; CANDF8 (Chronic mucocutaneous candidiasis) (TRAF3IP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAF3IP2	TRAF3IP2, C6orf5, ACT1, CIK5, C6orf4, C6orf6, PSORS13, CANDF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 8 (CANDF8) [MIM:615527]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:24120361). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kandidiasis, Ailesel, 8, 615527, Otozomal Resesif; CANDF8 (Kronik Mukokutanöz Kandidiyazi) (Prenatal) (TRAF3IP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Candidiasis, familial, 8, 615527, Autosomal recessive; CANDF8 (Chronic mucocutaneous candidiasis) (TRAF3IP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAF3IP2	TRAF3IP2, C6orf5, ACT1, CIK5, C6orf4, C6orf6, PSORS13, CANDF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Candidiasis, familial, 8 (CANDF8) [MIM:615527]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:24120361). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kandidiyazis, Ailesel, 9, 616445, Otozomal Resesif; CANDF9 (Kronik Mukokutanöz Kandidiyazi) (IL17RC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Candidiasis, familial, 9, 616445, Autosomal recessive; CANDF9 (Chronic mucocutaneous candidiasis) (IL17RC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL17RC	IL17RC, IL17RL, CANDF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 9 (CANDF9) [MIM:616445]: A disorder characterized by altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:25918342). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kandidiyazis, Ailesel, 9, 616445, Otozomal Resesif; CANDF9 (Kronik Mukokutanöz Kandidiyazi) (Prenatal) (IL17RC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Candidiasis, familial, 9, 616445, Autosomal recessive; CANDF9 (Chronic mucocutaneous candidiasis) (IL17RC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL17RC	IL17RC, IL17RL, CANDF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Candidiasis, familial, 9 (CANDF9) [MIM:616445]: A disorder characterized by altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. (ECO:0000269	PubMed:25918342). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cantu Sendromu (Hipertrikotik Osteokondrodisplazi, Cantu Tipi) (ABCC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CANTU SYNDROME (Hypertrichotic osteochondrodysplasia, Cantu type) (ABCC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC9	ABCC9, SUR2, CMD1O, ATPB12, CANTU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypertrichotic osteochondrodysplasia (HTOCD) [MIM:239850]: A rare disorder characterized by congenital hypertrichosis, neonatal macrosomia, a distinct osteochondrodysplasia, and cardiomegaly. The hypertrichosis leads to thick scalp hair, which extends onto the forehead, and a general increase in body hair. In addition, macrocephaly and coarse facial features, including a broad nasal bridge, epicanthal folds, a wide mouth, and full lips, can be suggestive of a storage disorder. About half of affected individuals are macrosomic and edematous at birth, whereas in childhood they usually have a muscular appearance with little subcutaneous fat. Thickened calvarium, narrow thorax, wide ribs, flattened or ovoid vertebral bodies, cova valga, osteopenia, enlarged medullary canals, and metaphyseal widening of long bones have been reported. Cardiac manifestations such as patent ductus arteriosus, ventricular hypertrophy, pulmonary hypertension, and pericardial effusions are present in approximately 80% of cases. Motor development is usually delayed due to hypotonia. Most patients have a mild speech delay, and a small percentage have learning difficulties or intellectual disability. (ECO:0000269|PubMed:22608503, ECO:0000269|PubMed:22610116). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CAP Miyopati 1, 609284, Otozomal Resesif, Otozomal Dominant (CAP Miyopati) (TPM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CAP myopathy 1, 609284, Autosomal recessive, Autosomal dominant (CAP myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAP Miyopati 1, 609284, Otozomal Resesif, Otozomal Dominant (CAP Miyopati) (Prenatal) (TPM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CAP myopathy 1, 609284, Autosomal recessive, Autosomal dominant (CAP myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CAP Miyopati 2, 609285, Otozomal Dominant (CAP Miyopati) (TPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CAP myopathy 2, 609285, Autosomal dominant (Cap myopathy) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAP Miyopati 2, 609285, Otozomal Dominant (CAP Miyopati) (Prenatal) (TPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CAP myopathy 2, 609285, Autosomal dominant (Cap myopathy) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kapiller Malformasyon-Arteriovenöz Malformasyonlar, 608354, Otozomal Dominant; CMAVM (Kapiller Malformasyon-Arteriovenöz Malformasyon Sendromu) (RASA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Capillary malformation-arteriovenous malformation, 608354, Autosomal dominant; CMAVM (Capillary malformation-arteriovenous malformation syndrome) (RASA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RASA1	RASA1, GAP, CMAVM, PKWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Capillary malformation-arteriovenous malformation (CMAVM) [MIM:608354]: A disorder characterized by atypical capillary malformations that are multiple, small, round to oval in shape and pinkish red in color. These capillary malformations are associated with either arteriovenous malformation, arteriovenous fistula, or Parkes Weber syndrome. (ECO:0000269|PubMed:14639529, ECO:0000269|PubMed:24038909). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kapiller Malformasyonlar, Konjenital, 1, Somatik, Mozaik, 163000; CMC (Ailesel Multipl Nevi Flammei) (GNAQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Capillary malformations, congenital, 1, somatic, mosaic, 163000; CMC (Familial multiple nevi flammei) (GNAQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAQ	GNAQ, SWS, CMC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Capillary malformations, congenital (CMC) [MIM:163000]: A form of vascular malformations that are present from birth, tend to grow with the individual, do not regress spontaneously, and show normal rates of endothelial cell turnover. Capillary malformations are distinct from capillary hemangiomas, which are highly proliferative lesions that appear shortly after birth and show rapid growth, slow involution, and endothelial hypercellularity. (ECO:0000269	PubMed:23656586). Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
CAPOS Sendromu, 601338, Otozomal Dominant; CAPOS (Serebellar Ataksi-Arefleksi-Pes Cavus-Optik Atrofi-Sensörinöral İşitme Kaybı Sendromu (CAPOS Sendromu)) (ATP1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CAPOS syndrome, 601338, Autosomal dominant; CAPOS (Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome) (ATP1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPOS Sendromu, 601338, Otozomal Dominant; CAPOS (Serebellar Ataksi-Arefleksi-Pes Cavus-Optik Atrofi-Sensörinöral İşitme Kaybı Sendromu (CAPOS Sendromu)) (MLPA)	CAPOS syndrome, 601338, Autosomal dominant; CAPOS (Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome) (MLPA)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CAPOS Sendromu, 601338, Otozomal Dominant; CAPOS (Serebellar Ataksi-Arefleksi-Pes Cavus-Optik Atrofi-Sensörinöral İşitme Kaybı Sendromu (CAPOS Sendromu)) (Prenatal) (ATP1A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CAPOS syndrome, 601338, Autosomal dominant; CAPOS (Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome) (ATP1A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CAPOS Sendromu, 601338, Otozomal Dominant; CAPOS (Serebellar Ataksi-Arefleksi-Pes Cavus-Optik Atrofi-Sensörinöral İşitme Kaybı Sendromu (CAPOS Sendromu)) (Prenatal) (MLPA)	CAPOS syndrome, 601338, Autosomal dominant; CAPOS (Cerebellar ataxia-areflexia-pes cavus-optic atrophy-sensorineural hearing loss syndrome) (Prenatal) (MLPA)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

CARASIL Sendromu, 600142, Otozomal Resesif (CARASIL) (HTRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CARASIL syndrome, 600142, Autosomal recessive (CARASIL) (HTRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA1	HTRA1, PRSS11, ARMD7, CARASIL, CADASIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy (CARASIL) [MIM:600142]: A cerebrovascular disease characterized by non-hypertensive arteriopathy of cerebral small vessels with subcortical infarcts, alopecia, and spondylosis. Small cerebral arteries show arteriosclerotic changes, fibrous intimal proliferation, and hyaline degeneration with splitting of the intima and/or the internal elastic membrane. Neurologic features include progressive dementia, gait disturbances, extrapyramidal and pyramidal signs, and demyelination of the cerebral white matter with sparing of U fibers. {ECO:0000269	PubMed:19387015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CARASIL Sendromu, 600142, Otozomal Resesif (CARASIL) (Prenatal) (HTRA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CARASIL syndrome, 600142, Autosomal recessive (CARASIL) (HTRA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HTRA1	HTRA1, PRSS11, ARMD7, CARASIL, CADASIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy (CARASIL) [MIM:600142]: A cerebrovascular disease characterized by non-hypertensive arteriopathy of cerebral small vessels with subcortical infarcts, alopecia, and spondylosis. Small cerebral arteries show arteriosclerotic changes, fibrous intimal proliferation, and hyaline degeneration with splitting of the intima and/or the internal elastic membrane. Neurologic features include progressive dementia, gait disturbances, extrapyramidal and pyramidal signs, and demyelination of the cerebral white matter with sparing of U fibers. {ECO:0000269	PubMed:19387015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karbamiloilfosfat Sentetaz 1 Eksikliği, 237300, Otozomal Resesif; CPT1 (Karbamoil-Fosfat Sentetaz 1 Eksikliği) (CPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carbamoylphosphate synthetase I deficiency, 237300, Autosomal recessive (Carbamoyl-phosphate synthetase 1 deficiency) (CPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPS1	CPS1, PHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carbamoyl phosphate synthetase 1 deficiency (CPS1D) [MIM:237300]: An autosomal recessive disorder of the urea cycle causing hyperammonemia. It can present as a devastating metabolic disease dominated by severe hyperammonemia in neonates or as a more insidious late-onset condition, generally manifesting as life-threatening hyperammonemic crises under catabolic situations. Clinical features include protein intolerance, intermittent ataxia, seizures, lethargy, developmental delay and mental retardation. {ECO:0000269|PubMed:11388595, ECO:0000269|PubMed:11474210, ECO:0000269|PubMed:12655559, ECO:0000269|PubMed:12955727, ECO:0000269|PubMed:15164414, ECO:0000269|PubMed:15617192, ECO:0000269|PubMed:16737834, ECO:0000269|PubMed:17310273, ECO:0000269|PubMed:20578160, ECO:0000269|PubMed:21120950, ECO:0000269|PubMed:9711878}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Karbamiloilfosfat Sentetaz 1 Eksikliği, 237300, Otozomal Resesif; CPT1 (Karbamoil-Fosfat Sentetaz 1 Eksikliği) (Prenatal) (CPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carbamoylphosphate synthetase I deficiency, 237300, Autosomal recessive (Carbamoyl-phosphate synthetase 1 deficiency) (CPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPS1	CPS1, PHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carbamoyl phosphate synthetase 1 deficiency (CPS1D) [MIM:237300]: An autosomal recessive disorder of the urea cycle causing hyperammonemia. It can present as a devastating metabolic disease dominated by severe hyperammonemia in neonates or as a more insidious late-onset condition, generally manifesting as life-threatening hyperammonemic crises under catabolic situations. Clinical features include protein intolerance, intermittent ataxia, seizures, lethargy, developmental delay and mental retardation. {ECO:0000269|PubMed:11388595, ECO:0000269|PubMed:11474210, ECO:0000269|PubMed:12655559, ECO:0000269|PubMed:12955727, ECO:0000269|PubMed:15164414, ECO:0000269|PubMed:15617192, ECO:0000269|PubMed:16737834, ECO:0000269|PubMed:17310273, ECO:0000269|PubMed:20578160, ECO:0000269|PubMed:21120950, ECO:0000269|PubMed:9711878}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karboksilesteraz 1 Eksikliği (CES1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carboxylesterase 1 deficiency (CES1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CES1	CES1, SES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Karboksilesteraz 1 Eksikliği (Prenatal) (CES1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carboxylesterase 1 deficiency (CES1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CES1	CES1, SES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karboksipeptidaz N Eksikliği, 212070, Otozomal Resesif (CPN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carboxypeptidase N deficiency, 212070, Autosomal recessive (CPN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPN1	CPN1, SCPN, CPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Karboksipeptidaz N Eksikliği, 212070, Otozomal Resesif (Prenatal) (CPN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carboxypeptidase N deficiency, 212070, Autosomal recessive (CPN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPN1	CPN1, SCPN, CPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akciğer Karsinoid Tümörü (Multipl Endokrin Neoplazi Tip 1) (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carcinoid tumor of lung (Multiple endocrine neoplasia type 1) (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Karsinoid Tümörü (Multipl Endokrin Neoplazi Tip 1) (MLPA)	Carcinoid tumor of lung (Multiple endocrine neoplasia type 1) (MLPA)	MEN1	MEN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Karsinoid Tümörler, Intestinal, 114900, Otozomal Dominant (Karsinoid Tümör ve Karsinoid Sendromi) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carcinoid tumors, intestinal, 114900, Autosomal dominant (Carcinoid tumor and carcinoid syndrome) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269	PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyak Aritmi, Ankirin-B İle İlişkili, 600919, Otozomal Dominant (Ailesel Uzun QT Sendromu) (ANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiac arrhythmia, ankyrin-B-related, 600919, Autosomal dominant (Familial long QT syndrome) (ANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANK2	ANK2, LQT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 4 (LQT4) [MIM:600919]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 4 shows many atypical features compared to classical long QT syndromes, including pronounced sinus bradycardia, polyphasic T waves and atrial fibrillation. Cardiac repolarization defects may be not as severe as in classical LQT syndromes and prolonged QT interval on EKG is not a consistent feature. {ECO:0000269|PubMed:12571597, ECO:0000269|PubMed:15178757}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyak İletim Defekti, Nonspesifik, 612838 (Ailesel Progresif Kardiyak İletim Defekti) (SCN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiac conduction defect, nonspecific, 612838 (Familial progressive cardiac conduction defect) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEE52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brugada syndrome 5 (BRGDA5) [MIM:612838]: A tachyarrhythmia characterized by right bundle branch block and ST segment elevation on an electrocardiogram (ECG). It can cause the ventricles to beat so fast that the blood is prevented from circulating efficiently in the body. When this situation occurs, the individual will faint and may die in a few minutes if the heart is not reset. {ECO:0000269	PubMed:18464934}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyak İletim Defekti, Yaşlanmış, 115080, Otozomal Dominant (Ailesel Progresif Kardiyak İletim Defekti) (AKAP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiac conduction defect, susceptibility to, 115080, Autosomal dominant (Familial progressive cardiac conduction defect) (AKAP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKAP10	AKAP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sudden cardiac death (SCD) [MIM:115080]: Unexpected rapid death due to cardiovascular collapse in a short time period, generally within one hour of initial symptoms. It is usually caused by the worsening of existing heart diseases. The sudden onset of symptoms, such as chest pain and cardiac arrhythmias, particularly ventricular tachycardia, can lead to the loss of consciousness and cardiac arrest followed by biological death. {ECO:0000269	PubMed:17485678}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Increased susceptibility to sudden cardiac death may be conferred by AKAP10 variants that are associated with markers of low vagus nerve sensitivity, e.g. fast basal heart rate and low heart rate variability.;	EDTA Blood Sample
Kalp İletim Hastalığı, ± Dilate Kardiyomiyopati, 616117, Otozomal Dominant; CCDD (Ailesel Atriyal Taşiaritmi-Ait-Hisian Kardiyak İletim Hastalığı) (TNNI3K geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiac conduction disease with or without dilated cardiomyopathy, 616117, Autosomal dominant; CCDD (Familial atrial tachyarrhythmia-infra-Hisian cardiac conduction disease) (TNNI3K gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI3K	TNNI3K, CCDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiac conduction disease with or without dilated cardiomyopathy (CCDD) [MIM:616117]: A cardiac disorder characterized by atrial tachyarrhythmia and conduction system disease. Some patients have dilated cardiomyopathy. {ECO:0000269	PubMed:24925317}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyak septal defekt (GATA4 8p23, GATA3 10p15) (MLPA)	Cardiac septal defect (GATA4 8p23, GATA3 10p15) (MLPA)	GATA4 8p23, GATA3 10p15	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyak septal defekt (GATA4 8p23, GATA3 10p15) (MLPA) (Prenatal)	Cardiac septal defect (GATA4 8p23, GATA3 10p15) (MLPA) (Prenatal)	GATA4 8p23, GATA3 10p15	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyak Valvüler Displazi, X'e Bağlı, 314400, X'e Bağlı Resesif; CVD1 (Konjenital Kapakçık Displazisi) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiac valvular dysplasia, X-linked, 314400, X-linked recessive; CVD1 (Congenital valvular dysplasia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyak Valvüler Displazi, X'e Bağlı, 314400, X'e Bağlı Resesif; CVD1 (Konjenital Kapakçık Displazisi) (MLPA)	Cardiac valvular dysplasia, X-linked, 314400, X-linked recessive; CVD1 (Congenital valvular dysplasia) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyoensefalomiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 1, 604377, Otozomal Resesif; SCO2 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (SCO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377, Autosomal recessive; SCO2 (Fatal infantile cytochrome C oxidase deficiency) (SCO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCO2	SCO2, CEMCOX1, MYP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyoensefalomiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 1, 604377, Otozomal Resesif; CEMCOX1 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (Prenatal) (SCO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 1, 604377, Autosomal recessive; CEMCOX1 (Fatal infantile cytochrome C oxidase deficiency) (SCO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCO2	SCO2, CEMCOX1, MYP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardioensefalomiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 2, 615119, Otozomal Resesif; COX15 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (COX15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119, Autosomal recessive; COX15 (Fatal infantile cytochrome C oxidase deficiency) (COX15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX15	COX15, CEMCOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardioensefalomiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 2, 615119, Otozomal Resesif; CEMCOX2 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 2, 615119, Autosomal recessive; CEMCOX2 (Fatal infantile cytochrome C oxidase deficiency) (COX15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX15	COX15, CEMCOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kardioensefalmiyopati Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 3, 616500, Otozomal Resesif; CEMCOX3 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (COA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500, Autosomal recessive; CEMCOX3 (Fatal infantile cytochrome C oxidase deficiency) (COA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COA5	COA5, C2orf64, PET191, CEMCOX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3 (CEMCOX3) [MIM:616500]: An infantile disorder with a fatal course in the first weeks of life, characterized by hypertrophic cardiomyopathy and mitochondrial complex IV deficiency. Postmortem microscopic investigations show accumulation of lipid droplets in cardiomyocytes and mitochondrial proliferation. {ECO:0000269	PubMed:21457908}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardioensefalmiyopati Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 3, 616500, Otozomal Resesif; CEMCOX3 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (Prenatal) (COA5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3, 616500, Autosomal recessive; CEMCOX3 (Fatal infantile cytochrome C oxidase deficiency) (COA5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COA5	COA5, C2orf64, PET191, CEMCOX3	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 3 (CEMCOX3) [MIM:616500]: An infantile disorder with a fatal course in the first weeks of life, characterized by hypertrophic cardiomyopathy and mitochondrial complex IV deficiency. Postmortem microscopic investigations show accumulation of lipid droplets in cardiomyocytes and mitochondrial proliferation. {ECO:0000269	PubMed:21457908}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardioensefalmiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 4, 616501, Otozomal Resesif; CEMCOX4 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (COA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501, Autosomal recessive; CEMCOX4 (Fatal infantile cytochrome C oxidase deficiency) (COA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COA6	COA6, C1orf31, CEMCOX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4 (CEMCOX4) [MIM:616501]: An infantile disorder with a fatal course in the first weeks of life, characterized by hypertrophic cardiomyopathy, left ventricular non-compaction, lactic acidosis, metabolic hypotonia, and mitochondrial complex IV deficiency. {ECO:0000269|PubMed:24549041, ECO:0000269|PubMed:25339201}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardioensefalmiyopati, Fatal İnfantil, Sitokrom C Oksidaz Eksikliğine Bağlı 4, 616501, Otozomal Resesif; CEMCOX4 (Fatal İnfantil Sitokrom C Oksidaz Eksikliği) (Prenatal) (COA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4, 616501, Autosomal recessive; CEMCOX4 (Fatal infantile cytochrome C oxidase deficiency) (COA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COA6	COA6, C1orf31, CEMCOX4	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency 4 (CEMCOX4) [MIM:616501]: An infantile disorder with a fatal course in the first weeks of life, characterized by hypertrophic cardiomyopathy, left ventricular non-compaction, lactic acidosis, metabolic hypotonia, and mitochondrial complex IV deficiency. {ECO:0000269	PubMed:24549041, ECO:0000269	PubMed:25339201}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 2, 615278; CFC2 (Kardiyofasiyokutanöz Sendrom) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiofaciocutaneous syndrome 2, 615278; CFC2 (Cardiofaciocutaneous syndrome) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 2, 615278; CFC2 (Kardiyofasiyokutanöz Sendromu) (MLPA)	Cardiofaciocutaneous syndrome 2, 615278; CFC2 (Cardiofaciocutaneous syndrome) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 2, 615278; CFC2 (Kardiyofasiyokutanöz Sendromu) (Prenatal) (KRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiofaciocutaneous syndrome 2, 615278; CFC2 (Cardiofaciocutaneous syndrome) (KRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 2, 615278; CFC2 (Kardiyofasiyokutanöz Sendromu) (Prenatal) (MLPA)	Cardiofaciocutaneous syndrome 2, 615278; CFC2 (Cardiofaciocutaneous syndrome) (Prenatal) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 3, 615279; CFC3 (Kardiyofasiyokutanöz Sendromu) (MAP2K1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiofaciocutaneous syndrome 3, 615279; CFC3 (Cardiofaciocutaneous syndrome) (MAP2K1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP2K1	MAP2K1, PRKMK1, MKK1, MEK1, CFC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiofaciocutaneous syndrome 3 (CFC3) [MIM:615279]: A form of cardiofaciocutaneous syndrome, a multiple congenital anomaly disorder characterized by a distinctive facial appearance, heart defects and mental retardation. Heart defects include pulmonic stenosis, atrial septal defects and hypertrophic cardiomyopathy. Some affected individuals present with ectodermal abnormalities such as sparse, friable hair, hyperkeratotic skin lesions and a generalized ichthyosis-like condition. Typical facial features are similar to Noonan syndrome. They include high forehead with bitemporal constriction, hypoplastic supraorbital ridges, downslanting palpebral fissures, a depressed nasal bridge, and posteriorly angulated ears with prominent helices. Distinctive features of CFC3 include macrostomia and horizontal shape of palpebral fissures. {ECO:0000269|PubMed:16439621, ECO:0000269|PubMed:18042262}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 3, 615279; CFC3 (Kardiyofasiyokutanöz Sendromu) (Prenatal) (MAP2K1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiofaciocutaneous syndrome 3, 615279; CFC3 (Cardiofaciocutaneous syndrome) (MAP2K1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP2K1	MAP2K1, PRKMK1, MKK1, MEK1, CFC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiofaciocutaneous syndrome 3 (CFC3) [MIM:615279]: A form of cardiofaciocutaneous syndrome, a multiple congenital anomaly disorder characterized by a distinctive facial appearance, heart defects and mental retardation. Heart defects include pulmonic stenosis, atrial septal defects and hypertrophic cardiomyopathy. Some affected individuals present with ectodermal abnormalities such as sparse, friable hair, hyperkeratotic skin lesions and a generalized ichthyosis-like condition. Typical facial features are similar to Noonan syndrome. They include high forehead with bitemporal constriction, hypoplastic supraorbital ridges, downslanting palpebral fissures, a depressed nasal bridge, and posteriorly angulated ears with prominent helices. Distinctive features of CFC3 include macrostomia and horizontal shape of palpebral fissures. {ECO:0000269|PubMed:16439621, ECO:0000269|PubMed:18042262}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 4, 615280; CFC4 (Kardiyofasiyokutanöz Sendromu) (MAP2K2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiofaciocutaneous syndrome 4, 615280; CFC4 (Cardiofaciocutaneous syndrome) (MAP2K2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP2K2	MAP2K2, PRKMK2, MEK2, MKK2, CFC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiofaciocutaneous syndrome 4 (CFC4) [MIM:615280]: A form of cardiofaciocutaneous syndrome, a multiple congenital anomaly disorder characterized by a distinctive facial appearance, heart defects and mental retardation. Heart defects include pulmonic stenosis, atrial septal defects and hypertrophic cardiomyopathy. Some affected individuals present with ectodermal abnormalities such as sparse, friable hair, hyperkeratotic skin lesions and a generalized ichthyosis-like condition. Typical facial features are similar to Noonan syndrome. They include high forehead with bitemporal constriction, hypoplastic supraorbital ridges, downslanting palpebral fissures, a depressed nasal bridge, and posteriorly angulated ears with prominent helices. {ECO:0000269|PubMed:16439621, ECO:0000269|PubMed:18042262, ECO:0000269|PubMed:20358587}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom 4, 615280; CFC4 (Kardiyofasiyokutanöz Sendromu) (Prenatal) (MAP2K2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiofaciocutaneous syndrome 4, 615280; CFC4 (Cardiofaciocutaneous syndrome) (MAP2K2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP2K2	MAP2K2, PRKMK2, MEK2, MKK2, CFC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiofaciocutaneous syndrome 4 (CFC4) [MIM:615280]: A form of cardiofaciocutaneous syndrome, a multiple congenital anomaly disorder characterized by a distinctive facial appearance, heart defects and mental retardation. Heart defects include pulmonic stenosis, atrial septal defects and hypertrophic cardiomyopathy. Some affected individuals present with ectodermal abnormalities such as sparse, friable hair, hyperkeratotic skin lesions and a generalized ichthyosis-like condition. Typical facial features are similar to Noonan syndrome. They include high forehead with bitemporal constriction, hypoplastic supraorbital ridges, downslanting palpebral fissures, a depressed nasal bridge, and posteriorly angulated ears with prominent helices. {ECO:0000269|PubMed:16439621, ECO:0000269|PubMed:18042262, ECO:0000269|PubMed:20358587}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom, 115150, Otozomal Dominant; CFC1 (Kardiyofasiyokutanöz Sendrom) (BRAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiofaciocutaneous syndrome, 115150, Autosomal dominant; CFC1 (Cardiofaciocutaneous syndrome) (BRAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyofasiyokutanöz Sendrom, 115150, Otozomal Dominant; CFC1 (Kardiyofasiyokutanöz Sendrom) (Prenatal) (BRAF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiofaciocutaneous syndrome, 115150, Autosomal dominant; CFC1 (Cardiofaciocutaneous syndrome) (BRAF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyomiyopati, Dilate, 1A, 115200, Otozomal Dominant; CMD1A (Ailesel İzole Dilate Kardiyomiyopati) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1A, 115200, Autosomal dominant; CMD1A (Familial isolated dilated cardiomyopathy) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1A, 115200, Otozomal Dominant; CMD1A (Ailesel İzole Dilate Kardiyomiyopati) (MLPA)	Cardiomyopathy, dilated, 1A, 115200, Autosomal dominant; CMD1A (Familial isolated dilated cardiomyopathy) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Kardiyomiyopati, Dilate,1AA, ± LVNC, 612158, Otozomal Dominant; CMD1AA (Allesel İzole Dilate Kardiyomiyopati) (ACTN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1AA, with or without LVNC, 612158, Autosomal dominant; CMD1AA (Familial isolated dilated cardiomyopathy) (ACTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN2	ACTN2, CMD1AA, CMH23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 23, with or without left ventricular non-compaction (CMH23) [MIM:612158]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:20022194, ECO:0000269|PubMed:25173926}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1BB, 612877; CMD1BB (Alleseİ İzole Dilate Kardiyomiyopati) (DSG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1BB, 612877; CMD1BB (Familial isolated dilated cardiomyopathy) (DSG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG2	DSG2, ARVD10, ARVC10, CMD1BB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1C ± LVNC, 601493, Otozomal Dominant; CMD1C (Alleseİ İzole Dilate Kardiyomiyopati) (LDB3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1C, with or without LVNC, 601493, Autosomal dominant; CMD1C (Familial isolated dilated cardiomyopathy) (LDB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDB3	LDB3, ZASP, CYPHER, KIAA0613, MFM4, CMD1C, CMH24, LVNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 3 (LVNC3) [MIM:601493]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1CC, 613122, Otozomal Dominant; CMD1CC (Alleseİ İzole Dilate Kardiyomiyopati) (NEXN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1CC, 613122, Autosomal dominant; CMD1CC (Familial isolated dilated cardiomyopathy) (NEXN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEXN	NEXN, NELIN, CMD1CC, CMH20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1D, 601494, Otozomal Dominant; CMD1D (Alleseİ İzole Dilate Kardiyomiyopati) (TNNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1D, 601494, Autosomal dominant; CMD1D (Familial isolated dilated cardiomyopathy) (TNNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT2	TNNT2, CMH2, CMD1D, RCM3, LVNC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1DD, 613172, Otozomal Dominant; CMD1DD (Alleseİ İzole Dilate Kardiyomiyopati) (RBM20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1DD, 613172, Autosomal dominant; CMD1DD (Familial isolated dilated cardiomyopathy) (RBM20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBM20	RBM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1E, 601154, Otozomal Dominant; CMD1E (Alleseİ İzole Dilate Kardiyomiyopati) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1E, 601154, Autosomal dominant; CMD1E (Familial isolated dilated cardiomyopathy) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1EE, 613252; CMD1EE (Alleseİ İzole Dilate Kardiyomiyopati) (MYH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1EE, 613252; CMD1EE (Familial isolated dilated cardiomyopathy) (MYH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH6	MYH6, ASD3, MYHCA, CMD1EE, CMH14, SSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1F ve Limb-Girdle Müskiler Distrofi Tip 1D, 602067 (DES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1F and limb-girdle muscular dystrophy type 1D, 602067 (DES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DES	CMD1F, CDCD3, LGMD1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1 FF; 613286; CMD1FF (Alleseİ İzole Dilate Kardiyomiyopati) (TNNI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1FF, 613286; CMD1FF (Familial isolated dilated cardiomyopathy) (TNNI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI3	TNNI3, CMH7, CMD2A, RCM1, CMD1FF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1G, 604145 (Alleseİ İzole Dilate Kardiyomiyopati) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1G, 604145 (Familial isolated dilated cardiomyopathy) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1G; Cmd1g (Alleseİ İzole Dilate Kardiyomiyopati) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	CARDIOMYOPATHY, DILATED, 1G; CMD1G (Familial isolated dilated cardiomyopathy) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1HH, 613881, Otozomal Dominant; CMD1HH (Alleseİ İzole Dilate Kardiyomiyopati) (BAG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1HH, 613881, Autosomal dominant; CMD1HH (Familial isolated dilated cardiomyopathy) (BAG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAG3	BAG3, MFM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1I, 604765; CMD1I (Alleseİ İzole Dilate Kardiyomiyopati) (DES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1I, 604765; CMD1I (Familial isolated dilated cardiomyopathy) (DES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DES	DES, CMD1I, MFM1, SCPNK, ARVD7, ARVC7, LGMD2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1II, 615184, Otozomal Dominant; CMD1II (Early-Onset Lamellar Cataract) (CRYAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1II, 615184, Autosomal dominant; CMD1II (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1II, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1J, 605362 (Sensorinöral Sağırlık-Dilate Kardiyomiyopati) (EYA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1J, 605362 (Sensorineural deafness with dilated cardiomyopathy) (EYA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA4	EYA4, DFNA10, CMD1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1J (CMD1J) [MIM:605362]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. CMD1J is characterized by the association of sensorineural hearing loss and dilated cardiomyopathy in the absence of other anomalies. {ECO:0000269|PubMed:15735644}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1JJ, 615235, Otozomal Dominant; CMD1JJ (Alleseİ İzole Dilate Kardiyomiyopati) (LAMA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1JJ, 615235, Autosomal dominant; CMD1JJ (Familial isolated dilated cardiomyopathy) (LAMA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA4	LAMA4, LAMA3, CMD1JJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1JJ (CMD1JJ) [MIM:615235]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:17646580}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1KK, 615248, Otozomal Dominant; CMD1KK (Alleseİ İzole Dilate Kardiyomiyopati) (MYPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1KK, 615248, Autosomal dominant; CMD1KK (Familial isolated dilated cardiomyopathy) (MYPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYPN	MYPN, CMD1DD, CMH22, RCM4, NEM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 22 (CMH22) [MIM:615248]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:22286171}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1L, 606685; CMD1L (Alleseİ İzole Dilate Kardiyomiyopati) (SGCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1L, 606685; CMD1L (Familial isolated dilated cardiomyopathy) (SGCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCD	SGCD, SGD, LGMD2F, CMD1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1L, 606685; CMD1L (Alleseİ İzole Dilate Kardiyomiyopati) (MLPA)	Cardiomyopathy, dilated, 1L, 606685; CMD1L (Familial isolated dilated cardiomyopathy) (MLPA)	SGCD	SGCD, SGD, LGMD2F, CMD1L	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1LL, 615373, Otozomal Dominant (Alleseİ İzole Dilate Kardiyomiyopati) (PRDM16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1LL, 615373, Autosomal dominant (Familial isolated dilated cardiomyopathy) (PRDM16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM16	PRDM16, MEL1, LVNC8, CMD1LL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1LL (CMD1LL) [MIM:615373]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:23768516}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1M, 607482; CMD1M (Alleseİ İzole Dilate Kardiyomiyopati) (CSRP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1M, 607482; CMD1M (Familial isolated dilated cardiomyopathy) (CSRP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSRP3	CSRP3, CRP3, CLP, CMD1M, CMH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1MM, 615396, Otozomal Dominant (Alleseİ İzole Dilate Kardiyomiyopati) (MYBPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1MM, 615396, Autosomal dominant (Familial isolated dilated cardiomyopathy) (MYBPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYBPC3	MYBPC3, CMH4, CMD1MM, LVNC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1NN, 615916, Otozomal Dominant; CMD1NN (Alleseİ İzole Dilate Kardiyomiyopati) (RAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1NN, 615916, Autosomal dominant; CMD1NN (Familial isolated dilated cardiomyopathy) (RAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAF1	RAF1, CRAF, NS5, CMD1NN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1O, 608569; CMD1O (Alleseİ İzole Dilate Kardiyomiyopati) (ABCC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1O, 608569; CMD1O (Familial isolated dilated cardiomyopathy) (ABCC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC9	ABCC9, SUR2, CMD1O, ATFB12, CANTU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1P, 609909; CMD1P (Alleseİ İzole Dilate Kardiyomiyopati) (PLN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1P, 609909; CMD1P (Familial isolated dilated cardiomyopathy) (PLN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLN	PLN, PLB, CMD1P, CMH18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1P (CMD1P) [MIM:609909]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:12610310, ECO:0000269|PubMed:16432188, ECO:0000269|PubMed:22137083}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1R, 613424, Otozomal Dominant; CMD1R (Alleseİ İzole Dilate Kardiyomiyopati) (ACTC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1R, 613424, Autosomal dominant; CMD1R (Familial isolated dilated cardiomyopathy) (ACTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTC1	ACTC1, CMD1R, CMH11, ASD5, LVNC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1S, 613426, Otozomal Dominant; CMD1S (Alleseİ İzole Dilate Kardiyomiyopati) (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1S, 613426, Autosomal dominant; CMD1S (Familial isolated dilated cardiomyopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1U, 613694, Otozomal Dominant; CMD1U (Alleseİ İzole Dilate Kardiyomiyopati) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1U, 613694, Autosomal dominant; CMD1U (Familial isolated dilated cardiomyopathy) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1V, 613697, Otozomal Dominant; CMD1V (Alleseİ İzole Dilate Kardiyomiyopati) (PSEN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1V, 613697, Autosomal dominant; CMD1V (Familial isolated dilated cardiomyopathy) (PSEN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN2	PSEN2, AD4, STM2, CMD1V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1W, 611407; CMD1W (Alleseİ İzole Dilate Kardiyomiyopati) (VCL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1W, 611407; CMD1W (Familial isolated dilated cardiomyopathy) (VCL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCL	VCL, CMD1W, CMH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1W (CMD1W) [MIM:611407]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:11815434, ECO:0000269|PubMed:16236538}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1X, 611615, Otozomal Resesif; CMD1X (Alleseİ İzole Dilate Kardiyomiyopati) (FKTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1X, 611615, Autosomal recessive; CMD1X (Familial isolated dilated cardiomyopathy) (FKTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kardiyomiyopati, Dilate, 1Y, 611878, Otozomal Dominant, CMD1Y (Aileseİ Izole Dilate Kardiyomiyopati) (TPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1Y, 611878, Autosomal dominant; CMD1Y (Familial isolated dilated cardiomyopathy) (TPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM1	TPM1, CMH3, CMD1Y , LVNC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 1Z, 611879; CMD1Z (Aileseİ Izole Dilate Kardiyomiyopati) (TNNC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 1Z, 611879; CMD1Z (Familial isolated dilated cardiomyopathy) (TNNC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNC1	TNNC1, CMD1Z, CMH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 2A, 611880, Otozomal Resesif, CMD2A (Aileseİ Izole Dilate Kardiyomiyopati) (TNNI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 2A, 611880, Autosomal recessive; CMD2A (Familial isolated dilated cardiomyopathy) (TNNI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI3	TNNI3, CMH7, CMD2A, RCM1, CMD1FF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 2B, 614672, Otozomal Resesif, CMD2B (Aileseİ Izole Dilate Kardiyomiyopati) (GATAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 2B, 614672, Autosomal recessive; CMD2B (Familial isolated dilated cardiomyopathy) (GATAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATAD1	GATAD1, ODAG, CMD2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 3B, 302045, X'e Bağlı; CMD3B (Aileseİ Izole Dilate Kardiyomiyopati) (DMD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, 3B, 302045, X-linked; CMD3B (Familial isolated dilated cardiomyopathy) (DMD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, 3B, 302045, X'e Bağlı; CMD3B (Aileseİ Izole Dilate Kardiyomiyopati) (MLPA)	Cardiomyopathy, dilated, 3B, 302045, X-linked; CMD3B (Familial isolated dilated cardiomyopathy) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, Hipergonadotropik Hipogonadizm (Dilate Kardiyomiyopati-Hipergonadotropik Hipogonadizm Sendromu (Malouf Sendromu)) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	CARDIOMYOPATHY, DILATED, WITH HYPERGONADOTROPIC HYPOGONADISM (Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Dilate, Hipergonadotropik Hipogonadizm (Dilate Kardiyomiyopati-Hipergonadotropik Hipogonadizm Sendromu (Malouf Sendromu)) (MLPA)	CARDIOMYOPATHY, DILATED, WITH HYPERGONADOTROPIC HYPOGONADISM (Dilated cardiomyopathy-hypergonadotropic hypogonadism syndrome) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Dilate Kardiyomiyopati, Yünsü Saç ve Keratoderma, 605676, Otozomal Resesif; DCWHK (Yünsü Saç-Palmoplantar Keratoderma-Dilate Kardiyomiyopati Sendromu) (DSP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676, Autosomal recessive; DCWHK (Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome) (DSP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated, with woolly hair and keratoderma (DCWHK) [MIM:605676]: An autosomal recessive cardiocutaneous syndrome characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. {ECO:0000269	PubMed:11063735}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dilate Kardiyomiyopati, Yünsü Saç ve Keratoderma, 605676, Otozomal Resesif; DCWHK (Yünsü Saç-Palmoplantar Keratoderma-Dilate Kardiyomiyopati Sendromu) (Prenatal) (DSP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676, Autosomal recessive; DCWHK (Woolly hair-palmoplantar keratoderma-dilated cardiomyopathy syndrome) (DSP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiomyopathy, dilated, with woolly hair and keratoderma (DCWHK) [MIM:605676]: An autosomal recessive cardiocutaneous syndrome characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. {ECO:0000269	PubMed:11063735}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kardiyomiyopati, Aileseİ Hipertrofik, 192600, Otozomal Dominant (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial hypertrophic, 192600, Autosomal dominant (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Hipertrofik, 192600, Otozomal Dominant (MLPA)	Cardiomyopathy, familial hypertrophic, 192600, Autosomal dominant (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Hipertrofik, 26; CMH26 (Filaminopati) (FLNC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial hypertrophic, 26; CMH26 (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Hipertrofik, 9, 613765; CMH9 (Tibial Müsküler Distrofi) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial hypertrophic, 9, 613765; CMH9 (Tibial muscular dystrophy) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Restriktif 5, 617047, Otozomal Dominant (Filaminopati) (FLNC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial restrictive 5, 617047, Autosomal dominant (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Restriktif, 1, 115210, Otozomal Dominant; RCM1 (Aileseİ Izole Restriktif Kardiyomiyopati) (TNNI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial restrictive, 1, 115210, Autosomal dominant; RCM1 (Familial isolated restrictive cardiomyopathy) (TNNI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI3	TNNI3, CMH7, CMD2A, RCM1, CMD1FF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial restrictive 1 (RCM1) [MIM:115210]: A heart disorder characterized by impaired filling of the ventricles with reduced diastolic volume, in the presence of normal or near normal wall thickness and systolic function. {ECO:0000269	PubMed:12531876}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati,Aileseİ Restriktif 3, 612422, Otozomal Dominant; RCM3 (Aileseİ Izole Restriktif Kardiyomiyopati) (TNNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial restrictive, 3, 612422, Autosomal dominant; RCM3 (Familial isolated restrictive cardiomyopathy) (TNNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT2	TNNT2, CMH2, CMD1D, RCM3, LVNC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Aileseİ Restriktif, 4, 615248, Otozomal Dominant (Aileseİ Izole Dilate Kardiyomiyopati) (MYPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, familial restrictive, 4, 615248, Autosomal dominant (Familial isolated restrictive cardiomyopathy) (MYPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYPN	MYPN, CMD1DD, CMH22, RCM4, NEM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 22 (CMH22) [MIM:615248]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269	PubMed:22286171}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik 6, 600858, Otozomal Dominant (PRKAG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic 6, 600858, Autosomal dominant (PRKAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAG2	PRKAG2, WPWS, CMH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 1, 192600, Otozomal Dominant (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 1, 192600, Autosomal dominant (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 1, Dijenik, 192600, Otozomal Dominant (MYLK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 1, digenic, 192600, Autosomal dominant (MYLK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYLK2	MYLK2, MLCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 10, 608758 (MYL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 10, 608758 (MYL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYL2	MYL2, CMH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 10 (CMH10) [MIM:608758]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. Rarely, patients present a variant of familial hypertrophic cardiomyopathy, characterized by mid-left ventricular chamber thickening. {ECO:0000269|PubMed:11102452, ECO:0000269|PubMed:12404107, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:12818575, ECO:0000269|PubMed:8673105, ECO:0000269|PubMed:9535554}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 11, 612098, Otozomal Dominant (ACTC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 11, 612098, Autosomal dominant (ACTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTC1	ACTC1, CMD1R, CMH11, ASD5, LVNC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 11 (CMH11) [MIM:612098]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:10330430, ECO:0000269|PubMed:10966831, ECO:0000269|PubMed:14729850, ECO:0000269|PubMed:18403758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 12, 612124, Otozomal Dominant (Aileseİ Izole Dilate Kardiyomiyopati) (CSRP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 12, 612124, Autosomal dominant (CSRP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSRP3	CSRP3, CRP3, CLP, CMD1M, CMH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 12 (CMH12) [MIM:612124]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:12642359, ECO:0000269|PubMed:18505755}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 13, 613243 (Aileseİ Izole Dilate Kardiyomiyopati) (TNNC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 13, 613243 (TNNC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNC1	TNNC1, CMD1Z, CMH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 13 (CMH13) [MIM:613243]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:11385718, ECO:0000269|PubMed:18572189}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 14, 613251 (MYH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 14, 613251 (MYH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH6	MYH6, ASD3, MYHCA, CMD1EE, CMH14, SSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 14 (CMH14) [MIM:613251]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:11815426, ECO:0000269|PubMed:15998695}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 15, 613255 (Aileseİ Izole Dilate Kardiyomiyopati) (VCL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 15, 613255 (VCL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCL	VCL, CMD1W, CMH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kardiyomiyopati, Hipertrofik, 16, 613838, Otozomal Dominant (MYOZ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 16, 613838, Autosomal dominant (MYOZ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYOZ2	MYOZ2, CMH16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 17, 613873, Otozomal Dominant (JPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 17, 613873, Autosomal dominant (JPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JPH2	JPH2, JP2, CMH17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 17 (CMH17) [MIM:613873]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269	PubMed:17509613}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 18, 613874, Otozomal Dominant (Allesel Izole Dilate Kardiyomiyopati) (PLN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 18, 613874, Autosomal dominant (PLN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLN	PLN, PLB, CMD1P, CMH18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 19, 613875, Otozomal Dominant (CALR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 19, 613875, Autosomal dominant (CALR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALR3	CALR3, CRT2, CMH19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 19 (CMH19) [MIM:613875]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269	PubMed:17655857}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 2, 115195, Otozomal Dominant (Allesel Izole Dilate Kardiyomiyopati) (TNNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 2, 115195, Autosomal dominant (TNNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT2	TNNT2, CMH2, CMD1D, RCM3, LVNC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 2 (CMH2) [MIM:115195]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:10525521, ECO:0000269|PubMed:11034944, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:15563892, ECO:0000269|PubMed:16199542, ECO:0000269|PubMed:21846512, ECO:0000269|PubMed:7898523, ECO:0000269|PubMed:8205619, ECO:0000269|PubMed:8989109, ECO:0000269|PubMed:9060892, ECO:0000269|PubMed:9140840, ECO:0000269|PubMed:9482583, ECO:0000269|Ref.19}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 20, 613876, Otozomal Dominant (Allesel Izole Dilate Kardiyomiyopati) (NEXN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 20, 613876, Autosomal dominant (NEXN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEXN	NEXN, NELIN, CMD1CC, CMH20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 20 (CMH20) [MIM:613876]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:20970104}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 22, 615248, Otozomal Dominant (Allesel Izole Restriktif Kardiyomiyopati) (MYPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 22, 615248, Autosomal dominant (Familial isolated restrictive cardiomyopathy) (MYPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYPN	MYPN, CMD1DD, CMH22, RCM4, NEM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 22 (CMH22) [MIM:615248]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:22286171}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 23, ± LVNC (Sol Ventrikül Nonkompaksiyon), 612158, Otozomal Dominant (ACTN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 23, with or without LVNC, 612158, Autosomal dominant (ACTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN2	ACTN2, CMD1AA, CMH23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 23, with or without left ventricular non-compaction (CMH23) [MIM:612158]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:20022194, ECO:0000269|PubMed:25173926}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 24, 601493, Otozomal Dominant (LDB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 24, 601493, Autosomal dominant (LDB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDB3	LDB3, ZASP, CYPHER, KIAA0613, MFM4, CMD1C, CMH24, LVNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 3 (LVNC3) [MIM:601493]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik 25, 607487, Otozomal Dominant (TCAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 25, 607487, Autosomal dominant (TCAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCAP	TCAP, LGMD2G, CMH25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 3, 115196, Otozomal Dominant (TPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 3, 115196, Autosomal dominant (TPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM1	TPM1, CMH3, CMD1Y , LVNC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 3 (CMH3) [MIM:115196]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:7898523, ECO:0000269|PubMed:8205619, ECO:0000269|PubMed:8523464, ECO:0000269|PubMed:9822100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 4, 115197, Otozomal Dominant (MYBPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 4, 115197, Autosomal dominant (MYBPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYBPC3	MYBPC3, CMH4, CMD1MM, LVNC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 4 (CMH4) [MIM:115197]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:11499718, ECO:0000269|PubMed:11499719, ECO:0000269|PubMed:11815426, ECO:0000269|PubMed:12379228, ECO:0000269|PubMed:12628722, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:12818575, ECO:0000269|PubMed:12951062, ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:14563344, ECO:0000269|PubMed:15114369, ECO:0000269|PubMed:15519027, ECO:0000269|PubMed:15563892, ECO:0000269|PubMed:16004897, ECO:0000269|PubMed:16199542, ECO:0000269|PubMed:18403758, ECO:0000269|PubMed:18929575, ECO:0000269|PubMed:18957093, ECO:0000269|PubMed:23840593, ECO:0000269|PubMed:7744002, ECO:0000269|PubMed:9048864, ECO:0000269|PubMed:9541104, ECO:0000269|PubMed:9541115, ECO:0000269|PubMed:9562578}. Note=The disease is caused	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 7, 613690, Otozomal Dominant (TNNI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 7, 613690, Autosomal dominant (TNNI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNI3	TNNI3, CMH7, CMD2A, RCM1, CMD1FF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyomiyopati, Hipertrofik, 8, 608751 (MYL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiomyopathy, hypertrophic, 8, 608751 (MYL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYL3	MYL3, CMH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 8 (CMH8) [MIM:608751]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. Rarely, patients present a variant of familial hypertrophic cardiomyopathy, characterized by mid-left ventricular chamber thickening. {ECO:0000269|PubMed:12021217, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:23594557, ECO:0000269|PubMed:8673105}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kardiyospondilokarpofasiyal Sendrom, 157800, Otozomal Dominant; CSCF (Kardiyospondilokarpofasiyal Sendromu) (MAP3K7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cardiospondylocarpofacial syndrome, 157800, Autosomal dominant; CSCF (Cardiospondylocarpofacial syndrome) (MAP3K7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP3K7	MAP3K7, TAK1, CSCF, FMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kardiyospondilokarpofasiyal Sendrom, 157800, Otozomal Dominant; CSCF (Kardiyospondilokarpofasiyal Sendromu) (Prenatal) (MAP3K7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cardiospondylocarpofacial syndrome, 157800, Autosomal dominant; CSCF (Cardiospondylocarpofacial syndrome) (MAP3K7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP3K7	MAP3K7, TAK1, CSCF, FMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Carney Kompleks Varyantı, 608837 (Carney Kompleks Trisma-Psödokamptodaktili Sendromu) (MYH8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carney complex variant, 608837 (Carney complex-trismus-pseudocamptodactyly syndrome) (MYH8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH8	MYH8, DA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Carney Kompleks Varyantı, 608837 (Carney Kompleks Trismus-Pseudocamptodaktili Sendromu) (Prenatal) (MYHB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carney complex variant, 608837 (Carney complex-trismus-pseudocamptodactyly syndrome) (MYHB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYHB	MYHB, DA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Carney Kompleksi, Tip 1, 160980, Otozomal Dominant; CNC1 (Carney Kompleksi) (PRKAR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carney complex, type 1, 160980, Autosomal dominant; CNC1 (Carney complex) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Carney Kompleksi, Tip 1, 160980, Otozomal Dominant; CNC1 (Carney Kompleksi) (PRKAR1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carney complex, type 1, 160980, Autosomal dominant; CNC1 (Carney complex) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karnitin Asetiltransferaz Eksikliği (CRAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carnitine acetyltransferase deficiency (CRAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRAT	CRAT, CAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Karnitin Asetiltransferaz Eksikliği (Prenatal) (CRAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carnitine acetyltransferase deficiency (CRAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRAT	CRAT, CAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karnitin Eksikliği, Sistemik Primer, 212140, Otozomal Resesif; CDSP (Sistemik Primer Karnitin Eksikliği) (SLC22A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carnitine deficiency, systemic primary, 212140, Autosomal recessive; CDSP (Systemic primary carnitine deficiency) (SLC22A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A5	SLC22A5, OCTN2, CDSP, SCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Karnitin Eksikliği, Sistemik Primer, 212140, Otozomal Resesif; CDSP (Sistemik Primer Karnitin Eksikliği) (Prenatal) (SLC22A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carnitine deficiency, systemic primary, 212140, Autosomal recessive; CDSP (Systemic primary carnitine deficiency) (SLC22A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC22A5	SLC22A5, OCTN2, CDSP, SCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karnitin Palmitoil Transferaz 2 Eksikliği, Letal Neonatal (Karnitin Palmitoil Transferaz 2 Eksikliği) (CPT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Karnitin Palmitoil Transferaz 2 Eksikliği, Letal Neonatal (Karnitin Palmitoil Transferaz 2 Eksikliği) (Prenatal) (CPT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, LETHAL NEONATAL (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karnitin Palmitoil Transferaz 2 Eksikliği, Miyopatik, Strese Bağlı (Karnitin Palmitoil Transferaz 2 Eksikliği) (CPT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Karnitin Palmitoil Transferaz 2 Eksikliği, Miyopatik, Strese Bağlı (Karnitin Palmitoil Transferaz 2 Eksikliği) (Prenatal) (CPT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CARNITINE PALMITOYLTRANSFERASE II DEFICIENCY, MYOPATHIC, STRESS-INDUCED (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karnitin-Açilkarnitin Translokaz Eksikliği, 212138, Otozomal Resesif; CACTD (Karnitin-Açilkarnitin Translokaz Eksikliği) (SLC25A20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carnitine-acylcarnitine translocase deficiency, 212138, Autosomal recessive; CACTD (Carnitine-acylcarnitine translocase deficiency) (SLC25A20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A20	SLC25A20, CACT, CAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carnitine-acylcarnitine translocase deficiency (CACTD) [MIM:212138]: A rare long-chain fatty acid oxidation disorder. Metabolic consequences include hypoketotic hypoglycemia under fasting conditions, hyperammonemia, elevated creatine kinase and transaminases, dicarboxylic aciduria, very low free carnitine and abnormal acylcarnitine profile with marked elevation of the long- chain acylcarnitines. Clinical features include neurologic abnormalities, cardiomyopathy, arrhythmias, skeletal muscle damage, liver dysfunction and episodes of life-threatening coma, which eventually lead to death. Most patients become symptomatic in the neonatal period with a rapidly progressive deterioration and a high mortality rate. {ECO:0000269	PubMed:12859414, ECO:0000269	PubMed:15057979, ECO:0000269	PubMed:15365988}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Karnitin-Açilkarnitin Translokaz Eksikliği, 212138, Otozomal Resesif; CACTD (Karnitin-Açilkarnitin Translokaz Eksikliği) (Prenatal) (SLC25A20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carnitine-acylcarnitine translocase deficiency, 212138, Autosomal recessive; CACTD (Carnitine-acylcarnitine translocase deficiency) (SLC25A20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A20	SLC25A20, CACT, CAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carnitine-acylcarnitine translocase deficiency (CACTD) [MIM:212138]: A rare long-chain fatty acid oxidation disorder. Metabolic consequences include hypoketotic hypoglycemia under fasting conditions, hyperammonemia, elevated creatine kinase and transaminases, dicarboxylic aciduria, very low free carnitine and abnormal acylcarnitine profile with marked elevation of the long- chain acylcarnitines. Clinical features include neurologic abnormalities, cardiomyopathy, arrhythmias, skeletal muscle damage, liver dysfunction and episodes of life-threatening coma, which eventually lead to death. Most patients become symptomatic in the neonatal period with a rapidly progressive deterioration and a high mortality rate. {ECO:0000269	PubMed:12859414, ECO:0000269	PubMed:15057979, ECO:0000269	PubMed:15365988}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karotis İntimal Medial Kalınlığı 1, 609338 (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carotid intimal medial thickness 1, 609338 (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CIMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Karpal Tünel Sendromu, Ailesel, 115430, Otozomal Dominant; CTS1 (TTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carpal tunnel syndrome, familial, 115430, Autosomal dominant; CTS1 (TTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTR	TTR, PALB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Carpenter Sendromu 2, 614976, Otozomal Resesif; CRPT2 (Carpenter Sendromu) (MEGF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carpenter syndrome 2, 614976, Autosomal recessive; CRPT2 (Carpenter syndrome) (MEGF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEGF8	MEGF8, EGFL4, CRPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carpenter syndrome 2 (CRPT2) [MIM:614976]: An autosomal recessive multiple congenital malformation disorder characterized by multisuture craniosynostosis and polysyndactyly of the hands and feet, in association with abnormal left-right patterning and other features, most commonly obesity, umbilical hernia, cryptorchidism, and congenital heart disease. {ECO:0000269	PubMed:23063620}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Carpenter Sendromu 2, 614976, Otozomal Resesif; CRPT2 (Carpenter Sendromu) (Prenatal) (MEGF8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carpenter syndrome 2, 614976, Autosomal recessive; CRPT2 (Carpenter syndrome) (MEGF8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MEGF8	MEGF8, EGFL4, CRPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carpenter syndrome 2 (CRPT2) [MIM:614976]: An autosomal recessive multiple congenital malformation disorder characterized by multisuture craniosynostosis and polysyndactyly of the hands and feet, in association with abnormal left-right patterning and other features, most commonly obesity, umbilical hernia, cryptorchidism, and congenital heart disease. (ECO:0000269	PubMed:23063620). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Carpenter Sendromu, 201000, Otozomal Resesif; CRPT1 (Carpenter Sendromu) (RAB23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Carpenter syndrome, 201000, Autosomal recessive; CRPT1 (Carpenter syndrome) (RAB23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB23	RAB23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carpenter syndrome 1 (CRPT1) [MIM:201000]: A rare autosomal recessive disorder characterized by acrocephaly with variable synostosis of the sagittal, lambdoid, and coronal sutures; peculiar facies; brachydactyly of the hands with syndactyly; preaxial polydactyly and syndactyly of the feet congenital heart defects; growth retardation; mental retardation; hypogenitalism; and obesity. In addition, cerebral malformations, oral and dental abnormalities, coxa valga, genu valgum, hydronephrosis, precocious puberty, and hearing loss may be observed. (ECO:0000269|PubMed:17503333, ECO:0000269|PubMed:21412941). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Carpenter Sendromu, 201000, Otozomal Resesif; CRPT1 (Carpenter Sendromu) (Prenatal) (RAB23 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Carpenter syndrome, 201000, Autosomal recessive; CRPT1 (Carpenter syndrome) (RAB23 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB23	RAB23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carpenter syndrome 1 (CRPT1) [MIM:201000]: A rare autosomal recessive disorder characterized by acrocephaly with variable synostosis of the sagittal, lambdoid, and coronal sutures; peculiar facies; brachydactyly of the hands with syndactyly; preaxial polydactyly and syndactyly of the feet congenital heart defects; growth retardation; mental retardation; hypogenitalism; and obesity. In addition, cerebral malformations, oral and dental abnormalities, coxa valga, genu valgum, hydronephrosis, precocious puberty, and hearing loss may be observed. (ECO:0000269|PubMed:17503333, ECO:0000269|PubMed:21412941). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kıkırdak Saç Hipoplazisi, 250250, Otozomal Resesif; CHH (Kıkırdak Saç Hipoplazisi) (RMRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cartilage-hair hypoplasia, 250250, Autosomal recessive; CHH (Cartilage-hair hypoplasia) (RMRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kıkırdak Saç Hipoplazisi, 250250, Otozomal Resesif; CHH (Kıkırdak Saç Hipoplazisi) (Prenatal) (RMRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cartilage-hair hypoplasia, 250250, Autosomal recessive; CHH (Cartilage-hair hypoplasia) (RMRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Katarakt 1, Çeşitli Tipler, 116200, Otozomal Dominant; CTRCT1 (Katarakt-Mikrokornea Sendromu) (GJA8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 1, multiple types, 116200, Autosomal dominant; CTRCT1 (Cataract-microcornea syndrome) (GJA8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA8	GJA8, CX50, CTRCT1, CZP1, CAE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Katarakt 10, Çeşitli Tipler, 600881, Otozomal Dominant; CTRCT10 (Early-Onset Lamellar Cataract) (CRYBA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 10, multiple types, 600881, Autosomal dominant; CTRCT10 (Early-onset lamellar cataract) (CRYBA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYBA1	CRYBA1, CRYB1, CTRCT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 10, multiple types (CTRCT10) [MIM:600881]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT10 includes congenital zonular with sutural opacities, among others. This is a form of zonular cataract with an erect Y-shaped anterior and an inverted Y-shaped posterior sutural opacities. Zonular or lamellar cataracts are opacities, broad or narrow, usually consisting of powdery white dots affecting only certain layers or zones between the cortex and nucleus of an otherwise clear lens. The opacity may be so dense as to render the entire central region of the lens completely opaque, or so translucent that vision is hardly if at all impeded. Zonular cataracts generally do not involve the embryonic nucleus, though sometimes they involve the fetal nucleus. Usually sharply separated from a clear cortex outside them, they may have projections from their outer edges known as riders or spokes. (ECO:0000269|PubMed:15016766, ECO:0000269|PubMed:9788845). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 11, Çeşitli Tipler, 610623, Otozomal Dominant; CTRCT11 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (PITX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 11, multiple types, 610623, Autosomal dominant; CTRCT11 (Early-onset non-syndromic cataract) (PITX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX3	PITX3, CTPP4, CTRCT11, ASGD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 11, multiple types (CTRCT11) [MIM:610623]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT11 includes posterior polar cataract, among others. Posterior polar cataract is a subcapsular opacity, usually disk-shaped, located at the back of the lens. Some CTRCT11 patients can present a severe phenotype including microphthalmia and neurological dysfunction. (ECO:0000269|PubMed:15286169, ECO:0000269|PubMed:9620774). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 12, Çeşitli Tipler, 611597, Otozomal Dominant; CTRCT12 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (BFSP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 12, multiple types, 611597, Autosomal dominant; CTRCT12 (Early-onset non-syndromic cataract) (BFSP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BFSP2	BFSP2, CP49, CP47, CTRCT12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 13, Yetişkin 1 Fenotipi, 116700, Otozomal Resesif; CTRCT13 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (GCNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 13 with adult i phenotype, 116700, Autosomal recessive; CTRCT13 (Early-onset non-syndromic cataract) (GCNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCNT2	GCNT2, Ii, CTRCT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 13, with adult i phenotype (CTRCT13) [MIM:116700]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT13 is associated with the rare adult i phenotype, in which adult red blood cells are rich in i antigen and contain low levels of I antigen. [ECO:0000269	PubMed:11739194). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 14, Çeşitli Tipler, 601885, Otozomal Dominant; CTRCT14 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (GJA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 14, multiple types, 601885, Autosomal dominant; CTRCT14 (Early-onset non-syndromic cataract) (GJA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA3	GJA3, CX46, CZP3, CAE3, CTRCT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 14, multiple types (CTRCT14) [MIM:601885]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT14 includes zonular pulverulent cataract, among others. Zonular or lamellar cataracts are opacities, broad or narrow, usually consisting of powdery white dots affecting only certain layers or zones between the cortex and nucleus of an otherwise clear lens. The opacity may be so dense as to render the entire central region of the lens completely opaque, or so translucent that vision is hardly if at all impeded. Usually sharply separated from a clear cortex outside them, they may have projections from their outer edges known as riders or spokes. (ECO:0000269|PubMed:10205266, ECO:0000269|PubMed:10746562, ECO:0000269|PubMed:14627959, ECO:0000269|PubMed:15208569, ECO:0000269|PubMed:15286166, ECO:0000269|PubMed:15448617, ECO:0000269|PubMed:16234473, ECO:0000269|PubMed:16254549, ECO:0000269|PubMed:16885921, ECO:0000269|PubMed:16971895, ECO:0000269|PubMed:17615540, ECO:0000269|PubMed:17893874, ECO:0000269|PubMed:20431721, ECO:0000269|PubMed:21552498, ECO:0000269|PubMed:21647269, ECO:0000269|PubMed:21681855, ECO:0000269|PubMed:21897748, ECO:0000269|PubMed:22312188, ECO:0000269|PubMed:24722942	EDTA Blood Sample
Katarakt 15, Çeşitli Tipler, 615274; CTRCT15 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (MIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 15, multiple types, 615274; CTRCT15 (Early-onset non-syndromic cataract) (MIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIP	MIP, AQP0, CTRCT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 15, multiple types (CTRCT15) [MIM:615274]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT15 includes polymorphic, progressive punctate lamellar, cortical, anterior and posterior polar, nonprogressive lamellar with sutural opacities, embryonic nuclear, and pulverulent cortical, among others. (ECO:0000269|PubMed:10802646, ECO:0000269|PubMed:11001937, ECO:0000269|PubMed:16564824, ECO:0000269|PubMed:17893667, ECO:0000269|PubMed:17960133, ECO:0000269|PubMed:20361015, ECO:0000269|PubMed:21245956). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 16, Çeşitli Tipler, 613763, Otozomal Resesif, Otozomal Dominant; CTRCT16 (Early-Onset Lamellar Cataract) (CRYAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 16, multiple types, 613763, Autosomal recessive, Autosomal dominant; CTRCT16 (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1B, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 17, Çeşitli Tipler, 611544; CTRCT17 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (CRYBB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 17, multiple types, 611544; CTRCT17 (Early-onset non-syndromic cataract) (CRYBB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYBB1	CRYBB1, CATCN3, CTRCT17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 17, multiple types (CTRCT17) [MIM:611544]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT17 includes nuclear and pulverulent cataracts, among others. Nuclear cataracts affect the central nucleus of the eye, are often not highly visually significant. The density of the opacities varies greatly from fine dots to a dense, white and chalk-like, central cataract. The condition is usually bilateral. Nuclear cataracts are often combined with opacified cortical fibers encircling the nuclear opacity, which are referred to as cortical riders. Pulverulent cataracts are characterized by a dust-like, 'pulverised' appearance of the opacities which can be found in any part of the lens. (ECO:0000269|PubMed:12360425, ECO:0000269|PubMed:17460281, ECO:0000269|PubMed:23508780). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Katarakt 18, Otozomal Resesif, 610019, Otozomal Resesif, CTRCT18 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (FYCO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 18, autosomal recessive, 610019, Autosomal recessive; CTRCT18 (Early-onset non-syndromic cataract) (FYCO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FYCO1	FYCO1, CATC2, CTRCT18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 19, Çeşitli Tipler, 615277, Otozomal Resesif; CTRCT19 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (LIM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 19, multiple types, 615277, Autosomal recessive; CTRCT19 (Early-onset non-syndromic cataract) (LIM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIM2	LIM2, MP19, CTRCT19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Katarakt 2, Çeşitli Tipler, 604307, Otozomal Dominant; CTRCT2 (Early-Onset Lamellar Cataract) (CRYGC gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 2, multiple types, 604307, Autosomal dominant; CTRCT2 (Early-onset lamellar cataract) (CRYGC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYGC	CRYGC, CRYG3, CTRCT2, CCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 2, multiple types (CTRCT2) [MIM:604307]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT2 includes Coppock-like cataract, among others. Coppock-like cataract is a congenital pulverulent disk-like opacity involving the embryonic nucleus with many tiny white dots in the lamellar portion of the lens. It is usually bilateral and dominantly inherited. In some cases, CTRCT2 is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. {ECO:0000269|PubMed:10521291, ECO:0000269|PubMed:10914683, ECO:0000269|PubMed:12011157, ECO:0000269|PubMed:22052681, ECO:0000269|PubMed:22876111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 20, Çeşitli Tipler, 116100, Otozomal Dominant; CTRCT20 (Erken Başlangıçlı Sendromik Olmayan Katarakt9 (CRYGS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 20, multiple types, 116100, Autosomal dominant; CTRCT20 (Early-onset non-syndromic cataract) (CRYGS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYGS	CRYGS, CRYG8, CTRCT20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 20, multiple types (CTRCT20) [MIM:116100]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT20 includes progressive polymorphic anterior, posterior, or peripheral cortical. {ECO:0000269	PubMed:16141006}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 21, Çeşitli Tipler, 610202, Otozomal Dominant; CTRCT21 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (MAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 21, multiple types, 610202, Autosomal dominant; CTRCT21 (Early-onset non-syndromic cataract) (MAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAF	MAF, CCA4, CTRCT21, AYGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 22, 609741, Otozomal Resesif, Otozomal Dominant; CTRCT22 (Early-Onset Lamellar Cataract) (CRYBB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 22, 609741, Autosomal recessive, Autosomal dominant; CTRCT22 (Early-onset lamellar cataract) (CRYBB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYBB3	CRYBB3, CRYB3, CATCN2, CTRCT22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 22, multiple types (CTRCT22) [MIM:609741]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT22 includes nuclear cataract among others. Nuclear cataracts affect the central nucleus of the eye, and are often not highly visually significant. The density of the opacities varies greatly from fine dots to a dense, white and chalk-like, central cataract. The condition is usually bilateral. Nuclear cataracts are often combined with opacified cortical fibers encircling the nuclear opacity, which are referred to as cortical riders. {ECO:0000269|PubMed:15914629, ECO:0000269|PubMed:23500780}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 23, 610425, CTRCT23 (Early-Onset Lamellar Cataract) (CRYBA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 23, 610425, CTRCT23 (Early-onset lamellar cataract) (CRYBA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYBA4	CRYBA4, CTRCT23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 23 (CTRCT23) [MIM:610425]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT23 is a zonular cataract. Zonular or lamellar cataracts are opacities, broad or narrow, usually consisting of powdery white dots affecting only certain layers or zones between the cortex and nucleus of an otherwise clear lens. The opacity may be so dense as to render the entire central region of the lens completely opaque, or so translucent that vision is hardly if at all impeded. Zonular cataracts generally do not involve the embryonic nucleus, though sometimes they involve the fetal nucleus. Usually sharply separated from a clear cortex outside them, they may have projections from their outer edges known as riders or spokes. {ECO:0000269	PubMed:16960806}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 30, Pulverulen, 116300, Otozomal Dominant; CTRCT30 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (VIM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 30, pulverulent, 116300, Autosomal dominant; CTRCT30 (Early-onset non-syndromic cataract) (VIM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VIM	VIM, CTRCT30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 30 (CTRCT30) [MIM:116300]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 31, Çeşitli Tipler, 605387; CTRCT31 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (CHMP4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 31, multiple types, 605387; CTRCT31 (Early-onset non-syndromic cataract) (CHMP4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHMP4B	CHMP4B, SNF7, CTPP3, CTRCT31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 31, multiple types (CTRCT31) [MIM:605387]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT31 includes posterior polar, progressive posterior subcapsular, nuclear, and anterior subcapsular cataracts. {ECO:0000269	PubMed:17701905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 33, 611391; CTRCT33 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (BFSP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 33, 611391; CTRCT33 (Early-onset non-syndromic cataract) (BFSP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BFSP1	BFSP1, CP115, CTRCT33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Katarakt 34, Çeşitli Tipler, 612968; CTRCT34 (Peters Anomalisi) (FOXE3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 34, multiple types, 612968; CTRCT34 (Peters anomaly) (FOXE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXE3	FOXE3, FKHL12, ASMD, CTRCT34, ASGD2, AAT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 36, 613887; CTRCT36 (TDRD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 36, 613887; CTRCT36 (TDRD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TDRD7	TDRD7, KIAA1529, TRAP, CATC4, CTRCT36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 36 (CTRCT36) [MIM:613887]: An opacification of the crystalline lens of the eye becoming evident at birth. It frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269	PubMed:21436445}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 38, Otozomal Resesif, 614691, Otozomal Resesif; CTRCT38 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (AGK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 38, autosomal recessive, 614691, Autosomal recessive; CTRCT38 (Early-onset non-syndromic cataract) (AGK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGK	AGK, MULK, MTDPS10, CATC5, CTRCT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 38 (CTRCT38) [MIM:614691]: An opacification of the crystalline lens of the eye becoming evident at birth. It frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269	PubMed:22415731}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 39, Çeşitli Tipler, Otozomal Dominant, 615188, Otozomal Dominant; CTRCT39 (Early-Onset Lamellar Cataract) (CRYGB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 39, multiple types, autosomal dominant, 615188, Autosomal dominant; CTRCT39 (Early-onset lamellar cataract) (CRYGB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYGB	CRYGB, CRYG2, CTRCT39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 39, multiple types (CTRCT39) [MIM:615188]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT39 includes lamellar, anterior polar, and complete cataracts. {ECO:0000269	PubMed:23288985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 4, Çeşitli Tipler, 115700, Otozomal Dominant; CTRCT4 (Katarakt-Mikrokornea Sendromu) (CRYGD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 4, multiple types, 115700, Autosomal dominant; CTRCT4 (Cataract-microcornea syndrome) (CRYGD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYGD	CRYGD, CRYG4, CTRCT4, CACA, CCA3, PCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 4, multiple types (CTRCT4) [MIM:115700]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT4 includes crystalline aculeiform, congenital cerulean and non-nuclear polymorphic cataracts, among others. Crystalline aculeiform cataract is characterized by fiberglass-like or needle-like crystals projecting in different directions, through or close to the axial region of the lens. Non-nuclear polymorphic cataract is a partial opacity with variable location between the fetal nucleus of the lens and the equator. The fetal nucleus is normal. The opacities are irregular and look similar to a bunch of grapes and may be present simultaneously in different lens layers. Congenital cerulean cataract is characterized by peripheral bluish and white opacifications organized in concentric layers with occasional central lesions arranged radially. The opacities are observed in the superficial layers of the fetal nucleus as well as the adult nucleus of the lens. Involvement is usually bilateral. Visual acuity is only mildly reduced in childhood. In adulthood, the opacifications may progress, making lens extraction necessary. Histologically the lesions are described as fusiform cavities between lens fibers which contain a deeply staining granular material. Although the lesions may take on various colors, a dull blue is the most common appearance and is responsible for the designation cerulean cataract. {ECO:0000269|PubMed:10521291, ECO:0000269|PubMed:10915766, ECO:0000269|PubMed:12011157, ECO:0000269|PubMed:12676897, ECO:0000269|PubMed:16943771, ECO:0000269|PubMed:17564961, ECO:0000269|PubMed:21051598	EDTA Blood Sample

Katarakt 40, X'e Bağlı, 302200, X'e Bağlı; CTRCT40 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (NHS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 40, X-linked, 302200, X-linked, CTRCT40 (Early-onset non-syndromic cataract) (NHS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NHS	NHS, CXN, CTRCT40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 40 (CTRCT40) [MIM:302200]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT40 manifests as a congenital nuclear opacity with severe visual impairment in affected males. Heterozygous females have suture cataracts and only slight reduction in vision. In some cases, cataract is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. {ECO:0000269	PubMed:19414485}. Note=The disease is caused by mutations affecting the gene represented in this entry. Caused by copy number variations predicted to result in altered transcriptional regulation of the NHS gene: a 0.8 Mb segmental duplication-triplication encompassing the NHS, SCML1 and RAI2 genes, and an 4.8 kb intragenic deletion in NHS intron 1.;	EDTA Blood Sample
Katarakt 41, 116400, Otozomal Dominant; CTRCT41 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (WFS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 41, 116400, Autosomal dominant; CTRCT41 (Early-onset non-syndromic cataract) (WFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 41 (CTRCT41) [MIM:116400]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. {ECO:0000269	PubMed:23531866}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 41, 116400, Otozomal Dominant; CTRCT41 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (MLPA)	Cataract 41, 116400, Autosomal dominant; CTRCT41 (Early-onset non-syndromic cataract) (MLPA)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cataract 41 (CTRCT41) [MIM:116400]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. {ECO:0000269	PubMed:23531866}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 42, 115900, Otozomal Dominant; CTRCT42 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (CRYBA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 42, 115900, Autosomal dominant; CTRCT42 (Early-onset non-syndromic cataract) (CRYBA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYBA2	CRYBA2, CTRCT42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 42 (CTRCT42) [MIM:115900]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269	PubMed:23508780}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 43, 616279, Otozomal Dominant; CTRCT43 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (UNC45B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 43, 616279, Autosomal dominant; CTRCT43 (Early-onset non-syndromic cataract) (UNC45B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNC45B	UNC45B, SMUNC45, CTRCT43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 43 (CTRCT43) [MIM:616279]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269	PubMed:24549050}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 44, 616509, Otozomal Resesif; CTRCT44 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (LSS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 44, 616509, Autosomal recessive; CTRCT44 (Early-onset non-syndromic cataract) (LSS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LSS	LSS, OSC, CTRCT44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 45, 616851, Otozomal Resesif; CTRCT45 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (SIPA1L3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 45, 616851, Autosomal recessive; CTRCT45 (Early-onset non-syndromic cataract) (SIPA1L3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIPA1L3	SIPA1L3, SPAL3, SPAR3, KIAA0545, CTRCT45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 46, Jüvenil Başlangıçlı, 212500, Otozomal Resesif; CTRCT46 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (LEMD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 46, juvenile-onset, 212500, Autosomal recessive; CTRCT46 (Early-onset non-syndromic cataract) (LEMD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEMD2	LEMD2, NET25, CTRCT42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 47, Jüvenil, Mikrokornea, 612018, Otozomal Dominant; CTRCT47 (Jüvenil Katarakt-Mikrokornea Renal Glikozüri Sendromu) (SLC16A12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 47, juvenile, with microcornea, 612018, Autosomal dominant; CTRCT47 (Juvenile cataract-microcornea-renal glucosuria syndrome) (SLC16A12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC16A12	SLC16A12, MCT12, CTRCT47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt 5, Çeşitli Tipler, 116800, Otozomal Dominant; CTRCT5 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (HSF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 5, multiple types, 116800, Autosomal dominant; CTRCT5 (Early-onset non-syndromic cataract) (HSF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSF4	HSF4, CTM, CTRCT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 5, multiple types (CTRCT5) [MIM:116800]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT5 includes infantile, lamellar, zonular, nuclear, anterior polar, stellate, and Marner-type cataracts, among others. Finger malformation is observed in some kindreds. {ECO:0000269|PubMed:12089525, ECO:0000269|PubMed:16878512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Katarakt 6, Çeşitli Tipler, 116600, Otozomal Dominant; CTRCT6 (Erken Başlangıçlı Sendromik Olmayan Katarakt) (EPHA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 6, multiple types, 116600, Autosomal dominant; CTRCT6 (Early-onset non-syndromic cataract) (EPHA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHA2	EPHA2, ECK, ARCC2, CTPP1, CTPA, ARCC2, CTRCT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 6, multiple types (CTRCT6) [MIM:116600]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT6 includes posterior polar and age- related cortical cataracts, among others. Posterior polar cataract is a subcapsular opacity, usually disk-shaped, located at the back of the lens. Age-related cortical cataract is a developmental punctate opacity restricted to the cortex. The cataract is white or cerulean, increases in number with age, but rarely affects vision. {ECO:0000269|PubMed:19005574, ECO:0000269|PubMed:19306328, ECO:0000269|PubMed:19649315}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Katarakt 9, Çeşitli Tipler, 604219, Otozomal Dominant; CTRCT9 (Katarakt-Mikrokornea Sendromu) (CRYAA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract 9, multiple types, 604219, Autosomal dominant; CTRCT9 (Katarakt-mikrokornea sendromu) (CRYAA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYAA	CRYAA, CRYA1, CTRCT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 9, multiple types (CTRCT9) [MIM:604219]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. CTRCT9 includes nuclear, zonular central nuclear, anterior polar, cortical, embryonal, anterior subcapsular, fan-shaped, and total cataracts, among others. In some cases cataract is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. {ECO:0000269|PubMed:14512969, ECO:0000269|PubMed:16453125, ECO:0000269|PubMed:18302245, ECO:0000269|PubMed:19407550, ECO:0000269|PubMed:23508780, ECO:0000269|PubMed:9467006}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kataraklt-Geç Başlangıçlı Kornea Distrofisi, 106210, Otozomal Dominant (İzole Aniridi) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract with late-onset corneal dystrophy, 106210, Autosomal dominant (isolated aniridia) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kataraklt-Geç Başlangıçlı Kornea Distrofisi, 106210, Otozomal Dominant (İzole Aniridi) (MLPA)	Cataract with late-onset corneal dystrophy, 106210, Autosomal dominant (isolated aniridia) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Katarakt, Konjenital, 182500 (SORD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataract, congenital, 182500 (SORD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SORD	SORD, SORD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Katarakt, Büyüme Hormonu Eksikliği, Duyusal Nöropati, Sensorinöral İşitme Kaybı ve İskelet Displazisi, 616007, Otozomal Resesif; CAGSSS (Katarakt-Büyüme Hormonu Eksikliği-Duyusal Nöropati-Sensorinöral İşitme Kaybı-İskelet Displazisi Sendromu) (IARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia, 616007, Autosomal recessive; CAGSSS (Cataract-growth hormone deficiency-sensory neuropathy-sensorineural hearing loss-skeletal dysplasia syndrome) (IARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IARS2	IARS2, CAGSSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataracts, growth hormone deficiency, sensory neuropathy, sensorineural hearing loss, and skeletal dysplasia (CAGSSS) [MIM:616007]: An autosomal recessive disorder characterized by cataracts, short-stature secondary to growth hormone deficiency, sensorineural hearing deficit, peripheral sensory neuropathy, skeletal dysplasia, scoliosis, and facial dysmorphism. {ECO:0000269	PubMed:25130867}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Catel-Manzke Sendromu, 616145, Otozomal Resesif; CATMANS (Catel-Manzke Sendromu) (TGDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Catel-Manzke syndrome, 616145, Autosomal recessive; CATMANS (Catel-Manzke syndrome) (TGDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGDS	TGDS, SDR2E1, CATMANS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Catel-Manzke syndrome (CATMANS) [MIM:616145]: A syndrome characterized by Pierre Robin sequence and a unique form of bilateral hyperphalangy causing a clinodactyly of the index finger. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). [ECO:0000269	PubMed:25480037]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Catel-Manzke Sendromu, 616145, Otozomal Resesif; CATMANS (Catel-Manzke Sendromu) (Prenatal) (TGDS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Catel-Manzke syndrome, 616145, Autosomal recessive; CATMANS (Catel-Manzke syndrome) (TGDS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGDS	TGDS, SDR2E1, CATMANS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Catel-Manzke syndrome (CATMANS) [MIM:616145]: A syndrome characterized by Pierre Robin sequence and a unique form of bilateral hyperphalangy causing a clinodactyly of the index finger. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). [ECO:0000269	PubMed:25480037]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CATSHL Sendromu, 610474, Otozomal Resesif, Otozomal Dominant; CATSHLS (Kamptodaktili-Uzun Boy-Skolyoz-İşitme Kaybı Sendromu) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CATSHL syndrome, 610474, Autosomal recessive, Autosomal dominant; CATSHLS (Camptodactyly-tall stature-scoliosis-hearing loss syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSHL Sendromu, 610474, Otozomal Resesif, Otozomal Dominant; CATSHLS (Kamptodaktili-Uzun Boy-Skolyoz-İşitme Kaybı Sendromu) (MLPA)	CATSHL syndrome, 610474, Autosomal recessive, Autosomal dominant; CATSHLS (Camptodactyly-tall stature-scoliosis-hearing loss syndrome) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CATSHL Sendromu, 610474, Otozomal Resesif, Otozomal Dominant; CATSHLS (Kamptodaktili-Uzun Boy-Skolyoz-İşitme Kaybı Sendromu) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CATSHL syndrome, 610474, Autosomal recessive, Autosomal dominant; CATSHLS (Camptodactyly-tall stature-scoliosis-hearing loss syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CATSHL Sendromu, 610474, Otozomal Resesif, Otozomal Dominant; CATSHLS (Kamptodaktili-Uzun Boy-Skolyoz-İşitme Kaybı Sendromu) (Prenatal) (MLPA)	CATSHL syndrome, 610474, Autosomal recessive, Autosomal dominant; CATSHLS (Camptodactyly-tall stature-scoliosis-hearing loss syndrome) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kaudal Duplikasyon Anomalisi, 607864 (Kaudal Duplikasyon) (AXIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Caudal duplication anomaly, 607864 (Caudal duplication) (AXIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AXIN1	AXIN1, AXIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kaudal Duplikasyon Anomalisi, 607864 (Kaudal Duplikasyon) (Prenatal) (AXIN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Caudal duplication anomaly, 607864 (Caudal duplication) (AXIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AXIN1	AXIN1, AXIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kaudal Regresyon Sendromu, 600145, Otozomal Dominant (Sirenomeli) (VANGL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Caudal regression syndrome, 600145, Autosomal dominant (Sirenomelia) (VANGL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VANGL1	VANGL1, STBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sacral defect with anterior meningocele (SDAM) [MIM:600145]: Form of caudal dysgenesis. It is present at birth and becomes symptomatic later in life, usually because of obstructive labor in females, chronic constipation, or meningitis. Inheritance is autosomal dominant. [ECO:0000269	PubMed:17409324]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kaudal Regresyon Sendromu, 600145, Otozomal Dominant (Sirenomeli) (Prenatal) (VANGL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Caudal regression syndrome, 600145, Autosomal dominant (Sirenomelia) (VANGL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VANGL1	VANGL1, STBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sacral defect with anterior meningocele (SDAM) [MIM:600145]: Form of caudal dysgenesis. It is present at birth and becomes symptomatic later in life, usually because of obstructive labor in females, chronic constipation, or meningitis. Inheritance is autosomal dominant. [ECO:0000269	PubMed:17409324]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Sinir Sistemi ve Retinanın Kavernöz Malformasyonları, 116860, Otozomal Dominant (Ailesel Serebral Kavernöz Malformasyon) (KRIT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cavernous malformations of CNS and retina, 116860, Autosomal dominant (Familial cerebral cavernous malformation) (KRIT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRIT1	CCM1, CAM, KRIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral cavernous malformations 1 (CCM1) [MIM:116860]: A congenital vascular anomaly of the central nervous system that can result in hemorrhagic stroke, seizures, recurrent headaches, and focal neurologic deficits. The lesions are characterized by grossly enlarged blood vessels consisting of a single layer of endothelium and without any intervening neural tissue, ranging in diameter from a few millimeters to several centimeters. {ECO:0000269	PubMed:12172908}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kavitelî Optik Disk Anomalileri, 611543, Otozomal Dominant; CODA (Ailesel Kavitelî Optik Disk Anomalisi) (MMP19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cavitary optic disc anomalies, 611543, Autosomal dominant; CODA (Familial cavitary optic disc anomaly) (MMP19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP19	MMP19, MMP18, CODA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD8 Eksikliği, Ailesel, 608957, Otozomal Resesif (Solunum Yolu Enfeksiyonlarına Yatkınlık, CD8 Alfa Zincir Mutasyonuna Bağlı) (CD8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	CD8 deficiency, familial, 608957, Autosomal recessive (Susceptibility to respiratory infections associated with CD8alpha chain mutation) (CD8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD8A	CD8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: CD8 deficiency, familial (CD8 deficiency) [MIM:608957]: An immunologic defect characterized by absence of CD8+ cells, leading to recurrent bacterial infections. {ECO:0000269	PubMed:11435463}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
CD8 Eksikliği, Ailesel, 608957, Otozomal Resesif (Solunum Yolu Enfeksiyonlarına Yatkınlık, CD8 Alfa Zincir Mutasyonuna Bağlı) (Prenatal) (CD8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	CD8 deficiency, familial, 608957, Autosomal recessive (Susceptibility to respiratory infections associated with CD8alpha chain mutation) (CD8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD8A	CD8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: CD8 deficiency, familial (CD8 deficiency) [MIM:608957]: An immunologic defect characterized by absence of CD8+ cells, leading to recurrent bacterial infections. {ECO:0000269	PubMed:11435463}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çölyak Hastalığı (Gluten enteropatisi) (HLA Analizi) (, geni) (HLA Analizi/ HLA Analysis) (Postnatal)	Celiac disease (Gluten enteropathy) (HLA analysis) (, gene) (HLA Analiz/ HLA Analysis) (Postnatal)	.	.	HLA Analizi/ HLA Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Çölyak Hastalığı, Yatkınlık, 212750, Otozomal Resesif, Multifaktöryel (HLA-DQA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Celiac disease, susceptibility to, 212750, Autosomal recessive, Multifactorial (HLA-DQA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DQA1	HLA-DQA1, CELIAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çölyak Hastalığı, Yatkınlık, 212750, Otozomal Resesif, Multifaktöryel (Sporadik Creutzfeldt-Jakob Hastalığı) (HLA-DQB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Celiac disease, susceptibility to, 212750, Autosomal recessive, Multifactorial (Sporadic Creutzfeldt-Jakob disease) (HLA-DQB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DQB1	HLA-DQB1, CELIAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çölyak Hastalığı, Yatkınlık, 3, 609755 (CTLA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Celiac disease, susceptibility to, 3, 609755 (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çölyak Hastalığı, Yatkınlık, 4, 609753 (MYO9B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Celiac disease, susceptibility to, 4, 609753 (MYO9B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO9B	MYO9B, MYR5, CELIAC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cenani-Lenz Sindaktili Sendromu, 212780, Otozomal Resesif; CLSS (Cenani-Lenz Sindaktili Sendromu) (LRP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cenani-Lenz syndactyly syndrome, 212780, Autosomal recessive; CLSS (Cenani-Lenz syndrome) (LRP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP4	LRP4, MEGF7, CLSS, SOST2, CMS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cenani-Lenz Sindaktili Sendromu, 212780, Otozomal Resesif; CLSS (Cenani-Lenz Sindaktili Sendromu) (Prenatal) (LRP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cenani-Lenz syndactyly syndrome, 212780, Autosomal recessive; CLSS (Cenani-Lenz syndrome) (LRP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP4	LRP4, MEGF7, CLSS, SOST2, CMS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Kor Hastalığı, 117000, Otozomal Resesif, Otozomal Dominant (Santral Kor Hastalığı - Miyopati; CCD (Homojen Tip 1)) (RYR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central core disease, 117000, Autosomal recessive, Autosomal dominant; CCD (Central core disease) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095	EDTA Blood Sample
Santral Kor Hastalığı, 117000, Otozomal Resesif, Otozomal Dominant (Santral Kor Hastalığı - Miyopati; CCD (Homojen Tip 1)) (Prenatal) (RYR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central core disease, 117000, Autosomal recessive, Autosomal dominant; CCD (Central core disease) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Santral Hipoventilasyon Sendromu, 209880, Otozomal Dominant (Haddad Sendromu) (GDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, 209880, Autosomal dominant (Haddad syndrome) (GDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDNF	GDNF, HSCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, 209880, Otozomal Dominant (Haddad Sendromu) (MLPA)	Central hypoventilation syndrome, 209880, Autosomal dominant (Haddad syndrome) (MLPA)	GDNF	GDNF, HSCR3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, 209880, Otozomal Dominant (Haddad Sendromu) (Prenatal) (GDNF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, 209880, Autosomal dominant (Haddad syndrome) (GDNF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDNF	GDNF, HSCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, 209880, Otozomal Dominant (Haddad Sendromu) (Prenatal) (MLPA)	Central hypoventilation syndrome, 209880, Autosomal dominant (Haddad syndrome) (Prenatal) (MLPA)	GDNF	GDNF, HSCR3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (EDN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:8696331). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (BDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (BDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:11840487). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (MLPA)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:11840487). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (MLPA)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (Prenatal) (RET geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (RET gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (Prenatal) (EDN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:8696331). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (Prenatal) (BDNF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (BDNF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:11840487). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (Prenatal) (MLPA)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (Prenatal) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant (Ondine Sendromu) (Prenatal) (MLPA)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant (Ondine syndrome) (Prenatal) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:8696331). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant; CCHS (Ondine Sendromu) (ASCL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant; CCHS (Ondine syndrome) (ASCL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASCL1	ASCL1, ASH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, 209880, Otozomal Dominant; CCHS (Ondine Sendromu) (Prenatal) (ASCL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, congenital, 209880, Autosomal dominant; CCHS (Ondine syndrome) (ASCL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASCL1	ASCL1, ASH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, ± Hirschsprung-Hastalığı 209880, Otozomal Dominant (Haddad Sendromu) (PHOX2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880, Autosomal dominant (Haddad syndrome) (PHOX2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHOX2B	PMX2B, NBPHOX, PHOX2B, N8LST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Santral Hipoventilasyon Sendromu, Konjenital, ± Hirschsprung Hastalığı 209880, Otozomal Dominant (Haddad Sendromu) (Prenatal) (PHOX2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, 209880, Autosomal dominant (Haddad syndrome) (PHOX2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHOX2B	PMX2B, NBPHOX, PHOX2B, N8LST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sentronükleer Miyopati 5, 615959, Otozomal Resesif (Otozomal Resesif Santronükleer Miyopati) (SPEG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Centronuclear myopathy 5, 615959, Autosomal recessive (Autosomal recessive centronuclear myopathy) (SPEG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPEG	SPEG, APEG1, CNM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, centronuclear, 5 (CNM5) [MIM:615959]: A form of centronuclear myopathy, a congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. CNM5 features include severe neonatal hypotonia with respiratory insufficiency, difficulty feeding, and delayed motor development. Some patients die in infancy, and some develop dilated cardiomyopathy. (ECO:0000269	PubMed:25087613). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sentronükleer Miyopati 5, 615959, Otozomal Resesif (Otozomal Resesif Santronükleer Miyopati) (Prenatal) (SPEG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Centronuclear myopathy 5, 615959, Autosomal recessive (Autosomal recessive centronuclear myopathy) (SPEG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPEG	SPEG, APEG1, CNM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, centronuclear, 5 (CNM5) [MIM:615959]: A form of centronuclear myopathy, a congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. CNM5 features include severe neonatal hypotonia with respiratory insufficiency, difficulty feeding, and delayed motor development. Some patients die in infancy, and some develop dilated cardiomyopathy. (ECO:0000269	PubMed:25087613). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sentronükleer Miyopati, Otozomal, Değiştirici, 160150, Otozomal Dominant (Otozomal Dominant Santronükleer Miyopati) (MTMR14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Centronuclear myopathy, autosomal, modifier of, 160150, Autosomal dominant (Autosomal dominant centronuclear myopathy) (MTMR14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTMR14	MTMR14, C3orf29, HJUMPY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sentronükleer Miyopati, Otozomal, Değiştirici, 160150, Otozomal Dominant (Otozomal Dominant Santronükleer Miyopati) (Prenatal) (MTMR14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Centronuclear myopathy, autosomal, modifier of, 160150, Autosomal dominant (Autosomal dominant centronuclear myopathy) (MTMR14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTMR14	MTMR14, C3orf29, HJUMPY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi ve Hipogonadotropik Hipogonadizm, 212840, Otozomal Resesif; GDHS (Serebeller Ataksi-Hipogonadizm Sendromu) (RNF216 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840, Autosomal recessive; GDHS (Cerebellar ataxia-hypogonadism syndrome) (RNF216 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF216	RNF216, TRIAD3, ZIN, CAHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gordon Holmes syndrome (GDHS) [MIM:212840]: A disease characterized by cerebellar symptoms and signs of sex steroid deficiency. Clinical features include cerebellar and brain stem atrophy, cerebellar ataxia, hypothalamic LHRH deficiency, hypogonadotrophic hypogonadism, lack of secondary sexual characteristics, and infertility. {ECO:0000269|PubMed:23656589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 3, 613227, Otozomal Resesif; CAMRQ3 (Disequilibrium Sendromu) (CA8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227, Autosomal recessive; CAMRQ3 (Dysequilibrium syndrome) (CA8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CA8	CA8, CALS, CARP, CAMRQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 3 (CMARQ3) [MIM:613227]: A congenital cerebellar ataxia associated with dysarthia, quadrupedal gait and mental retardation. {ECO:0000269|PubMed:19461874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 3, 613227, Otozomal Resesif; CAMRQ3 (Disequilibrium Sendromu) (Prenatal) (CA8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia and mental retardation with or without quadrupedal locomotion 3, 613227, Autosomal recessive; CAMRQ3 (Dysequilibrium syndrome) (CA8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CA8	CA8, CALS, CARP, CAMRQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 3 (CMARQ3) [MIM:613227]: A congenital cerebellar ataxia associated with dysarthia, quadrupedal gait and mental retardation. {ECO:0000269|PubMed:19461874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, 604290, Otozomal Resesif (Aseruloplasminemi) (CP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia, 604290, Autosomal recessive (Aceruloplasminemia) (CP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CP	CP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebellar Ataksi, 604290, Otozomal Resesif (Aseruloplasminemi) (Prenatal) (CP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia, 604290, Autosomal recessive (Aceruloplasminemia) (CP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CP	CP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, Sağırlık ve Narkolepsi, Otozomal Dominant, 604121, Otozomal Dominant; ADCADN (Otozomal Dominant Serebellar Ataksi-Sağırlık-Narkolepsi Sendromu) (DNMT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121, Autosomal dominant; ADCADN (Autosomal dominant cerebellar ataxia, deafness and narcolepsy) (DNMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNMT1	DNMT1, MCMT, HSN1E, ADCADN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (ADCADN) [MIM:604121]: An autosomal dominant neurologic disorder characterized by adult onset of progressive cerebellar ataxia, narcolepsy, cataplexy, sensorineural deafness, and dementia. More variable features include optic atrophy, sensory neuropathy, psychosis, and depression. {ECO:0000269|PubMed:22328086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi, Sağırlık ve Narkolepsi, Otozomal Dominant, 604121, Otozomal Dominant; ADCADN (Otozomal Dominant Serebellar Ataksi-Sağırlık-Narkolepsi Sendromu) (Prenatal) (DNMT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant, 604121, Autosomal dominant; ADCADN (Autosomal dominant cerebellar ataxia, deafness and narcolepsy) (DNMT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNMT1	DNMT1, MCMT, HSN1E, ADCADN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (ADCADN) [MIM:604121]: An autosomal dominant neurologic disorder characterized by adult onset of progressive cerebellar ataxia, narcolepsy, cataplexy, sensorineural deafness, and dementia. More variable features include optic atrophy, sensory neuropathy, psychosis, and depression. {ECO:0000269|PubMed:22328086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 1; Camrq1 (Disequilibrium Sendromu) (VLDLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 (Dysequilibrium syndrome) (VLDLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VLDLR	VLDLR, CAMRQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 1; Camrq1 (Disequilibrium Sendromu) (MLPA)	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 (Dysequilibrium syndrome) (MLPA)	VLDLR	VLDLR, CAMRQ1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 1; Camrq1 (Disequilibrium Sendromu) (Prenatal) (VLDLR geni) (Tüm Gen Dizi Analizi) (Prenatal)	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 (Dysequilibrium syndrome) (VLDLR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VLDLR	VLDLR, CAMRQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 1; Camrq1 (Disequilibrium Sendromu) (Prenatal) (MLPA)	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 1; CAMRQ1 (Dysequilibrium syndrome) (Prenatal) (MLPA)	VLDLR	VLDLR, CAMRQ1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dysequilibrium Sendromu 2, 610185, Otozomal Resesif; CAMRQ2 (Disequilibrium Sendromu) (WDR81 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185, Autosomal recessive; CAMRQ2 (Dysequilibrium syndrome) (WDR81 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR81	WDR81, CAMRQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dysequilibrium Sendromu 2, 610185, Otozomal Resesif; CAMRQ2 (Disequilibrium Sendromu) (Prenatal) (WDR81 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 2, 610185, Autosomal recessive; CAMRQ2 (Dysequilibrium syndrome) (WDR81 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR81	WDR81, CAMRQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 4, 615268, Otozomal Resesif; CAMRQ4 (Disequilibrium Sendromu) (ATP8A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268, Autosomal recessive; CAMRQ4 (Dysequilibrium syndrome) (ATP8A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP8A2	ATP8A2, ATP1B, CAMRQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebellar Ataksi, Mental Retardasyon ve Dikselülitrium Sendromu 4, 615268, Otozomal Resesif; CAMRQ4 (Disequilibrium Sendromu) (Prenatal) (ATP8A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 4, 615268, Autosomal recessive; CAMRQ4 (Dysequilibrium syndrome) (ATP8A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP8A2	ATP8A2, ATP1B, CAMRQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi,Nonprogresif, Mental Retardasyon, 614756, Otozomal Dominant (Progresif Olmayan Serebellar Ataksi-Entelektüel Yeterszlik) (CAMTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar ataxia, nonprogressive, with mental retardation, 614756, Autosomal dominant (Non-progressive cerebellar ataxia with intellectual disability) (CAMTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAMTA1	CAMTA1, KIAA0833, CANPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, non-progressive, with mental retardation (CANPMR) [MIM:614756]: A neurodevelopmental disorder characterized by mildly delayed psychomotor development, early onset of cerebellar ataxia, and intellectual disability later in childhood and adult life. Other features may include neonatal hypotonia, dysarthria, and dysmetria. Brain imaging in some patients shows cerebellar atrophy. Dysmorphic facial features are variable. {ECO:0000269|PubMed:22693284}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi,Nonprogresif, Mental Retardasyon, 614756, Otozomal Dominant (Progresif Olmayan Serebellar Ataksi-Entelektüel Yeterszlik) (Prenatal) (CAMTA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar ataxia, nonprogressive, with mental retardation, 614756, Autosomal dominant (Non-progressive cerebellar ataxia with intellectual disability) (CAMTA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAMTA1	CAMTA1, KIAA0833, CANPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, non-progressive, with mental retardation (CANPMR) [MIM:614756]: A neurodevelopmental disorder characterized by mildly delayed psychomotor development, early onset of cerebellar ataxia, and intellectual disability later in childhood and adult life. Other features may include neonatal hypotonia, dysarthria, and dysmetria. Brain imaging in some patients shows cerebellar atrophy. Dysmorphic facial features are variable. {ECO:0000269|PubMed:22693284}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Atrofi, Görme Bozukluğu ve Psikomotor Retardasyon, 616875, Otozomal Resesif; CAVIPMR (EMC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875, Autosomal recessive; CAVIPMR (EMC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EMC1	EMC1, KIAA0090, CAVIPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebellar Atrofi, Görme Bozukluğu ve Psikomotor Retardasyon, 616875, Otozomal Resesif; CAVIPMR (Prenatal) (EMC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar atrophy, visual impairment, and psychomotor retardation, 616875, Autosomal recessive; CAVIPMR (EMC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EMC1	EMC1, KIAA0090, CAVIPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 1, 613227, Otozomal Resesif (Disequilibrium Sendromu) (VLDLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050, Autosomal recessive (Dysequilibrium syndrome) (VLDLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VLDLR	VLDLR, CAMRQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 1, 613227, Otozomal Resesif (Disequilibrium Sendromu) (MLPA)	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050, Autosomal recessive (Dysequilibrium syndrome) (MLPA)	VLDLR	VLDLR, CAMRQ1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 1, 613227, Otozomal Resesif (Disequilibrium Sendromu) (Prenatal) (VLDLR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050, Autosomal recessive (Dysequilibrium syndrome) (VLDLR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VLDLR	VLDLR, CAMRQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Serebellar Ataksi ve Mental Retardasyon ± Quadrupedal Lokomosyon 1, 613227, Otozomal Resesif (Disequilibrium Sendromu) (Prenatal) (MLPA)	Cerebellar hypoplasia and mental retardation with or without quadrupedal locomotion 1, 224050, Autosomal recessive (Dysequilibrium syndrome) (Prenatal) (MLPA)	VLDLR	VLDLR, CAMRQ1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellar ataxia, mental retardation, and dysequilibrium syndrome 1 (CAMRQ1) [MIM:224050]: A congenital, non-progressive cerebellar ataxia associated with disturbed equilibrium, delayed ambulation, mental retardation, cerebellar hypoplasia and mild cerebral gyral simplification. Additional features include short stature, strabismus, pes planus and, rarely, seizures. {ECO:0000269|PubMed:16080122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrofasiyodental Sendrom, 616202, Otozomal Resesif, CFDS (Serebellar-Fasiyal -Dental Sendromu) (BRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebellofacidental syndrome, 616202, Autosomal recessive; CFDS (Cerebellar-facial-dental syndrome) (BRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRF1	BRF1, TAF3C, GTF3B, TF3B90, CFDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellofacidental syndrome (CFDS) [MIM:616202]: An autosomal recessive disorder characterized by cerebellar hypoplasia, delayed development and intellectual disability, as well as facial dysmorphic features, short stature, microcephaly, and dental anomalies. {ECO:0000269|PubMed:25561519}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebrofasiyodental Sendrom, 616202, Otozomal Resesif, CFDS (Serebellar-Fasiyal -Dental Sendromu) (Prenatal) (BRF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebellofacidental syndrome, 616202, Autosomal recessive; CFDS (Cerebellar-facial-dental syndrome) (BRF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRF1	BRF1, TAF3C, GTF3B, TF3B90, CFDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebellofacidental syndrome (CFDS) [MIM:616202]: An autosomal recessive disorder characterized by cerebellar hypoplasia, delayed development and intellectual disability, as well as facial dysmorphic features, short stature, microcephaly, and dental anomalies. {ECO:0000269|PubMed:25561519}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Amiloid Anjiyopati, 105150, Otozomal Dominant (Kalıtsal Serebral Hemoraji-Amiloidoz) (CST3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral amyloid angiopathy, 105150, Autosomal dominant (Hereditary cerebral hemorrhage with amyloidosis) (CST3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CST3	CST3, ARMD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 6 (AMYL6) [MIM:105150]: A hereditary generalized amyloidosis due to cystatin C amyloid deposition. Cystatin C amyloid accumulates in the walls of arteries, arterioles, and sometimes capillaries and veins of the brain, and in various organs including lymphoid tissue, spleen, salivary glands, and seminal vesicles. Amyloid deposition in the cerebral vessels results in cerebral amyloid angiopathy, cerebral hemorrhage and premature stroke. Cystatin C levels in the cerebrospinal fluid are abnormally low. {ECO:0000269|PubMed:1352265, ECO:0000269|PubMed:2541223}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Serebral Amiloid Anjiyopati, Hollanda, İtalyan, Iowa, Flaman, Arktik Türevleri, 605714, Otozomal Dominant (Abeta21g Amiloidoz) (APP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral amyloid angiopathy, Dutch, Italian, Iowa, Flemish, Arctic variants, 605714, Autosomal dominant (Abeta21g amyloidosis) (APP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APP	APP, AAA, CVAP, AD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Serebral Amiloid Anjiyopati, Itm2b-İlgili, 1 (ITM2B Amiloidoz) (ITM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	CEREBRAL AMYLOID ANGIOPATHY, ITM2B-RELATED, 1 (ITM2B amyloidosis) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. {ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362}. Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. {ECO:0000269|PubMed:10781099}.	EDTA Blood Sample
Serebral Amiloid Anjiyopati, Itm2b-İlgili, 2 (ITM2B Amiloidoz) (ITM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	CEREBRAL AMYLOID ANGIOPATHY, ITM2B-RELATED, 2 (ITM2B amyloidosis) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. {ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362}. Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. {ECO:0000269|PubMed:10781099}.	EDTA Blood Sample
Serebral Amiloid Anjiyopati, PRNP İle İlişkili, 137440, Otozomal Dominant (Gerstmann-Straussler-Scheinker Sendromu) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral amyloid angiopathy, PRNP-related, 137440, Autosomal dominant (Gerstmann-Straussler-Scheinker syndrome) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Arteriopati, Subkortikal Enfarktüs ve Lökoensefalopati 1, 125310, Otozomal Dominant; CADASIL1 (CADASIL) (NOTCH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1, 125310, Autosomal dominant; CADASIL1 (CADASIL) (NOTCH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH3	NOTCH3, CADASIL1, CASIL, IMF2, LMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, autosomal dominant (CADASIL) [MIM:125310]: A cerebrovascular disease characterized by multiple subcortical infarcts, pseudobulbar palsy, dementia, and the presence of granular deposits in small cerebral arteries producing ischemic stroke. {ECO:0000269|PubMed:10227618, ECO:0000269|PubMed:10371548, ECO:0000269|PubMed:10802807, ECO:0000269|PubMed:10854111, ECO:0000269|PubMed:11058919, ECO:0000269|PubMed:11102981, ECO:0000269|PubMed:11559313, ECO:0000269|PubMed:11755616, ECO:0000269|PubMed:11810186, ECO:0000269|PubMed:12136071, ECO:0000269|PubMed:12146805, ECO:0000269|PubMed:12589106, ECO:0000269|PubMed:12810003, ECO:0000269|PubMed:15229130, ECO:0000269|PubMed:15300988, ECO:0000269|PubMed:15364702, ECO:0000269|PubMed:15378071, ECO:0000269|PubMed:15818833, ECO:0000269|PubMed:16009764, ECO:0000269|PubMed:24000151, ECO:0000269|PubMed:9388399}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Serebral Arteriyopati, Otozomal Dominant, Subkortikal Enfarktüsler ve Lökoensefalopati, Tip 2, Otozomal Dominant, 616779 (CARASIL); CADASIL2 (HTRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779, Autosomal dominant (CARASIL); CADASIL2 (HTRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA1	HTRA1, PRSS11, ARMD7, CARASIL, CADASIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy (CARASIL) [MIM:600142]: A cerebrovascular disease characterized by non-hypertensive arteriopathy of cerebral small vessels with subcortical infarcts, alopecia, and spondylosis. Small cerebral arteries show arteriosclerotic changes, fibrous intimal proliferation, and hyaline degeneration with splitting of the intima and/or the internal elastic membrane. Neurologic features include progressive dementia, gait disturbances, extrapyramidal and pyramidal signs, and demyelination of the cerebral white matter with sparing of U fibers. {ECO:0000269|PubMed:19387015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Kavernöz Malformasyonlar 3, 603285; CCM3 (Alleael Serebral Kavernöz Malformasyon) (PDCD10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral cavernous malformations 3, 603285; CCM3 (Familial cerebral cavernous malformation) (PDCD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDCD10	PDCD10, TFAR15, CCM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Kavernomatöz Malformasyonlar-1, 116860, Otozomal Dominant (Alleael Serebral Kavernöz Malformasyon) (KRIT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral cavernous malformations-1, 116860, Autosomal dominant (Familial cerebral cavernous malformation) (KRIT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRIT1	CCM1, CAM, KRIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral cavernous malformations 1 (CCM1) [MIM:116860]: A congenital vascular anomaly of the central nervous system that can result in hemorrhagic stroke, seizures, recurrent headaches, and focal neurologic deficits. The lesions are characterized by grossly enlarged blood vessels consisting of a single layer of endothelium and without any intervening neural tissue, ranging in diameter from a few millimeters to several centimeters. {ECO:0000269|PubMed:12172908}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Kavernöz Malformasyonlar-2, 603284, Otozomal Dominant (Alleael Serebral Kavernöz Malformasyon) (CCM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral cavernous malformations-2, 603284, Autosomal dominant (Familial cerebral cavernous malformation) (CCM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCM2	C7orf22, CCM2, MGC4067	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral cavernous malformations 2 (CCM2) [MIM:603284]: A congenital vascular anomaly of the central nervous system that can result in hemorrhagic stroke, seizures, recurrent headaches, and focal neurologic deficits. The lesions are characterized by grossly enlarged blood vessels consisting of a single layer of endothelium and without any intervening neural tissue, ranging in diameter from a few millimeters to several centimeters. {ECO:0000269|PubMed:14624391, ECO:0000269|PubMed:14740320, ECO:0000269|PubMed:22415356}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 1, 300352, X'e Bağlı Resesif; CCDS1 (X'e Bağlı Kreatin Transportör Eksikliği) (SLC6A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral creatine deficiency syndrome 1, 300352, X-linked recessive; CCDS1 (X-linked creatine transporter deficiency) (SLC6A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A8	SLC6A8, CRTR, CCDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 1, 300352, X'e Bağlı Resesif; CCDS1 (X'e Bağlı Kreatin Transporter Eksikliği) (MLPA)	Cerebral creatine deficiency syndrome 1, 300352, X-linked recessive; CCDS1 (X-linked creatine transporter deficiency) (MLPA)	SLC6A8	SLC6A8, CRTR, CCDS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 1, 300352, X'e Bağlı Resesif; CCDS1 (X'e Bağlı Kreatin Transporter Eksikliği) (Prenatal) (SLC6A8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral creatine deficiency syndrome 1, 300352, X-linked recessive; CCDS1 (X-linked creatine transporter deficiency) (SLC6A8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A8	SLC6A8, CRTR, CCDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 1, 300352, X'e Bağlı Resesif; CCDS1 (X'e Bağlı Kreatin Transporter Eksikliği) (Prenatal) (MLPA)	Cerebral creatine deficiency syndrome 1, 300352, X-linked recessive; CCDS1 (X-linked creatine transporter deficiency) (Prenatal) (MLPA)	SLC6A8	SLC6A8, CRTR, CCDS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 2, 612736, Otozomal Resesif; CCDS2 (Guanidinoasetat Metil Transferaz Eksikliği) (GAMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral creatine deficiency syndrome 2, 612736, Autosomal recessive; CCDS2 (Guanidinoacetate methyltransferase deficiency) (GAMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAMT	GAMT, CCDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 2, 612736, Otozomal Resesif; CCDS2 (Guanidinoasetat Metil Transferaz Eksikliği) (Prenatal) (GAMT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral creatine deficiency syndrome 2, 612736, Autosomal recessive; CCDS2 (Guanidinoacetate methyltransferase deficiency) (GAMT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GAMT	GAMT, CCDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Serebral Kreatin Eksikliği Sendromu 3, 612718, Otozomal Resesif; CCDS3 (L-Arginin: Glisin Amidinotransferase Eksikliği (Serebral Kreatin Eksikliği Sendromu 3)) (GATM gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral creatine deficiency syndrome 3, 612718, Autosomal recessive; CCDS3 (L-Arginine:glycine amidinotransferase deficiency) (GATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATM	GATM, AGAT, CCDS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral creatine deficiency syndrome 3 (CCDS3) [MIM:612718]: An autosomal recessive disorder characterized by developmental delay/regression, mental retardation, severe disturbance of expressive and cognitive speech, and severe depletion of creatine/phosphocreatine in the brain. Most patients develop a myopathy characterized by muscle weakness and atrophy later in life. (ECO:0000269|PubMed:11555793, ECO:0000269|PubMed:20682460, ECO:0000269|PubMed:22386973, ECO:0000269|PubMed:23660394, ECO:0000269|PubMed:23770102). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Kreatin Eksikliği Sendromu 3, 612718, Otozomal Resesif; CCDS3 (L-Arginin: Glisin Amidinotransferase Eksikliği (Serebral Kreatin Eksikliği Sendromu 3)) (Prenatal) (GATM gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral creatine deficiency syndrome 3, 612718, Autosomal recessive; CCDS3 (L-Arginine:glycine amidinotransferase deficiency) (GATM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATM	GATM, AGAT, CCDS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebral creatine deficiency syndrome 3 (CCDS3) [MIM:612718]: An autosomal recessive disorder characterized by developmental delay/regression, mental retardation, severe disturbance of expressive and cognitive speech, and severe depletion of creatine/phosphocreatine in the brain. Most patients develop a myopathy characterized by muscle weakness and atrophy later in life. (ECO:0000269|PubMed:11555793, ECO:0000269|PubMed:20682460, ECO:0000269|PubMed:22386973, ECO:0000269|PubMed:23660394, ECO:0000269|PubMed:23770102). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Disgenezis, Nöropati, İktiyozis, ve Palmoplantar Keratoderma Sendromu, 609528, Otozomal Resesif (CEDNIK Sendromu (Serebral Disgenesis, Nöropati, İktiyozis, ve Palmoplantar Keratoderma Sendromu)) (SNAP29 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528, Autosomal recessive (CEDNIK syndrome) (SNAP29 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNAP29	SNAP29, CEDNIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Disgenezis, Nöropati, İktiyozis, ve Palmoplantar Keratoderma Sendromu, 609528, Otozomal Resesif (CEDNIK Sendromu (Serebral Disgenesis, Nöropati, İktiyozis, ve Palmoplantar Keratoderma Sendromu)) (Prenatal) (SNAP29 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528, Autosomal recessive (CEDNIK syndrome) (SNAP29 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNAP29	SNAP29, CEDNIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Enfarktüs, Yatkınlık, 601367, Multifaktöryel (PRKCH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral infarction, susceptibility to, 601367, Multifactorial (PRKCH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCH	PRKCH, PKCL, PRKCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ischemic stroke (ISCHSTR) [MIM:601367]: A stroke is an acute neurologic event leading to death of neural tissue of the brain and resulting in loss of motor, sensory and/or cognitive function. Ischemic strokes, resulting from vascular occlusion, is considered to be a highly complex disease consisting of a group of heterogeneous disorders with multiple genetic and environmental risk factors. (ECO:0000269	PubMed:17206144). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 1, 603513, Otozomal Resesif; CPSQ1 (Kalssal Konjenital Spastik Tetraplejj) (GAD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral palsy, spastic quadriplegic, 1, 603513, Autosomal recessive; CPSQ1 (Inherited congenital spastic tetraplegia) (GAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAD1	GAD1, SCP, CPSQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 1, 603513, Otozomal Resesif; CPSQ1 (Kalssal Konjenital Spastik Tetraplejj) (Prenatal) (GAD1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral palsy, spastic quadriplegic, 1, 603513, Autosomal recessive; CPSQ1 (Inherited congenital spastic tetraplegia) (GAD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GAD1	GAD1, SCP, CPSQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 2, 612900, CPSQ2 (Kalıtsal Konjenital Spastik Tetraplejj) (KANK1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral palsy, spastic quadriplegic, 2, 612900, CPSQ2 (Inherited congenital spastic tetraplegia) (KANK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KANK1	KANK1, KANK, ANKRD15, KIAA0172, CPSQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 2, 612900, CPSQ2 (Kalıtsal Konjenital Spastik Tetraplejj) (Prenatal) (KANK1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral palsy, spastic quadriplegic, 2, 612900, CPSQ2 (Inherited congenital spastic tetraplegia) (KANK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KANK1	KANK1, KANK, ANKRD15, KIAA0172, CPSQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 3, 617008, Otozomal Resesif; CPSQ3 (Kalıtsal Konjenital Spastik Tetraplejj) (ADD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebral palsy, spastic quadriplegic, 3, 617008, Autosomal recessive; CPSQ3 (Inherited congenital spastic tetraplegia) (ADD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADD3	ADD3, ADDL, CPSQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebral Palsi, Spastik Kuadriplejik, 3, 617008, Otozomal Resesif; CPSQ3 (Kalıtsal Konjenital Spastik Tetraplejj) (Prenatal) (ADD3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebral palsy, spastic quadriplegic, 3, 617008, Autosomal recessive; CPSQ3 (Inherited congenital spastic tetraplegia) (ADD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADD3	ADD3, ADDL, CPSQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrokostomandibuler Sendrom, 117650, Otozomal Dominant; CCMS (Serebro-Kosto-Mandibular Sendromu) (SNRPB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrocostomandibular syndrome, 117650, Autosomal dominant; CCMS (Cerebro-costo-mandibular syndrome) (SNRPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNRPB	SNRPB, CCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebrocostomandibular syndrome (CCMS) [MIM:117650]: A syndrome characterized by severe micrognathia, rib defects ranging from a few dorsal rib segments to complete absence of ossification, and mental retardation. (ECO:0000269|PubMed:25047197, ECO:0000269|PubMed:25504470). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebrokostomandibuler Sendrom, 117650, Otozomal Dominant; CCMS (Serebro-Kosto-Mandibular Sendromu) (Prenatal) (SNRPB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrocostomandibular syndrome, 117650, Autosomal dominant; CCMS (Cerebro-costo-mandibular syndrome) (SNRPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNRPB	SNRPB, CCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebrocostomandibular syndrome (CCMS) [MIM:117650]: A syndrome characterized by severe micrognathia, rib defects ranging from a few dorsal rib segments to complete absence of ossification, and mental retardation. (ECO:0000269|PubMed:25047197, ECO:0000269|PubMed:25504470). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 1, 214150, Otozomal Resesif; COFS1 (Cockayne Sendromu) (ERCC6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrooculofacioskeletal syndrome 1, 214150, Autosomal recessive; COFS1 (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 1, 214150, Otozomal Resesif; COFS1 (Cockayne Sendromu) (Prenatal) (ERCC6 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrooculofacioskeletal syndrome 1, 214150, Autosomal recessive; COFS1 (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 2, 610756, COFS2 (Cockayne Sendromu) (ERCC2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrooculofacioskeletal syndrome 2, 610756, COFS2 (ERCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC2	ERCC2, EM9, XPD, COFS2, TTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 2, 610756, COFS2 (Cockayne Sendromu) (Prenatal) (ERCC2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrooculofacioskeletal syndrome 2, 610756, COFS2 (ERCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC2	ERCC2, EM9, XPD, COFS2, TTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 3, 616570, Otozomal Resesif; COFS3 (COFS) (ERCC5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrooculofacioskeletal syndrome 3, 616570, Autosomal recessive; COFS3 (COFS) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 3, 616570, Otozomal Resesif; COFS3 (COFS) (Prenatal) (ERCC5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrooculofacioskeletal syndrome 3, 616570, Autosomal recessive; COFS3 (COFS) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 4, 610758, Otozomal Resesif; COFS4 (Cockayne Sendromu) (ERCC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrooculofacioskeletal syndrome 4, 610758, Autosomal recessive; COFS4 (ERCC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC1	ERCC1, UV20, COFS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebro-oculo-facio-skeletal syndrome 4 (COFS4) [MIM:610758]: A disorder of prenatal onset characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic arthrogryposis, growth failure and severe psychomotor retardation. COFS is considered to be part of the nucleotide-excision repair disorders spectrum that include also xeroderma pigmentosum, trichothiodystrophy and Cockayne syndrome. (ECO:0000269|PubMed:17273966, ECO:0000269|PubMed:23623389). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebrookülofasiyoskeletal Sendrom 4, 610758, Otozomal Resesif; COFS4 (Cockayne Sendromu) (Prenatal) (ERCC1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrooculofacioskeletal syndrome 4, 610758, Autosomal recessive; COFS4 (Cockayne syndrome) (ERCC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC1	ERCC1, UV20, COFS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebro-oculo-facio-skeletal syndrome 4 (COFS4) [MIM:610758]: A disorder of prenatal onset characterized by microcephaly, congenital cataracts, facial dysmorphism, neurogenic arthrogryposis, growth failure and severe psychomotor retardation. COFS is considered to be part of the nucleotide-excision repair disorders spectrum that include also xeroderma pigmentosum, trichothiodystrophy and Cockayne syndrome. (ECO:0000269|PubMed:17273966, ECO:0000269|PubMed:23623389). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebroretinal Mikroanjiyopati, Kalsifikasyon ve Kistler 2, 617341, Otozomal Resesif; CRMCC2 (Coats Arı Sendromu) (STN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341, Autosomal recessive; CRMCC2 (Coats plus syndrome) (STN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STN1	STN1, OBFC1, AAF44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Serebroretinal Mikroanjiyopati, Kalsifikasyon ve Kistler 2, 617341, Otozomal Resesif; CRMCC2 (Coats Arı Sendromu) (Prenatal) (STN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebroretinal microangiopathy with calcifications and cysts 2, 617341, Autosomal recessive; CRMCC2 (Coats plus syndrome) (STN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STN1	STN1, OBFC1, AAF44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebroretinal Mikroanjiyopati, Kalsifikasyon ve Kistlerler, 612199, Otozomal Resesif (Coats Arı Sendromu) (CTC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebroretinal microangiopathy with calcifications and cysts, 612199, Autosomal recessive (Coats plus syndrome) (CTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTC1	CTC1, CRMCC, C17orf68, AAF132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebroretinal microangiopathy with calcifications and cysts (CRMCC) [MIM:612199]: An autosomal recessive pleiomorphic disorder characterized primarily by intracranial calcifications, leukodystrophy, and brain cysts, resulting in spasticity, ataxia, dystonia, seizures, and cognitive decline. Patients also have retinal telangiectasia and exudates (Coats disease) as well as extraneurologic manifestations, including osteopenia with poor bone healing and a high risk of gastrointestinal bleeding and portal hypertension caused by vasculature ectasias in the stomach, small intestine, and liver. Some individuals also have hair, skin, and nail changes, as well as anemia and thrombocytopenia. (ECO:0000269|PubMed:22267198, ECO:0000269|PubMed:22387016). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Serebroretinal Mikroanjiyopati,Kalsifikasyon ve Kistlerler, 612199, Otozomal Resesif (Coats Arı Sendromu) (Prenatal) (CTC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebroretinal microangiopathy with calcifications and cysts, 612199, Autosomal recessive (Coats plus syndrome) (CTC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTC1	CTC1, CRMCC, C17orf68, AAF132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebroretinal microangiopathy with calcifications and cysts (CRMCC) [MIM:612199]: An autosomal recessive pleiomorphic disorder characterized primarily by intracranial calcifications, leukodystrophy, and brain cysts, resulting in spasticity, ataxia, dystonia, seizures, and cognitive decline. Patients also have retinal telangiectasia and exudates (Coats disease) as well as extraneurologic manifestations, including osteopenia with poor bone healing and a high risk of gastrointestinal bleeding and portal hypertension caused by vasculature ectasias in the stomach, small intestine, and liver. Some individuals also have hair, skin, and nail changes, as well as anemia and thrombocytopenia. {ECO:0000269|PubMed:22267198, ECO:0000269|PubMed:22387016}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrotendinöz Ksantomatozis, 213700, Otozomal Resesif, CTX (Serebrotendinöz Ksantomatoz) (CYP27A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrotendinous xanthomatosis, 213700, Autosomal recessive; CTX (Cerebrotendinous xanthomatosis) (CYP27A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP27A1	CYP27A1, CYP27, CTX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebrotendinous xanthomatosis (CTX) [MIM:213700]: Rare sterol storage disorder characterized clinically by progressive neurologic dysfunction, premature atherosclerosis, and cataracts. {ECO:0000269|PubMed:12000359, ECO:0000269|PubMed:2019602, ECO:0000269|PubMed:7915755, ECO:0000269|PubMed:9186905, ECO:0000269|PubMed:9790667}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebrotendinöz Ksantomatozis, 213700, Otozomal Resesif, CTX (Serebrotendinöz Ksantomatoz) (Prenatal) (CYP27A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cerebrotendinous xanthomatosis, 213700, Autosomal recessive; CTX (Cerebrotendinous xanthomatosis) (CYP27A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP27A1	CYP27A1, CYP27, CTX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebrotendinous xanthomatosis (CTX) [MIM:213700]: Rare sterol storage disorder characterized clinically by progressive neurologic dysfunction, premature atherosclerosis, and cataracts. {ECO:0000269|PubMed:12000359, ECO:0000269|PubMed:2019602, ECO:0000269|PubMed:7915755, ECO:0000269|PubMed:9186905, ECO:0000269|PubMed:9790667}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serebrovasküler Hastalıklar, Tikayıcı (SERPINA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cerebrovascular disease, occlusive (SERPINA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA3	SERPINA3, AACT, ACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 1, 256730, Otozomal Resesif, CLN1 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (PPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 1, 256730, Autosomal recessive; CLN1 (Late infantile neuronal ceroid lipofuscinosis) (PPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPT1	PPT1, CLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 1 (CLN1) [MIM:256730]: A form of neuronal ceroid lipofuscinosis with variable age at onset. Infantile, late-infantile, juvenile, and adult onset have been reported. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment pattern seen most often in CLN1 is referred to as granular osmiophilic deposits (GROD). {ECO:0000269|PubMed:11506414, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:7637805, ECO:0000269|PubMed:9425237, ECO:0000269|PubMed:9664077}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 1, 256730, Otozomal Resesif, CLN1 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (PPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 1, 256730, Autosomal recessive; CLN1 (Late infantile neuronal ceroid lipofuscinosis) (PPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPT1	PPT1, CLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 1 (CLN1) [MIM:256730]: A form of neuronal ceroid lipofuscinosis with variable age at onset. Infantile, late-infantile, juvenile, and adult onset have been reported. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment pattern seen most often in CLN1 is referred to as granular osmiophilic deposits (GROD). {ECO:0000269|PubMed:11506414, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:7637805, ECO:0000269|PubMed:9425237, ECO:0000269|PubMed:9664077}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 10, 610127, Otozomal Resesif, CLN10 (CLN10) (NCL10) (CTSD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 10, 610127, Autosomal recessive; CLN10 (CLN10) (NCL10) (CTSD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSD	CTSD, CPSD, CLN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 10 (CLN10) [MIM:610127]: A form of neuronal ceroid lipofuscinosis with onset at birth or early childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. {ECO:0000269|PubMed:16670177, ECO:0000269|PubMed:16685649, ECO:0000269|PubMed:21990111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 10, 610127, Otozomal Resesif, CLN10 (CLN10) (NCL10) (Prenatal) (CTSD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 10, 610127, Autosomal recessive; CLN10 (CLN10) (NCL10) (CTSD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTSD	CTSD, CPSD, CLN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 10 (CLN10) [MIM:610127]: A form of neuronal ceroid lipofuscinosis with onset at birth or early childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. {ECO:0000269|PubMed:16670177, ECO:0000269|PubMed:16685649, ECO:0000269|PubMed:21990111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 11, 614706, Otozomal Resesif (CLN11) (NCL11) (GRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 11, 614706, Autosomal recessive (CLN11) (NCL11) (GRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRN	GRN, CLN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 11, 614706, Otozomal Resesif (CLN11) (NCL11) (Prenatal) (GRN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 11, 614706, Autosomal recessive (CLN11) (NCL11) (GRN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRN	GRN, CLN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 13, Kufs Tipi, 615362, Otozomal Resesif; CLN13 (CLN13 Hastalığı) (CTSF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362, Autosomal recessive; CLN13 (CLN13 disease) (CTSF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSF	CTSF, CLN13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 13, Kufs Tipi, 615362, Otozomal Resesif; CLN13 (CLN13 Hastalığı) (Prenatal) (CTSF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 13, Kufs type, 615362, Autosomal recessive; CLN13 (CLN13 disease) (CTSF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTSF	CTSF, CLN13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 2, 204500, Otozomal Resesif, CLN2 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (TPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 2, 204500, Autosomal recessive; CLN2 (Late infantile neuronal ceroid lipofuscinosis) (TPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPP1	TPP1, CLN2, SCAR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 2 (CLN2) [MIM:204500]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment pattern seen most often in CLN2 consists of curvilinear profiles. {ECO:0000269|PubMed:10330339, ECO:0000269|PubMed:10665500, ECO:0000269|PubMed:11241479, ECO:0000269|PubMed:11339651, ECO:0000269|PubMed:11589012, ECO:0000269|PubMed:12376936, ECO:0000269|PubMed:12414822, ECO:0000269|PubMed:12698559, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:20340139, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:9295267}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 2, 204500, Otozomal Resesif, CLN2 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (TPP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 2, 204500, Autosomal recessive; CLN2 (Late infantile neuronal ceroid lipofuscinosis) (TPP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPP1	TPP1, CLN2, SCAR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 2 (CLN2) [MIM:204500]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment pattern seen most often in CLN2 consists of curvilinear profiles. {ECO:0000269|PubMed:10330339, ECO:0000269|PubMed:10665500, ECO:0000269|PubMed:11241479, ECO:0000269|PubMed:11339651, ECO:0000269|PubMed:11589012, ECO:0000269|PubMed:12376936, ECO:0000269|PubMed:12414822, ECO:0000269|PubMed:12698559, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:20340139, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:9295267}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 3, 204200, Otozomal Resesif, CLN3 (CLN3 Hastalığı) (CLN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 3, 204200, Autosomal recessive; CLN3 (CLN3 disease) (CLN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN3	CLN3, BTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 3 (CLN3) [MIM:204200]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The hallmark of CLN3 is the ultrastructural pattern of lipopigment with a fingerprint profile, which can have 3 different appearances: pure within a lysosomal residual body; in conjunction with curvilinear or rectilinear profiles; and as a small component within large membrane-bound lysosomal vacuoles. The combination of fingerprint profiles within lysosomal vacuoles is a regular feature of blood lymphocytes from patients with neuronal ceroid lipofuscinosis type 3. {ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:9311735, ECO:0000269|PubMed:9490299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Seroid Lipofuksinoz, Nöronal, 3, 204200, Otozomal Resesif, CLN3 (CLN3 Hastalığı) (Prenatal) (CLN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 3, 204200, Autosomal recessive; CLN3 (CLN3 disease) (CLN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN3	CLN3, BTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 3 (CLN3) [MIM:204200]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent lipsosomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The hallmark of CLN3 is the ultrastructural pattern of lipopigment with a fingerprint profile, which can have 3 different appearances: pure within a lysosomal residual body; in conjunction with curvilinear or rectilinear profiles; and as a small component within large membrane-bound lysosomal vacuoles. The combination of fingerprint profiles within lysosomal vacuoles is a regular feature of blood lymphocytes from patients with neuronal ceroid lipofuscinosis type 3. (ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:9311735, ECO:0000269|PubMed:9490299). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 4, Parry Tipi, 162350, Otozomal Dominant; CLN4B (CLN4B Hastalığı) (DNAJC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350, Autosomal dominant; CLN4B (CLN4B disease) (DNAJC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC5	DNAJC5, DNAJCSA, CSP, CLN4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4B (CLN4B) [MIM:162350]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4B has no visual involvement and is characterized by seizures and other neurologic symptoms. (ECO:0000269|PubMed:21820099, ECO:0000269|PubMed:22073189, ECO:0000269|PubMed:22235333, ECO:0000269|PubMed:22978711). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 4, Parry Tipi, 162350, Otozomal Dominant; CLN4B (CLN4B Hastalığı) (Prenatal) (DNAJC5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 4, Parry type, 162350, Autosomal dominant; CLN4B (CLN4B disease) (DNAJC5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNAJC5	DNAJC5, DNAJCSA, CSP, CLN4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4B (CLN4B) [MIM:162350]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4B has no visual involvement and is characterized by seizures and other neurologic symptoms. (ECO:0000269|PubMed:21820099, ECO:0000269|PubMed:22073189, ECO:0000269|PubMed:22235333, ECO:0000269|PubMed:22978711). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 4a, Otozomal Resesif, Cln4a (CLN4A Hastalığı) (CLN6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CEROID LIPOFUSCINOSIS, NEURONAL, 4A, AUTOSOMAL RECESSIVE; CLN4A (CLN4A disease) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4A (CLN4A) [MIM:204300]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4A has no visual involvement and is characterized by progressive myoclonic epilepsy. (ECO:0000269|PubMed:21549341). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 4a, Otozomal Resesif, Cln4a (CLN4A Hastalığı) (Prenatal) (CLN6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CEROID LIPOFUSCINOSIS, NEURONAL, 4A, AUTOSOMAL RECESSIVE; CLN4A (CLN4A disease) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4A (CLN4A) [MIM:204300]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4A has no visual involvement and is characterized by progressive myoclonic epilepsy. (ECO:0000269|PubMed:21549341). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 5, 256731, Otozomal Resesif; CLN5 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (CLN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 5, 256731, Autosomal recessive; CLN5 (Late infantile neuronal ceroid lipofuscinosis) (CLN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN5	CLN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 5 (CLN5) [MIM:256731]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 5 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. (ECO:0000269|PubMed:15728307, ECO:0000269|PubMed:16814585, ECO:0000269|PubMed:17607606, ECO:0000269|PubMed:19309691, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:9662406). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 5, 256731, Otozomal Resesif; CLN5 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (CLN5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 5, 256731, Autosomal recessive; CLN5 (Late infantile neuronal ceroid lipofuscinosis) (CLN5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN5	CLN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 5 (CLN5) [MIM:256731]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposmal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 5 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. (ECO:0000269|PubMed:15728307, ECO:0000269|PubMed:16814585, ECO:0000269|PubMed:17607606, ECO:0000269|PubMed:19309691, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:9662406). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 6, 601780, Otozomal Resesif; CLN6 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (CLN6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 6, 601780, Autosomal recessive; CLN6 (Late infantile neuronal ceroid lipofuscinosis) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 6, 601780, Otozomal Resesif; CLN6 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (CLN6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 6, 601780, Autosomal recessive; CLN6 (Late infantile neuronal ceroid lipofuscinosis) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 7, 610951, Otozomal Resesif; CLN7 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (MFSD8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 7, 610951, Autosomal recessive; CLN7 (Late infantile neuronal ceroid lipofuscinosis) (MFSD8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFSD8	MFSD8, MGC33302, CLN7, CCMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 7, 610951, Otozomal Resesif; CLN7 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (MFSD8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 7, 610951, Autosomal recessive; CLN7 (Late infantile neuronal ceroid lipofuscinosis) (MFSD8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MFSD8	MFSD8, MGC33302, CLN7, CCMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 8, 600143, Otozomal Resesif; CLN8 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (CLN8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 8, 600143, Autosomal recessive; CLN8 (Late infantile neuronal ceroid lipofuscinosis) (CLN8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN8	CLN8, EPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 8 (CLN8) [MIM:600143]: A form of neuronal ceroid lipofuscinosis with onset in childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 8 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. (ECO:0000269|PubMed:15024724, ECO:0000269|PubMed:16570191, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:19431184, ECO:0000269|PubMed:21990111). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 8, 600143, Otozomal Resesif; CLN8 (Geç İnfantil Nöronal Seroid Lipofuksinozis) (Prenatal) (CLN8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 8, 600143, Autosomal recessive; CLN8 (Late infantile neuronal ceroid lipofuscinosis) (CLN8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN8	CLN8, EPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 8 (CLN8) [MIM:600143]: A form of neuronal ceroid lipofuscinosis with onset in childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 8 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. (ECO:0000269|PubMed:15024724, ECO:0000269|PubMed:16570191, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:19431184, ECO:0000269|PubMed:21990111). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, 8, Northern Epilepsi Varyantı, 610003, Otozomal Resesif (Progresif Epilepsi-Entelektüel Yetersizlik Sendromu, Fin Tipi) (CLN8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003, Autosomal recessive (Progressive epilepsy-intellectual disability syndrome, Finnish type) (CLN8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN8	CLN8, EPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 8 (CLN8) [MIM:600143]: A form of neuronal ceroid lipofuscinosis with onset in childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 8 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. (ECO:0000269|PubMed:15024724, ECO:0000269|PubMed:16570191, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:19431184, ECO:0000269|PubMed:21990111). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Seroid Lipofuksinoz, Nöronal, 8, Northern Epilepsi Varyantı, 610003, Otozomal Resesif (Progresif Epilepsi-Entelektüel Yetersizlik Sendromu, Fin Tipi) (Prenatal) (CLN8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, 8, Northern epilepsy variant, 610003, Autosomal recessive (Progressive epilepsy-intellectual disability syndrome, Finnish type) (CLN8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN8	CLN8, EPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 8 (CLN8) [MIM:600143]: A form of neuronal ceroid lipofuscinosis with onset in childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 8 comprise mixed combinations of granular, curvilinear, and fingerprint profiles. {ECO:0000269|PubMed:1502472A, ECO:0000269|PubMed:16570191, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:19431184, ECO:0000269|PubMed:21990111}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, Kufs Türü, Erişkin Başlangıçlı, 204300, Otozomal Resesif (CLN4A Hastalığı) (CLN6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300, Autosomal recessive (CLN4A disease) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4A (CLN4A) [MIM:204300]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4A has no visual involvement and is characterized by progressive myoclonic epilepsy. {ECO:0000269	PubMed:21549341}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seroid Lipofuksinoz, Nöronal, Kufs Türü, Erişkin Başlangıçlı, 204300, Otozomal Resesif (CLN4A Hastalığı) (Prenatal) (CLN6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ceroid lipofuscinosis, neuronal, Kufs type, adult onset, 204300, Autosomal recessive (CLN4A disease) (CLN6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLN6	CLN6, CLN4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ceroid lipofuscinosis, neuronal, 4A (CLN4A) [MIM:204300]: An adult-onset neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. CLN4A has no visual involvement and is characterized by progressive myoclonic epilepsy. {ECO:0000269	PubMed:21549341}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serviks Kanseri, Somatik, 603956 (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cervical cancer, somatic, 603956 (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Chanarin-Dorfman Sendromu, 275630, Otozomal Resesif; CDS (Nötral Lipid Depo Hastalığı) (Dorfman-Chanarin Hastalığı) ; (ABHD5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chanarin-Dorfman syndrome, 275630, Autosomal recessive; CDS (Neutral Lipid Storage Disorder) (Dorfman-Chanarin disease) (ABHD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABHD5	ABHD5, CGI58, IECN2, NCIE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chanarin-Dorfman syndrome (CDS) [MIM:275630]: An autosomal recessive inborn error of lipid metabolism with multisystemic accumulation of triglycerides although plasma concentrations are normal. Clinical characteristics are congenital generalized ichthyosis, vacuolated leukocytes, hepatomegaly, myopathy, cataracts, neurosensory hearing loss and developmental delay. The disorder presents at birth with generalized, fine, white scaling of the skin and a variable degree of erythema resembling non-bullous congenital ichthyosiform erythroderma. {ECO:0000269|PubMed:11590543, ECO:0000269|PubMed:17495960}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Chanarin-Dorfman Sendromu, 275630, Otozomal Resesif; CDS (Nötral Lipid Depo Hastalığı) (Dorfman-Chanarin Hastalığı) (Prenatal) (ABHD5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chanarin-Dorfman syndrome, 275630, Autosomal recessive; CDS (Neutral Lipid Storage Disorder) (Dorfman-Chanarin disease) (ABHD5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABHD5	ABHD5, CGI58, IECN2, NCIE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Chanarin-Dorfman syndrome (CDS) [MIM:275630]: An autosomal recessive inborn error of lipid metabolism with multisystemic accumulation of triglycerides although plasma concentrations are normal. Clinical characteristics are congenital generalized ichthyosis, vacuolated leukocytes, hepatomegaly, myopathy, cataracts, neurosensory hearing loss and developmental delay. The disorder presents at birth with generalized, fine, white scaling of the skin and a variable degree of erythema resembling non-bullous congenital ichthyosiform erythroderma. {ECO:0000269|PubMed:11590543, ECO:0000269|PubMed:17495960}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Char Sendromu, 169100, Otozomal Dominant; CHAR (Char Sendromu) (TFAP2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Char syndrome, 169100, Autosomal dominant; CHAR (Char syndrome) (TFAP2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFAP2B	TFAP2B, CHAR, PDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Char syndrome (CHAR) [MIM:169100]: An autosomal dominant disorder characterized by patent ductus arteriosus (PDA), facial dysmorphism and hand anomalies. {ECO:0000269|PubMed:10802654, ECO:0000269|PubMed:11505339}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Char Sendromu, 169100, Otozomal Dominant; CHAR (Char Sendromu) (Prenatal) (TFAP2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Char syndrome, 169100, Autosomal dominant; CHAR (Char syndrome) (TFAP2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TFAP2B	TFAP2B, CHAR, PDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Char syndrome (CHAR) [MIM:169100]: An autosomal dominant disorder characterized by patent ductus arteriosus (PDA), facial dysmorphism and hand anomalies. {ECO:0000269|PubMed:10802654, ECO:0000269|PubMed:11505339}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı ve Sağırlık(Charcot-Marie-Tooth Hastalığı Tip 1E) (CMT1E) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS (Charcot-Marie-Tooth disease type 1E) (CMT1E) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı ve Sağırlık(Charcot-Marie-Tooth Hastalığı Tip 1E) (CMT1E) (MLPA)	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS (Charcot-Marie-Tooth disease type 1E) (CMT1E) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı tip 2A2, Otozomal dominant (MFN2 1p36.22, MP2 1q23.3) (MLPA)	Charcot-Marie-Tooth Disease type 2A2, Autosomal dominant (MFN2 1p36.22, MP2 1q23.3) (MLPA)	MFN2 1p36.22, MP2 1q23.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, otozomal resesif demiyelinizan tip paneli (SH3TC2; NEFL, GDAP1; EGR2; SBF2; MTMR2; PRX) (MLPA)	Charcot-Marie-Tooth Disease, autosomal recessive demyelinating type panel (SH3TC2; NEFL; GDAP1; EGR2; SBF2; MTMR2; PRX	SH3TC2; NEFL; GDAP1; EGR2; SBF2; MTMR2; PRX	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Otozomal Dominant, Tip 2K, 607831, Otozomal Resesif, Otozomal Dominant; CMT2K (Otozomal Resesif Charcot-Marie-Tooth Hastalığı-Sea Kısırlığı) (JPH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, autosomal dominant, type 2K, 607831, Autosomal recessive, Autosomal dominant; CMT2K (Autosomal recessive Charcot-Marie-Tooth disease with hoarseness) (JPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JPH1	JPH1, JP1, CMT2K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2O, 614228, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2O) (DYNC1H1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2O, 614228, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2O) (DYNC1H1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYNC1H1	DYNC1H1, DNCL, DNECL, CMT20, MRD13, SMALED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2A2A, 609260, Otozomal Dominant; CMT2A2A (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2A2) (MFN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260, Autosomal dominant; CMT2A2A (Autosomal dominant Charcot-Marie-Tooth disease type 2A2) (MFN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2A2A, 609260, Otozomal Dominant; CMT2A2A (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2A2) (MLPA)	Charcot-Marie-Tooth disease, axonal, type 2A2A, 609260, Autosomal dominant; CMT2A2A (Autosomal dominant Charcot-Marie-Tooth disease type 2A2) (MLPA)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2A2B, 617087, Otozomal Resesif (Kalıtsal Motor ve Duyusal Nöropati Tip 6) (MFN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087, Autosomal recessive (Hereditary motor and sensory neuropathy type 6) (MFN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2A2B, 617087, Otozomal Resesif (Kalsital Motor ve Duyusal Nöropati Tip 6) (MLPA)	Charcot-Marie-Tooth disease, axonal, type 2A2B, 617087, Autosomal recessive (Hereditary motor and sensory neuropathy type 6) (MLPA)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2CC, 616924, Otozomal Dominant; CMT2CC (Amyotrofik Lateral Skleroz) (NEFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2CC, 616924, Autosomal dominant; CMT2CC (Amyotrophic lateral sclerosis) (NEFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEFH	NEFH, CMT2CC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2e, Cmt2e (Charcot-Marie-Tooth Hastalığı Tip 2E) (NEFL geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E; CMT2E (Autosomal dominant Charcot-Marie-Tooth disease type 2E) (NEFL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEFL	NEFL, CMT2E, CMT1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2F, 606595, Otozomal Dominant; CMT2F (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2F) (HSPB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2F, 606595, Autosomal dominant; CMT2F (Autosomal dominant Charcot-Marie-Tooth disease type 2F) (HSPB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPB1	HSPB1, HSP27, CMT2F, HMN2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2F (CMT2F) [MIM:606595]: A dominant axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. Onset of Charcot-Marie-Tooth disease type 2F is between 15 and 25 years with muscle weakness and atrophy usually beginning in feet and legs (peroneal distribution). Upper limb involvement occurs later. {ECO:0000269|PubMed:15122254, ECO:0000269|PubMed:22206013}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2i, Cmt2i (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2 I) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I; CMT2I (Autosomal dominant Charcot-Marie-Tooth disease type 2I) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2i, Cmt2i (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2I) (MLPA)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I; CMT2I (Autosomal dominant Charcot-Marie-Tooth disease type 2I) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2j, Cmt2j (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2 J) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J; CMT2J (Autosomal dominant Charcot-Marie-Tooth disease type 2J) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2j, Cmt2j (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2J) (MLPA)	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J; CMT2J (Autosomal dominant Charcot-Marie-Tooth disease type 2J) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2K, 607831, Otozomal Resesif, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2K) (GDAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2K, 607831, Autosomal recessive, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2K) (GDAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2K, 607831, Otozomal Resesif, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2K) (MLPA)	Charcot-Marie-Tooth disease, axonal, type 2K, 607831, Autosomal recessive, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2K) (MLPA)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2L, 608673, Otozomal Dominant, CMT2L (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2L) (HSPB8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2L, 608673, Autosomal dominant; CMT2L (Autosomal dominant Charcot-Marie-Tooth disease type 2L) (HSPB8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPB8	HSPB8, H11, E2IG1, DHMN2, CMT2L, HMN2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2M, 606482, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2M) (DNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2M, 606482, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2M) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, I.CCSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2N, 613287, Otozomal Dominant; CMT2N (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2N) (AARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2N, 613287, Autosomal dominant; CMT2N (Autosomal dominant Charcot-Marie-Tooth disease type 2N) (AARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AARS	AARS, CMT2N, EIEE29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2N (CMT2N) [MIM:613287]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. {ECO:0000269|PubMed:20045102, ECO:0000269|PubMed:22009580, ECO:0000269|PubMed:22206013}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2Q, 615025, Otozomal Dominant; CMT2Q (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2Q) (DHTKD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2Q, 615025, Autosomal dominant; CMT2Q (Autosomal dominant Charcot-Marie-Tooth disease type 2Q) (DHTKD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHTKD1	DHTKD1, KIAA1630, AMOXAD, CMT2Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2S, 616155, Otozomal Resesif; CMT2S (Charcot-Marie-Tooth Hastalığı Tip 2S) (IGHMBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2S, 616155, Autosomal recessive; CMT2S (Charcot-Marie-Tooth disease type 2S) (IGHMBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGHMBP2	IGHMBP2, SMUBP2, CATF1, SMARD1, HMN6, CMT2S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2S (CMT2S) [MIM:616155]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. {ECO:0000269|PubMed:25439726}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2T, 617017, Otozomal Resesif, Otozomal Dominant; CMT2T (Charcot-Marie-Tooth Hastalığı Tip 2T) (MME geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2T, 617017, Autosomal recessive, Autosomal dominant; CMT2T (Charcot-Marie-Tooth disease type 2T) (MME gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MME	MME, CD10, CALLA, NEP, CMT2T, SCA43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2U, 616280, Otozomal Dominant; CMT2U (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2U) (MARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2U, 616280, Autosomal dominant; CMT2U (Autosomal dominant Charcot-Marie-Tooth disease type 2U) (MARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARS	MARS, MTRNS, METRS, ILLD, CMT2U	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2V, 616491, Otozomal Dominant; CMT2V (Charcot-Marie-Tooth Hastalığı, Otozomal Dominant Tip 2V) (NAGLU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2V, 616491, Autosomal dominant; CMT2V (Autosomal dominant Charcot-Marie-Tooth disease type 2V) (NAGLU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGLU	NAGLU, MPS3B, CMT2V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2V (CMT2V) [MIM:616491]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. CMT2V is an autosomal dominant sensory neuropathy with late onset. The main clinical feature is recurrent leg pain that progresses to constant painful paraesthesias in the feet and later the hands. As it evolves, some patients develop a mild sensory ataxia. {ECO:0000269|PubMed:25818967}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2W, 616625, Otozomal Dominant; CMT2W (HARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2W, 616625, Autosomal dominant; CMT2W (HARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HARS	HARS, USH3B, CMT2W	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2W (CMT2W) [MIM:616625]: An autosomal dominant, axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. CMT2W patients manifest a peripheral neuropathy mainly affecting the lower limbs and resulting in gait difficulties and distal sensory impairment. Most patients also have upper limb involvement. {ECO:0000269|PubMed:22930593, ECO:0000269|PubMed:26072516}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2X, 616668, Otozomal Resesif; CMT2X (Otozomal Resesif Charcot-Marie-Tooth Hastalığı Tip 2X) (SPG11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2X, 616668, Autosomal recessive; CMT2X (Autosomal recessive Charcot Marie Tooth disease type 2X) (SPG11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG11	SPG11, KIAA1840, FLJ21439, ALS5, CMT2X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Tip 2Z, 616688, Otozomal Dominant, CMT2Z (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2Z) (MORC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, type 2Z, 616688, Autosomal dominant; CMT2Z (Autosomal dominant Charcot-Marie-Tooth disease type 2Z) (MORC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MORC2	MORC2, ZCW3, ZCWCC1, KIAA0852, CMT2Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Charcot-Marie-Tooth Hastalığı, Aksonal, Vokal Kord Parezisi, 607706, Otozomal Resesif (Otozomal Resesif Charcot-Marie-Tooth Hastalığı-Sesi Kısalığı) (GDAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706, Autosomal recessive (Autosomal recessive Charcot-Marie-Tooth disease with hoarseness) (GDAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Aksonal, Vokal Kord Parezisi, 607706, Otozomal Resesif (Otozomal Resesif Charcot-Marie-Tooth Hastalığı-Ses Kısalığı) (MLPA)	Charcot-Marie-Tooth disease, axonal, with vocal cord paresis, 607706, Autosomal recessive (Autosomal recessive Charcot-Marie-Tooth disease with hoarseness) (MLPA)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Demiyelinizan, Tip 1b; Cmt1b (Charcot-Marie-Tooth Hastalığı Tip 1B) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B; CMT1B (Charcot-Marie-Tooth disease type 1B) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Demiyelinizan, Tip 1b; Cmt1b (Charcot-Marie-Tooth Hastalığı Tip 1B) (MLPA)	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B; CMT1B (Charcot-Marie-Tooth disease type 1B) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form B, 606482, Otozomal Dominant; CMTDIB (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip B) (DNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, dominant intermediate B, 606482, Autosomal dominant; CMTDIB (Autosomal dominant intermediate Charcot-Marie-Tooth disease type B) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, LCCSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form C, 608323, Otozomal Dominant; CMTDIC (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip C) (YARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, dominant intermediate C, 608323, Autosomal dominant; CMTDIC (Autosomal dominant intermediate Charcot-Marie-Tooth disease type C) (YARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YARS	YARS, CMTDIC, TYRRS, YTS, YRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, dominant, intermediate type, C (CMTDIC) [MIM:608323]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. The dominant intermediate type C is characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:16429158}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form D, 607791, Otozomal Dominant; CMTDID (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip D) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, dominant intermediate D, 607791, Autosomal dominant; CMTDID (Autosomal dominant intermediate Charcot-Marie-Tooth disease type D) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form D, 607791, Otozomal Dominant; CMTDID (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip D) (MLPA)	Charcot-Marie-Tooth disease, dominant intermediate D, 607791, Autosomal dominant; CMTDID (Autosomal dominant intermediate Charcot-Marie-Tooth disease type D) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form E, 614455, Otozomal Dominant; CMTDIE (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip E) (INF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, dominant intermediate E, 614455, Autosomal dominant; CMTDIE (Autosomal dominant intermediate Charcot-Marie-Tooth disease type E) (INF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INF2	INF2, FSGSS, C14orf173, CMTDIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Dominant Intermediate Form F, 615185, Otozomal Dominant; CMTDIF (Otozomal Dominant Intermediate Charcot-Marie-Tooth Hastalığı Tip F) (GNB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, dominant intermediate F, 615185, Autosomal dominant; CMTDIF (Autosomal dominant intermediate Charcot-Marie-Tooth disease type F) (GNB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB4	GNB4, CMTD1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Ayak Deformitesi, 192950, Otozomal Dominant (HOXD10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, foot deformity of, 192950, Autosomal dominant (HOXD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD10	HOXD10, HOX4D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vertical talus, congenital (CVT) [MIM:192950]: A rare malformation characterized by vertical orientation of the talus with a rigid dorsal dislocation of the navicular, equinus deformity of the calcaneus, abduction deformity of the forefoot, and contracture of the soft tissues of the hind- and mid-foot. This condition is usually associated with multiple other congenital deformities and only rarely is an isolated deformity with familial occurrence. {ECO:0000269|PubMed:15146389}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Resesif Intermediate C, 615376, Otozomal Resesif; CMTRIC (Otozomal Resesif Intermediate Charcot-Marie-Tooth Hastalığı Tip C) (PLEKHG5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, recessive intermediate C, 615376, Autosomal recessive; CMTRIC (Autosomal recessive intermediate Charcot-Marie-Tooth disease type C) (PLEKHG5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEKHG5	PLEKHG5, KIAA0720, DSMAA, CMTRIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, recessive, intermediate type, C (CMTRIC) [MIM:615376]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:23777631, ECO:0000269|PubMed:23844677}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Resesif Dominant Intermediate Form D, 616039, Otozomal Resesif; CMTRID (Otozomal Resesif Intermediate Charcot-Marie-Tooth Hastalığı Tip D) (COX6A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, recessive intermediate D, 616039, Autosomal recessive; CMTRID (Autosomal recessive intermediate Charcot-Marie-Tooth disease type D) (COX6A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX6A1	COX6A1, CMTRID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, recessive, intermediate type, D (CMTRID) [MIM:616039]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:25152455}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Resesif Intermediate Form A, 608340, Otozomal Resesif (Otozomal Resesif Intermediate Charcot-Marie-Tooth Hastalığı Tip A) (GDAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340, Autosomal recessive (Autosomal recessive intermediate Charcot-Marie-Tooth disease type A) (GDAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Resesif Intermediate Form A, 608340, Otozomal Resesif (Otozomal Resesif Intermediate Charcot-Marie-Tooth Hastalığı Tip A) (MLPA)	Charcot-Marie-Tooth disease, recessive intermediate, A, 608340, Autosomal recessive (Autosomal recessive intermediate Charcot-Marie-Tooth disease type A) (MLPA)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Resesif Intermediate, B, 613641, Otozomal Resesif (Otozomal Resesif Intermediate Charcot-Marie-Tooth Hastalığı Tip B) (KARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, recessive intermediate, B, 613641, Autosomal recessive (Autosomal recessive intermediate Charcot-Marie-Tooth disease type B) (KARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KARS	KARS, CMTRIB, DFNB89	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, recessive, intermediate type, B (CMTRIB) [MIM:613641]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:20920668}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1A, 118220, Otozomal Dominant; CMT1A (Charcot-Marie-Tooth Hastalığı Tip 1A) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1A, 118220, Autosomal dominant; CMT1A (Charcot-Marie-Tooth disease type 1A) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Charcot-Marie-Tooth Hastalığı, Tip 1A, 118220, Otozomal Dominant; CMT1A (Charcot-Marie-Tooth Hastalığı Tip 1A) (MLPA)	Charcot-Marie-Tooth disease, type 1A, 118220, Autosomal dominant; CMT1A (Charcot-Marie-Tooth disease type 1A) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12407641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1B, 118200, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 1B) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1B, 118200, Autosomal dominant (Charcot-Marie-Tooth disease type 1B) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1B, 118200, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 1B) (MLPA)	Charcot-Marie-Tooth disease, type 1B, 118200, Autosomal dominant (Charcot-Marie-Tooth disease type 1B) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1C, 601098, Otozomal Dominant; CMT1C (Charcot-Marie-Tooth Hastalığı Tip 1C) (LITAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1C, 601098, Autosomal dominant; CMT1C (Charcot-Marie-Tooth disease type 1C) (LITAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LITAF	LITAF, CMT1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1C (CMT1C) [MIM:601098]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:12525712, ECO:0000269|PubMed:15776429, ECO:0000269|PubMed:15786462}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1D, 607678, Otozomal Dominant; CMT1D (Charcot-Marie-Tooth Hastalığı Tip 1D) (EGR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1D, 607678, Autosomal dominant; CMT1D (Charcot-Marie-Tooth disease type 1D) (EGR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGR2	EGR2, KROX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1D, 607678, Otozomal Dominant; CMT1D (Charcot-Marie-Tooth Hastalığı Tip 1D) (MLPA)	Charcot-Marie-Tooth disease, type 1D, 607678, Autosomal dominant; CMT1D (Charcot-Marie-Tooth disease type 1D) (MLPA)	EGR2	EGR2, KROX20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1E, 118300, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 1E) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1E, 118300, Autosomal dominant (Charcot-Marie-Tooth disease type 1E) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1E, 118300, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 1E) (MLPA)	Charcot-Marie-Tooth disease, type 1E, 118300, Autosomal dominant (Charcot-Marie-Tooth disease type 1E) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 1F, 607734, Otozomal Resesif, Otozomal Dominant; CMT1F (Charcot-Marie-Tooth Hastalığı Tipi 1F) (NEFL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 1F, 607734, Autosomal recessive, Autosomal dominant; CMT1F (Charcot-Marie-Tooth disease type 1F) (NEFL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEFL	NEFL, CMT2E, CMT1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2A1, 118210, Otozomal Dominant; CMT2A1 (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2A1) (KIF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2A1, 118210, Autosomal dominant; CMT2A1 (Autosomal dominant Charcot-Marie-Tooth disease type 2A1) (KIF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1B	KIF1B, CMT2A, CMT2A1, NBLST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2B, 600882, Otozomal Dominant; CMT2B (Charcot-Marie-Tooth Hastalığı Tip 2B) (RAB7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2B, 600882, Autosomal dominant; CMT2B (Autosomal dominant Charcot-Marie-Tooth disease type 2B) (RAB7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB7A	RAB7, CMT2B, PSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2B1, 605588, Otozomal Resesif; CMT2B1 (Charcot-Marie-Tooth Hastalığı Tip 2B1) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2B1, 605588, Autosomal recessive; CMT2B1 (Charcot-Marie-Tooth disease type 2B1) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2B1, 605588, Otozomal Resesif; CMT2B1 (Charcot-Marie-Tooth Hastalığı Tip 2B1) (MLPA)	Charcot-Marie-Tooth disease, type 2B1, 605588, Autosomal recessive; CMT2B1 (Charcot-Marie-Tooth disease type 2B1) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2B2, 605589, Otozomal Resesif; CMT2B2 (Charcot-Marie-Tooth Hastalığı Tip 2B2) (MED25 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2B2, 605589, Autosomal recessive; CMT2B2 (Charcot-Marie-Tooth disease type 2B2) (MED25 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED25	MED25, PTOV2, ARC92, CMT2B2, BVSYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2D, 601472, Otozomal Dominant; CMT2D (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tipi 2D) (GARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2D, 601472, Autosomal dominant; CMT2D (Autosomal dominant Charcot-Marie-Tooth disease type 2D) (GARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GARS	GARS, SMAD1, CMT2D, HMN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2E, 607684, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 2E) (NEFL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2E, 607684, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2E) (NEFL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEFL	NEFL, CMT2E, CMT1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2E (CMT2E) [MIM:607684]: A dominant axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. {ECO:0000269|PubMed:10841809, ECO:0000269|PubMed:11220745, ECO:0000269|PubMed:12481988, ECO:0000269|PubMed:15241803, ECO:0000269|PubMed:22206013}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2I, 607677, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2I) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2I, 607677, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2I) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2I, 607677, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2I) (MLPA)	Charcot-Marie-Tooth disease, type 2I, 607677, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2I) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2J, 607736, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2J) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2J, 607736, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2J) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2J, 607736, Otozomal Dominant (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2J) (MLPA)	Charcot-Marie-Tooth disease, type 2J, 607736, Autosomal dominant (Autosomal dominant Charcot-Marie-Tooth disease type 2J) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2R, 615490, Otozomal Resesif (Charcot-Marie-Tooth Hastalığı Tip 2R) (TRIM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2R, 615490, Autosomal recessive (Charcot-Marie-Tooth disease type 2R) (TRIM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIM2	TRIM2, KIAA0517, CMT2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2R (CMT2R) [MIM:615490]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. {ECO:0000269|PubMed:23562820}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 2Y, 616687, Otozomal Dominant; CMT2Y (Otozomal Dominant Charcot-Marie-Tooth Hastalığı Tip 2, VCP Mutasyonuna Bağlı) (VCP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 2Y, 616687, Autosomal dominant; CMT2Y (Autosomal dominant Charcot-Marie-Tooth disease type 2 due to VCP mutation) (VCP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCP	VCP, IBMPFD1, ALS14, CMT2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Charcot-Marie-Tooth Hastalığı, Tip 4A, 214400, Otozomal Resesif; CMT4A (Charcot-Marie-Tooth Hastalığı Tip 4A) (GDAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4A, 214400, Autosomal recessive; CMT4A (Charcot-Marie-Tooth disease type 4A) (GDAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4A (CMT4A) [MIM:214400]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. CMT4A is a severe form characterized by early age of onset and rapid progression leading to inability to walk in late childhood or adolescence. {ECO:0000269|PubMed:11743579, ECO:0000269|PubMed:12601710, ECO:0000269|PubMed:15772096, ECO:0000269|PubMed:16172208}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4A, 214400, Otozomal Resesif; CMT4A (Charcot-Marie-Tooth Hastalığı Tip 4A) (MLPA)	Charcot-Marie-Tooth disease, type 4A, 214400, Autosomal recessive; CMT4A (Charcot-Marie-Tooth disease type 4A) (MLPA)	GDAP1	GDAP1, CMT4A, CMT2K, CMTRIA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4A (CMT4A) [MIM:214400]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. CMT4A is a severe form characterized by early age of onset and rapid progression leading to inability to walk in late childhood or adolescence. {ECO:0000269|PubMed:11743579, ECO:0000269|PubMed:12601710, ECO:0000269|PubMed:15772096, ECO:0000269|PubMed:16172208}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4B1, 601382, Otozomal Resesif; CMT4B1 (Charcot-Marie-Tooth Hastalığı Tip 4B1) (MTMR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4B1, 601382, Autosomal recessive; CMT4B1 (Charcot-Marie-Tooth disease type 4B1) (MTMR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTMR2	MTMR2, CMT4B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4B1 (CMT4B1) [MIM:601382]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. {ECO:0000269|PubMed:10802647, ECO:0000269|PubMed:12398840}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4B1, 601382, Otozomal Resesif; CMT4B1 (Charcot-Marie-Tooth Hastalığı Tip 4B1) (MLPA)	Charcot-Marie-Tooth disease, type 4B1, 601382, Autosomal recessive; CMT4B1 (Charcot-Marie-Tooth disease type 4B1) (MLPA)	MTMR2	MTMR2, CMT4B1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4B1 (CMT4B1) [MIM:601382]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. {ECO:0000269|PubMed:10802647, ECO:0000269|PubMed:12398840}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4B2, 604563, Otozomal Resesif; CMT4B2 (Charcot-Marie-Tooth Hastalığı Tip 4B2) (SBF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4B2, 604563, Autosomal recessive; CMT4B2 (Charcot-Marie-Tooth disease type 4B2) (SBF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SBF2	SBF2, MTMR13, CMT4B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4B2, 604563, Otozomal Resesif; CMT4B2 (Charcot-Marie-Tooth Hastalığı Tip 4B2) (MLPA)	Charcot-Marie-Tooth disease, type 4B2, 604563, Autosomal recessive; CMT4B2 (Charcot-Marie-Tooth disease type 4B2) (MLPA)	SBF2	SBF2, MTMR13, CMT4B2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4B3, 615284, Otozomal Resesif; CMT4B3 (Charcot-Marie-Tooth Hastalığı Tip 4B3) (SBF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4B3, 615284, Autosomal recessive; CMT4B3 (Charcot-Marie-Tooth disease type 4B3) (SBF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SBF1	SBF1, MTMR5, CMT4B3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4B3 (CMT4B3) [MIM:615284]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. {ECO:0000269|PubMed:23749797}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4C, 601596, Otozomal Resesif; CMT4C (Charcot-Marie-Tooth Hastalığı Tip 4C) (SH3TC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4C, 601596, Autosomal recessive; CMT4C (Charcot-Marie-Tooth disease type 4C) (SH3TC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH3TC2	SH3TC2, KIAA1985, MNMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4C, 601596, Otozomal Resesif; CMT4C (Charcot-Marie-Tooth Hastalığı Tip 4C) (MLPA)	Charcot-Marie-Tooth disease, type 4C, 601596, Autosomal recessive; CMT4C (Charcot-Marie-Tooth disease type 4C) (MLPA)	SH3TC2	SH3TC2, KIAA1985, MNMN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4D, 601455, Otozomal Resesif; CMT4D (Charcot-Marie-Tooth Hastalığı Tip 4D) (NDRG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4D, 601455, Autosomal recessive; CMT4D (Charcot-Marie-Tooth disease type 4D) (NDRG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDRG1	NDRG1, HMSNL, CMT4D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4D (CMT4D) [MIM:601455]: A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. {ECO:0000269|PubMed:10831399}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4F, 614895, Otozomal Resesif; CMT4F (Charcot-Marie-Tooth Hastalığı Tip 4F) (PRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4F, 614895, Autosomal recessive; CMT4F (Charcot-Marie-Tooth disease type 4F) (PRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRX	PRX, CMT4F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4F, 614895, Otozomal Resesif; CMT4F (Charcot-Marie-Tooth Hastalığı Tip 4F) (MLPA)	Charcot-Marie-Tooth disease, type 4F, 614895, Autosomal recessive; CMT4F (Charcot-Marie-Tooth disease type 4F) (MLPA)	PRX	PRX, CMT4F	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Charcot-Marie-Tooth Hastalığı, Tip 4H, 609311, Otozomal Resesif; CMT4H (Charcot-Marie-Tooth Hastalığı Tip 4H) (FGD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4H, 609311, Autosomal recessive; CMT4H (Charcot-Marie-Tooth disease type 4H) (FGD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGD4	FGD4, FRABIN, CMT4H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 4H (CMT4H) [MIM:609311] A recessive demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. By convention autosomal recessive forms of demyelinating Charcot-Marie-Tooth disease are designated CMT4. {ECO:0000269|PubMed:17564959, ECO:0000269|PubMed:17564972}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4J, 611228, Otozomal Resesif; CMT4J (Charcot-Marie-Tooth Hastalığı Tip 4J) (FIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4J, 611228, Autosomal recessive; CMT4J (Charcot-Marie-Tooth disease type 4J) (FIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIG4	FIG4, KIAA0274, SAC3, ALS11, YVS, BTOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, Tip 4K, 616684, Otozomal Resesif; CMT4K (SURF1 İlişkili Charcot-Marie-Tooth Hastalığı Tip 4) (SURF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, type 4K, 616684, Autosomal recessive; CMT4K (SURF1-related Charcot-Marie-Tooth disease type 4) (SURF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SURF1	SURF1, CMT4K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, 300905, X'e Bağlı Dominant, 6, X'e Bağlı Dominant; CMTX6 (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 6) (PDK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, X-linked dominant, 6, 300905, X-linked dominant; CMTX6 (X-linked Charcot-Marie-Tooth disease type 6) (PDK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDK3	PDK3, CMTX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, X-linked dominant, 6 (CMTX6) [MIM:300905]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies characterized by severely reduced motor nerve conduction velocities (NCVs) (less than 38m/s) and segmental demyelination and remyelination, and primary peripheral axonal neuropathies characterized by normal or mildly reduced NCVs and chronic axonal degeneration and regeneration on nerve biopsy. {ECO:0000269	PubMed:23297365}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Charcot-Marie-Tooth Hastalığı, X'e Bağlı Resesif, 5, 311070, X'e Bağlı Resesif; CMTX5 (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 5) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth disease, X-linked recessive, 5, 311070, X-linked recessive; CMTX5 (X-linked Charcot-Marie-Tooth disease type 5) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Charcot-Marie-Tooth hastalığı tip 1A/ Ailesel basınca duyarlı nöropati (Kalıtsal neuropathy with liability to pressure palsies-HNPP) (CMT/HNPP bölgesi 17p12) (MLPA)	Charcot-Marie-Tooth hastalığı tip 1A/ Hereditary neuropathy with liability to pressure palsies-HNPP) (CMT / HNPP region 17p12) (MLPA)	CMT/HNPP bölgesi 17p12	.	MLPA	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Nöropati, X'e Bağlı Dominant 1, 302800, X'e Bağlı Dominant; CMTX1 (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 1) (GJB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800, X-linked dominant; CMTX1 (X-linked Charcot-Marie-Tooth disease type 1) (GJB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB1	GJB1, CX32, CMTX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Tooth Nöropati, X'e Bağlı Dominant 1, 302800, X'e Bağlı Dominant; CMTX1 (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 1) (MLPA)	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800, X-linked dominant; CMTX1 (X-linked Charcot-Marie-Tooth disease type 1) (MLPA)	GJB1	GJB1, CX32, CMTX1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Charcot-Marie-Toothe Hastalığı, Aksonal, Tip 2P, 614436, Otozomal Resesif, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 2P) (LRSAM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Charcot-Marie-Toothe disease, axonal, type 2P, 614436, Autosomal recessive, Autosomal dominant (Charcot-Marie-Tooth disease type 2P) (LRSAM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRSAM1	LRSAM1, TAL, RIFLE, CMT2P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2P (CMT2P) [MIM:614436]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. {ECO:0000269|PubMed:20865121, ECO:0000269|PubMed:22012984}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CHARGE Sendromu (8q12.1 Mikrodelesyonu) (FISH)	CHARGE syndrome (8q12.1 microdeletion) (FISH)	8q12.1	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
CHARGE Sendromu (8q12.1 Mikrodelesyonu) (FISH) (Prenatal)	CHARGE syndrome (8q12.1 microdeletion) (Prenatal) (FISH)	8q12.1	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (SEMA3E geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (SEMA3E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEMA3E	SEMA3E, SEMAH, KIAA0331	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (CHD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (CHD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHD7	CHD7, HH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (MLPA)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (MLPA)	CHD7	CHD7, HH5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (Prenatal) (SEMA3E geni) (Tüm Gen Dizi Analizi) (Prenatal)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (SEMA3E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEMA3E	SEMA3E, SEMAH, KIAA0331	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (Prenatal) (CHD7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (CHD7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHD7	CHD7, HH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CHARGE Sendromu, 214800, Otozomal Dominant (CHARGE Sendromu) (Prenatal) (MLPA)	CHARGE syndrome, 214800, Autosomal dominant (CHARGE syndrome) (Prenatal) (MLPA)	CHD7	CHD7, HH5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Chediak-Higashi Sendromu, 214500, Otozomal Resesif; CHS (Chédiak-Higashi Sendromu) (LYST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chediak-Higashi syndrome, 214500, Autosomal recessive; CHS (Chédiak-Higashi syndrome) (LYST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LYST	LYST, CHS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Chediak-Higashi Sendromu, 214500, Otozomal Resesif; CHS (Chédiak-Higashi Sendromu) (Prenatal) (LYST geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chediak-Higashi syndrome, 214500, Autosomal recessive; CHS (Chédiak-Higashi syndrome) (LYST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LYST	LYST, CHS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çerubizm 118400, Otozomal Dominant (Çerubizm) (SH3BP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cherubism, 118400, Autosomal dominant (Cherubism) (SH3BP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH3BP2	SH3BP2, CRPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cherubism (CRBM) [MIM:118400]: An autosomal dominant syndrome characterized by excessive bone degradation of the upper and lower jaws, which often begins around three years of age. It is followed by development of fibrous tissue masses, which causes a characteristic facial swelling. {ECO:0000269|PubMed:11381256, ECO:0000269|PubMed:12900899, ECO:0000269|PubMed:14577811}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Chilblain Lupus 2, 614415, Otozomal Dominant; CHBL2 (Chilblain Lupus) (SAMHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chilblain lupus 2, 614415, Autosomal dominant; CHBL2 (Chilblain lupus) (SAMHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAMHD1	SAMHD1, AGS5, DCIP, CHBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Chilblain Lupus, 610448, Otozomal Dominant; CHBL1 (Chilblain Lupus) (TREX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chilblain lupus, 610448, Autosomal dominant; CHBL1 (Chilblain lupus) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHILD Sendromu, 308050, X'e Bağlı Dominant (CHILD Sendromu) (NSDHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHILD syndrome, 308050, X-linked dominant (CHILD syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHILD Sendromu, 308050, X'e Bağlı Dominant (CHILD Sendromu) (Prenatal) (NSDHL geni) (Tüm Gen Dizi Analizi) (Prenatal)	CHILD syndrome, 308050, X-linked dominant (CHILD syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CHIME Sendromu, 280000, Otozomal Resesif (CHIME Sendromu) (PIGL geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHIME syndrome, 280000, Autosomal recessive (CHIME syndrome) (PIGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGL	PIGL, CHIME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation and ear anomalies syndrome (CHIME) [MIM:280000]: An extremely rare autosomal recessive multisystem disorder clinically characterized by colobomas, congenital heart defects, migratory ichthyosiform dermatosis, mental retardation, and ear anomalies including conductive hearing loss. Other clinical features include distinctive facial features, abnormal growth, genitourinary abnormalities, seizures, and feeding difficulties. {ECO:0000269	PubMed:22444671}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CHIME Sendromu, 280000, Otozomal Resesif (CHIME Sendromu) (Prenatal) (PIGL geni) (Tüm Gen Dizi Analizi) (Prenatal)	CHIME syndrome, 280000, Autosomal recessive (CHIME syndrome) (PIGL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGL	PIGL, CHIME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation and ear anomalies syndrome (CHIME) [MIM:280000]: An extremely rare autosomal recessive multisystem disorder clinically characterized by colobomas, congenital heart defects, migratory ichthyosiform dermatosis, mental retardation, and ear anomalies including conductive hearing loss. Other clinical features include distinctive facial features, abnormal growth, genitourinary abnormalities, seizures, and feeding difficulties. {ECO:0000269	PubMed:22444671}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kimerizm (FISH Analizi) (Aynı Cinsiyet)	Chimerism (FISH) (Same gender)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Kimerizm (STR Analizi) (Aynı ve Aynı Cinsiyet) (HER BİR BİREY İÇİN)	Chimerism (STR analysis)	.	.	STR analizi	EDTA'lı Kan (2-4 ml)		10	45	.	.	45	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow
Chitayat Sendromu, 617180, Otozomal Dominant (İzole Yonca Yaprağı Skalp Sendromu) (ERF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chitayat syndrome, 617180, Autosomal dominant (Isolated cloverleaf skull syndrome) (ERF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERF	ERF, PE2, CRS4, CHYTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Chitayat Sendromu, 617180, Otozomal Dominant (İzole Yonca Yaprağı Skalp Sendromu) (Prenatal) (ERF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chitayat syndrome, 617180, Autosomal dominant (Isolated cloverleaf skull syndrome) (ERF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERF	ERF, PE2, CRS4, CHYTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Koanal Atrezi ve Lenfödem, 613611, Otozomal Resesif (Lenfödem-Posterior Koanal Atrezi Sendromu) (PTPN14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Choanal atresia and lymphedema, 613611, Autosomal recessive (Lymphedema-posterior choanal atresia syndrome) (PTPN14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN14	PTPN14, PEZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Choanal atresia and lymphedema (CHATLY) [MIM:613611]: A disease characterized by posterior choanal atresia and lymphedema. Additional features are a high-arched palate, hypoplastic nipples, and mild pectus excavatum. {ECO:0000269	PubMed:20826270}. Note=The disease is caused by mutations affecting the gene represented in this entry. A homozygous deletion in PTPN14 predicted to result in frameshift and premature truncation, has been shown to be the cause of choanal atresia and lymphedema in one family.	EDTA Blood Sample
Koanal Atrezi ve Lenfödem, 613611, Otozomal Resesif (Lenfödem-Posterior Koanal Atrezi Sendromu) (Prenatal) (PTPN14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Choanal atresia and lymphedema, 613611, Autosomal recessive (Lymphedema-posterior choanal atresia syndrome) (PTPN14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPN14	PTPN14, PEZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Choanal atresia and lymphedema (CHATLY) [MIM:613611]: A disease characterized by posterior choanal atresia and lymphedema. Additional features are a high-arched palate, hypoplastic nipples, and mild pectus excavatum. {ECO:0000269	PubMed:20826270}. Note=The disease is caused by mutations affecting the gene represented in this entry. A homozygous deletion in PTPN14 predicted to result in frameshift and premature truncation, has been shown to be the cause of choanal atresia and lymphedema in one family.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Benign Tekrarlayıcı Intrahepatik, 2, 605479, Otozomal Resesif, BRIC2 (Benign Tekrarlayan Intrahepatik Kolestaz) (ABCB11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, benign recurrent intrahepatic, 2, 605479, Autosomal recessive; BRIC2 (Benign recurrent intrahepatic cholestasis) (ABCB11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB11	ABCB11, BSEP, SPGP, PFIC2, BRIC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolestaz, Benign Tekrarlayıcı Intrahepatik, 243300, Otozomal Resesif; BRIC1 (Benign Tekrarlayan Intrahepatik Kolestaz) (ATP8B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, benign recurrent intrahepatic, 243300, Autosomal recessive; BRIC1 (Benign recurrent intrahepatic cholestasis) (ATP8B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP8B1	ATP8B1, FIC1, BRIC, PFIC1, ICP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kolestaz, İntrahepatik, Gebelik, 1, 147480, Otozomal Dominant, ICP1 (Gebelik İntrahepatik Kolestazı) (ATP8B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, intrahepatic, of pregnancy, 1, 147480, Autosomal dominant; ICP1 (intrahepatic cholestasis of pregnancy) (ATP8B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP8B1	ATP8B1, FIC1, BRIC, PFIC1, ICP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cholestasis of pregnancy, intrahepatic 1 (ICP1) [MIM:147480]: A liver disorder of pregnancy. It presents during the second or, more commonly, the third trimester of pregnancy with intense pruritus which becomes more severe with advancing gestation and cholestasis. Cholestasis results from abnormal biliary transport from the liver into the small intestine. ICP1 causes fetal distress, spontaneous premature delivery and intrauterine death. ICP1 patients have spontaneous and progressive disappearance of cholestasis after delivery. {ECO:0000269|PubMed:15657619, ECO:0000269|PubMed:15888793}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kolestaz, İntrahepatik, Gebelik, 3, 614972, Otozomal Resesif, Otozomal Dominant (Gebelik İntrahepatik Kolestazı) (ABCB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, intrahepatic, of pregnancy, 3, 614972, Autosomal recessive, Autosomal dominant (Intrahepatic cholestasis of pregnancy) (ABCB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB4	ABCB4, PGY3, MDR3, ICP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolestaz, İntrahepatik, Gebelik, 3, 614972, Otozomal Resesif, Otozomal Dominant (Gebelik İntrahepatik Kolestazı) (MLPA)	Cholestasis, intrahepatic, of pregnancy, 3, 614972, Autosomal recessive, Autosomal dominant (Intrahepatic cholestasis of pregnancy) (MLPA)	ABCB4	ABCB4, PGY3, MDR3, ICP3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolestaz, Progresif Kanaliküler (VIL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive canalicular (VIL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VIL1	VIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Biliary atresia is a chronic and progressive cholestatic liver disease of childhood characterized by an abnormal villin gene expression and severe malformation of canalicular microvillus structure.	EDTA Blood Sample
Kolestaz, Progresif Kanalikuler (Prenatal), (VIL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive canalicular (VIL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VIL1	VIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Biliary atresia is a chronic and progressive cholestatic liver disease of childhood characterized by an abnormal villin gene expression and severe malformation of canalicular microvillus structure.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 1, 211600, Otozomal Resesif; PFIC1 (Progresif Ailesel İntrahepatik Kolestaz) (ATP8B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive familial intrahepatic 1, 211600, Autosomal recessive; PFIC1 (Progressive familial intrahepatic cholestasis) (ATP8B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP8B1	ATP8B1, FIC1, BRIC, PFIC1, ICP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cholestasis of pregnancy, intrahepatic 1 (ICP1) [MIM:147480]: A liver disorder of pregnancy. It presents during the second or, more commonly, the third trimester of pregnancy with intense pruritus which becomes more severe with advancing gestation and cholestasis. Cholestasis results from abnormal biliary transport from the liver into the small intestine. ICP1 causes fetal distress, spontaneous premature delivery and intrauterine death. ICP1 patients have spontaneous and progressive disappearance of cholestasis after delivery. {ECO:0000269|PubMed:15657619, ECO:0000269|PubMed:15888793}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 1, 211600, Otozomal Resesif; PFIC1 (Progresif Ailesel İntrahepatik Kolestaz) (Prenatal) (ATP8B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive familial intrahepatic 1, 211600, Autosomal recessive; PFIC1 (Progressive familial intrahepatic cholestasis) (ATP8B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP8B1	ATP8B1, FIC1, BRIC, PFIC1, ICP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cholestasis of pregnancy, intrahepatic 1 (ICP1) [MIM:147480]: A liver disorder of pregnancy. It presents during the second or, more commonly, the third trimester of pregnancy with intense pruritus which becomes more severe with advancing gestation and cholestasis. Cholestasis results from abnormal biliary transport from the liver into the small intestine. ICP1 causes fetal distress, spontaneous premature delivery and intrauterine death. ICP1 patients have spontaneous and progressive disappearance of cholestasis after delivery. {ECO:0000269|PubMed:15657619, ECO:0000269|PubMed:15888793}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 2, 601847, Otozomal Resesif; PFIC2 (Progresif Ailesel İntrahepatik Kolestaz) (ABCB11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive familial intrahepatic 2, 601847, Autosomal recessive; PFIC2 (Progressive familial intrahepatic cholestasis) (ABCB11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB11	ABCB11, BSEP, SPGP, PFIC2, BRIC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cholestasis, progressive familial intrahepatic 2 (PFIC2) [MIM:601847]: A disorder characterized by early onset of cholestasis that progresses to hepatic fibrosis, cirrhosis, and end-stage liver disease before adulthood. {ECO:0000269|PubMed:10579978, ECO:0000269|PubMed:11815775, ECO:0000269|PubMed:24969679, ECO:0000269|PubMed:9806540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 2, 601847, Otozomal Resesif; PFIC2 (Progresif Ailesel İntrahepatik Kolestaz) (Prenatal) (ABCB11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive familial intrahepatic 2, 601847, Autosomal recessive; PFIC2 (Progressive familial intrahepatic cholestasis) (ABCB11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCB11	ABCB11, BSEP, SPGP, PFIC2, BRIC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cholestasis, progressive familial intrahepatic 2 (PFIC2) [MIM:601847]: A disorder characterized by early onset of cholestasis that progresses to hepatic fibrosis, cirrhosis, and end-stage liver disease before adulthood. {ECO:0000269|PubMed:10579978, ECO:0000269|PubMed:11815775, ECO:0000269|PubMed:24969679, ECO:0000269|PubMed:9806540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 3, 602347, Otozomal Resesif; PFIC3 (Progresif Ailesel İntrahepatik Kolestaz) (ABCB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive familial intrahepatic 3, 602347, Autosomal recessive; PFIC3 (Progressive familial intrahepatic cholestasis) (ABCB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB4	ABCB4, PGY3, MDR3, ICP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 3, 602347, Otozomal Resesif; PFIC3 (Progresif Ailesel İntrahepatik Kolestaz) (MLPA)	Cholestasis, progressive familial intrahepatic 3, 602347, Autosomal recessive; PFIC3 (Progressive familial intrahepatic cholestasis) (MLPA)	ABCB4	ABCB4, PGY3, MDR3, ICP3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 3, 602347, Otozomal Resesif; PFIC3 (Progresif Ailesel İntrahepatik Kolestaz) (Prenatal) (ABCB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive familial intrahepatic 3, 602347, Autosomal recessive; PFIC3 (Progressive familial intrahepatic cholestasis) (ABCB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCB4	ABCB4, PGY3, MDR3, ICP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 3, 602347, Otozomal Resesif; PFIC3 (Progresif Ailesel İntrahepatik Kolestaz) (Prenatal) (MLPA)	Cholestasis, progressive familial intrahepatic 3, 602347, Autosomal recessive; PFIC3 (Progressive familial intrahepatic cholestasis) (Prenatal) (MLPA)	ABCB4	ABCB4, PGY3, MDR3, ICP3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 4, 615878, Otozomal Resesif; PFIC4 (Progresif Ailesel İntrahepatik Kolestaz) (TJP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive familial intrahepatic 4, 615878, Autosomal recessive; PFIC4 (Progressive familial intrahepatic cholestasis) (TJP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TJP2	TJP2, ZO2, PFIC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cholestasis, progressive familial intrahepatic 4 (PFIC4) [MIM:615878]: A disorder characterized by early onset of cholestasis that progresses to hepatic fibrosis, cirrhosis, and end-stage liver disease before adulthood. {ECO:0000269	PubMed:24614073}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 4, 615878, Otozomal Resesif; PFIC4 (Progresif Ailesel İntrahepatik Kolestaz) (Prenatal) (TJP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive familial intrahepatic 4, 615878, Autosomal recessive; PFIC4 (Progressive familial intrahepatic cholestasis) (TJP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TJP2	TJP2, ZO2, PFIC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cholestasis, progressive familial intrahepatic 4 (PFIC4) [MIM:615878]: A disorder characterized by early onset of cholestasis that progresses to hepatic fibrosis, cirrhosis, and end-stage liver disease before adulthood. {ECO:0000269	PubMed:24614073}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik 5, 617049, Otozomal Resesif; NR1H4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholestasis, progressive familial intrahepatic 5, 617049, Autosomal recessive (NR1H4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR1H4	NR1H4, FXR, RIP14, PFIC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kolestaz, Progresif Ailesel İntrahepatik, 5, 617049, Otozomal Resesif (Prenatal) (NR1H4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholestasis, progressive familial intrahepatic, 5, 617049, Autosomal recessive (NR1H4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR1H4	NR1H4, FXR, RIP14, PFIC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolesteril Ester Depo Hastalığı, 278000, Otozomal Resesif (Lizozomal Asit Lipaz Eksikliği) (LIPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cholesteryl ester storage disease, 278000, Autosomal recessive (Lysosomal acid lipase deficiency) (LIPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPA	LIPA, CESD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Wolman disease (WOD) [MIM:278000]: A severe manifestation of LIPA deficiency, leading to the accumulation of cholesteryl esters and triglycerides in most tissues of the body. WD occurs in infancy and is nearly always fatal before the age of 1 year. {ECO:0000269	PubMed:8146180}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kolesteril Ester Depo Hastalığı, 278000, Otozomal Resesif (Lizozomal Asit Lipaz Eksikliği) (Prenatal) (LIPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cholesteryl ester storage disease, 278000, Autosomal recessive (Lysosomal acid lipase deficiency) (LIPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIPA	LIPA, CESD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Wolman disease (WOD) [MIM:278000]: A severe manifestation of LIPA deficiency, leading to the accumulation of cholesteryl esters and triglycerides in most tissues of the body. WD occurs in infancy and is nearly always fatal before the age of 1 year. {ECO:0000269	PubMed:8146180}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrokalsinoz 2, 118600, Otozomal Dominant; CCAL2 (Ailesel Kalsiyum Pirofosfat Depotlayonu) (ANKH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrocalcinosis 2, 118600, Autosomal dominant; CCAL2 (Familial calcium pyrophosphate deposition) (ANKH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKH	ANKH, HANK, ANK, CMDJ, CCAL2, CPPDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrodisplazia Punktata, X'e Bağlı Dominant, 302960, X'e Bağlı Dominant; CDPX2 (X'e Bağlı Dominant Kondrodisplazia Punktata) (EBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia punctata, X-linked dominant, 302960, X-linked dominant; CDPX2 (X-linked dominant chondrodysplasia punctata) (EBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EBP	EBP, CDPX2, CPXD, CPX, MEND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrodisplazia Punktata, X'e Bağlı Dominant, 302960, X'e Bağlı Dominant; CDPX2 (X'e Bağlı Dominant Kondrodisplazia Punktata) (Prenatal) (EBP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia punctata, X-linked dominant, 302960, X-linked dominant; CDPX2 (X-linked dominant chondrodysplasia punctata) (EBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EBP	EBP, CDPX2, CPXD, CPX, MEND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kondrodisplazia Punktata, X'e Bağlı, 302950, X'e Bağlı CDPX1 (Brakitelefalanjik Kondrodisplazia Punktata) (ARSE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia punctata, X-linked recessive, 302950, X-linked recessive; CDPX1 (Brachytelephalangic chondrodysplasia punctata) (ARSE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARSE	ARSE, CDPX1, CDPXR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chondrodysplasia punctata 1, X-linked recessive (CDPX1) [MIM:302950]: A clinically and genetically heterogeneous disorder characterized by punctiform calcification of the bones. CDPX1 is a congenital defect of bone and cartilage development characterized by aberrant bone mineralization, severe underdevelopment of nasal cartilage, and distal phalangeal hypoplasia. This disease can also be induced by inhibition with the drug warfarin. (ECO:0000269|PubMed:12567415, ECO:0000269|PubMed:7720070, ECO:0000269|PubMed:9409863). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kondrodisplazia Punktata, X'e Bağlı, 302950, X'e Bağlı CDPX1 (Brakitelefalanjik Kondrodisplazia Punktata) (Prenatal) (ARSE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia punctata, X-linked recessive, 302950, X-linked recessive; CDPX1 (Brachytelephalangic chondrodysplasia punctata) (ARSE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARSE	ARSE, CDPX1, CDPXR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Chondrodysplasia punctata 1, X-linked recessive (CDPX1) [MIM:302950]: A clinically and genetically heterogeneous disorder characterized by punctiform calcification of the bones. CDPX1 is a congenital defect of bone and cartilage development characterized by aberrant bone mineralization, severe underdevelopment of nasal cartilage, and distal phalangeal hypoplasia. This disease can also be induced by inhibition with the drug warfarin. (ECO:0000269|PubMed:12567415, ECO:0000269|PubMed:7720070, ECO:0000269|PubMed:9409863). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrodisplazi, Ektem Displokasyonlar, GPAPP Tipi, 614078, Otozomal Resesif (Kondrodisplazi-Ektem Çıkıklar, Gpapp Tipi) (IMPAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia with joint dislocations, GPAPP type, 614078, Autosomal recessive (Chondrodysplasia with joint dislocations, gPAPP type) (IMPAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPAD1	IMPAD1, GPAPP, IMPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chondrodysplasia with joint dislocations, GPAPP type (CDP-GPAPP) [MIM:614078]: A condition consisting of congenital joint dislocations, chondrodysplasia with short stature, micrognathia and cleft palate, and a distinctive face. (ECO:0000269	PubMed:21548340). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kondrodisplazi, Ektem Displokasyonlar, GPAPP Tipi, 614078, Otozomal Resesif (Kondrodisplazi-Ektem Çıkıklar, Gpapp Tipi) (Prenatal) (IMPAD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia with joint dislocations, GPAPP type, 614078, Autosomal recessive (Chondrodysplasia with joint dislocations, gPAPP type) (IMPAD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IMPAD1	IMPAD1, GPAPP, IMPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Chondrodysplasia with joint dislocations, GPAPP type (CDP-GPAPP) [MIM:614078]: A condition consisting of congenital joint dislocations, chondrodysplasia with short stature, micrognathia and cleft palate, and a distinctive face. (ECO:0000269	PubMed:21548340). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrodisplazi, Platispondili, Belirgin Brakidaktili, Hidrosefali ve Mikroftalmi, 300863, X'e Bağlı Dominant (X'e Bağlı Dominant Kondrodisplazi, Chassaing-Lacombe Tipi) (HDAC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863, X-linked dominant (X-linked dominant chondrodysplasia, Chassaing-Lacombe type) (HDAC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HDAC6	HDAC6, CPBHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia (CDP-PBHM) [MIM:300863]: A disease characterized by chondrodysplasia, severe platyspondyly, hydrocephaly, and facial features with microphthalmia. Bone abnormalities include a distinctive metaphyseal cupping of the metacarpals, metatarsals, and phalanges. Affected females show a milder phenotype with small stature, sometimes associated with body asymmetry and mild mental retardation. (ECO:0000269	PubMed:20181727). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kondrodisplazi, Platispondili, Belirgin Brakidaktili, Hidrosefali ve Mikroftalmi, 300863, X'e Bağlı Dominant (X'e Bağlı Dominant Kondrodisplazi, Chassaing-Lacombe Tipi) (Prenatal) (HDAC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia, 300863, X-linked dominant (X-linked dominant chondrodysplasia, Chassaing-Lacombe type) (HDAC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HDAC6	HDAC6, CPBHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Chondrodysplasia with platyspondyly, distinctive brachydactyly, hydrocephaly, and microphthalmia (CDP-PBHM) [MIM:300863]: A disease characterized by chondrodysplasia, severe platyspondyly, hydrocephaly, and facial features with microphthalmia. Bone abnormalities include a distinctive metaphyseal cupping of the metacarpals, metatarsals, and phalanges. Affected females show a milder phenotype with small stature, sometimes associated with body asymmetry and mild mental retardation. (ECO:0000269	PubMed:20181727). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrodisplazi, Blomstrand Tipi, 215045, Otozomal Resesif; BOCD (Blomstrand Letal Kondrodisplazi) (PTH1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia, Blomstrand type, 215045, Autosomal recessive; BOCD (Blomstrand lethal chondrodysplasia) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrodisplazi, Blomstrand Tipi, 215045, Otozomal Resesif; BOCD (Blomstrand Letal Kondrodisplazi) (Prenatal) (PTH1R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia, Blomstrand type, 215045, Autosomal recessive; BOCD (Blomstrand lethal chondrodysplasia) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrodisplazia, Grebe Tipi, 200700, Otozomal Resesif (Akromezomelik Displazi, Grebe Tipi) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrodysplasia, Grebe type, 200700, Autosomal recessive (Acromesomelic dysplasia, Grebe type) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrodisplazia, Grebe Tipi, 200700, Otozomal Resesif (Akromezomelik Displazi, Grebe Tipi) (Prenatal) (GDF5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chondrodysplasia, Grebe type, 200700, Autosomal recessive (Acromesomelic dysplasia, Grebe type) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kondrosarkom, 215300, Otozomal Resesif (Kondrosarkoma) (EXT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrosarcoma, 215300, Autosomal recessive (Chondrosarcoma) (EXT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXT1	EXT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrosarkom, 215300, Otozomal Resesif (Kondrosarkoma) (MLPA)	Chondrosarcoma, 215300, Autosomal recessive (Chondrosarcoma) (MLPA)	EXT1	EXT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kondrosarkoma, İskelet Dışı Miksoid, 612237 (İskelet Dışı Miksoid Kondrosarkom) (TAF15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrosarcoma, extraskeletal myxoid, 612237 (Extraskeletal myxoid chondrosarcoma) (TAF15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF15	TAF15, TAF2N, RBP56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kondrosarkom, Ekstraskeletal Miksoid, 612237 (İskelet Dışı Miksoid Kondrosarkom) (NR4A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chondrosarcoma, extraskeletal myxoid, 612237 (Extraskeletal myxoid chondrosarcoma) (NR4A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR4A3	CSMF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHOPS Sendromu, 616368, Otozomal Dominant (Kognitif Bozukluk-Kaba Yüz-Kalp Defekti-Obezite-Pulmoner Tutulum-Boy Kısalığı-İskelet Displazisi Sendromu) (AFF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHOPS syndrome, 616368, Autosomal dominant (Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome) (AFF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFF4	AFF4, AF5Q31, CHOPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: CHOPS syndrome (CHOPS) [MIM:616368]: A syndrome characterized by cognitive impairment, coarse facies, heart defects, obesity, pulmonary involvement, short stature, and skeletal dysplasia. (ECO:0000269	PubMed:25730767). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CHOPS Sendromu, 616368, Otozomal Dominant (Kognitif Bozukluk-Kaba Yüz-Kalp Defekti-Obezite-Pulmoner Tutulum-Boy Kısalığı-İskelet Displazisi Sendromu) (Prenatal) (AFF4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CHOPS syndrome, 616368, Autosomal dominant (Cognitive impairment-coarse facies-heart defects-obesity-pulmonary involvement-short stature-skeletal dysplasia syndrome) (AFF4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AFF4	AFF4, AF5Q31, CHOPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: CHOPS syndrome (CHOPS) [MIM:616368]: A syndrome characterized by cognitive impairment, coarse facies, heart defects, obesity, pulmonary involvement, short stature, and skeletal dysplasia. (ECO:0000269	PubMed:25730767). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kore, Çocuklukta Başlayan, Psikomotor Retardasyon, 616939, Otozomal Resesif; COCPMR (GPR88 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chorea, childhood-onset, with psychomotor retardation, 616939, Autosomal recessive; COCPMR (GPR88 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR88	GPR88, STRG, COCPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kore, Kalıtsal İyi Huylu, 118700, Otozomal Dominant; BHC (Benign Aliesel Kore) (NKX2-1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chorea, hereditary benign, 118700, Autosomal dominant; BHC (Benign familial chorea) (NKX2-1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chorea, hereditary benign (BHC) [MIM:118700]: A rare autosomal dominant movement disorder, defined by early onset in childhood, a stable or non-progressive course of chorea, and no mental deterioration. Chorea is characterized by involuntary, forcible, rapid, jerky movements that may be subtle or become confluent, markedly altering normal patterns of movement. (ECO:0000269|PubMed:11971878, ECO:0000269|PubMed:15955952). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kore, Kalıtsal İyi Huylu, 118700, Otozomal Dominant; BHC (Benign Aliesel Kore) (MLPA)	Chorea, hereditary benign, 118700, Autosomal dominant; BHC (Benign familial chorea) (MLPA)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Chorea, hereditary benign (BHC) [MIM:118700]: A rare autosomal dominant movement disorder, defined by early onset in childhood, a stable or non-progressive course of chorea, and no mental deterioration. Chorea is characterized by involuntary, forcible, rapid, jerky movements that may be subtle or become confluent, markedly altering normal patterns of movement. (ECO:0000269|PubMed:11971878, ECO:0000269|PubMed:15955952). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Koreoakantositozis, 200150, Otozomal Resesif; CHAC (Koreoakantositoz) (VPS13A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Choreoacanthocytosis, 200150, Autosomal recessive; CHAC (Choreoacanthocytosis) (VPS13A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS13A	VPS13A, CHAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koreoatetoz, Hipotiroidizm ve Neonatal Solunum Zorluğu, 610978, Otozomal Dominant (Beyin, Akciğer-Tiroid Sendromu) (NKX2-1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978, Autosomal dominant (Brain-lung-thyroid syndrome) (NKX2-1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koreoatetoz, Hipotiroidizm ve Neonatal Solunum Zorluğu, 610978, Otozomal Dominant (Beyin, Akciğer-Tiroid Sendromu) (Postnatal)	Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978, Autosomal dominant (Brain-lung-thyroid syndrome) (NKX2-1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Koreoatetoz, Hipotiroidizm ve Neonatal Solunum Zorluğu, 610978, Otozomal Dominant (Beyin, Akciğer-Tiroid Sendromu) (Prenatal) (NKX2-1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978, Autosomal dominant (Brain-lung-thyroid syndrome) (NKX2-1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koreoatetoz, Hipotiroidizm ve Neonatal Solunum Zorluğu, 610978, Otozomal Dominant (Beyin, Akciğer-Tiroid Sendromu) (Prenatal) (MLPA)	Choreoathetosis, hypothyroidism, and neonatal respiratory distress, 610978, Autosomal dominant (Brain-lung-thyroid syndrome) (Prenatal) (MLPA)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koroidal Distrofi, Santral Areolar 2, 613105, Otozomal Dominant; CACD2 (Santral Areolar Koroidal Distrofi) (PRPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Choroidal dystrophy, central areolar 2, 613105, Autosomal dominant; CACD2 (Central areolar choroidal dystrophy) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroidal Distrofi, Santral Areolar 2, 613105, Otozomal Dominant; CACD2 (Santral Areolar Koroidal Distrofi) (Prenatal) (PRPH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Choroidal dystrophy, central areolar 2, 613105, Autosomal dominant; CACD2 (Central areolar choroidal dystrophy) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koryonik Villus Örneği (CVS) Kromozom Analizi	Chorionic Villus Sample (CVS)- Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Merkezimizden temin edilen transport besi yeri içinde	*	.	.	15	30	20	Prenatal	.	Transport Medium suppliedfrom our center
Koroid Pleksus Papillomu, 260500, Otozomal Dominant (Koroid Pleksus Karsinomu) (MLPA)	Choroid plexus papilloma, 260500, Autosomal dominant (Choroid plexus carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Koroideremi, 303100, X'e Bağlı Dominant; CHM (Koroideremi) (CHM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Choroideremia, 303100, X-linked dominant; CHM (Choroideremia) (CHM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHM	CHM, TCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Choroideremia (CHM) [MIM:303100]: An X-linked recessive disease characterized by a slowly progressive degeneration of the choroid, photoreceptors, and retinal pigment epithelium. Affected males develop night blindness in their teenage years followed by loss of peripheral vision and complete blindness at middle age. Carrier females are generally asymptomatic but funduscopic examination often shows patchy areas of chorioretinal atrophy. (ECO:0000269|PubMed:19427510, ECO:0000269|PubMed:21905166, ECO:0000269|PubMed:7951216). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koroideremi, Sağırlık ve Mental Retardasyon, 303110, X'e Bağlı Resesif (Koroideremi-Sağırlık-Obezite Sendromu) (2200)	Choroideremia, deafness, and mental retardation, 303110, X-linked recessive (Choroideremia-deafness-obesity syndrome) (440)	.	DELXq21, CXDELq21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Koroideremi, Sağırlık ve Mental Retardasyon, 303110, X'e Bağlı Resesif (Koroideremi-Sağırlık-Obezite Sendromu) (Prenatal)	Choroideremia, deafness, and mental retardation, 303110, X-linked recessive (Choroideremia-deafness-obesity syndrome) (Prenatal)	.	DELXq21, CXDELq21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 10q22.3-Q23.2 Delesyon Sendromu, 612242 (Jüvenil Polipozis Sendromu) (2200)	Chromosome 10q22.3-q23.2 deletion syndrome, 612242 (Juvenile polyposis syndrome) (440)	.	DEL10q23, C10DELq23	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 10q22.3-Q23.2 Delesyon Sendromu, 612242 (Jüvenil Polipozis Sendromu) (Prenatal)	Chromosome 10q22.3-q23.2 deletion syndrome, 612242 (Juvenile polyposis syndrome) (Prenatal)	.	DEL10q23, C10DELq23	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 10q26 Delesyon Sendromu, 609625, Otozomal Dominant (Distal Monozomi 10q) (2200)	Chromosome 10q26 deletion syndrome, 609625, Autosomal dominant (Distal monosomy 10q) (440)	.	DEL10q26, C10q26DEL	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 10q26 Delesyon Sendromu, 609625, Otozomal Dominant (Distal Monozomi 10q) (Prenatal)	Chromosome 10q26 deletion syndrome, 609625, Autosomal dominant (Distal monosomy 10q) (Prenatal)	.	DEL10q26, C10q26DEL	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 11p13 Delesyon Sendromu, Distal, 616902, Otozomal Dominant (2200)	Chromosome 11p13 deletion syndrome, distal, 616902, Autosomal dominant (440)	.	DEL11p13, C11DELp13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 11p13 Delesyon Sendromu, Distal, 616902, Otozomal Dominant (Prenatal)	Chromosome 11p13 deletion syndrome, distal, 616902, Autosomal dominant (Prenatal)	.	DEL11p13, C11DELp13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 11p15-P14 Delesyon Sendromu, 606528, Otozomal Resesif (2200)	Chromosome 11p15-p14 deletion syndrome, 606528, Autosomal recessive (440)	.	DEL11p15p14, C11DELp15p14	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 11p15-P14 Delesyon Sendromu, 606528, Otozomal Resesif (Prenatal)	Chromosome 11p15-p14 deletion syndrome, 606528, Autosomal recessive (Prenatal)	.	DEL11p15p14, C11DELp15p14	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
13/ 21 Kromozom Alfa Satellit (FISH)	Chromosome 13/21 Alpha Satellite (FISH)	13cent-21cent	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
13/ 21 Kromozom Alfa Satellit (FISH) (Prenatal)	Chromosome 13/21 Alpha Satellite (FISH) (Prenatal)	13cent-21cent	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 13q14 Delesyon Sendromu, 613884, Otozomal Dominant, Sporadik Vakalar (Monozomi 13q14) (2200)	Chromosome 13q14 deletion syndrome, 613884, Autosomal dominant, Isolated cases (Monosomy 13q14) (440)	.	DEL13q14, C13DELq14	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 13q14 Delesyon Sendromu, 613884, Otozomal Dominant, Sporadik Vakalar (Monozomi 13q14) (Prenatal)	Chromosome 13q14 deletion syndrome, 613884, Autosomal dominant, Isolated cases (Monosomy 13q14) (Prenatal)	.	DEL13q14, C13DELq14	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
14/ 22 Kromozom Alfa Satellit (FISH)	Chromosome 14/22 Alpha Satellite (FISH)	14cent-22cent	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
14/ 22 Kromozom Alfa Satellit (FISH) (Prenatal)	Chromosome 14/22 Alpha Satellite (FISH) (Prenatal)	14cent-22cent	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 14q11-Q22 Delesyon Sendromu, 613457, Sporadik Vakalar (14q11.2 Mikrodelesyon Sendromu) (2200)	Chromosome 14q11-q22 deletion syndrome, 613457, Isolated cases (14q11.2 microdeletion syndrome) (440)	.	DEL14q11q22, C14DELq11q22	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 14q11-Q22 Delesyon Sendromu, 613457, Sporadik Vakalar (14q11.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 14q11-q22 deletion syndrome, 613457, Isolated cases (14q11.2 microdeletion syndrome) (Prenatal)	.	DEL14q11q22, C14DELq11q22	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
15. Kromozom Alfa Satellit (FISH)	Chromosome 15 Alpha Satellite (FISH)	15cent	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
15. Kromozom Alfa Satellit (FISH) (Prenatal)	Chromosome 15 Alpha Satellite (FISH) (Prenatal)	15cent	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 15q11.2 Delesyon Sendromu, 615656, Otozomal Dominant (15q11.2 Mikrodelesyon Sendromu) (2200)	Chromosome 15q11.2 deletion syndrome, 615656, Autosomal dominant (15q11.2 microdeletion syndrome) (440)	.	DEL15q11.2, C15DELq11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 15q11.2 Delesyon Sendromu, 615656, Otozomal Dominant (15q11.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 15q11.2 deletion syndrome, 615656, Autosomal dominant (15q11.2 microdeletion syndrome) (Prenatal)	.	DEL15q11.2, C15DELq11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 15q13.3 Mikrodelesyon Sendromu, 612001 (15q13.3 Mikrodelesyon Sendromu) (2200)	Chromosome 15q13.3 microdeletion syndrome, 612001 (15q13.3 microdeletion syndrome) (440)	.	DEL15q13.3, MICRODEL15q13.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 15q13.3 Mikrodelesyon Sendromu, 612001 (15q13.3 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 15q13.3 microdeletion syndrome, 612001 (15q13.3 microdeletion syndrome) (Prenatal)	.	DEL15q13.3, MICRODEL15q13.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 15q14 Delesyon Sendromu, 616898, Otozomal Dominant (15q14 Mikrodelesyon Sendromu) (2200)	Chromosome 15q14 deletion syndrome, 616898, Autosomal dominant (15q14 microdeletion syndrome) (440)	.	DEL15q14, C15DELq14	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 15q14 Delesyon Sendromu, 616898, Otozomal Dominant (15q14 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 15q14 deletion syndrome, 616898, Autosomal dominant (15q14 microdeletion syndrome) (Prenatal)	.	DEL15q14, C15DELq14	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 15q25 Delesyon Sendromu, 614294, Otozomal Dominant (2200)	Chromosome 15q25 deletion syndrome, 614294, Autosomal dominant (440)	.	DEL15q25, C15DELq25	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 15q25 Delesyon Sendromu, 614294, Otozomal Dominant (Prenatal)	Chromosome 15q25 deletion syndrome, 614294, Autosomal dominant (Prenatal)	.	DEL15q25, C15DELq25	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 15q26-Qter Delesyon Sendromu, 612626, Sporadik Vakalar (Distal Monozomi 15Q) (2200)	Chromosome 15q26-qter deletion syndrome, 612626, Isolated cases (Distal monosomy 15q) (440)	.	DEL15q26qter, C15DELq26qter	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 15q26-Qter Delesyon Sendromu, 612626, Sporadik Vakalar (Distal Monozomi 15Q) (Prenatal)	Chromosome 15q26-qter deletion syndrome, 612626, Isolated cases (Distal monosomy 15q) (Prenatal)	.	DEL15q26qter, C15DELq26qter	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p11.2 Delesyon Sendromu, 220kb, 613444 (Distal 16p11.2 Mikrodelesyon Sendromu) (2200)	Chromosome 16p11.2 deletion syndrome, 220kb, 613444 (Distal 16p11.2 microdeletion syndrome) (440)	.	BMIQ16, DEL16p11.2, C16DELp11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p11.2 Delesyon Sendromu, 220kb, 613444 (Distal 16p11.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 16p11.2 deletion syndrome, 220kb, 613444 (Distal 16p11.2 microdeletion syndrome) (Prenatal)	.	BMIQ16, DEL16p11.2, C16DELp11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p11.2 Delesyon Sendromu, 593kb, 611913 (Proksimal 16p11.2 Mikrodelesyon Sendromu) (2200)	Chromosome 16p11.2 deletion syndrome, 593kb, 611913 (Proximal 16p11.2 microdeletion syndrome) (440)	.	DEL16p11.2, C16DELp11.2, AUTS14A	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p11.2 Delesyon Sendromu, 593kb, 611913 (Proksimal 16p11.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 16p11.2 deletion syndrome, 593kb, 611913 (Proximal 16p11.2 microdeletion syndrome) (Prenatal)	.	DEL16p11.2, C16DELp11.2, AUTS14A	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p11.2 Duplikasyon Sendromu, 614671 (Proksimal 16p11.2 Mikroduplikasyon Sendromu) (2200)	Chromosome 16p11.2 duplication syndrome, 614671 (Proximal 16p11.2 microduplication syndrome) (440)	.	DUP16p11.2, C16DUPp11.2, AUTS14B	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p11.2 Duplikasyon Sendromu, 614671 (Proksimal 16p11.2 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 16p11.2 duplication syndrome, 614671 (Proximal 16p11.2 microduplication syndrome) (Prenatal)	.	DUP16p11.2, C16DUPp11.2, AUTS14B	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p12.1 Delesyon Sendromu, 520kb, 136570 (2200)	Chromosome 16p12.1 deletion syndrome, 520kb, 136570 (440)	.	DEL16p12.1, C16DELp12.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p12.1 Delesyon Sendromu, 520kb, 136570 (Prenatal)	Chromosome 16p12.1 deletion syndrome, 520kb, 136570 (Prenatal)	.	DEL16p12.1, C16DELp12.1	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p12.2-P11.2 Delesyon Sendromu, 613604, Sporadik Vakalar (16p11.2p12.2 Mikrodelesyon Sendromu) (2200)	Chromosome 16p12.2-p11.2 deletion syndrome, 613604, Isolated cases (16p11.2p12.2 microdeletion syndrome) (440)	.	DEL16p12.1p11.2, C16DELp12.1p11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p12.2-P11.2 Delesyon Sendromu, 613604, Sporadik Vakalar (16p11.2p12.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 16p12.2-p11.2 deletion syndrome, 613604, Isolated cases (16p11.2p12.2 microdeletion syndrome) (Prenatal)	.	DEL16p12.1p11.2, C16DELp12.1p11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p13.2 Delesyon Sendromu, 616863, Otozomal Dominant (2200)	Chromosome 16p13.2 deletion syndrome, 616863, Autosomal dominant (440)	.	DEL16p13.2, C16DELp13.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p13.2 Delesyon Sendromu, 616863, Otozomal Dominant (Prenatal)	Chromosome 16p13.2 deletion syndrome, 616863, Autosomal dominant (Prenatal)	.	DEL16p13.2, C16DELp13.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p13.3 Delesyon Sendromu, 610543 (Rubinstein-Taybi Sendromu) (2200)	Chromosome 16p13.3 deletion syndrome, 610543 (Rubinstein-Taybi syndrome) (440)	.	DEL16p13.3, RSTSS	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p13.3 Delesyon Sendromu, 610543 (Rubinstein-Taybi Sendromu) (Prenatal)	Chromosome 16p13.3 deletion syndrome, 610543 (Rubinstein-Taybi syndrome) (Prenatal)	.	DEL16p13.3, RSTSS	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p13.3 Duplikasyon Sendromu, 613458, Otozomal Dominant, Sporadik Vakalar (16p13.3 Mikroduplikasyon Sendromu) (2200)	Chromosome 16p13.3 duplication syndrome, 613458, Autosomal dominant, Isolated cases (16p13.3 microduplication syndrome) (440)	.	DUP16p13.3, C16DUPq13.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p13.3 Duplikasyon Sendromu, 613458, Otozomal Dominant, Sporadik Vakalar (16p13.3 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 16p13.3 duplication syndrome, 613458, Autosomal dominant, Isolated cases (16p13.3 microduplication syndrome) (Prenatal)	.	DUP16p13.3, C16DUPq13.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 16p22 Delesyon Sendromu, 614541, Sporadik Vakalar (2200)	Chromosome 16p22 deletion syndrome, 614541, Isolated cases (440)	.	C16DELq22, DEL16q22	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 16p22 Delesyon Sendromu, 614541, Sporadik Vakalar (Prenatal)	Chromosome 16p22 deletion syndrome, 614541, Isolated cases (Prenatal)	.	C16DELq22, DEL16q22	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17p13.1 Delesyon Sendromu, 613776, Otozomal Dominant (2200)	Chromosome 17p13.1 deletion syndrome, 613776, Autosomal dominant (440)	.	DEL17p13.1, C17DELp13.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 17p13.1 Delesyon Sendromu, 613776, Otozomal Dominant (Prenatal)	Chromosome 17p13.1 deletion syndrome, 613776, Autosomal dominant (Prenatal)	.	DEL17p13.1, C17DELp13.1	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17p13.3 Duplikasyon Sendromu, 613215 (17p13.3 Mikroduplikasyon Sendromu) (2200)	Chromosome 17p13.3 duplication syndrome, 613215 (17p13.3 microduplication syndrome) (440)	.	DUP17p13.3, C17DUPp13.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 17p13.3 Duplikasyon Sendromu, 613215 (17p13.3 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 17p13.3 duplication syndrome, 613215 (17p13.3 microduplication syndrome) (Prenatal)	.	DUP17p13.3, C17DUPp13.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
KROMOZOM 17p13.3, Telomerik, DUPLİKASYON SENDROMU (Tibiyal Aplazi-Ektrodaktili Sendromu) (2200)	CHROMOSOME 17p13.3, TELOMERIC, DUPLICATION SYNDROME (Tibial aplasia-ectrodactyly syndrome) (440)	.	SHFLD3, DUP17p13.3, C17DUPp13.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
KROMOZOM 17p13.3, Telomerik, DUPLİKASYON SENDROMU (Tibiyal Aplazi-Ektrodaktili Sendromu) (Prenatal)	CHROMOSOME 17p13.3, TELOMERIC, DUPLICATION SYNDROME (Tibial aplasia-ectrodactyly syndrome) (Prenatal)	.	SHFLD3, DUP17p13.3, C17DUPp13.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17q11.2 Delesyon Sendromu, 1.4Mb, 613675, Otozomal Dominant (Nörofibromatozis Tip 1) (2200)	Chromosome 17q11.2 deletion syndrome, 1.4Mb, 613675, Autosomal dominant (Neurofibromatosis type 1) (440)	.	DEL17q11.2, C17DELq11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 17q11.2 Delesyon Sendromu, 1.4Mb, 613675, Otozomal Dominant (Nörofibromatozis Tip 1) (Prenatal)	Chromosome 17q11.2 deletion syndrome, 1.4Mb, 613675, Autosomal dominant (Neurofibromatosis type 1) (Prenatal)	.	DEL17q11.2, C17DELq11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17q12 Delesyon Sendromu, 614527, Otozomal Dominant (17q12 Mikrodelesyon Sendromu) (2200)	Chromosome 17q12 deletion syndrome, 614527, Autosomal dominant (17q12 microdeletion syndrome) (440)	.	DEL17q12, C17DELq12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 17q12 Delesyon Sendromu, 614527, Otozomal Dominant (17q12 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 17q12 deletion syndrome, 614527, Autosomal dominant (17q12 microdeletion syndrome) (Prenatal)	.	DEL17q12, C17DELq12	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kromozom 17q12 Duplikasyon Sendromu, 614526, Otozomal Dominant (17q12 Mikroduplikasyon Sendromu) (2200)	Chromosome 17q12 duplication syndrome, 614526, Autosomal dominant (17q12 microduplication syndrome) (440)		DUP17q12, C17DUPq12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 17q12 Duplikasyon Sendromu, 614526, Otozomal Dominant (17q12 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 17q12 duplication syndrome, 614526, Autosomal dominant (17q12 microduplication syndrome) (Prenatal)		DUP17q12, C17DUPq12	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17q21.31 Duplikasyon Sendromu, 613533 (17q21.31 Mikroduplikasyon Sendromu) (2200)	Chromosome 17q21.31 duplication syndrome, 613533 (17q21.31 microduplication syndrome) (440)		DUP17q21.31, C17DUPq21.31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 17q21.31 Duplikasyon Sendromu, 613533 (17q21.31 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 17q21.31 duplication syndrome, 613533 (17q21.31 microduplication syndrome) (Prenatal)		DUP17q21.31, C17DUPq21.31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17q23.1-Q23.2 Delesyon Sendromu, 613355, Sporadik Vakalar (17q23.1q23.2 Mikrodelesyon Sendromu) (2200)	Chromosome 17q23.1-q23.2 deletion syndrome, 613355, Isolated cases (17q23.1q23.2 microdeletion syndrome) (440)		DEL17q23.1q23.2, C17DELq23.1q23.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 17q23.1-Q23.2 Delesyon Sendromu, 613355, Sporadik Vakalar (17q23.1q23.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 17q23.1-q23.2 deletion syndrome, 613355, Isolated cases (17q23.1q23.2 microdeletion syndrome) (Prenatal)		DEL17q23.1q23.2, C17DELq23.1q23.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 17q23.1-Q23.3 Duplikasyon Sendromu, 613618, Otozomal Dominant (Ailesel Çarpık Ayak ± Eşlik Eden Alt Ekstremite Anomalileri) (2200)	Chromosome 17q23.1-q23.3 duplication syndrome, 613618, Autosomal dominant (Familial clubfoot with or without associated lower limb anomalies) (440)		DUP17q23.1q23.2, C17DUPq23.1q23.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 17q23.1-Q23.3 Duplikasyon Sendromu, 613618, Otozomal Dominant (Ailesel Çarpık Ayak ± Eşlik Eden Alt Ekstremite Anomalileri) (Prenatal)	Chromosome 17q23.1-q23.3 duplication syndrome, 613618, Autosomal dominant (Familial clubfoot with or without associated lower limb anomalies) (Prenatal)		DUP17q23.1q23.2, C17DUPq23.1q23.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 18p Delesyon Sendromu, 146390, Otozomal Dominant (Monozomi 18p) (2200)	Chromosome 18p deletion syndrome, 146390, Autosomal dominant (Monosomy 18p) (440)		DEL18p, C18DELp	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 18p Delesyon Sendromu, 146390, Otozomal Dominant (Monozomi 18p) (Prenatal)	Chromosome 18p deletion syndrome, 146390, Autosomal dominant (Monosomy 18p) (Prenatal)		DEL18p, C18DELp	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 18q Delesyon Sendromu, 601808, Otozomal Dominant (Monozomi 18q) (2200)	Chromosome 18q deletion syndrome, 601808, Autosomal dominant (Monosomy 18q) (440)		DEL18q	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 18q Delesyon Sendromu, 601808, Otozomal Dominant (Monozomi 18q) (Prenatal)	Chromosome 18q deletion syndrome, 601808, Autosomal dominant (Monosomy 18q) (Prenatal)		DEL18q	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 19p13.13 Delesyon Sendromu, 613638 (19p13 13 Mikrodelesyon Sendromu) (2200)	Chromosome 19p13.13 deletion syndrome, 613638 (19p13.13 microdeletion syndrome) (440)		DEL19p13.13, C19DEL p13 13, DUP19p13 13, C19DUPp13 13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 19p13.13 Delesyon Sendromu, 613638 (19p13 13 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 19p13.13 deletion syndrome, 613638 (19p13.13 microdeletion syndrome) (Prenatal)		DEL19p13.13, C19DEL p13 13, DUP19p13 13, C19DUPp13 13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 19p13.13 Duplikasyon Sendromu, 613638 (19p13.13 Mikrodelesyon Sendromu) (2200)	Chromosome 19p13.13 duplication syndrome, 613638 (19p13.13 microdeletion syndrome) (440)		DEL19p13.13, C19DEL p13 13, DUP19p13 13, C19DUPp13 13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 19p13.13 Duplikasyon Sendromu, 613638 (19p13.13 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 19p13.13 duplication syndrome, 613638 (19p13.13 microdeletion syndrome) (Prenatal)		DEL19p13.13, C19DEL p13 13, DUP19p13 13, C19DUPp13 13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 19p13.11 Delesyon Sendromu, Distal, 613026, Otozomal Dominant (19q13.11 Mikrodelesyon Sendromu) (2200)	Chromosome 19q13.11 deletion syndrome, distal, 613026, Autosomal dominant (19q13.11 microdeletion syndrome) (440)		DEL19q13.11d, C19DELq13.11d	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 19q13.11 Delesyon Sendromu, Distal, 613026, Otozomal Dominant (19q13.11 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 19q13.11 deletion syndrome, distal, 613026, Autosomal dominant (19q13.11 microdeletion syndrome) (Prenatal)		DEL19q13.11d, C19DELq13.11d	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 19q13.11 Delesyon Sendromu, Proksimal, 617219, Otozomal Dominant (2200)	Chromosome 19q13.11 deletion syndrome, proximal, 617219, Autosomal dominant (440)		DEL19q13.11p, C19DELq13.11p	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 19q13.11 Delesyon Sendromu, Proksimal, 617219, Otozomal Dominant (Prenatal)	Chromosome 19q13.11 deletion syndrome, proximal, 617219, Autosomal dominant (Prenatal)		DEL19q13.11p, C19DELq13.11p	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 1p32-P31 Delesyon Sendromu, 613735, Sporadik Vakalar (1p31p32 Mikrodelesyon Sendromu) (2200)	Chromosome 1p32-p31 deletion syndrome, 613735, Isolated cases (1p31p32 microdeletion syndrome) (440)		DEL1p32p31, C1DELp32p31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 1p32-P31 Delesyon Sendromu, 613735, Sporadik Vakalar (1p31p32 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 1p32-p31 deletion syndrome, 613735, Isolated cases (1p31p32 microdeletion syndrome) (Prenatal)		DEL1p32p31, C1DELp32p31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 1p36 Delesyon Sendromu, 607872, Sporadik Vakalar (1p36 Delesyon Sendromu) (2200)	Chromosome 1p36 deletion syndrome, 607872, Isolated cases (1p36 deletion syndrome) (440)		DEL1p36, C1DELp36	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 1p36 Delesyon Sendromu, 607872, Sporadik Vakalar (1p36 Delesyon Sendromu) (Prenatal)	Chromosome 1p36 deletion syndrome, 607872, Isolated cases (1p36 deletion syndrome) (Prenatal)		DEL1p36, C1DELp36	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 1q21.1 Delesyon Sendromu, 612474, Otozomal Dominant, Sporadik Vakalar (1q21.1 Mikrodelesyon Sendromu) (2200)	Chromosome 1q21.1 deletion syndrome, 612474, Autosomal dominant, Isolated cases (1q21.1 microdeletion syndrome) (440)		DEL1q21, C1DELq21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 1q21.1 Delesyon Sendromu, 612474, Otozomal Dominant, Sporadik Vakalar (1q21.1 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 1q21.1 deletion syndrome, 612474, Autosomal dominant, Isolated cases (1q21.1 microdeletion syndrome) (Prenatal)		DEL1q21, C1DELq21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 1q21.1 Duplikasyon Sendromu, 612475, Otozomal Dominant, Sporadik Vakalar (1q21.1 Mikroduplikasyon Sendromu) (2200)	Chromosome 1q21.1 duplication syndrome, 612475, Autosomal dominant, Isolated cases (1q21.1 microduplication syndrome) (440)		DUP1q21, C1DUPq21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 1q21.1 Duplikasyon Sendromu, 612475, Otozomal Dominant, Sporadik Vakalar (1q21.1 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 1q21.1 duplication syndrome, 612475, Autosomal dominant, Isolated cases (1q21.1 microduplication syndrome) (Prenatal)		DUP1q21, C1DUPq21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 1q41-Q42 Delesyon Sendromu, 612530, Sporadik Vakalar (Holoprosensefali) (2200)	Chromosome 1q41-q42 deletion syndrome, 612530, Isolated cases (Holoprosencephaly) (440)		DEL1q41q42, C1DELq41q42	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 1q41-Q42 Delesyon Sendromu, 612530, Sporadik Vakalar (Holoprosensefali) (Prenatal)	Chromosome 1q41-q42 deletion syndrome, 612530, Isolated cases (Holoprosencephaly) (Prenatal)		DEL1q41q42, C1DELq41q42	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 22q11.2 Delesyon Sendromu, Distal, 611867 (Distal 22q11.2 Mikrodelesyon Sendromu) (2200)	Chromosome 22q11.2 deletion syndrome, distal, 611867 (Distal 22q11.2 microdeletion syndrome) (440)		DEL22q11.2, C22DELq11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 22q11.2 Delesyon Sendromu, Distal, 611867 (Distal 22q11.2 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 22q11.2 deletion syndrome, distal, 611867 (Distal 22q11.2 microdeletion syndrome) (Prenatal)		DEL22q11.2, C22DELq11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 22q11.2 Mikrodüplikasyon Sendromu, 608363, Otozomal Dominant, Sporadik Vakalar (22q11.2 Mikroduplikasyon Sendromu) (2200)	Chromosome 22q11.2 microduplication syndrome, 608363, Autosomal dominant, Isolated cases (22q11.2 microduplication syndrome) (440)		DUP22q11 2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 22q11.2 Mikrodüplikasyon Sendromu, 608363, Otozomal Dominant, Sporadik Vakalar (22q11.2 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 22q11.2 microduplication syndrome, 608363, Autosomal dominant, Isolated cases (22q11.2 microduplication syndrome) (Prenatal)		DUP22q11 2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 22q13 Duplikasyon Sendromu, 615538, Sporadik Vakalar (2200)	Chromosome 22q13 duplication syndrome, 615538, Isolated cases (440)		DUP22q13, C22DUPq13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 22q13 Duplikasyon Sendromu, 615538, Sporadik Vakalar (Prenatal)	Chromosome 22q13 duplication syndrome, 615538, Isolated cases (Prenatal)		DUP22q13, C22DUPq13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 2p12-P11.2 Delesyon Sendromu, 613564 (2200)	Chromosome 2p12-p11.2 deletion syndrome, 613564 (440)		DEL2p12p11.2, C2DELp12p11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 2p12-P11.2 Delesyon Sendromu, 613564 (Prenatal)	Chromosome 2p12-p11.2 deletion syndrome, 613564 (Prenatal)		DEL2p12p11.2, C2DELp12p11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 2p16.1-P15 Delesyon Sendromu, 612513, Sporadik Vakalar (2p15p16.1 Mikrodelesyon Sendromu) (2200)	Chromosome 2p16.1-p15 deletion syndrome, 612513, Isolated cases (2p15p16.1 microdeletion syndrome) (440)		DEL2p16.1-p15, C2DELp161-p15	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 2p16.1-P15 Delesyon Sendromu, 612513, Sporadik Vakalar (2p15p16.1 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 2p16.1-p15 deletion syndrome, 612513, Isolated cases (2p15p16.1 microdeletion syndrome) (Prenatal)		DEL2p16.1-p15, C2DELp161-p15	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
KROMOZOM 2p16.3 Delesyon Sendromu (2200)	CHROMOSOME 2p16.3 DELETION SYNDROME (440)			Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
KROMOZOM 2p16.3 Delesyon Sendromu (Prenatal)	CHROMOSOME 2p16.3 DELETION SYNDROME (Prenatal)			Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 2q31.1 Duplikasyon Sendromu, 613681, Otozomal Dominant (Mezomelik Displazi, Kantaputra Tipi) (2200)	Chromosome 2q31.1 duplication syndrome, 613681, Autosomal dominant (Mesomelic dysplasia, Kantaputra type) (440)		DUP2q31.1, C2DUPq31.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 2q31.1 Duplikasyon Sendromu, 613681, Otozomal Dominant (Mezomelik Displazi, Kantaputra Tipi) (Prenatal)	Chromosome 2q31.1 duplication syndrome, 613681, Autosomal dominant (Mesomelic dysplasia, Kantaputra type) (Prenatal)		DUP2q31.1, C2DUPq31.1	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 2q31.2 Delesyon Sendromu, 612345 (2200)	Chromosome 2q31.2 deletion syndrome, 612345 (440)		DEL2q31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 2q31.2 Delesyon Sendromu, 612345 (Prenatal)	Chromosome 2q31.2 deletion syndrome, 612345 (Prenatal)		DEL2q31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
KROMOZOM 2q35 DUPLİKASYON SENDROMU (Kraniyosinostozis, Filadelfia Tipi) (2200)	CHROMOSOME 2q35 DUPLICATION SYNDROME (Craniosynostosis, Philadelphia type) (440)		DUP2q35, C2DUPq35, SDTY1, SD1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
KROMOZOM 2q35 DUPLİKASYON SENDROMU (Kraniyosinostozis, Filadelfia Tipi) (Prenatal)	CHROMOSOME 2q35 DUPLICATION SYNDROME (Craniosynostosis, Philadelphia type) (Prenatal)		DUP2q35, C2DUPq35, SDTY1, SD1	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 2q37 Delesyon Sendromu, 600430, Otozomal Dominant (2q37 Mikrodelesyon Sendromu) (2200)	Chromosome 2q37 deletion syndrome, 600430, Autosomal dominant (2q37 microdeletion syndrome) (440)		BDMR, C2DELq37, DEL2q37	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 2q37 Delesyon Sendromu, 600430, Otozomal Dominant (2q37 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 2q37 deletion syndrome, 600430, Autosomal dominant (2q37 microdeletion syndrome) (Prenatal)		BDMR, C2DELq37, DEL2q37	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 3q13.31 Delesyon Sendromu, 615433, Otozomal Dominant (3q13 Mikrodelesyon Sendromu) (2200)	Chromosome 3q13.31 deletion syndrome, 615433, Autosomal dominant (3q13 microdeletion syndrome) (440)		DEL3q13.31, C13DELq13.31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 3q13.31 Delesyon Sendromu, 615433, Otozomal Dominant (3q13 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 3q13.31 deletion syndrome, 615433, Autosomal dominant (3q13 microdeletion syndrome) (Prenatal)		DEL3q13.31, C13DELq13.31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 3q29 Mikrodelesyon Sendromu, 609425, Sporadik Vakalar (3q29 Mikrodelesyon Sendromu) (2200)	Chromosome 3q29 microdeletion syndrome, 609425, Isolated cases (3q29 microdeletion syndrome) (440)		DEL3q29, MICRODEL3q29	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120			60	Postnatal			EDTA Blood Sample and Heparinized blood sample
Kromozom 3q29 Mikrodelesyon Sendromu, 609425, Sporadik Vakalar (3q29 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 3q29 microdeletion syndrome, 609425, Isolated cases (3q29 microdeletion syndrome) (Prenatal)		DEL3q29, MICRODEL3q29	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN				15	30	20	Prenatal			Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kromozom 3q29 Mikroduplikasyon Sendromu, 611936, Otozomal Dominant (3q29 Mikroduplikasyon) (2200)	Chromosome 3q29 microduplication syndrome, 611936, Autosomal dominant (3q29 microduplication) (440)	.	.	DUP3q29, MICRODUP3q29	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 3q29 Mikroduplikasyon Sendromu, 611936, Otozomal Dominant (3q29 Mikroduplikasyon) (Prenatal)	Chromosome 3q29 microduplication syndrome, 611936, Autosomal dominant (3q29 microduplication) (440)	.	.	DUP3q29, MICRODUP3q29	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 4q21 Delesyon Sendromu, 613509, Sporadik Vakalar (4q21 Mikrodelesyon Sendromu) (2200)	Chromosome 4q21 deletion syndrome, 613509, Isolated cases (4q21 microdeletion syndrome) (440)	.	.	DEL4q21, C4DEL q21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 4q21 Delesyon Sendromu, 613509, Sporadik Vakalar (4q21 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 4q21 deletion syndrome, 613509, Isolated cases (4q21 microdeletion syndrome) (Prenatal)	.	.	DEL4q21, C4DEL q21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 4q32.1-Q32.2 Triplication Sendromu, 613603, Otozomal Dominant (2200)	Chromosome 4q32.1-q32.2 triplication syndrome, 613603, Autosomal dominant (440)	.	.	TRIP4q32.1q32.2, C4TRIPq32.1q32.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 4q32.1-Q32.2 Triplication Sendromu, 613603, Otozomal Dominant (Prenatal)	Chromosome 4q32.1-q32.2 triplication syndrome, 613603, Autosomal dominant (Prenatal)	.	.	TRIP4q32.1q32.2, C4TRIPq32.1q32.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 5p13 Duplikasyon Sendromu, 613174, Sporadik Vakalar (5p13 Mikroduplikasyon Sendromu) (2200)	Chromosome 5p13 duplication syndrome, 613174, Isolated cases (5p13 microduplication syndrome) (440)	.	.	DUP5p13, CSDUPp13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 5p13 Duplikasyon Sendromu, 613174, Sporadik Vakalar (5p13 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 5p13 duplication syndrome, 613174, Isolated cases (5p13 microduplication syndrome) (Prenatal)	.	.	DUP5p13, CSDUPp13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
KROMOZOM 5Q Delesyon Sendromu (Izole Del (5q) Kromozom Anomalili ile İlişkili Miyelodisplastik Sendrom) (2200)	CHROMOSOME 5q DELETION SYNDROME (Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality) (440)	RPS14	RPS14, EMTB	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
KROMOZOM 5Q Delesyon Sendromu (Izole Del (5q) Kromozom Anomalili ile İlişkili Miyelodisplastik Sendrom) (Prenatal)	CHROMOSOME 5q DELETION SYNDROME (Myelodysplastic syndrome associated with isolated del(5q) chromosome abnormality) (Prenatal)	RPS14	RPS14, EMTB	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 5q12 Delesyon Sendromu, 615668, Otozomal Dominant (PDE4D Haploinsufficiency Sendromu) (2200)	Chromosome 5q12 deletion syndrome, 615668, Autosomal dominant (PDE4D haploinsufficiency syndrome) (440)	.	.	DEL5q12, CSDEL q12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 5q12 Delesyon Sendromu, 615668, Otozomal Dominant (PDE4D Haploinsufficiency Sendromu) (Prenatal)	Chromosome 5q12 deletion syndrome, 615668, Autosomal dominant (PDE4D haploinsufficiency syndrome) (Prenatal)	.	.	DEL5q12, CSDEL q12	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 5q14.3 Delesyon Sendromu, 613443, Otozomal Dominant (5q14.3 Mikrodelesyon Sendromu) (2200)	Chromosome 5q14.3 deletion syndrome, 613443, Autosomal dominant (5q14.3 microdeletion syndrome) (440)	MEF2C	MEF2C, CSDEL q14.3, DEL5q14.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. [ECO:0000269	PubMed:19592390]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample and Heparinized blood sample
Kromozom 5q14.3 Delesyon Sendromu, 613443, Otozomal Dominant (5q14.3 Mikrodelesyon Sendromu) (MLPA)	Chromosome 5q14.3 deletion syndrome, 613443, Autosomal dominant (5q14.3 microdeletion syndrome) (MLPA)	MEF2C	MEF2C, CSDEL q14.3, DEL5q14.3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. [ECO:0000269	PubMed:19592390]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kromozom 5q14.3 Delesyon Sendromu, 613443, Otozomal Dominant (5q14.3 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 5q14.3 deletion syndrome, 613443, Autosomal dominant (5q14.3 microdeletion syndrome) (Prenatal)	MEF2C	MEF2C, CSDEL q14.3, DEL5q14.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. [ECO:0000269	PubMed:19592390]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 5q14.3 Delesyon Sendromu, 613443, Otozomal Dominant (5q14.3 Mikrodelesyon Sendromu) (Prenatal) (MLPA)	Chromosome 5q14.3 deletion syndrome, 613443, Autosomal dominant (5q14.3 microdeletion syndrome) (Prenatal) (MLPA)	MEF2C	MEF2C, CSDEL q14.3, DEL5q14.3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. [ECO:0000269	PubMed:19592390]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 6pter-P24 Delesyon Sendromu, 612582, Sporadik Vakalar (Distal Monozomi 6p) (2200)	Chromosome 6pter-p24 deletion syndrome, 612582, Isolated cases (Distal monosomy 6p) (440)	.	.	DEL6pter, C6DEL pter	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 6pter-P24 Delesyon Sendromu, 612582, Sporadik Vakalar (Distal Monozomi 6p) (Prenatal)	Chromosome 6pter-p24 deletion syndrome, 612582, Isolated cases (Distal monosomy 6p) (Prenatal)	.	.	DEL6pter, C6DEL pter	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 6q11-Q14 Delesyon Sendromu, 613544, Sporadik Vakalar (2200)	Chromosome 6q11-q14 deletion syndrome, 613544, Isolated cases (440)	.	.	DEL6q11q14, C6DEL q11q14	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 6q11-Q14 Delesyon Sendromu, 613544, Sporadik Vakalar (Prenatal)	Chromosome 6q11-q14 deletion syndrome, 613544, Isolated cases (Prenatal)	.	.	DEL6q11q14, C6DEL q11q14	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 6q25-Q25 Delesyon Sendromu, 612863 (6q25 Mikrodelesyon Sendromu) (2200)	Chromosome 6q25-q25 deletion syndrome, 612863 (6q25 microdeletion syndrome) (440)	.	.	DEL6q24q25, C6DEL q25q25	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 6q25-Q25 Delesyon Sendromu, 612863 (6q25 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 6q25-q25 deletion syndrome, 612863 (6q25 microdeletion syndrome) (Prenatal)	.	.	DEL6q24q25, C6DEL q25q25	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 7q11.23 Delesyon Sendromu, Distal, 1.2Mb, 613729 (Distal 7q11.23 Mikrodelesyon Sendromu) (2200)	Chromosome 7q11.23 deletion syndrome, distal, 1.2Mb, 613729 (Distal 7q11.23 microdeletion syndrome) (440)	.	.	DEL7q11.23, C7DEL q11.23	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 7q11.23 Delesyon Sendromu, Distal, 1.2Mb, 613729 (Distal 7q11.23 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 7q11.23 deletion syndrome, distal, 1.2Mb, 613729 (Distal 7q11.23 microdeletion syndrome) (Prenatal)	.	.	DEL7q11.23, C7DEL q11.23	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 7q11.23 Duplikasyon Sendromu, 609757, Otozomal Dominant (7q11.23 Mikroduplikasyon Sendromu) (2200)	Chromosome 7q11.23 duplication syndrome, 609757, Autosomal dominant (7q11.23 microduplication syndrome) (440)	.	.	DUP7q11.23, C7DUPq11.23	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 7q11.23 Duplikasyon Sendromu, 609757, Otozomal Dominant (7q11.23 Mikroduplikasyon Sendromu) (Prenatal)	Chromosome 7q11.23 duplication syndrome, 609757, Autosomal dominant (7q11.23 microduplication syndrome) (Prenatal)	.	.	DUP7q11.23, C7DUPq11.23	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 8p11 Miyeloproliferatif Sendrom, 613523 (FGFR1 Yeniden Düzenlenmesi İle İlişkili Miyeloid Neoplazm) (2200)	Chromosome 8p11 myeloproliferative syndrome, 613523 (Myeloid neoplasm associated with FGFR1 rearrangement) (440)	.	.	SCLL	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 8p11 Miyeloproliferatif Sendrom, 613523 (FGFR1 Yeniden Düzenlenmesi İle İlişkili Miyeloid Neoplazm) (Prenatal)	Chromosome 8p11 myeloproliferative syndrome, 613523 (Myeloid neoplasm associated with FGFR1 rearrangement) (Prenatal)	.	.	SCLL	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 8p21.11 Delesyon Sendromu, 614230, Otozomal Dominant, Sporadik Vakalar (8p21.11 Mikrodelesyon Sendromu) (2200)	Chromosome 8p21.11 deletion syndrome, 614230, Autosomal dominant, Isolated cases (8p21.11 microdeletion syndrome) (440)	.	.	DEL8p21.11, C8DEL q21.11	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 8p21.11 Delesyon Sendromu, 614230, Otozomal Dominant, Sporadik Vakalar (8p21.11 Mikrodelesyon Sendromu) (Prenatal)	Chromosome 8p21.11 deletion syndrome, 614230, Autosomal dominant, Isolated cases (8p21.11 microdeletion syndrome) (Prenatal)	.	.	DEL8p21.11, C8DEL q21.11	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom 9p Delesyon Sendromu, 158170, Otozomal Dominant (Monozomi 9p) (2200)	Chromosome 9p deletion syndrome, 158170, Autosomal dominant (Monosomy 9p) (440)	.	.	DEL9p, C9DEL p	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom 9p Delesyon Sendromu, 158170, Otozomal Dominant (Monozomi 9p) (Prenatal)	Chromosome 9p deletion syndrome, 158170, Autosomal dominant (Monosomy 9p) (Prenatal)	.	.	DEL9p, C9DEL p	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xp11.22 Duplikasyon Sendromu (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (2200)	CHROMOSOME Xp11.22 DUPLICATION SYNDROME (X-linked non-syndromic intellectual disability) (440)	.	.	DUPXp11.22, MRX17, MRX31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xp11.22 Duplikasyon Sendromu (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal)	CHROMOSOME Xp11.22 DUPLICATION SYNDROME (X-linked non-syndromic intellectual disability) (Prenatal)	.	.	DUPXp11.22, MRX17, MRX31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xp11.23-P11.22 Duplikasyon Sendromu, 300801, Xe Bağlı Dominant (Mikroduplikasyon Xp11.22-P11.23 Sendromu) (2200)	Chromosome Xp11.23-p11.22 duplication syndrome, 300801, X-linked dominant (Microduplication Xp11.22-p11.23 syndrome) (440)	.	.	DUPXp11.23p11.22, CXDUPp11.23p11.22	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xp11.23-P11.22 Duplikasyon Sendromu, 300801, Xe Bağlı Dominant (Mikroduplikasyon Xp11.22-P11.23 Sendromu) (Prenatal)	Chromosome Xp11.23-p11.22 duplication syndrome, 300801, X-linked dominant (Microduplication Xp11.22-p11.23 syndrome) (Prenatal)	.	.	DUPXp11.23p11.22, CXDUPp11.23p11.22	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xp11.3 Delesyon Sendromu, 300578, X-linked recessive (X`e Bağlı Resesif (Xe Bağlı Mental Retardasyon-Retinit Pigmentosa Sendromu) (2200)	Chromosome Xp11.3 deletion syndrome, 300578, X-linked recessive (X-linked intellectual disability-retinitis pigmentosa syndrome) (440)	.	.	DELXp11.3, CXDELp11.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xp11.3 Delesyon Sendromu, 300578, X`e Bağlı Resesif (Xe Bağlı Mental Retardasyon-Retinit Pigmentosa Sendromu) (Prenatal)	Chromosome Xp11.3 deletion syndrome, 300578, X-linked recessive (X-linked intellectual disability-retinitis pigmentosa syndrome) (Prenatal)	.	.	DELXp11.3, CXDELp11.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xp21 Delesyon Sendromu, 300679 (Monozomi Xp21) (2200)	Chromosome Xp21 deletion syndrome, 300679 (Monosomy Xp21) (440)	.	.	DELXp21, CXDELp21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xp21 Delesyon Sendromu, 300679 (Monozomi Xp21) (Prenatal)	Chromosome Xp21 deletion syndrome, 300679 (Monosomy Xp21) (Prenatal)	.	.	DELXp21, CXDELp21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xp26.3 Duplikasyon Sendromu, 300942, Xe Bağlı Dominant (Ailesel İnfantil Spazmlar) (2200)	Chromosome Xp26.3 duplication syndrome, 300942, X-linked dominant (Familial infantile spasms) (440)	.	.	CXDUPp26.3, DUPXp26.3, XLAG	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xp26.3 Duplikasyon Sendromu, 300942, Xe Bağlı Dominant (Ailesel İnfantil Spazmlar) (Prenatal)	Chromosome Xp26.3 duplication syndrome, 300942, X-linked dominant (Familial infantile spasms) (Prenatal)	.	.	CXDUPp26.3, DUPXp26.3, XLAG	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xq27.3-Q28 Duplikasyon Sendromu, 300869, Xe Bağlı Resesif (Xq27.3q28 Duplikasyon Sendromu) (2200)	Chromosome Xq27.3-q28 duplication syndrome, 300869, X-linked recessive (Xq27.3q28 duplication syndrome) (440)	.	.	DUPXq27.3q28, CXDUPq27.3q28	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xq27.3-Q28 Duplikasyon Sendromu, 300869, X'e Bağlı Resesif (Xq27.3q28 Duplikasyon Sendromu) (Prenatal)	Chromosome Xq27.3-q28 duplication syndrome, 300869, X-linked recessive (Xq27.3q28 duplication syndrome) (Prenatal)	.	.	DUPXq27.3q28, CXDUPq27.3q28	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kromozom Xq28 Duplikasyon Sendromu, 300815 (Trisomi Xq28) (2200)	Chromosome Xq28 duplication syndrome, 300815 (Trisomy Xq28) (440)	.	.	DUPXq28, CXq28	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Kromozom Xq28 Duplikasyon Sendromu, 300815 (Trisomi Xq28) (Prenatal)	Chromosome Xq28 duplication syndrome, 300815 (Trisomy Xq28) (Prenatal)	.	.	DUPXq28, CXq28	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Atriyal ve İntestinal Rhm Bozukluğu 616201, Otozomal Resesif, CAID (Kronik Atriyal ve İntestinal Disritmi Sendromu) (SGO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chronic atrial and intestinal dysrhythmia, 616201, Autosomal recessive; CAID (Chronic atrial and intestinal dysrhythmia syndrome) (SGO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGO1	SGOL1, SGO, SGO1, CAID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kronik Granülomatöz Hastalık, NCF-1 Eksikliğine Bağlı, 233710, Otozomal Resesif (Kronik Granülomatöz Hastalık) (NCF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chronic granulomatous disease due to deficiency of NCF-2, 233710, Autosomal recessive; CDG2 (Chronic granulomatous disease) (NCF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NCF2	NCF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-positive 2, autosomal recessive (CGD2) [MIM:233710]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10498624, ECO:0000269|PubMed:10598813, ECO:0000269|PubMed:11123388, ECO:0000269|PubMed:16937026, ECO:0000269|PubMed:18625437, ECO:0000269|PubMed:19624736, ECO:0000269|PubMed:20167518, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8286749, ECO:0000269|PubMed:9070911}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kronik Granülomatöz Hastalık, NCF-1 Eksikliğine Bağlı, 233710, Otozomal Resesif (Kronik Granülomatöz Hastalık) (Prenatal) (NCF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chronic granulomatous disease due to deficiency of NCF-2, 233710, Autosomal recessive; CDG2 (Chronic granulomatous disease) (NCF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NCF2	NCF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-positive 2, autosomal recessive (CGD2) [MIM:233710]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10498624, ECO:0000269|PubMed:10598813, ECO:0000269|PubMed:11123388, ECO:0000269|PubMed:16937026, ECO:0000269|PubMed:18625437, ECO:0000269|PubMed:19624736, ECO:0000269|PubMed:20167518, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8286749, ECO:0000269|PubMed:9070911}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık Paneli (Xp21.1; 1q25.3; 16q24.3; 22q13.1) (MLPA)	Chronic Granulomatous Disease Panel (Xp21.1; 1q25.3; 16q24.3; 22q13.1) (MLPA)	Xp21.1; 1q25.3; 16q24.3; 22q13.1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kronik Granülomatöz Hastalık Paneli (Xp21.1; 1q25.3; 16q24.3; 22q13.1) (MLPA) (Prenatal)	Chronic Granulomatous Disease Panel (Xp21.1; 1q25.3; 16q24.3; 22q13.1) (MLPA) (Prenatal)	Xp21.1; 1q25.3; 16q24.3; 22q13.1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal, CYBA Eksikliğine Bağlı, 233690, Otozomal Resesif (Kronik Granülomatöz Hastalık) (CYBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690, Autosomal recessive (Chronic granulomatous disease) (CYBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYBA	CYBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-negative, autosomal recessive (ARCGD) [MIM:233690]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10759707, ECO:0000269|PubMed:10910929, ECO:0000269|PubMed:10914676, ECO:0000269|PubMed:1415254, ECO:0000269|PubMed:1763037, ECO:0000269|PubMed:18422995, ECO:0000269|PubMed:2243141, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8168815}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal, CYBA Eksikliğine Bağlı, 233690, Otozomal Resesif (Kronik Granülomatöz Hastalık) (Prenatal) (CYBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690, Autosomal recessive (Chronic granulomatous disease) (CYBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYBA	CYBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-negative, autosomal recessive (ARCGD) [MIM:233690]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10759707, ECO:0000269|PubMed:10910929, ECO:0000269|PubMed:10914676, ECO:0000269|PubMed:1415254, ECO:0000269|PubMed:1763037, ECO:0000269|PubMed:18422995, ECO:0000269|PubMed:2243141, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8168815}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık, X'e Bağlı, 306400, X'e Bağlı (Kronik Granülomatöz Hastalık) (CYBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chronic granulomatous disease, X-linked, 306400, X-linked recessive (Chronic granulomatous disease) (CYBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. {ECO:0000269|PubMed:21278736}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kronik Granülomatöz Hastalık, X'e Bağlı, 306400, X'e Bağlı (Kronik Granülomatöz Hastalık) (CYBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chronic granulomatous disease, X-linked, 306400, X-linked recessive (Chronic granulomatous disease) (CYBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. {ECO:0000269|PubMed:21278736}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MBL Yetersizliğine Bağlı Kronik Enfeksiyonlar, 614372, Otozomal Dominant; MBLD (İmmün Sistemi Baskılanmış Hastada Enfeksiyon Duyarlılığı) (MBL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chronic infections, due to MBL deficiency, 614372, Autosomal dominant; MBLD (Susceptibility to infection in immunocompromised patient) (MBL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBL2	MBL2, MBL, MBP1, MBL2D, MBPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBL Yetersizliğine Bağlı Kronik Enfeksiyonlar, 614372, Otozomal Dominant; MBLD (İmmün Sistemi Baskılanmış Hastada Enfeksiyon Duyarlılığı) (Prenatal) (MBL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chronic infections, due to MBL deficiency, 614372, Autosomal dominant; MBLD (Susceptibility to infection in immunocompromised patient) (MBL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MBL2	MBL2, MBL, MBP1, MBL2D, MBPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Chudley-Mccullough Sendromu, 604213, Otozomal Resesif; CMCS (Chudley-Mccullough Sendromu) (GPSM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chudley-McCullough syndrome, 604213, Autosomal recessive; CMCS (Chudley-McCullough syndrome) (GPSM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPSM2	GPSM2, LGN, PINS, DFNB82, CMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Chudley-Mccullough Sendromu, 604213, Otozomal Resesif; CMCS (Chudley-Mccullough Sendromu) (Prenatal) (GPSM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Chudley-McCullough syndrome, 604213, Autosomal recessive; CMCS (Chudley-McCullough syndrome) (GPSM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPSM2	GPSM2, LGN, PINS, DFNB82, CMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şilomikron Retansiyon Hastalığı, 246700, Otozomal Resesif; CMRD (Şilomikron Retansiyon Hastalığı) (SAR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Chylomicron retention disease, 246700, Autosomal recessive; CMRD (Chylomicron retention disease) (SAR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAR1B	SAR1B, CMRD, SARA2, ANDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kimezin Psödogeni; Cymp (CYMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	CHYMOSIN PSEUDOGENE; CYMP (CYMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYMP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 1, x Situs İnversus, 244400, Otozomal Resesif; CILD1 (Primer Siliyer Diskinezi) (DNAI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400, Autosomal recessive; CILD1 (Primary ciliary dyskinesia) (DNAI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAI1	DNAI1, CILD1, ICS, PCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 1 (CILD1) [MIM:244400]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269|PubMed:25186273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 1, x Situs İnversus, 244400, Otozomal Resesif; CILD1 (Primer Siliyer Diskinezi) (MLPA)	Ciliary dyskinesia, primary, 1, with or without situs inversus, 244400, Autosomal recessive; CILD1 (Primary ciliary dyskinesia) (MLPA)	DNAI1	DNAI1, CILD1, ICS, PCD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ciliary dyskinesia, primary, 1 (CILD1) [MIM:244400]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269|PubMed:25186273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 10, 612518; CILD10 (Primer Siliyer Diskinezi) (DNAAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 10, 612518; CILD10 (Primary ciliary dyskinesia) (DNAAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAAF2	KTU, C14orf104, CILD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 10 (CILD10) [MIM:612518]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269|PubMed:19052621, ECO:0000269|PubMed:25186273}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Siliyer Diskinezi, Primer, 11, 612649; CILD11 (Primer Siliyer Diskinezi) (RSPH4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 11, 612649; CILD11 (Primary ciliary dyskinesia) (RSPH4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPH4A	RSPH4A, CILD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 12, 612650; CILD12 (Primer Siliyer Diskinezi) (RSPH9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 12, 612650; CILD12 (Primary ciliary dyskinesia) (RSPH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPH9	RSPH9, CILD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 12 (CILD12) [MIM:612650]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit situs inversus, due to dysfunction of monocilia at the embryonic node and randomization of left-right body asymmetry. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269|PubMed:19200523}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Silliyer Diskinezi, Primer, 13, 613193, Otozomal Resesif, CILD13 (Primer Silliyer Diskinezi) (DNAAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 13, 613193, Autosomal recessive; CILD13 (Primary ciliary dyskinesia) (DNAAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAAF1	DNAAF1, LRRC50, ODA7, CILD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 14, 613807, CILD14 (Primer Silliyer Diskinezi) (CCDC39 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 14, 613807, CILD14 (Primary ciliary dyskinesia) (CCDC39 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC39	CCDC39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 15, 613808, CILD15 (Primer Silliyer Diskinezi) (CCDC40 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 15, 613808, CILD15 (Primary ciliary dyskinesia) (CCDC40 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC40	CCDC40, KIAA1640	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 15 (CILD15) [MIM:613808]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:21131974, ECO:0000269|PubMed:22693285, ECO:0000269|PubMed:23255504, ECO:0000269|PubMed:23402890, ECO:0000269|PubMed:25186273). Note=The disease is caused by mutations affecting the gene represented in this entry. The disease is characterized by primary ciliary dyskinesia with inner dynein arm (IDA) defects and axonemal disorganisation: defects in CCDC39 and CCDC40 constitute the major cause of this phenotype. (ECO:0000269|PubMed:22693285, ECO:0000269|PubMed:23255504).;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 16, 614017, Otozomal Resesif, CILD16 (Primer Silliyer Diskinezi) (DNAL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 16, 614017, Autosomal recessive; CILD16 (Primary ciliary dyskinesia) (DNAL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAL1	DNAL1, C14orf168, CILD16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 17, 614679, Otozomal Resesif, CILD17 (Primer Silliyer Diskinezi) (CCDC103 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 17, 614679, Autosomal recessive; CILD17 (Primary ciliary dyskinesia) (CCDC103 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC103	CCDC103, SMH, PR46B, CILD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 17 (CILD17) [MIM:614679]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:22581229, ECO:0000269|PubMed:25186273). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 18, 614874, Otozomal Resesif, CILD18 (Primer Silliyer Diskinezi) (DNAAF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 18, 614874, Autosomal recessive; CILD18 (Primary ciliary dyskinesia) (DNAAF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAAF5	HEATR2, CILD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 18 (CILD18) [MIM:614874]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:23040496, ECO:0000269|PubMed:25232951). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 19, 614935, Otozomal Resesif, CILD19 (Primer Silliyer Diskinezi) (LRRC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 19, 614935, Autosomal recessive; CILD19 (Primary ciliary dyskinesia) (LRRC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRRC6	LRRC6, LRTP, CILD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 2, 606763, Otozomal Resesif, CILD2 (Primer Silliyer Diskinezi) (DNAAF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 2, 606763, Autosomal recessive; CILD2 (Primary ciliary dyskinesia) (DNAAF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAAF3	DNAAF3, PF22, DAB1, CILD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 2 (CILD2) [MIM:606763]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus., due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269	PubMed:22387996). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 20, 615067, Otozomal Resesif, CILD20 (Primer Silliyer Diskinezi) (CCDC114 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 20, 615067, Autosomal recessive; CILD20 (Primary ciliary dyskinesia) (CCDC114 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC114	CCDC114, CILD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 20 (CILD20) [MIM:615067]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. Unlike other forms of CILD characterized by reduced fertility, patients with CILD20 do not appear to be infertile. (ECO:0000269|PubMed:23261302, ECO:0000269|PubMed:23261303). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 21, 615294, Otozomal Resesif, CILD21 (Primer Silliyer Diskinezi) (DRC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 21, 615294, Autosomal recessive; CILD21 (Primary ciliary dyskinesia) (DRC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRC1	DRC1, CCDC164, C2orf39, CILD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 21 (CILD21) [MIM:615294]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:23354437, ECO:0000269|PubMed:25186273). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 22, 615444, Otozomal Resesif, CILD22 (Primer Silliyer Diskinezi) (ZMYND10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 22, 615444, Autosomal recessive; CILD22 (Primary ciliary dyskinesia) (ZMYND10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZMYND10	ZMYND10, BLU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 22 (CILD22) [MIM:615444]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:23891469, ECO:0000269|PubMed:23891471, ECO:0000269|PubMed:25186273). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 23, 615451, Otozomal Resesif, CILD23 (Primer Silliyer Diskinezi) (ARMC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 23, 615451, Autosomal recessive; CILD23 (Primary ciliary dyskinesia) (ARMC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARMC4	ARMC4, CILD23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Silliyer Diskinezi, Primer, 24, 615481, Otozomal Resesif, CILD24 (Primer Silliyer Diskinezi) (RSPH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 24, 615481, Autosomal recessive; CILD24 (Primary ciliary dyskinesia) (RSPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPH1	RSPH1, TSGA2, TSA2, CILD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 25, 615482, Otozomal Resesif, CILD25 (Primer Silliyer Diskinezi) (DYX1C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 25, 615482, Autosomal recessive; CILD25 (Primary ciliary dyskinesia) (DYX1C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYX1C1	DYX1C1, DYXC1, DYX1, CILD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 25 (CILD25) [MIM:615482]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269|PubMed:23872636, ECO:0000269|PubMed:25186273). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 26, 615500, Otozomal Resesif, CILD26 (Primer Silliyer Diskinezi) (C21orf59 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 26, 615500, Autosomal recessive; CILD26 (Primary ciliary dyskinesia) (C21orf59 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C21orf59	C21ORF59, CILD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 26 (CILD26) [MIM:615500]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. (ECO:0000269	PubMed:24094744). Note=The disease is caused by mutations affecting the gene represented in this entry. Cilia in nasal epithelia show the absence of both outer and inner dynein- arm components and complete paralysis.;	EDTA Blood Sample
Silliyer Diskinezi, Primer, 27, 615504, Otozomal Resesif, CILD27 (Primer Silliyer Diskinezi) (CCDC65 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 27, 615504, Autosomal recessive; CILD27 (Primary ciliary dyskinesia) (CCDC65 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC65	CCDC65, CILD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 28, 615505, Otozomal Resesif, CILD28 (Primer Silliyer Diskinezi) (SPAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 28, 615505, Autosomal recessive; CILD28 (Primary ciliary dyskinesia) (SPAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPAG1	SPAG1, CILD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silliyer Diskinezi, Primer, 29, 615872, Otozomal Resesif, CILD29 (Primer Silliyer Diskinezi) (CCNO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 29, 615872, Autosomal recessive; CILD29 (Primary ciliary dyskinesia) (CCNO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCNO	CCNO, UNG2, CILD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 29 (CILD29) [MIM:615872]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. CILD29 patients do not exhibit situs inversus, a congenital abnormality in which visceral organs are opposite to their normal positions (situs solitus) due to lateral transposition. (ECO:0000269|PubMed:24747639). Note=The disease is caused by mutations affecting the gene represented in this entry. Marked reduction of cilia in multiciliate cells due to defective mother centriole generation and placement. Remaining cilia correctly express axonemal motor proteins, are motile and do not show beating defects. Defects are probably caused by a strong reduction in the number of multiple motile cilia covering the cell surface in respiratory epithelial cells (PubMed:24747639). (ECO:0000269|PubMed:24747639).;	EDTA Blood Sample

Siliyer Diskinezi, Primer, 3, Situs İnversus 608644; CILD3 (Primer Siliyer Diskinezi) (DNAH5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 3, with or without situs inversus, 608644; CILD3 (Primary ciliary dyskinesia) (DNAH5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAH5	DNAH5, HL1, PCD, CILD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kartagener syndrome (KTGS) [MIM:244400]: An autosomal recessive disorder characterized by the association of primary ciliary dyskinesia with situs inversus. Clinical features include recurrent respiratory infections, bronchiectasis, infertility, and lateral transposition of the viscera of the thorax and abdomen. The situs inversus is most often total, although it can be partial in some cases (isolated dextrocardia or isolated transposition of abdominal viscera). {ECO:0000269	PubMed:11788826}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siliyer Diskinezi, Primer, 30, 616037, Otozomal Resesif; CILD30 (Primer Siliyer Diskinezi) (CCDC151 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 30, 616037, Autosomal recessive; CILD30 (Primary ciliary dyskinesia) (CCDC151 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC151	CCDC151, CILD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 30 (CILD30) [MIM:616037]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269|PubMed:25192045, ECO:0000269|PubMed:25224326}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siliyer Diskinezi, Primer, 32, 616481, Otozomal Resesif; CILD32 (Primer Siliyer Diskinezi) (RSPH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 32, 616481, Autosomal recessive; CILD32 (Primary ciliary dyskinesia) (RSPH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPH3	RSPH3, RSHL2, RSP3, CILD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 32 (CILD32) [MIM:616481]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. {ECO:0000269	PubMed:26073779}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siliyer Diskinezi, Primer, 33, 616726, Otozomal Resesif; CILD33 (Primer Siliyer Diskinezi) (GAS8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 33, 616726, Autosomal recessive; CILD33 (Primary ciliary dyskinesia) (GAS8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAS8	GAS8, GAS11, CILD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 34, 617091, Otozomal Resesif; CILD34 (Primer Siliyer Diskinezi) (DNAJB13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 34, 617091, Autosomal recessive; CILD34 (Primary ciliary dyskinesia) (DNAJB13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJB13	DNAJB13, TSARG6, CILD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Siliyer Diskinezi, Primer, 35, 617092, Otozomal Resesif; CILD35 (Primer Siliyer Diskinezi) (TTC25 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 35, 617092, Autosomal recessive; CILD35 (Primary ciliary dyskinesia) (TTC25 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC25	TTC25, CILD35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 36, X'e Bağlı, 300991, X'e Bağlı Resesif; CILD36 (Primer Siliyer Diskinezi) (PIH1D3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 36, X-linked, 300991, X-linked recessive; CILD36 (Primary ciliary dyskinesia) (PIH1D3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIH1D3	PIH1D3, CİL.D36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 5, 608647, Otozomal Resesif; CILD5 (Primer Siliyer Diskinezi) (HYDIN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 5, 608647, Autosomal recessive; CILD5 (Primary ciliary dyskinesia) (HYDIN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HYDIN	HYDIN, HYDIN1, CILD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 5 (CILD5) [MIM:608647]: An autosomal recessive form of primary dyskinesia, a disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. CILD5 is characterized by early onset of a progressive decline in lung function due to an inability to clear mucus and particles from the airways. Affected individuals have recurrent infections of the sinuses, ears, airways, and lungs. Sperm motility is also decreased. Individuals with CILD5 do not have situs inversus. {ECO:0000269|PubMed:23022101, ECO:0000269|PubMed:25188273}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siliyer Diskinezi, Primer, 6, 610852, Otozomal Resesif; CILD6 (Primer Siliyer Diskinezi) (NME8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 6, 610852, Autosomal recessive; CILD6 (Primary ciliary dyskinesia) (NME8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NME8	NME8, TXNDC3, SPTRX2, CILD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 6 (CILD6) [MIM:610852]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia; reduced fertility is often observed in male patients due to abnormalities of sperm tails. Half of the patients exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269	PubMed:17360646}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Siliyer Diskinezi, Primer, 7, x Situs İnversus, 611884, Otozomal Resesif; CILD7 (Primer Siliyer Diskinezi) (DNAH11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 7, with or without situs inversus, 611884; Autosomal recessive; CILD7 (Primary ciliary dyskinesia) (DNAH11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAH11	DNAH11, DNAHC11, CILD7, DNAH8L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Siliyer Diskinezi, Primer, 9, x Situs İnversus, 612444; CILD9 (Primer Siliyer Diskinezi) (DNAI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ciliary dyskinesia, primary, 9, with or without situs inversus, 612444; CILD9 (Primary ciliary dyskinesia) (DNAI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAI2	DNAI2, CILD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
CINCA Sendromu, 607115, Otozomal Dominant (CINCA Sendromu) (NLRP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CINCA syndrome, 607115, Autosomal dominant (CINCA syndrome) (NLRP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP3	NLRP3, CIAS1, FCU, FCAS1, NALP3, PYPAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CINCA Sendromu, 607115, Otozomal Dominant (CINCA Sendromu) (Prenatal) (NLRP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CINCA syndrome, 607115, Autosomal dominant (CINCA syndrome) (NLRP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLRP3	NLRP3, CIAS1, FCU, FCAS1, NALP3, PYPAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı Siroz (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (PHKG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cirrhosis due to liver phosphorylase kinase deficiency (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKG2	PHKG2, GSD9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9C (GSD9C) [MIM:613027]: A metabolic disorder manifesting in infancy with hepatomegaly, growth retardation, hypotonia, liver dysfunction, and elevated plasma aminotransferases and lipids. These symptoms improve with age in most cases; however, some patients may develop hepatic fibrosis or cirrhosis. {ECO:0000269|PubMed:12930917, ECO:0000269|PubMed:8896567, ECO:0000269|PubMed:9245685}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı Siroz (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (Prenatal) (PHKG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cirrhosis due to liver phosphorylase kinase deficiency (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKG2	PHKG2, GSD9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9C (GSD9C) [MIM:613027]: A metabolic disorder manifesting in infancy with hepatomegaly, growth retardation, hypotonia, liver dysfunction, and elevated plasma aminotransferases and lipids. These symptoms improve with age in most cases; however, some patients may develop hepatic fibrosis or cirrhosis. {ECO:0000269|PubMed:12930917, ECO:0000269|PubMed:8896567, ECO:0000269|PubMed:9245685}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Siroz, Kriptojenik, 215600, Otozomal Resesif (İdiopatik Bakır İlişkili Siroz) (KRT8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cirrhosis, cryptogenic, 215600, Autosomal recessive (Idiopathic copper-associated cirrhosis) (KRT8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT8	KRT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cirrhosis (CIRRH) [MIM:215600]: A liver disease characterized by severe panlobular liver-cell swelling with Mallory body formation, prominent pericellular fibrosis, and marked deposits of copper. Clinical features include abdomen swelling, jaundice and pulmonary hypertension. {ECO:0000269	PubMed:12724528}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siroz, Kriptojenik, 215600, Otozomal Resesif (İdiopatik Bakır İlişkili Siroz) (KRT18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cirrhosis, cryptogenic, 215600, Autosomal recessive (Idiopathic copper-associated cirrhosis) (KRT18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT18	KRT18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cirrhosis (CIRRH) [MIM:215600]: A liver disease characterized by severe panlobular liver-cell swelling with Mallory body formation, prominent pericellular fibrosis, and marked deposits of copper. Clinical features include abdomen swelling, jaundice and pulmonary hypertension. {ECO:0000269|PubMed:12724528, ECO:0000269|PubMed:9011570}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Siroz İçin Nonkriptojenik, Yatkınlık, 215600, Otozomal Resesif (İdiopatik Bakır İlişkili Siroz) (KRT8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cirrhosis, noncryptogenic, susceptibility to, 215600, Autosomal recessive (Idiopathic copper-associated cirrhosis) (KRT8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT8	KRT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cirrhosis (CIRRH) [MIM:215600]: A liver disease characterized by severe panlobular liver-cell swelling with Mallory body formation, prominent pericellular fibrosis, and marked deposits of copper. Clinical features include abdomen swelling, jaundice and pulmonary hypertension. {ECO:0000269	PubMed:12724528}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Siroz İçin Nonkriptojenik, Yatkınlık, 215600, Otozomal Resesif (İdiopatik Bakır İlişkili Siroz) (KRT18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cirrhosis, noncryptogenic, susceptibility to, 215600, Autosomal recessive (Idiopathic copper-associated cirrhosis) (KRT18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT18	KRT18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cirrhosis (CIRRH) [MIM:215600]: A liver disease characterized by severe panlobular liver-cell swelling with Mallory body formation, prominent pericellular fibrosis, and marked deposits of copper. Clinical features include abdomen swelling, jaundice and pulmonary hypertension. {ECO:0000269|PubMed:12724528, ECO:0000269|PubMed:9011570}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sitrüllinemi, 215700, Otozomal Resesif (Sitrüllinemi Tip 1) (ASS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Citrullinemia, 215700, Autosomal recessive (Citrullinemia type I) (ASS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASS1	ASS1, ASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Citrullinemia 1 (CTLN1) [MIM:215700]: The classic form of citrullinemia, an autosomal recessive disease characterized primarily by elevated serum and urine citrulline levels. Ammonia intoxication is another manifestation. It is a disorder of the urea cycle, usually manifesting in the first few days of life. Affected infants appear normal at birth, but as ammonia builds up in the body they present symptoms such as lethargy, poor feeding, vomiting, seizures and loss of consciousness. Less commonly, a milder form can develop later in childhood or adulthood. {ECO:0000269|PubMed:11708871, ECO:0000269|PubMed:11941481, ECO:0000269|PubMed:12815590, ECO:0000269|PubMed:14680976, ECO:0000269|PubMed:16475226, ECO:0000269|PubMed:1943692, ECO:0000269|PubMed:2358466, ECO:0000269|PubMed:25179242, ECO:0000269|PubMed:7977368}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Sitrüllinemi, 215700, Otozomal Resesif (Sitrüllinemi Tip 1) (Prenatal) (ASS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Citrullinemia, 215700, Autosomal recessive (Citrullinemia type I) (ASS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASS1	ASS1, ASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Citrullinemia 1 (CTLN1) [MIM:215700]: The classic form of citrullinemia, an autosomal recessive disease characterized primarily by elevated serum and urine citrulline levels. Ammonia intoxication is another manifestation. It is a disorder of the urea cycle, usually manifesting in the first few days of life. Affected infants appear normal at birth, but as ammonia builds up in the body they present symptoms such as lethargy, poor feeding, vomiting, seizures and loss of consciousness. Less commonly, a milder form can develop later in childhood or adulthood. {ECO:0000269|PubMed:11708871, ECO:0000269|PubMed:11941481, ECO:0000269|PubMed:12815590, ECO:0000269|PubMed:14680976, ECO:0000269|PubMed:16475226, ECO:0000269|PubMed:1943692, ECO:0000269|PubMed:2358466, ECO:0000269|PubMed:25179242, ECO:0000269|PubMed:7977368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sitrüllinemi, Yetişkin Başlangıçlı Tip 2, 603471, Otozomal Resesif; CTLN2 (Sitrüllinemi Tip 2) (SLC25A13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Citrullinemia, adult-onset type II, 603471, Autosomal recessive; CTLN2 (Citrullinemia type II) (SLC25A13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A13	SLC25A13, CTLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sitrüllinemi, Yetişkin Başlangıçlı Tip 2, 603471, Otozomal Resesif; CTLN2 (Sitrüllinemi Tip 2) (Prenatal) (SLC25A13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Citrullinemia, adult-onset type II, 603471, Autosomal recessive; CTLN2 (Citrullinemia type II) (SLC25A13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A13	SLC25A13, CTLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sitrüllinemi, Tip 2, Neonatal Başlangıçlı, 605814, Otozomal Resesif (Neonatal Intrahepatik Kolestaz, Sitrin Eksikliğine Bağlı) (SLC25A13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Citrullinemia, type II, neonatal-onset, 605814, Autosomal recessive (Neonatal intrahepatic cholestasis due to citrin deficiency) (SLC25A13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A13	SLC25A13, CTLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Cholestasis, neonatal intrahepatic, caused by citrin deficiency (NICCD) [MIM:605814]: A form of citrullinemia type 2 with neonatal onset, characterized by suppression of the bile flow, hepatic fibrosis, low birth weight, growth retardation, hyperproteinemia, variable liver dysfunction. Neonatal intrahepatic cholestasis due to citrin deficiency is generally not severe and symptoms disappear by one year of age with an appropriate diet. Years or even decades later, however, some individuals develop the characteristic features of citrullinemia type 2 with neuropsychiatric symptoms. {ECO:0000269|PubMed:11793471}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sitrüllinemi, Tip 2, Neonatal Başlangıçlı, 605814, Otozomal Resesif (Neonatal Intrahepatik Kolestaz, Sitrin Eksikliğine Bağlı) (Prenatal) (SLC25A13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Citrullinemia, type II, neonatal-onset, 605814, Autosomal recessive (Neonatal intrahepatic cholestasis due to citrin deficiency) (SLC25A13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A13	SLC25A13, CTLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cholestasis, neonatal intrahepatic, caused by citrin deficiency (NICCD) [MIM:605814]: A form of citrullinemia type 2 with neonatal onset, characterized by suppression of the bile flow, hepatic fibrosis, low birth weight, growth retardation, hyperproteinemia, variable liver dysfunction. Neonatal intrahepatic cholestasis due to citrin deficiency is generally not severe and symptoms disappear by one year of age with an appropriate diet. Years or even decades later, however, some individuals develop the characteristic features of citrullinemia type 2 with neuropsychiatric symptoms. {ECO:0000269|PubMed:11793471}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CK Sendromu, 300831, X'e Bağlı Resesif (CK Sendromu) (NSDHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	CK syndrome, 300831, X-linked recessive (CK syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: CK syndrome (CKS) [MIM:300831]: A disorder characterized by mild to severe cognitive impairment, seizures, microcephaly, cerebral cortical malformations, dysmorphic facial features, and thin body habitus. {ECO:0000269|PubMed:21129721}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CK Sendromu, 300831, X'e Bağlı Resesif (CK Sendromu) (Prenatal) (NSDHL geni) (Tüm Gen Dizi Analizi) (Prenatal)	CK syndrome, 300831, X-linked recessive (CK syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: CK syndrome (CKS) [MIM:300831]: A disorder characterized by mild to severe cognitive impairment, seizures, microcephaly, cerebral cortical malformations, dysmorphic facial features, and thin body habitus. {ECO:0000269|PubMed:21129721}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
c-KIT (Ekzon 9 - 11 - 13 ve 17) (Dizi Analizi) (KIT geni) (Dizi Analizi) (Postnatal)	cKIT (Exon9,11,13,17) (Sequence analysis) (KIT gene) (Dizi Analizi) (Postnatal)	KIT	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
CKS1B/CDKN2C (P18) (FISH)	CKS1B/CDKN2C (P18) (FISH)	1q21	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Yarık Dudak / Damak-Ektodermal Displazi Sendromu, 225060, Otozomal Resesif; CLPED1 (Yarık Dudak / Damak) (NECTIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleft lip/palate-ectodermal dysplasia syndrome, 225060, Autosomal recessive; CLPED1 (Cleft lip/palate) (NECTIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NECTIN1	NECTIN1, PVRL1, HVEC, PVRR1, PRR1, EDH, OFC7, CLPED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Ectodermal dysplasia, Margarita Island type (EDMI) [MIM:225060]: An autosomal recessive form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is a syndrome characterized by the association of cleft lip/palate, ectodermal dysplasia (sparse short and dry scalp hair, sparse eyebrows and eyelashes), and partial syndactyly of the fingers and/or toes. Two thirds of the patients do not manifest oral cleft but present with abnormal teeth and nails. {ECO:0000269|PubMed:10932188}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Yarık Dudak / Damak-Ektodermal Displazi Sendromu, 225060, Otozomal Resesif; CLPED1 (Yarık Dudak / Damak) (Prenatal) (NECTIN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cleft lip/palate-ectodermal dysplasia syndrome, 225060, Autosomal recessive; CLPED1 (Cleft lip/palate) (NECTIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NECTIN1	NECTIN1, PVRL1, HVEC, PVRR1, PRR1, EDH, OFC7, CLPED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, Margarita Island type (EDMI) [MIM:225060]: An autosomal recessive form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is a syndrome characterized by the association of cleft lip/palate, ectodermal dysplasia (sparse short and dry scalp hair, sparse eyebrows and eyelashes), and partial syndactyly of the fingers and/or toes. Two thirds of the patients do not manifest oral cleft but present with abnormal teeth and nails. {ECO:0000269|PubMed:10932188}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık Damak-Ankiloglossi, 303400, X'e Bağlı; CPX (X'e Bağlı Yarık Damak ve Ankiloglossi) (TBX22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleft palate with ankyloglossia, 303400, X-linked; CPX (X-linked cleft palate and ankyloglossia) (TBX22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX22	TBX22, CPX, ABERS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Yarık Damak-Ankiloglossi, 303400, X'e Bağlı; CPX (X'e Bağlı Yarık Damak ve Ankiloglossi) (Prenatal) (TBX22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cleft palate with ankyloglossia, 303400, X-linked; CPX (X-linked cleft palate and ankyloglossia) (TBX22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX22	TBX22, CPX, ABERS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık Damak, İzole, 119540, Otozomal Dominant; CPI (Yarık Dudak) (UBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleft palate, isolated, 119540, Autosomal dominant; CPI (Cleft palate) (UBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBB	UBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Yarık Damak, İzole, 119540, Otozomal Dominant; CPI (Yarık Dudak) (Prenatal) (UBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cleft palate, isolated, 119540, Autosomal dominant; CPI (Cleft palate) (UBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBB	UBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık Damak, Psikomotor Retardasyon ve Ayrı Edici Yüz Özellikleri, 616728, Otozomal Dominant; CPRF (Damak Anomalileri-Geniş Aralıklı Dişler-Fasiyel Dismorfizm-Gelişimsel Gecikme Sendromu) (KDM1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleft palate, psychomotor retardation, and distinctive facial features, 616728, Autosomal dominant; CPRF (Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome) (KDM1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDM1A	KDM1A, LSD1, AOF2, BHC110, KIAA0601, CPRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Yarık Damak, Psikomotor Retardasyon ve Ayrı Edici Yüz Özellikleri, 616728, Otozomal Dominant; CPRF (Damak Anomalileri-Geniş Aralıklı Dişler-Fasiyel Dismorfizm-Gelişimsel Gecikme Sendromu) (Prenatal) (KDM1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cleft palate, psychomotor retardation, and distinctive facial features, 616728, Autosomal dominant; CPRF (Palatal anomalies-widely spaced teeth-facial dysmorphism-developmental delay syndrome) (KDM1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KDM1A	KDM1A, LSD1, AOF2, BHC110, KIAA0601, CPRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kleidokranial Displazi, 119600, Otozomal Dominant (Kleidokranial Displazi) (RUNX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleidocranial dysplasia, 119600, Autosomal dominant (Cleidocranial dysplasia) (RUNX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kleidokranial Displazi, 119600, Otozomal Dominant (Kleidokranial Displazi) (MLPA)	Cleidocranial dysplasia, 119600, Autosomal dominant (Cleidocranial dysplasia) (MLPA)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Kleidokranial Displazi, Forme Fruste, Sadece Dış Anomalileri, 119600, Otozomal Dominant (Kleidokranial Displazi) (RUNX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600, Autosomal dominant (Cleidocranial dysplasia) (RUNX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kleidokranial Displazi, Forme Fruste, Sadece Dış Anomalileri, 119600, Otozomal Dominant (Kleidokranial Displazi) (MLPA)	Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600, Autosomal dominant (Cleidocranial dysplasia) (MLPA)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hasta Muayenesi	Clinical evaluation and examination (.)	.	.	Klinik değerlendirme/ Clinical Evaluation	Tüm Tıbbi Dokümanlar ve Ale Fotoğrafları		0	0	.	.	0	Postnatal	.	Medical Documents and Family Photos
Klopidogrel İçin Bozulmuş Yanıt, 609535, Otozomal Resesif (Klopidogrel Direnci) (CYP2C19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Clopidogrel, impaired responsiveness to, 609535, Autosomal recessive (Resistance to clopidogrel) (CYP2C19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C19	CYP2C, CYP2C19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Klopidogrel İçin Bozulmuş Yanıt, 609535, Otozomal Resesif (Klopidogrel Direnci) (MLPA)	Clopidogrel, impaired responsiveness to, 609535, Autosomal recessive (Resistance to clopidogrel) (MLPA)	CYP2C19	CYP2C, CYP2C19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Clouston Sendromu (Hidrotik Ektodermal Displazi) (GJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CLOUSTON SYNDROME (Hidrotic ectodermal dysplasia) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 2, Clouston type (ECTD2) [MIM:129500]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD2 is an autosomal dominant condition characterized by atrichosis, nail hypoplasia and deformities, hyperpigmentation of the skin, normal teeth, normal sweat and sebaceous gland function. Palmoplantar hyperkeratosis is a frequent feature. Hearing impairment has been detected in few cases. {ECO:0000269|PubMed:11017065, ECO:0000269|PubMed:11874494}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Clouston Sendromu (Hidrotik Ektodermal Displazi) (Prenatal) (GJB6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CLOUSTON SYNDROME (Hidrotic ectodermal dysplasia) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 2, Clouston type (ECTD2) [MIM:129500]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD2 is an autosomal dominant condition characterized by atrichosis, nail hypoplasia and deformities, hyperpigmentation of the skin, normal teeth, normal sweat and sebaceous gland function. Palmoplantar hyperkeratosis is a frequent feature. Hearing impairment has been detected in few cases. {ECO:0000269|PubMed:11017065, ECO:0000269|PubMed:11874494}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CLOVE Sendromu, Somatik, 612918 (CLOVES Sendromu) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	CLOVE syndrome, somatic, 612918 (CLOVES syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLOVE Sendromu, Somatik, 612918 (CLOVES Sendromu) (Prenatal) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Prenatal)	CLOVE syndrome, somatic, 612918 (CLOVES syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Clubfoot, Konjenital, ± Uzun Kemik Eksikliği Ve/Veya Ayna (Mirror-İmage) Polidaktili, 119800, Otozomal Dominant; CCF (Ailesel Çarpık Ayak ± Eşlik Eden Alt Ekstremite Anomalileri) (PITX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800, Autosomal dominant; CCF (Familial clubfoot with or without associated lower limb anomalies) (PITX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX1	PITX1, PTX1, BFT, POTX, CCF, LBNBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly (CCF) [MIM:119800]: A congenital limb deformity defined as fixation of the foot in cavus, adductus, varus, and equinus (i.e., inclined inwards, axially rotated outwards, and pointing downwards) with concomitant soft tissue abnormalities. Clubfoot may occur in isolation or as part of a syndrome. Some patients present tibial hemimelia, bilateral patellar hypoplasia, and preaxial mirror-image polydactyly. {ECO:0000269|PubMed:18950742, ECO:0000269|PubMed:22258522}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Clubfoot, Konjenital, ± Uzun Kemik Eksikliği Ve/Veya Ayna (Mirror-İmage) Polidaktili, 119800, Otozomal Dominant; CCF (Ailesel Çarpık Ayak ± Eşlik Eden Alt Ekstremite Anomalileri) (Prenatal) (PITX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly, 119800, Autosomal dominant; CCF (Familial clubfoot with or without associated lower limb anomalies) (PITX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PITX1	PITX1, PTX1, BFT, POTX, CCF, LBNBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly (CCF) [MIM:119800]: A congenital limb deformity defined as fixation of the foot in cavus, adductus, varus, and equinus (i.e., inclined inwards, axially rotated outwards, and pointing downwards) with concomitant soft tissue abnormalities. Clubfoot may occur in isolation or as part of a syndrome. Some patients present tibial hemimelia, bilateral patellar hypoplasia, and preaxial mirror-image polydactyly. {ECO:0000269|PubMed:18950742, ECO:0000269|PubMed:22258522}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
cMYC ile IGH, IGL ve IGK Translokasyonları (Burkit Lenfoma) t(8;14) , t(8;22) ve t(2;8) (FISH)	cMYC/ IGH, IGL and IGK Translocation (Burkit Lymphoma) t(8;14) , t(8;22) and t(2;8) (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	COACH syndrome, 216360, Autosomal recessive (Joubert syndrome with hepatic defect) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269|PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	COACH syndrome, 216360, Autosomal recessive (Joubert syndrome with hepatic defect) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269|PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (Prenatal) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	COACH syndrome, 216360, Autosomal recessive (Joubert syndrome with hepatic defect) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269|PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (Prenatal) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	COACH syndrome, 216360, Autosomal recessive (Joubert syndrome with hepatic defect) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269|PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Postnatal)	COACH syndrome, 216360, Autosomal recessive; JBTS6 (Joubert syndrome with hepatic defect) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COACH Sendromu, 216360, Otozomal Resesif (Joubert Sendromu-Hepatik Defekt (COACH Sendromu)) (Prenatal) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Prenatal)	COACH syndrome, 216360, Autosomal recessive; JBTS6 (Joubert syndrome with hepatic defect) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Cockayne Sendromu, Tip A, 216400, Otozomal Resesif; CSA (Cockayne Sendromu) (ERCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cockayne syndrome, type A, 216400, Autosomal recessive; CSA (Cockayne syndrome) (ERCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC8	ERCC8, CKN1, CSA, UVSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome A (CSA) [MIM:216400]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:14661080, ECO:0000269|PubMed:15744458, ECO:0000269|PubMed:19894250}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Cockayne Sendromu, Tip A, 216400, Otozomal Resesif; CSA (Cockayne Sendromu) (Prenatal) (ERCC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cockayne syndrome, type A, 216400, Autosomal recessive; CSA (Cockayne syndrome) (ERCC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC8	ERCC8, CKN1, CSA, UVSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cockayne syndrome A (CSA) [MIM:216400]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:14661080, ECO:0000269|PubMed:15744458, ECO:0000269|PubMed:19894250}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cockayne Sendromu, Tip B, 133540, Otozomal Resesif; CSB (Cockayne Sendromu) (ERCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cockayne syndrome, type B, 133540, Autosomal recessive; CSB (Cockayne syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:9443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Cockayne Sendromu, Tip B, 133540, Otozomal Resesif; CSB (Cockayne Sendromu) (Prenatal) (ERCC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cockayne syndrome, type B, 133540, Autosomal recessive; CSB (Cockayne syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:9443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koza (Cocoon) Sendromu, 613630 (Fetal Kapatma Sendromu) (CHUK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cocoon syndrome, 613630 (Fetal encasement syndrome) (CHUK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHUK	CHUK, IKBKA, NFKBIKA, IKKA, IKK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cocoon syndrome (COCOS) [MIM:613630]: A lethal syndrome characterized by multiple fetal malformations including defective face and seemingly absent limbs, which are bound to the trunk and encased under the skin. {ECO:0000269|PubMed:20961246}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koza (Cocoon) Sendromu, 613630 (Fetal Kapatma Sendromu) (Prenatal) (CHUK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cocoon syndrome, 613630 (Fetal encasement syndrome) (CHUK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHUK	CHUK, IKBKA, NFKBIKA, IKKA, IKK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cocoon syndrome (COCOS) [MIM:613630]: A lethal syndrome characterized by multiple fetal malformations including defective face and seemingly absent limbs, which are bound to the trunk and encased under the skin. {ECO:0000269|PubMed:20961246}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CODAS Sendromu, 600373, Otozomal Resesif (CODAS Sendromu) (LONP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CODAS syndrome, 600373, Autosomal recessive (CODAS syndrome) (LONP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LONP1	LONP1, PRSS15, LON, CODASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CODAS Sendromu, 600373, Otozomal Resesif (CODAS Sendromu) (Prenatal) (LONP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CODAS syndrome, 600373, Autosomal recessive (CODAS syndrome) (LONP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LONP1	LONP1, PRSS15, LON, CODASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kodein Yatkınlık, 608902, Otozomal Resesif (CYP2D6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Codeine sensitivity, 608902, Autosomal recessive (CYP2D6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2D6	CYP2D6, CPD6, P450DB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kodein Yatkınlık, 608902, Otozomal Resesif (MLPA)	Codeine sensitivity, 608902, Autosomal recessive (MLPA)	CYP2D6	CYP2D6, CPD6, P450DB1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 1, 607426, Otozomal Resesif; COQ10D1 (Leigh Sendromu-Nefrotik Sendrom) (COQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 1, 607426, Autosomal recessive; COQ10D1 (Leigh syndrome with nephrotic syndrome) (COQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ2	COQ2, COQ10D1, MSA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 1, 607426, Otozomal Resesif; COQ10D1 (Leigh Sendromu-Nefrotik Sendrom) (Prenatal) (COQ2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 1, 607426, Autosomal recessive; COQ10D1 (Leigh syndrome with nephrotic syndrome) (COQ2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ2	COQ2, COQ10D1, MSA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 2, 614651, Otozomal Resesif; COQ10D2 (Sağırlık-Ensefalonöropati-Obezite-Valvülopati Sendromu) (PDSS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 2, 614651, Autosomal recessive; COQ10D2 (Deafness-encephaloneuropathy-obesity-valvulopathy syndrome) (PDSS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDSS1	PDSS1, TPT, COQ1, COQ10D2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coenzyme Q10 deficiency, primary, 2 (COQ10D2) [MIM:614651]: An autosomal recessive multisystem disorder characterized by early-onset deafness, optic atrophy, mild mental retardation, peripheral neuropathy, obesity, liveds reticularis, and cardiac valvulopathy. {ECO:0000269|PubMed:17332895}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 2, 614651, Otozomal Resesif; COQ10D2 (Sağırlık-Ensefalonöropati-Obezite-Valvülopati Sendromu) (Prenatal) (PDSS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 2, 614651, Autosomal recessive; COQ10D2 (Deafness-encephaloneuropathy-obesity-valvulopathy syndrome) (PDSS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDSS1	PDSS1, TPT, COQ1, COQ10D2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coenzyme Q10 deficiency, primary, 2 (COQ10D2) [MIM:614651]: An autosomal recessive multisystem disorder characterized by early-onset deafness, optic atrophy, mild mental retardation, peripheral neuropathy, obesity, liveds reticularis, and cardiac valvulopathy. {ECO:0000269|PubMed:17332895}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 3, 614652, Otozomal Resesif; COQ10D3 (Leigh Sendromu-Nefrotik Sendrom) (PDSS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 3, 614652, Autosomal recessive; COQ10D3 (Leigh syndrome with nephrotic syndrome) (PDSS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDSS2	PDSS2, DLP1, C6orf210, COQ10D3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coenzyme Q10 deficiency, primary, 3 (COQ10D3) [MIM:614652]: A fatal encephalomyopathic form of coenzyme Q10 deficiency with nephrotic syndrome. Coenzyme Q10 deficiency is an autosomal recessive disorder with variable manifestations consistent with 5 major phenotypes. The phenotypes include an encephalomyopathic form with seizures and ataxia; a multisystem infantile form with encephalopathy, cardiomyopathy and renal failure; a predominantly cerebellar form with ataxia and cerebellar atrophy; Leigh syndrome with growth retardation; and an isolated myopathic form. {ECO:0000269|PubMed:17186472}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 3, 614652, Otozomal Resesif; COQ10D3 (Leigh Sendromu-Nefrotik Sendrom) (Prenatal) (PDSS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 3, 614652, Autosomal recessive; COQ10D3 (Leigh syndrome with nephrotic syndrome) (PDSS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDSS2	PDSS2, DLP1, C6orf210, COQ10D3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coenzyme Q10 deficiency, primary, 3 (COQ10D3) [MIM:614652]: A fatal encephalomyopathic form of coenzyme Q10 deficiency with nephrotic syndrome. Coenzyme Q10 deficiency is an autosomal recessive disorder with variable manifestations consistent with 5 major phenotypes. The phenotypes include an encephalomyopathic form with seizures and ataxia; a multisystem infantile form with encephalopathy, cardiomyopathy and renal failure; a predominantly cerebellar form with ataxia and cerebellar atrophy; Leigh syndrome with growth retardation; and an isolated myopathic form. {ECO:0000269|PubMed:17186472}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 4, 612016, Otozomal Resesif; COQ10D4 (Otozomal Resesif Ataksi, Ubikinon Eksikliğine Bağlı) (COQ8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 4, 612016, Autosomal recessive; COQ10D4 (Autosomal recessive ataxia due to ubiquinone deficiency) (COQ8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ8A	ADCK3, COQ8, CABC1, SCAR9, ARCA2, COQ10D4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 4, 612016, Otozomal Resesif; COQ10D4 (Otozomal Resesif Ataksi, Ubikinon Eksikliğine Bağlı) (Prenatal) (COQ8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 4, 612016, Autosomal recessive; COQ10D4 (Autosomal recessive ataxia due to ubiquinone deficiency) (COQ8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ8A	ADCK3, COQ8, CABC1, SCAR9, ARCA2, COQ10D4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Koenzim Q10 Eksikliği, Primer, 5, 614654, Otozomal Resesif; COQ10D5 (Ensefalopati-Hipertrofik Kardiyomiyopati Renal Tübüler Hastalık Sendromu) (COQ9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 5, 614654, Autosomal recessive; COQ10D5 (Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome) (COQ9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ9	COQ9, C16orf49, COQ10D5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coenzyme Q10 deficiency, primary, 5 (COQ10D5) [MIM:614654]: A form of coenzyme Q10 deficiency, an autosomal recessive disorder with variable manifestations consistent with 5 major phenotypes. The phenotypes include an encephalomyopathic form with seizures and ataxia; a multisystem infantile form with encephalopathy, cardiomyopathy and renal failure; a predominantly cerebellar form with ataxia and cerebellar atrophy; Leigh syndrome with growth retardation; and an isolated myopathic form. {ECO:0000269|PubMed:19375058}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 5, 614654, Otozomal Resesif; COQ10D5 (Ensefalopati-Hipertrofik Kardiyomiyopati Renal Tübüler Hastalık Sendromu) (Prenatal) (COQ9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 5, 614654, Autosomal recessive; COQ10D5 (Encephalopathy-hypertrophic cardiomyopathy-renal tubular disease syndrome) (COQ9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ9	COQ9, C16orf49, COQ10D5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coenzyme Q10 deficiency, primary, 5 (COQ10D5) [MIM:614654]: A form of coenzyme Q10 deficiency, an autosomal recessive disorder with variable manifestations consistent with 5 major phenotypes. The phenotypes include an encephalomyopathic form with seizures and ataxia; a multisystem infantile form with encephalopathy, cardiomyopathy and renal failure; a predominantly cerebellar form with ataxia and cerebellar atrophy; Leigh syndrome with growth retardation; and an isolated myopathic form. {ECO:0000269|PubMed:19375058}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 6, 614650, Otozomal Resesif; COQ10D6 (Ailesel Steroide Dirençli Nefrotik Sendrom-Sensorinöral Sağırlık) (COQ6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 6, 614650, Autosomal recessive; COQ10D6 (Familial steroid-resistant nephrotic syndrome with sensorineural deafness) (COQ6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ6	COQ6, CGI10, COQ10D6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 6, 614650, Otozomal Resesif; COQ10D6 (Ailesel Steroide Dirençli Nefrotik Sendrom-Sensorinöral Sağırlık) (Prenatal) (COQ6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 6, 614650, Autosomal recessive; COQ10D6 (Familial steroid-resistant nephrotic syndrome with sensorineural deafness) (COQ6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ6	COQ6, CGI10, COQ10D6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 7, 616276, Otozomal Resesif; COQ10D7 (Neonatal Ensefalomyopati-Kardiyomiyopati ve Solunum Zorluğu Sendromu) (COQ4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 7, 616276, Autosomal recessive; COQ10D7 (Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome) (COQ4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ4	COQ4, COQ10D7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 7, 616276, Otozomal Resesif; COQ10D7 (Neonatal Ensefalomyopati-Kardiyomiyopati ve Solunum Zorluğu Sendromu) (Prenatal) (COQ4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 7, 616276, Autosomal recessive; COQ10D7 (Neonatal encephalomyopathy-cardiomyopathy-respiratory distress syndrome) (COQ4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ4	COQ4, COQ10D7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 8, 616733, Otozomal Resesif; COQ10D8 (COQ7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coenzyme Q10 deficiency, primary, 8, 616733, Autosomal recessive; COQ10D8 (COQ7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ7	COQ7, CLK1, COQ10D8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koenzim Q10 Eksikliği, Primer, 8, 616733, Otozomal Resesif; COQ10D8 (COQ7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coenzyme Q10 deficiency, primary, 8, 616733, Autosomal recessive; COQ10D8 (COQ7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ7	COQ7, CLK1, COQ10D8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Lowry Sendromu, 303600 X'e Bağlı Dominant, Sporadik Vakalar; CLS (Coffin-Lowry Sendromu) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Lowry syndrome, 303600, X-linked dominant, isolated cases; CLS (Coffin-Lowry syndrome) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coffin-Lowry Sendromu, 303600 X'e Bağlı Dominant, Sporadik Vakalar; CLS (Coffin-Lowry Sendromu) (Prenatal) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Lowry syndrome, 303600, X-linked dominant, isolated cases; CLS (Coffin-Lowry syndrome) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 1, 135900, Otozomal Dominant; CSS1 (Coffin-Siris Sendromu) (ARID1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Siris syndrome 1, 135900, Autosomal dominant; CSS1 (Coffin-Siris syndrome) (ARID1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARID1B	ARID1B, BAF250B, KIAA1235, MRD12, CSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 1, 135900, Otozomal Dominant; CSS1 (Coffin-Siris Sendromu) (MLPA)	Coffin-Siris syndrome 1, 135900, Autosomal dominant; CSS1 (Coffin-Siris syndrome) (MLPA)	ARID1B	ARID1B, BAF250B, KIAA1235, MRD12, CSS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 1, 135900, Otozomal Dominant; CSS1 (Coffin-Siris Sendromu) (Prenatal) (ARID1B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Siris syndrome 1, 135900, Autosomal dominant; CSS1 (Coffin-Siris syndrome) (ARID1B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARID1B	ARID1B, BAF250B, KIAA1235, MRD12, CSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 1, 135900, Otozomal Dominant; CSS1 (Coffin-Siris Sendromu) (Prenatal) (MLPA)	Coffin-Siris syndrome 1, 135900, Autosomal dominant; CSS1 (Coffin-Siris syndrome) (MLPA)	ARID1B	ARID1B, BAF250B, KIAA1235, MRD12, CSS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 2, 614607, Otozomal Dominant; CSS2 (Coffin-Siris Sendromu) (ARID1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Siris syndrome 2, 614607, Autosomal dominant; CSS2 (Coffin-Siris syndrome) (ARID1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARID1A	ARID1A, C1orf4, B120, SMARCF1, MRD14, CSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 2, 614607, Otozomal Dominant; CSS2 (Coffin-Siris Sendromu) (MLPA)	Coffin-Siris syndrome 2, 614607, Autosomal dominant; CSS2 (Coffin-Siris syndrome) (MLPA)	ARID1A	ARID1A, C1orf4, B120, SMARCF1, MRD14, CSS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 2, 614607, Otozomal Dominant; CSS2 (Coffin-Siris Sendromu) (Prenatal) (ARID1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Siris syndrome 2, 614607, Autosomal dominant; CSS2 (Coffin-Siris syndrome) (ARID1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARID1A	ARID1A, C1orf4, B120, SMARCF1, MRD14, CSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 2, 614607, Otozomal Dominant; CSS2 (Coffin-Siris Sendromu) (Prenatal) (MLPA)	Coffin-Siris syndrome 2, 614607, Autosomal dominant; CSS2 (Coffin-Siris syndrome) (Prenatal) (MLPA)	ARID1A	ARID1A, C1orf4, B120, SMARCF1, MRD14, CSS2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 3, 614608, Otozomal Dominant; CSS3 (Coffin-Siris Sendromu) (SMARCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Siris syndrome 3, 614608, Autosomal dominant; CSS3 (Coffin-Siris syndrome) (SMARCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCB1	SMARCB1, SNF5, INI1, RDT, RTPS1, MRD15, SWNTS1, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 3, 614608, Otozomal Dominant; CSS3 (Coffin-Siris Sendromu) (Prenatal) (SMARCB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Siris syndrome 3, 614608, Autosomal dominant; CSS3 (Coffin-Siris syndrome) (SMARCB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCB1	SMARCB1, SNF5, INI1, RDT, RTPS1, MRD15, SWNTS1, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 4, 614609, Otozomal Dominant; CSS4 (Coffin-Siris Sendromu) (SMARCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Siris syndrome 4, 614609, Autosomal dominant; CSS4 (Coffin-Siris syndrome) (SMARCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCA4	SMARCA4, BRG1, RTPS2, MRD16, CSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 16 (MRD16) [MIM:614609]: A disease characterized by multiple congenital anomalies and mental retardation. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD16 patients manifest developmental delay, absent or hypoplastic fifth fingernails or toenails, thick eyebrows and long eyelashes, hirsutism. Additional findings include hypotonia, microcephaly, seizures, a Dandy-Walker malformation, and vision and hearing problems. {ECO:0000269|PubMed:22426308}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Coffin-Siris Sendromu 4, 614609, Otozomal Dominant; CSS4 (Coffin-Siris Sendromu) (Prenatal) (SMARCA4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Siris syndrome 4, 614609, Autosomal dominant; CSS4 (Coffin-Siris syndrome) (SMARCA4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCA4	SMARCA4, BRG1, RTPS2, MRD16, CSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 16 (MRD16) [MIM:614609]: A disease characterized by multiple congenital anomalies and mental retardation. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD16 patients manifest developmental delay, absent or hypoplastic fifth fingernails or toenails, thick eyebrows and long eyelashes, hirsutism. Additional findings include hypotonia, microcephaly, seizures, a Dandy-Walker malformation, and vision and hearing problems. {ECO:0000269|PubMed:22426308}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris Sendromu 5, 616938, Otozomal Dominant; CSS5 (Coffin-Siris Sendromu) (SMARCE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coffin-Siris syndrome 5, 616938, Autosomal dominant; CSS5 (Coffin-Siris syndrome) (SMARCE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCE1	SMARCE1, BAF57, CSS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coffin-Siris Sendromu 5, 616938, Otozomal Dominant; CSS5 (Coffin-Siris Sendromu) (Prenatal) (SMARCE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coffin-Siris syndrome 5, 616938, Autosomal dominant; CSS5 (Coffin-Siris syndrome) (SMARCE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCE1	SMARCE1, BAF57, CSS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coffin-Siris sendromu panel-Nöroblastoma, over kanseri + çeşitli diğer tümör tipleri (ARID1A, ARID1B) (MLPA)	Coffin-Siris syndrome panel- Neuroblastoma, ovarian cancer + various other tumour types (ARID1A, ARID1B) (MLPA)	ARID1A, ARID1B	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Coffin-Siris sendromu panel-Nöroblastoma, over kanseri + çeşitli diğer tümör tipleri (ARID1A, ARID1B) (MLPA) (Prenatal)	Coffin-Siris syndrome panel- Neuroblastoma, ovarian cancer + various other tumour types (ARID1A, ARID1B) (MLPA) (Prenatal)	ARID1A, ARID1B	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kognitif Yetersizlik, ± Serebellar Ataksi, 614306, Otozomal Dominant; CIAT (SCN8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cognitive impairment with or without cerebellar ataxia, 614306, Autosomal dominant; CIAT (SCN8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN8A	SCN8A, CIAT, EIEE13, BFIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cognitive impairment with or without cerebellar ataxia (CIAT) [MIM:614306]: A disorder characterized by markedly delayed cognitive and motor development, attention deficit disorder, and cerebellar ataxia. Features include bilateral esophoria, strabismatic amblyopia, unsustained gaze evoked nystagmus on horizontal gaze, ataxic gait, dysmetria in the upper limbs and dysarthria, with normal strength, tone, and reflexes. {ECO:0000269|PubMed:16236810}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kognitif Yetersizlik, ± Serebellar Ataksi, 614306, Otozomal Dominant; CIAT (Prenatal) (SCN8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cognitive impairment with or without cerebellar ataxia, 614306, Autosomal dominant; CIAT (SCN8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN8A	SCN8A, CIAT, EIEE13, BFIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cognitive impairment with or without cerebellar ataxia (CIAT) [MIM:614306]: A disorder characterized by markedly delayed cognitive and motor development, attention deficit disorder, and cerebellar ataxia. Features include bilateral esophoria, strabismatic amblyopia, unsustained gaze evoked nystagmus on horizontal gaze, ataxic gait, dysmetria in the upper limbs and dysarthria, with normal strength, tone, and reflexes. {ECO:0000269|PubMed:16236810}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cohen Sendromu, 216550, Otozomal Resesif; COH1 (Cohen Sendromu) (VPS13B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cohen syndrome, 216550, Autosomal recessive; COH1 (Cohen syndrome) (VPS13B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS13B	VPS13B, KIAA0532, COH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cohen Sendromu, 216550, Otozomal Resesif; COH1 (Cohen Sendromu) (Prenatal) (VPS13B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cohen syndrome, 216550, Autosomal recessive; COH1 (Cohen syndrome) (VPS13B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS13B	VPS13B, KIAA0532, COH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Colchicine Direnci, 120080 (ABCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colchicine resistance, 120080 (ABCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB1	ABCB1, PGY1, MDR1, IBD13, CLCs	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Soğukla Uyarılan Terleme Sendromu 1, 272430, Otozomal Resesif; CISS1 (Soğukla Uyarılan Terleme Sendromu) (CRLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cold-induced sweating syndrome 1, 272430, Autosomal recessive; CISS1 (Cold-induced sweating syndrome) (CRLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRLF1	CRLF1, CISS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cold-induced sweating syndrome 1 (CISS1) [MIM:272430]: An autosomal recessive disorder characterized by profuse sweating induced by cool surroundings (temperatures of 7 to 18 degrees Celsius). Patients manifest, in the neonatal period, orofacial weakness with impaired sucking and swallowing, resulting in poor feeding. Affected infants show a tendency to startle, with contractions of the facial muscles in response to tactile stimuli or during crying, trismus, abundant salivation, and opisthotonus. These features are referred to as Crisponi syndrome and can result in early death in infancy. Patients who survive into childhood have hyperhidrosis, mainly of the upper body, in response to cold temperatures, and sweat very little with heat. Additional abnormalities include a high-arched palate, nasal voice, depressed nasal bridge, inability to fully extend the elbows and kyphoscoliosis. {ECO:0000269|PubMed:12509788, ECO:0000269|PubMed:16952376, ECO:0000269|PubMed:17436251, ECO:0000269|PubMed:17436252, ECO:0000269|PubMed:21326283, ECO:0000269|PubMed:23026229, ECO:0000269|PubMed:24488861}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Soğukla Uyarılan Terleme Sendromu 1, 272430, Otozomal Resesif; CISS1 (Soğukla Uyarılan Terleme Sendromu) (Prenatal) (CRLF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cold-induced sweating syndrome 1, 272430, Autosomal recessive; CISS1 (Cold-induced sweating syndrome) (CRLF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRLF1	CRLF1, CISS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cold-induced sweating syndrome 1 (CISS1) [MIM:272430]: An autosomal recessive disorder characterized by profuse sweating induced by cool surroundings (temperatures of 7 to 18 degrees Celsius). Patients manifest, in the neonatal period, orofacial weakness with impaired sucking and swallowing, resulting in poor feeding. Affected infants show a tendency to startle, with contractions of the facial muscles in response to tactile stimuli or during crying, trismus, abundant salivation, and opisthotonus. These features are referred to as Crisponi syndrome and can result in early death in infancy. Patients who survive into childhood have hyperhidrosis, mainly of the upper body, in response to cold temperatures, and sweat very little with heat. Additional abnormalities include a high-arched palate, nasal voice, depressed nasal bridge, inability to fully extend the elbows and kyphoscoliosis. {ECO:0000269|PubMed:12509788, ECO:0000269|PubMed:16952376, ECO:0000269|PubMed:17436251, ECO:0000269|PubMed:17436252, ECO:0000269|PubMed:21326283, ECO:0000269|PubMed:23026229, ECO:0000269|PubMed:24488861}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Soğukla Uyarılan Terleme Sendromu 2, 610313, Otozomal Resesif; CISS2 (Soğukla Uyarılan Terleme Sendromu) (CLCF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cold-induced sweating syndrome 2, 610313, Autosomal recessive; CISS2 (Cold-induced sweating syndrome) (CLCF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCF1	CLCF1, BSF3, CLC, CISS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Soğukla Uyarılan Terleme Sendromu 2, 610313, Otozomal Resesif; CISS2 (Soğukla Uyarılan Terleme Sendromu) (Prenatal) (CLCF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cold-induced sweating syndrome 2, 610313, Autosomal recessive; CISS2 (Cold-induced sweating syndrome) (CLCF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCF1	CLCF1, BSF3, CLC, CISS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Soğukla Uyarılan Terleme Sendromu 3, 617055; CISS3 (Soğukla Uyarılan Terleme Sendromu) (KLHL7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cold-induced sweating syndrome 3, 617055; CISS3 (Cold-induced sweating syndrome) (KLHL7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL7	KLHL7, RP42, CISS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Soğukla Uyarılan Terleme Sendromu 3, 617055; CISS3 (Soğukla Uyarılan Terleme Sendromu) (Prenatal) (KLHL7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cold-induced sweating syndrome 3, 617055; CISS3 (Cold-induced sweating syndrome) (KLHL7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLHL7	KLHL7, RP42, CISS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cole Hastalığı, 615522, Otozomal Dominant; COLED (Hipopigmentasyon-Punktat Palmoplantar Keratoderma Sendromu) (ENPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cole disease, 615522, Autosomal dominant; COLED (Hypopigmentation-punctate palmoplantar keratoderma syndrome) (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENPP1	ENPP1, PDNP1, NPPS, M6S1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cole Hastalığı, 615522, Otozomal Dominant; COLED (Hipopigmentasyon-Punktat Palmoplantar Keratoderma Sendromu) (Prenatal) (ENPP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cole disease, 615522, Autosomal dominant; COLED (Hypopigmentation-punctate palmoplantar keratoderma syndrome) (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ENPP1	ENPP1, PDNP1, NPPS, M6S1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cole-Carpenter Sendromu 1, 112240, Otozomal Dominant; CLCRP1 (Cole-Carpenter Sendromu) (P4HB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cole-Carpenter syndrome 1, 112240, Autosomal dominant; CLCRP1 (Cole-Carpenter syndrome) (P4HB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P4HB	P4HB, PROHB, CLCRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cole-Carpenter syndrome 1 (CLCRP1) [MIM:112240]: A form of Cole-Carpenter syndrome, a disorder characterized by features of osteogenesis imperfecta such as bone deformities and severe bone fragility with frequent fractures, in association with craniosynostosis, ocular proptosis, hydrocephalus, growth failure and distinctive facial features. Craniofacial findings include marked frontal bossing, midface hypoplasia, and micrognathia. Despite the craniosynostosis and hydrocephalus, intellectual development is normal. CLCRP1 inheritance is autosomal dominant. {ECO:0000269|PubMed:25683117}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cole-Carpenter Sendromu 1, 112240, Otozomal Dominant; CLCRP1 (Cole-Carpenter Sendromu) (Prenatal) (P4HB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cole-Carpenter syndrome 1, 112240, Autosomal dominant; CLCRP1 (Cole-Carpenter syndrome) (P4HB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	P4HB	P4HB, PROHB, CLCRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cole-Carpenter syndrome 1 (CLCRP1) [MIM:112240]: A form of Cole-Carpenter syndrome, a disorder characterized by features of osteogenesis imperfecta such as bone deformities and severe bone fragility with frequent fractures, in association with craniosynostosis, ocular proptosis, hydrocephalus, growth failure and distinctive facial features. Craniofacial findings include marked frontal bossing, midface hypoplasia, and micrognathia. Despite the craniosynostosis and hydrocephalus, intellectual development is normal. CLCRP1 inheritance is autosomal dominant. {ECO:0000269|PubMed:25683117}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cole-Carpenter Sendromu 2, 616294, Otozomal Resesif; CLCRP2 (Cole-Carpenter Sendromu) (SEC24D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cole-Carpenter syndrome 2, 616294, Autosomal recessive; CLCRP2 (Cole-Carpenter syndrome) (SEC24D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC24D	SEC24D, KIAA0755, CLCRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cole-Carpenter Sendromu 2, 616294, Otozomal Resesif; CLCRP2 (Cole-Carpenter Sendromu) (Prenatal) (SEC24D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cole-Carpenter syndrome 2, 616294, Autosomal recessive; CLCRP2 (Cole-Carpenter syndrome) (SEC24D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEC24D	SEC24D, KIAA0755, CLCRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Sinir Kolobomu, 120430, Otozomal Dominant (Morning Glory Sendromu) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coloboma of optic nerve, 120430, Autosomal dominant (Morning glory syndrome) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Optik Sinir Kolobomu, 120430, Otozomal Dominant (Morning Glory Sendromu) (MLPA)	Coloboma of optic nerve, 120430, Autosomal dominant (Morning glory syndrome) (MLPA)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolobom, Konjenital Kalp Hastalığı, İktiyoziform Dermatoz, Mental Retardasyon ve Kulak Anomalileri Sendromu; CHIME (CHIME Sendromu) (PIGL geni) (Tüm Gen Dizi Analizi) (Postnatal)	COLOBOMA, CONGENITAL HEART DISEASE, ICHTHYOSIFORM DERMATOSIS, MENTAL RETARDATION, AND EAR ANOMALIES SYNDROME, CHIME (CHME syndrome) (PIGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGL	PIGL, CHIME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation and ear anomalies syndrome (CHME) [MIM:280000]: An extremely rare autosomal recessive multisystem disorder clinically characterized by colobomas, congenital heart defects, migratory ichthyosiform dermatosis, mental retardation, and ear anomalies including conductive hearing loss. Other clinical features include distinctive facial features, abnormal growth, genitourinary abnormalities, seizures, and feeding difficulties. {ECO:0000269|PubMed:22444671}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kolobom, Konjenital Kalp Hastalığı, İktiyoziform Dermatoz, Mental Retardasyon ve Kulak Anomalileri Sendromu; CHIME (CHIME Sendromu) (Prenatal) (PIGL geni) (Tüm Gen Dizi Analizi) (Prenatal)	COLOBOMA, CONGENITAL HEART DISEASE, ICHTHYOSIFORM DERMATOSIS, MENTAL RETARDATION, AND EAR ANOMALIES SYNDROME, CHIME (CHME syndrome) (PIGL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGL	PIGL, CHIME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coloboma, congenital heart disease, ichthyosiform dermatosis, mental retardation and ear anomalies syndrome (CHME) [MIM:280000]: An extremely rare autosomal recessive multisystem disorder clinically characterized by colobomas, congenital heart defects, migratory ichthyosiform dermatosis, mental retardation, and ear anomalies including conductive hearing loss. Other clinical features include distinctive facial features, abnormal growth, genitourinary abnormalities, seizures, and feeding difficulties. {ECO:0000269|PubMed:22444671}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolobom, Oküler, 120200, Otozomal Dominant (Oküler Kolobomu) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coloboma, ocular, 120200, Autosomal dominant (Ocular coloboma) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolobom, Oküler, 120200, Otozomal Dominant (Oküler Kolobomu) (MLPA)	Coloboma, ocular, 120200, Autosomal dominant (Ocular coloboma) (MLPA)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolobom, Oküler, 120433, Otozomal Dominant (Uveal Koloboma-Yarık Dudak ve Damak-Entelektüel Yetersizlik) (YAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coloboma, ocular, 120433, Autosomal dominant (Uveal coloboma-cleft lip and palate-intellectual disability) (YAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YAP1	YAP1, COB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation (COB1) [MIM:120433]: An autosomal dominant disease characterized by uveal colobomata, microphthalmia, cataract and cleft lip/palate. Considerable variability is observed among patients. uveal colobomata being the most constant feature. Some patients manifest mental retardation of varying degree and/or sensorineural, mid-frequency hearing loss. {ECO:0000269|PubMed:24462371}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kolobom, Oküler, Otozomal Resesif, 216820, Otozomal Resesif (Oküler Koloboma) (SALL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coloboma, ocular, autosomal recessive, 216820, Autosomal recessive (Ocular coloboma) (SALL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SALL2	SALL2, HSAL2, COLB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coloboma, ocular, autosomal recessive (COAR) [MIM:216820]: An ocular anomaly resulting from abnormal morphogenesis of the optic cup and stalk, and incomplete fusion of the fetal intra-ocular fissure during gestation. The clinical presentation is variable. Some individuals may present with minimal defects in the anterior iris leaf without other ocular defects. More complex malformations create a combination of iris, uveoretinal and/or optic nerve defects without or with microphthalmia or even anophthalmia. {ECO:0000269|PubMed:24412933}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kolobom, Oküler, ± İşitme Engeli, Yank Dudak / Damak ve / Veya Mental Retardasyon, 120433, Otozomal Dominant; COB1 (Uveal Koloboma-Yank Dudak ve Damak-Entelektüel Yetersizlik) (YAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433, Autosomal dominant; COB1 (Uveal coloboma-cleft lip and palate-intellectual disability) (YAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YAP1	YAP1, COB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation (COB1) [MIM:120433]: An autosomal dominant disease characterized by uveal colobomata, microphthalmia, cataract and cleft lip/palate. Considerable variability is observed among patients, uveal colobomata being the most constant feature. Some patients manifest mental retardation of varying degree and/or sensorineural, mid-frequency hearing loss. {ECO:0000269	PubMed:24462371}; Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Kolobom, Oküler, ± İşitme Engeli, Yank Dudak / Damak ve / Veya Mental Retardasyon, 120433, Otozomal Dominant; COB1 (Uveal Koloboma-Yank Dudak ve Damak-Entelektüel Yetersizlik) (Prenatal) (YAP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation, 120433, Autosomal dominant; COB1 (Uveal coloboma-cleft lip and palate-intellectual disability) (YAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	YAP1	YAP1, COB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Coloboma, ocular, with or without hearing impairment, cleft lip/palate, and/or mental retardation (COB1) [MIM:120433]: An autosomal dominant disease characterized by uveal colobomata, microphthalmia, cataract and cleft lip/palate. Considerable variability is observed among patients, uveal colobomata being the most constant feature. Some patients manifest mental retardation of varying degree and/or sensorineural, mid-frequency hearing loss. {ECO:0000269	PubMed:24462371}; Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kolon Kanseri, İleri Evre, Somatik, 114500 (SRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colon cancer, advanced, somatic, 114500 (SRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRC	SRC, ASV, SRC1, THC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Note=SRC kinase activity has been shown to be increased in several tumor tissues and tumor cell lines such as colon carcinoma cells.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolon Kanseri, Somatik, 114500 (RAD54B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colon cancer, somatic, 114500 (RAD54B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD54B	RAD54B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolon Kanseri, Somatik, 114500 (PTPRJ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colon cancer, somatic, 114500 (PTPRJ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRJ	PTPRJ, DEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolon Kanseri, Somatik, 114500 (PTPN12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colon cancer, somatic, 114500 (PTPN12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN12	PTPN12, PTPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolon Kanseri, Yatkınlık, 114500, Otozomal Dominant (AURKA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colon cancer, susceptibility to, 114500, Autosomal dominant (AURKA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AURKA	AURKA, STK15, AURORA2, BTAK, ARK1, STK6, AIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolonik Adenoma Rekürrensi, Azalmış Risk, 114500, Otozomal Dominant (ODC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colonic adenoma recurrence, reduced risk of, 114500, Autosomal dominant (ODC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ODC1	ODC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renk Körlüğü, Deutan, 303800, X'e Bağlı, CBD (Mavi Kon Monokromatizmi) (OPN1MW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorblindness, deutan, 303800, X-linked; CBD (Blue cone monochromatism) (OPN1MW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPN1MW	OPN1MW, GCP, CBD, CBBM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renk Körlüğü, Protan, 303900, X'e Bağlı; CBP (Mavi Kon Monokromatizmi) (OPN1LW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorblindness, protan, 303900, X-linked; CBP (Blue cone monochromatism) (OPN1LW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPN1LW	OPN1LW, RCP, CBP, CBBM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renk Körlüğü, Tritan, 190900, Otozomal Dominant (Tritanopia) (OPN1SW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorblindness, tritan, 190900, Autosomal dominant (Tritanopia) (OPN1SW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPN1SW	OPN1SW, BCP, CBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tritan color blindness (CBT) [MIM:190900]: A disorder of vision characterized by a selective deficiency of blue spectral sensitivity. {ECO:0000269|PubMed:1386496, ECO:0000269|PubMed:1531728}; Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kolorektal Adenomatöz Polipozis, Otozomal Resesif, Pikomatriksmatar, 132600, Somatik Mutasyon (Pikomatriksoma) (MUTYH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600, Somatic mutation (Pilomatrixoma) (MUTYH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUTYH	MUTYH, MYH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser-Kromozomal Instabilite, Somatik (BUB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer with chromosomal instability, somatic (BUB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BUB1	BUB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, 114500, Otozomal Dominant (PLA2G2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, 114500, Autosomal dominant (PLA2G2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G2A	PLA2G2A, PLA2B, PLA2L, MOM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, 114500, Otozomal Dominant (Kalıtsal Meme Kanseri) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, 114500, Autosomal dominant (Hereditary breast cancer) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, 114500, Otozomal Dominant (Kalıtsal Meme Kanseri) (MLPA)	Colorectal cancer, 114500, Autosomal dominant (Hereditary breast cancer) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Otozomal Dominant Tip 1, 120435 (Lynch Sendromu) (MSH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 1, 120435, Autosomal dominant (Lynch syndrome) (MSH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH2	MSH2, COCA1, FCC1, HNPCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11930458,	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Otozomal Dominant Tip 1, 120435 (Lynch Sendromu) (MLPA)	Colorectal cancer, hereditary nonpolyposis, type 1, 120435, Autosomal dominant (Lynch syndrome) (MLPA)	MSH2	MSH2, COCA1, FCC1, HNPCC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11930458,	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 2, 609310; HNPCC2 (Lynch Sendromu) (MLH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 2, 609310; HNPCC2 (Lynch syndrome) (MLH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH1	MLH1, COCA2, HNPCC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 2, 609310; HNPCC2 (Lynch Sendromu) (MLPA)	Colorectal cancer, hereditary nonpolyposis, type 2, 609310; HNPCC2 (Lynch syndrome) (MLPA)	MLH1	MLH1, COCA2, HNPCC2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 4, 614337; HNPCC4 (Lynch Sendromu) (PMS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 4, 614337; HNPCC4 (Lynch syndrome) (PMS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMS2	PMS2, PMSL2, HNPCC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 4, 614337; HNPCC4 (Lynch Sendromu) (MLPA)	Colorectal cancer, hereditary nonpolyposis, type 4, 614337; HNPCC4 (Lynch syndrome) (MLPA)	PMS2	PMS2, PMSL2, HNPCC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 5, 614350, Otozomal Dominant; HNPCC5 (Lynch Sendromu) (MSH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 5, 614350, Autosomal dominant; HNPCC5 (Lynch syndrome) (MSH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH6	MSH6, GTBP, HNPCC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 5, 614350, Otozomal Dominant; HNPCC5 (Lynch Sendromu) (MLPA)	Colorectal cancer, hereditary nonpolyposis, type 5, 614350, Autosomal dominant; HNPCC5 (Lynch syndrome) (MLPA)	MSH6	MSH6, GTBP, HNPCC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 6, 614331; HNPCC6 (Lynch Sendromu) (TGFBR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 6, 614331; HNPCC6 (Lynch syndrome) (TGFBR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBR2	TGFBR2, HNPCC6, AAT3, MFS2, LDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Kalıtsal Nonpolipozis, Tip 7, 614385; HNPCC7 (Lynch Sendromu) (MLH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 7, 614385; HNPCC7 (Lynch syndrome) (MLH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH3	MLH3, HNPCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Allesel Non-Polipozis, Tip 8, 613244; HNPCC8 (Lynch Sendromu) (EPCAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, hereditary nonpolyposis, type 8, 613244; HNPCC8 (Lynch syndrome) (EPCAM gene) (Sequence Analysis) (Postnatal)	EPCAM	EPCAM, ACSTD1, TROP1, M4S1, MIC18, DIARS, HNPCC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Somatik (BRAF V600E - 599 ve 601. kodonların mutasyonları - Ekzon 15 mutasyonları) (BRAF geni) (Dizi Analizi) (Postnatal)	Colorectal cancer, somatic (BRAF V600E - 599 ve 601. codons mutations - Exon 15 mutations) (BRAF gene) (Sequence Analysis) (Postnatal)	BRAF	BRAF	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24465489}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Kolorektal Kanser, Somatik, 114500 (FLCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (FLCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLCN	FLCN, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hepatocellular carcinoma (HCC) [MIM:114550]: A primary malignant neoplasm of epithelial liver cells. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. Note=The gene represented in this entry may be involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (MLH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (MLH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH3	MLH3, HNPCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:11317354}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (PDGFRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (PDGFRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRL	PDGFRL, PDGRL, PRLTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (MCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (MCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCC	MCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (DLC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (DLC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLC1	DLC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (DCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (DCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCC	DCC, MRMV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:9065402}. Note=The gene represented in this entry may be involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (BUB1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (BUB1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BUB1B	BUB1B, BUBR1, MVA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (BAX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (BAX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAX	BAX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (Ekzon 4) (AKT1 geni) (Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (Exon 4) (AKT1 gene) (Sequence Analysis) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (AXIN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (AXIN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AXIN2	AXIN2, ODCRCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:11917067}. Note=The gene represented in this entry is involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Somatik, 114500 (EP300 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, somatic, 114500 (EP300 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EP300	EP300, RSTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kolorektal Kanser, Yatkınlık, 1, 608812 (GALNT12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 1, 608812 (GALNT12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALNT12	GALNT12, CRCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer 1 (CRCS1) [MIM:608812]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:19617566, ECO:0000269|PubMed:22461326, ECO:0000269|PubMed:24115450}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The role of GALNT12 in colon cancer susceptibility is however subject to discussion: studies on 103 probants with colorectal cancer 1 (CRCS1) suggest that it does not act as a major contributor of CRCS1 (PubMed:24115450). {ECO:0000269|PubMed:24115450}..	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 10, 612591, Otozomal Dominant, CRCS1 (Alenüle Alesel Adenomatöz Polipozis) (POLD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 10, 612591, Autosomal dominant; CRCS1 (Attenuated familial adenomatous polyposis) (POLD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLD1	POLD1, CRCS10, MDPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer 10 (CRCS10) [MIM:612591]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24501277}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 114500, Otozomal Dominant (TLR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 114500, Autosomal dominant (TLR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR2	TLR2, TIL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 114500, Otozomal Dominant (CCND1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 114500, Autosomal dominant (CCND1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCND1	CCND1, PRAD1, BCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 12, 615083, Otozomal Dominant (Alenüle Alesel Adenomatöz Polipozis) (POLE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 12, 615083, Autosomal dominant (Attenuated familial adenomatous polyposis) (POLE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLE	POLE1, CRCS12, FILS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer 12 (CRCS12) [MIM:615083]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. CRCS12 is characterized by a high-penetrance predisposition to the development of colorectal adenomas and carcinomas, with a variable tendency to develop multiple and large tumors. Onset is usually before age 40 years. The histologic features of the tumors are unremarkable. {ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24501277}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 3, 612229 (SMAD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Colorectal cancer, susceptibility to, 3, 612229 (SMAD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD7	SMAD7, MADH7, CRCS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer 3 (CRCS3) [MIM:612229]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:17934461}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry..	EDTA Blood Sample
Kolorektal Kanser, Yatkınlık, 4, 601228, Otozomal Dominant (Kalıtsal Karışık Polipozis Sendromu) (Kromozom 15q13-q14 duplikasyon sendromu, 40-KB) (2200	Colorectal cancer, susceptibility to, 4, 601228, Autosomal dominant (Hereditary mixed polyposis syndrome) (CHROMOSOME 15q13-q14 DUPLICATION SYNDROME, 40-KB) (4400	.	HMPS1, CRAC1, CRCS4, DUP15q, CI5DUPq	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal		EDTA Blood Sample and Heparinized blood sample
Kombine C6 / C7 Eksikliği (Immun Yetmezlik, Kompleman Geç Bileşen Eksikliğine Bağlı) (C6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined C6/C7 deficiency (Immunodeficiency due to a late component of complement deficiency) (C6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C6	C6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 6 deficiency (C6D) [MIM:612446]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269	PubMed:15565285}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine C6 / C7 Eksikliği (Immun Yetmezlik, Kompleman Geç Bileşen Eksikliğine Bağlı) (Prenatal) (C6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined C6/C7 deficiency (Immunodeficiency due to a late component of complement deficiency) (C6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C6	C6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement component 6 deficiency (C6D) [MIM:612446]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections, predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. {ECO:0000269	PubMed:15565285}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Hücresel ve Humoral İmmün Yetmezlik, Granüloma, 233650, Otozomal Resesif (Kombine İmmün Yetmezlik-Cilt Granülomaları) (RAG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined cellular and humoral immune defects with granulomas, 233650, Autosomal recessive (Combined immunodeficiency with skin granulomas) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B cells, and sparse thymic tissue on ultrasonography. {ECO:0000269	PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Hücresel ve Humoral İmmün Yetmezlik, Granüloma, 233650, Otozomal Resesif (Kombine İmmün Yetmezlik-Cilt Granülomaları) (RAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined cellular and humoral immune defects with granulomas, 233650, Autosomal recessive (Combined immunodeficiency with skin granulomas) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B cells, and sparse thymic tissue on ultrasonography. {ECO:0000269	PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Hücresel ve Humoral İmmün Yetmezlik, Granüloma, 233650, Otozomal Resesif (Kombine İmmün Yetmezlik-Cilt Granülomaları) (Prenatal) (RAG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined cellular and humoral immune defects with granulomas, 233650, Autosomal recessive (Combined immunodeficiency with skin granulomas) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B cells, and sparse thymic tissue on ultrasonography. {ECO:0000269	PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kombine Hücresel ve Humoral İmmün Yetmezlik, Granülomа, 233650, Otozomal Resesif (Kombine İmmün Yetmezlik-Cilt Granülomaları) (Prenatal) (RAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined cellular and humoral immune defects with granulomas, 233650, Autosomal recessive (Combined immunodeficiency with skin granulomas) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B cells, and sparse thymic tissue on ultrasonography. (ECO:0000269	PubMed:18463379). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine D-2 ve L-2-Hidroksiglutarik Asidüri, 615182, Otozomal Resesif; D2L2AD (D, L-2-Hidroksi Glutarik Asidüri) (SLC25A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182, Autosomal recessive; D2L2AD (D,L-2-hydroxyglutaric aciduria) (SLC25A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A1	SLC25A1, SLC20A3, CTP, D2L2AD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined D-2- and L-2-hydroxyglutaric aciduria (D2L2AD) [MIM:615182]: An autosomal recessive neurometabolic disorder characterized by neonatal-onset encephalopathy with severe muscular weakness, intractable seizures, respiratory distress, and lack of psychomotor development resulting in early death. Brain imaging shows abnormalities including enlarged ventricles, delayed myelination, and germinal layer cysts. (ECO:0000269	PubMed:23561848). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine D-2 ve L-2-Hidroksiglutarik Asidüri, 615182, Otozomal Resesif; D2L2AD (D, L-2-Hidroksi Glutarik Asidüri) (Prenatal) (SLC25A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined D-2- and L-2-hydroxyglutaric aciduria, 615182, Autosomal recessive; D2L2AD (D,L-2-hydroxyglutaric aciduria) (SLC25A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A1	SLC25A1, SLC20A3, CTP, D2L2AD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined D-2- and L-2-hydroxyglutaric aciduria (D2L2AD) [MIM:615182]: An autosomal recessive neurometabolic disorder characterized by neonatal-onset encephalopathy with severe muscular weakness, intractable seizures, respiratory distress, and lack of psychomotor development resulting in early death. Brain imaging shows abnormalities including enlarged ventricles, delayed myelination, and germinal layer cysts. (ECO:0000269	PubMed:23561848). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Faktör 5 ve 8 Eksikliği, 227300, Otozomal Resesif; F5F8D1 (Kombine Faktör 5 ve Faktör 8 Eksikliği) (LMAN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined factor V and VIII deficiency, 227300, Autosomal recessive; F5F8D1 (Combined deficiency of factor V and factor VIII) (LMAN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMAN1	LMAN1, ERGIC53, F5F8D, MCFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor V and factor VII combined deficiency 1 (F5F8D1) [MIM:227300]: A blood coagulation disorder characterized by bleeding symptoms similar to those in hemophilia or parahemophilia, that are caused by single deficiency of FV or FVIII, respectively. The most common symptoms are epistaxis, menorrhagia, and excessive bleeding during or after trauma. Plasma levels of coagulation factors V and VIII are in the range of 5 to 30% of normal. (ECO:0000269	PubMed:10090935, ECO:0000269	PubMed:19787799). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Faktör 5 ve 8 Eksikliği, 227300, Otozomal Resesif; F5F8D1 (Kombine Faktör 5 ve Faktör 8 Eksikliği) (Prenatal) (LMAN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined factor V and VIII deficiency, 227300, Autosomal recessive; F5F8D1 (Combined deficiency of factor V and factor VIII) (LMAN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMAN1	LMAN1, ERGIC53, F5F8D, MCFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Factor V and factor VII combined deficiency 1 (F5F8D1) [MIM:227300]: A blood coagulation disorder characterized by bleeding symptoms similar to those in hemophilia or parahemophilia, that are caused by single deficiency of FV or FVIII, respectively. The most common symptoms are epistaxis, menorrhagia, and excessive bleeding during or after trauma. Plasma levels of coagulation factors V and VIII are in the range of 5 to 30% of normal. (ECO:0000269	PubMed:10090935, ECO:0000269	PubMed:19787799). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Hiperlipidemi, Ailesel, 144250, Otozomal Dominant (Ailesel Lipoprotein Lipaz Eksikliği) (LPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined hyperlipidemia, familial, 144250, Autosomal dominant (Familial lipoprotein lipase deficiency) (LPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPL	LPL, LIPD, HDLCQ11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Hiperlipidemi, Ailesel, 144250, Otozomal Dominant (Ailesel Lipoprotein Lipaz Eksikliği) (MLPA)	Combined hyperlipidemia, familial, 144250, Autosomal dominant (Familial lipoprotein lipase deficiency) (MLPA)	LPL	LPL, LIPD, HDLCQ11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Kombine İmmün Yetmezlik, Orta X'e Bağlı, 312863, X'e Bağlı; CIDX (İmmun Yetmezlik, Ağır Kombine, T-B + Gama Zinciri Eksikliğine Bağlı) (IL2RG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined immunodeficiency, X-linked, moderate, 312863, X-linked recessive; CIDX (T-B+ severe combined immunodeficiency due to gamma chain deficiency) (IL2RG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL2RG	IL2RG, SCIDX1, SCIDX, IMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kombine İmmün Yetmezlik, Orta X'e Bağlı, 312863, X'e Bağlı; CIDX (İmmun Yetmezlik, Ağır Kombine, T-B + Gama Zinciri Eksikliğine Bağlı) (Prenatal) (IL2RG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined immunodeficiency, X-linked, moderate, 312863, X-linked recessive; CIDX (T-B+ severe combined immunodeficiency due to gamma chain deficiency) (IL2RG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL2RG	IL2RG, SCIDX1, SCIDX, IMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Malonik ve Metilmalonik Asidüri, 614265; CMAMMA (Kombine Malonik ve Metilmalonik Asidemi) (ACSF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined malonic and methylmalonic aciduria, 614265; CMAMMA (Combined malonic and methylmalonic acidemia) (ACSF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACSF3	ACSF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined malonic and methylmalonic aciduria (CMAMMA) [MIM:614265]: A metabolic disease characterized by malonic and methylmalonic aciduria, with urinary excretion of much larger amounts of methylmalonic acid than malonic acid, in the presence of normal malonyl-CoA decarboxylase activity. Clinical features include coma, ketoacidosis, hypoglycemia, failure to thrive, microcephaly, dystonia, axial hypotonia and/or developmental delay, and neurologic manifestations including seizures, psychiatric disease and/or cognitive decline. (ECO:0000269	PubMed:21841779). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Malonik ve Metilmalonik Asidüri, 614265; CMAMMA (Kombine Malonik ve Metilmalonik Asidemi) (Prenatal) (ACSF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined malonic and methylmalonic aciduria, 614265; CMAMMA (Combined malonic and methylmalonic acidemia) (ACSF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACSF3	ACSF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined malonic and methylmalonic aciduria (CMAMMA) [MIM:614265]: A metabolic disease characterized by malonic and methylmalonic aciduria, with urinary excretion of much larger amounts of methylmalonic acid than malonic acid, in the presence of normal malonyl-CoA decarboxylase activity. Clinical features include coma, ketoacidosis, hypoglycemia, failure to thrive, microcephaly, dystonia, axial hypotonia and/or developmental delay, and neurologic manifestations including seizures, psychiatric disease and/or cognitive decline. (ECO:0000269	PubMed:21841779). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 1, 609060, Otozomal Resesif; COXPD1 (Hepatoensefalopati, Kombine Oksidatif Fosforilasyon Eksikliği Tip 1 E Bağlı) (GFM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 1, 609060, Autosomal recessive; COXPD1 (Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1) (GFM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFM1	GFM1, EFG1, GFM, COXPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 1 (COXPD1) [MIM:609060]: A mitochondrial disease resulting in early rapidly progressive hepatoencephalopathy. (ECO:0000269	PubMed:15537906, ECO:0000269	PubMed:17160893). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 1, 609060, Otozomal Resesif; COXPD1 (Hepatoensefalopati, Kombine Oksidatif Fosforilasyon Eksikliği Tip 1 E Bağlı) (Prenatal) (GFM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 1, 609060, Autosomal recessive; COXPD1 (Hepatoencephalopathy due to combined oxidative phosphorylation deficiency type 1) (GFM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFM1	GFM1, EFG1, GFM, COXPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 1 (COXPD1) [MIM:609060]: A mitochondrial disease resulting in early rapidly progressive hepatoencephalopathy. (ECO:0000269	PubMed:15537906, ECO:0000269	PubMed:17160893). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 10, 614702, Otozomal Resesif; COXPD10 (Mitokondriyal Hipertrofik Kardiyomiyopati-Laktik Asidoz, MTO1 Eksikliğine Bağlı) (MTO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 10, 614702, Autosomal recessive; COXPD10 (Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency) (MTO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTO1	MTO1, COXPD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 10, 614702, Otozomal Resesif; COXPD10 (Mitokondriyal Hipertrofik Kardiyomiyopati-Laktik Asidoz, MTO1 Eksikliğine Bağlı) (Prenatal) (MTO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 10, 614702, Autosomal recessive; COXPD10 (Mitochondrial hypertrophic cardiomyopathy with lactic acidosis due to MTO1 deficiency) (MTO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTO1	MTO1, COXPD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 11, 614922, Otozomal Resesif; COXPD11 (Kombine Oksidatif Fosforilasyon Defekti Tip 11) (RMND1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 11, 614922, Autosomal recessive; COXPD11 (Combined oxidative phosphorylation defect type 11) (RMND1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RMND1	RMND1, COXPD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 11 (COXPD11) [MIM:614922]: A severe, multisystemic, autosomal recessive, disorder characterized by deficiencies of multiple mitochondrial respiratory enzymes leading to neonatal hypotonia and lactic acidosis. Affected individuals may have respiratory insufficiency, foot deformities, or seizures. (ECO:0000269|PubMed:23022098, ECO:0000269|PubMed:23022099, ECO:0000269|PubMed:25604853, ECO:0000269|PubMed:26238252). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 11, 614922, Otozomal Resesif; COXPD11 (Kombine Oksidatif Fosforilasyon Defekti Tip 11) (Prenatal) (RMND1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 11, 614922, Autosomal recessive; COXPD11 (Combined oxidative phosphorylation defect type 11) (RMND1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RMND1	RMND1, COXPD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 11 (COXPD11) [MIM:614922]: A severe, multisystemic, autosomal recessive, disorder characterized by deficiencies of multiple mitochondrial respiratory enzymes leading to neonatal hypotonia and lactic acidosis. Affected individuals may have respiratory insufficiency, foot deformities, or seizures. (ECO:0000269|PubMed:23022098, ECO:0000269|PubMed:23022099, ECO:0000269|PubMed:25604853, ECO:0000269|PubMed:26238252). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 12, 614924, Otozomal Resesif; COXPD12 (Lökoensefalopati-Talamus ve Beyin Anomalileri-Yüksek Laktat Sendromu) (EARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 12, 614924, Autosomal recessive; COXPD12 (Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome) (EARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EARS2	EARS2, KIAA1970, COXPD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 12 (COXPD12) [MIM:614924]: An autosomal recessive, mitochondrial, neurologic disorder characterized by onset in infancy of hypotonia and delayed psychomotor development, or early developmental regression, associated with T2-weighted hyperintensities in the deep cerebral white matter, brainstem, and cerebellar white matter. Serum lactate is increased due to a defect in mitochondrial respiration. There are 2 main phenotypic groups: those with a milder disease course and some recovery of skills after age 2 years, and those with a severe disease course resulting in marked disability. (ECO:0000269	PubMed:22492562, ECO:0000269	PubMed:23008233). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 12, 614924, Otozomal Resesif; COXPD12 (Lökoensefalopati-Talamus ve Beyin Anomalileri-Yüksek Laktat Sendromu) (Prenatal) (EARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 12, 614924, Autosomal recessive; COXPD12 (Leukoencephalopathy-thalamus and brainstem anomalies-high lactate syndrome) (EARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EARS2	EARS2, KIAA1970, COXPD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 12 (COXPD12) [MIM:614924]: An autosomal recessive, mitochondrial, neurologic disorder characterized by onset in infancy of hypotonia and delayed psychomotor development, or early developmental regression, associated with T2-weighted hyperintensities in the deep cerebral white matter, brainstem, and cerebellar white matter. Serum lactate is increased due to a defect in mitochondrial respiration. There are 2 main phenotypic groups: those with a milder disease course and some recovery of skills after age 2 years, and those with a severe disease course resulting in marked disability. (ECO:0000269	PubMed:22492562, ECO:0000269	PubMed:23008233). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kombine Oksidatif Fosforilasyon Eksikliği 13, 614932, Otozomal Resesif; COXPD13 (Kombine Oksidatif Fosforilasyon Defekti Tip 13) (PNPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 13, 614932, Autosomal recessive; COXPD13 (Combined oxidative phosphorylation defect type 13) (PNPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPT1	PNPT1, OLD3S, COXPD13, DFNB70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 13 (COXPD13) [MIM:614932]: A mitochondrial disorder characterized by early onset severe encephalomyopathy, dystonia, choreoathetosis, bucofacial dyskinesias and combined mitochondrial respiratory chain deficiency. Nerve conductions velocities are decreased. Levels of plasma and cerebrospinal fluid lactate are increased. {ECO:0000269	PubMed:23084291}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 13, 614932, Otozomal Resesif; COXPD13 (Kombine Oksidatif Fosforilasyon Defekti Tip 13) (Prenatal) (PNPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 13, 614932, Autosomal recessive; COXPD13 (Combined oxidative phosphorylation defect type 13) (PNPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPT1	PNPT1, OLD3S, COXPD13, DFNB70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 13 (COXPD13) [MIM:614932]: A mitochondrial disorder characterized by early onset severe encephalomyopathy, dystonia, choreoathetosis, bucofacial dyskinesias and combined mitochondrial respiratory chain deficiency. Nerve conductions velocities are decreased. Levels of plasma and cerebrospinal fluid lactate are increased. {ECO:0000269	PubMed:23084291}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 14, 614946, Otozomal Resesif; COXPD14 (Kombine Oksidatif Fosforilasyon Defekti Tip 14) (FARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 14, 614946, Autosomal recessive; COXPD14 (Combined oxidative phosphorylation defect type 14) (FARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FARS2	FARS2, FARS1, COXPD14, SPG77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 14 (COXPD14) [MIM:614946]: A severe multisystemic autosomal recessive disorder characterized by neonatal onset of global developmental delay, refractory seizures, and lactic acidosis. Biochemical studies show deficiencies of multiple mitochondrial respiratory enzymes. {ECO:0000269|PubMed:22493341, ECO:0000269|PubMed:22833457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 14, 614946, Otozomal Resesif; COXPD14 (Kombine Oksidatif Fosforilasyon Defekti Tip 14) (Prenatal) (FARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 14, 614946, Autosomal recessive; COXPD14 (Combined oxidative phosphorylation defect type 14) (FARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FARS2	FARS2, FARS1, COXPD14, SPG77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 14 (COXPD14) [MIM:614946]: A severe multisystemic autosomal recessive disorder characterized by neonatal onset of global developmental delay, refractory seizures, and lactic acidosis. Biochemical studies show deficiencies of multiple mitochondrial respiratory enzymes. {ECO:0000269|PubMed:22493341, ECO:0000269|PubMed:22833457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 15, 614947, Otozomal Resesif; COXPD15 (Kombine Oksidatif Fosforilasyon Defekti Tip 15) (MTFMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 15, 614947, Autosomal recessive; COXPD15 (Combined oxidative phosphorylation defect type 15) (MTFMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTFMT	MTFMT, COXPD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 15, 614947, Otozomal Resesif; COXPD15 (Kombine Oksidatif Fosforilasyon Defekti Tip 15) (Prenatal) (MTFMT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 15, 614947, Autosomal recessive; COXPD15 (Combined oxidative phosphorylation defect type 15) (MTFMT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTFMT	MTFMT, COXPD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 16, 615395, Otozomal Resesif; COXPD16 (İnfantil Hipertrofik Kardiyomyopati, MRPL44 Eksikliğine Bağlı) (MRPL44 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 16, 615395, Autosomal recessive; COXPD16 (Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency) (MRPL44 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRPL44	MRPL44, COXPD16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 16 (COXPD16) [MIM:615395]: An autosomal recessive, mitochondrial disorder characterized by hypertrophic cardiomyopathy, liver steatosis, and decreased levels of mitochondrial complexes I and IV in heart and skeletal muscle. {ECO:0000269	PubMed:23315540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 16, 615395, Otozomal Resesif; COXPD16 (İnfantil Hipertrofik Kardiyomyopati, MRPL44 Eksikliğine Bağlı) (Prenatal) (MRPL44 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 16, 615395, Autosomal recessive; COXPD16 (Infantile hypertrophic cardiomyopathy due to MRPL44 deficiency) (MRPL44 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MRPL44	MRPL44, COXPD16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 16 (COXPD16) [MIM:615395]: An autosomal recessive, mitochondrial disorder characterized by hypertrophic cardiomyopathy, liver steatosis, and decreased levels of mitochondrial complexes I and IV in heart and skeletal muscle. {ECO:0000269	PubMed:23315540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 17, 615440, Otozomal Resesif; COXPD17 (Kombine Oksidatif Fosforilasyon Defekti Tip 17) (ELAC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 17, 615440, Autosomal recessive; COXPD17 (Combined oxidative phosphorylation defect type 17) (ELAC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELAC2	ELAC2, HPC2, COXPD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 17 (COXPD17) [MIM:615440]: An autosomal recessive disorder of mitochondrial dysfunction characterized by onset of severe hypertrophic cardiomyopathy in the first year of life. Other features include hypotonia, poor growth, lactic acidosis, and failure to thrive. The disorder may be fatal in early childhood. {ECO:0000269	PubMed:23849775}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 17, 615440, Otozomal Resesif; COXPD17 (Kombine Oksidatif Fosforilasyon Defekti Tip 17) (Prenatal) (ELAC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 17, 615440, Autosomal recessive; COXPD17 (Combined oxidative phosphorylation defect type 17) (ELAC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELAC2	ELAC2, HPC2, COXPD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 17 (COXPD17) [MIM:615440]: An autosomal recessive disorder of mitochondrial dysfunction characterized by onset of severe hypertrophic cardiomyopathy in the first year of life. Other features include hypotonia, poor growth, lactic acidosis, and failure to thrive. The disorder may be fatal in early childhood. {ECO:0000269	PubMed:23849775}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 18, 615578, Otozomal Resesif; COXPD18 (Büyüme ve Gelişme Gerilği-Hipotoni-Görme Bozukluğu-Laktik Asidoz Sendromu) (SFXN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 18, 615578, Autosomal recessive; COXPD18 (Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome) (SFXN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFXN4	SFXN4, COXPD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 18 (COXPD18) [MIM:615578]: An autosomal recessive disorder of mitochondrial dysfunction characterized by intrauterine growth retardation, hypotonia, visual impairment, speech delay, and lactic acidosis associated with decreased mitochondrial respiratory chain activity. Affected patients may also show hematologic abnormalities, mainly macrocytic anemia. {ECO:0000269	PubMed:24119684}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 18, 615578, Otozomal Resesif; COXPD18 (Büyüme ve Gelişme Gerilği-Hipotoni-Görme Bozukluğu-Laktik Asidoz Sendromu) (Prenatal) (SFXN4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 18, 615578, Autosomal recessive; COXPD18 (Growth and developmental delay-hypotonia-vision impairment-lactic acidosis syndrome) (SFXN4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SFXN4	SFXN4, COXPD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 18 (COXPD18) [MIM:615578]: An autosomal recessive disorder of mitochondrial dysfunction characterized by intrauterine growth retardation, hypotonia, visual impairment, speech delay, and lactic acidosis associated with decreased mitochondrial respiratory chain activity. Affected patients may also show hematologic abnormalities, mainly macrocytic anemia. {ECO:0000269	PubMed:24119684}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 19, 615595, Otozomal Resesif; COXPD19 (Ağır Neonatal Laktik Asidoz, NFS1-ISD11 Kompleks Eksikliğine Bağlı) (LYRM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 19, 615595, Autosomal recessive; COXPD19 (Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency) (LYRM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LYRM4	LYRM4, ISD11, C6orf149, COXPD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 19, 615595, Otozomal Resesif; COXPD19 (Ağır Neonatal Laktik Asidoz, NFS1-ISD11 Kompleks Eksikliğine Bağlı) (Prenatal) (LYRM4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 19, 615595, Autosomal recessive; COXPD19 (Severe neonatal lactic acidosis due to NFS1-ISD11 complex deficiency) (LYRM4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LYRM4	LYRM4, ISD11, C6orf149, COXPD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 2, 610498, Otozomal Resesif; COXPD2 (Kombine Oksidatif Fosforilasyon Defekti Tip 2) (MRPS16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 2, 610498, Autosomal recessive; COXPD2 (Combined oxidative phosphorylation defect type 2) (MRPS16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRPS16	MRPS16, COXPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 2 (COXPD2) [MIM:610498]: A mitochondrial disease resulting in fatal neonatal metabolic acidosis with agenesis of the corpus callosum. {ECO:0000269	PubMed:15505824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 2, 610498, Otozomal Resesif; COXPD2 (Kombine Oksidatif Fosforilasyon Defekti Tip 2) (Prenatal) (MRPS16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 2, 610498, Autosomal recessive; COXPD2 (Combined oxidative phosphorylation defect type 2) (MRPS16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MRPS16	MRPS16, COXPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 2 (COXPD2) [MIM:610498]: A mitochondrial disease resulting in fatal neonatal metabolic acidosis with agenesis of the corpus callosum. {ECO:0000269	PubMed:15505824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 20, 615917, Otozomal Resesif; COXPD20 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 20) (VARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 20, 615917, Autosomal recessive; COXPD20 (Combined oxidative phosphorylation deficiency type 20) (VARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VARS2	VARS2, KIAA1885, COXPD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 20 (COXPD20) [MIM:615917]: A disorder due to mitochondrial respiratory chain complex defects. Clinical features are variable and include muscle weakness with hypotonia, central neurological disease with progressive external ophthalmoplegia, ptosis and ataxia, delayed psychomotor development, cardiomyopathy, abnormal liver function, facial dysmorphism, microcephaly and epilepsy. {ECO:0000269|PubMed:24827421, ECO:0000269|PubMed:25058219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 20, 615917, Otozomal Resesif; COXPD20 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 20) (Prenatal) (VARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 20, 615917, Autosomal recessive; COXPD20 (Combined oxidative phosphorylation deficiency type 20) (VARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VARS2	VARS2, KIAA1885, COXPD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 20 (COXPD20) [MIM:615917]: A disorder due to mitochondrial respiratory chain complex defects. Clinical features are variable and include muscle weakness with hypotonia, central neurological disease with progressive external ophthalmoplegia, ptosis and ataxia, delayed psychomotor development, cardiomyopathy, abnormal liver function, facial dysmorphism, microcephaly and epilepsy. {ECO:0000269|PubMed:24827421, ECO:0000269|PubMed:25058219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 21, 615918, Otozomal Resesif; COXPD21 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 21) (TARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 21, 615918, Autosomal recessive; COXPD21 (Combined oxidative phosphorylation deficiency type 21) (TARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TARS2	TARS2, COXPD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 21 (COXPD21) [MIM:615918]: A mitochondrial disorder characterized by a lethal encephalomyopathy. Shortly after birth, affected individuals manifest axial hypotonia, limb hypertonia, psychomotor delay, and increased serum lactate. Additional features include subsarcolemmal lipofuscin-positive deposits in muscle, cerebral spongiosis, and hepatic steatosis. {ECO:0000269	PubMed:24827421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 21, 615918, Otozomal Resesif; COXPD21 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 21) (Prenatal) (TARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 21, 615918, Autosomal recessive; COXPD21 (Combined oxidative phosphorylation deficiency type 21) (TARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TARS2	TARS2, COXPD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 21 (COXPD21) [MIM:615918]: A mitochondrial disorder characterized by a lethal encephalomyopathy. Shortly after birth, affected individuals manifest axial hypotonia, limb hypertonia, psychomotor delay, and increased serum lactate. Additional features include subsarcolemmal lipofuscin-positive deposits in muscle, cerebral spongiosis, and hepatic steatosis. {ECO:0000269	PubMed:24827421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 22, 616045, Otozomal Resesif; COXPD22 (ATP5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 22, 616045, Autosomal recessive; COXPD22 (ATP5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP5A1	ATP5A1, ATPM, ATP5A, ORM, MC5DN4, COXPD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 22, 616045, Otozomal Resesif; COXPD22 (Prenatal) (ATP5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 22, 616045, Autosomal recessive; COXPD22 (ATP5A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP5A1	ATP5A1, ATPM, ATP5A, ORM, MC5DN4, COXPD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kombine Oksidatif Fosforilasyon Eksikliği 23, 616198, Otozomal Resesif; COXPD23 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 23) (GTPBP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 23, 616198, Autosomal recessive; COXPD23 (Combined oxidative phosphorylation deficiency type 23) (GTPBP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GTPBP3	GTPBP3, MSS1, COXPD23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 23 (COXPD23) [MIM:616198]: An autosomal recessive mitochondrial disorder characterized by hypertrophic cardiomyopathy and/or neurologic symptoms with onset in early childhood. Disease features include hypertrophic cardiomyopathy, hypotonia, delayed psychomotor development, lactic acidosis, impaired activities of respiratory complexes I and IV, and defective translation of mitochondrial proteins. Disease severity is variable, ranging from death in early infancy to survival into the second decade of life. {ECO:0000269|PubMed:25434004}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 23, 616198, Otozomal Resesif; COXPD23 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 23) (Prenatal) (GTPBP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 23, 616198, Autosomal recessive; COXPD23 (Combined oxidative phosphorylation deficiency type 23) (GTPBP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GTPBP3	GTPBP3, MSS1, COXPD23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 23 (COXPD23) [MIM:616198]: An autosomal recessive mitochondrial disorder characterized by hypertrophic cardiomyopathy and/or neurologic symptoms with onset in early childhood. Disease features include hypertrophic cardiomyopathy, hypotonia, delayed psychomotor development, lactic acidosis, impaired activities of respiratory complexes I and IV, and defective translation of mitochondrial proteins. Disease severity is variable, ranging from death in early infancy to survival into the second decade of life. {ECO:0000269|PubMed:25434004}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 24, 616239, Otozomal Resesif; COXPD24 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 24) (NARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 24, 616239, Autosomal recessive; COXPD24 (Combined oxidative phosphorylation deficiency type 24) (NARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NARS2	NARS2, COXPD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 24, 616239, Otozomal Resesif; COXPD24 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 24) (Prenatal) (NARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 24, 616239, Autosomal recessive; COXPD24 (Combined oxidative phosphorylation deficiency type 24) (NARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NARS2	NARS2, COXPD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 25, 616430, Otozomal Resesif; COXPD25 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 25) (MARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 25, 616430, Autosomal recessive; COXPD25 (Combined oxidative phosphorylation deficiency type 25) (MARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARS2	MARS2, SPAX3, COXPD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 25, 616430, Otozomal Resesif; COXPD25 (Kombine Oksidatif Fosforilasyon Eksikliği Tip 25) (Prenatal) (MARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 25, 616430, Autosomal recessive; COXPD25 (Combined oxidative phosphorylation deficiency type 25) (MARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MARS2	MARS2, SPAX3, COXPD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 26, 616539, Otozomal Resesif; COXPD2 (Kombine Oksidatif Fosforilasyon Defekti Tip 26) (TRMT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 26, 616539, Autosomal recessive; COXPD2 (Combined oxidative phosphorylation defect type 26) (TRMT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRMT5	TRMT5, TRM5, KIAA1393, COXPD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 26, 616539, Otozomal Resesif; COXPD2 (Kombine Oksidatif Fosforilasyon Defekti Tip 26) (Prenatal) (TRMT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 26, 616539, Autosomal recessive; COXPD2 (Combined oxidative phosphorylation defect type 26) (TRMT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRMT5	TRMT5, TRM5, KIAA1393, COXPD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 27 616672, Otozomal Resesif; COXPD27 (Kombine Oksidatif Fosforilasyon Defekti Tip 27) (CARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 27, 616672, Autosomal recessive; COXPD27 (Combined oxidative phosphorylation defect type 27) (CARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARS2	CARS2, COXPD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 27 616672, Otozomal Resesif; COXPD27 (Kombine Oksidatif Fosforilasyon Defekti Tip 27) (Prenatal) (CARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 27, 616672, Autosomal recessive; COXPD27 (Combined oxidative phosphorylation defect type 27) (CARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CARS2	CARS2, COXPD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 28 616794, Otozomal Resesif; COXPD28 (Neonatal Ağır Kardiyopulmoner Yetmezlik, Mitokondriyal Metilasyon Defektine Bağlı) (SLC25A26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 28, 616794, Autosomal recessive; COXPD28 (Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect) (SLC25A26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A26	SLC25A26, SAMC, COXPD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 28 616794, Otozomal Resesif; COXPD28 (Neonatal Ağır Kardiyopulmoner Yetmezlik, Mitokondriyal Metilasyon Defektine Bağlı) (Prenatal) (SLC25A26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 28, 616794, Autosomal recessive; COXPD28 (Neonatal severe cardiopulmonary failure due to mitochondrial methylation defect) (SLC25A26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A26	SLC25A26, SAMC, COXPD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 29 616811, Otozomal Resesif; COXPD29 (TXN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 29, 616811, Autosomal recessive; COXPD29 (TXN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TXN2	TXN2, TRX2, MTRX, COXPD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 29 616811, Otozomal Resesif; COXPD29 (Prenatal) (TXN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 29, 616811, Autosomal recessive; COXPD29 (TXN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TXN2	TXN2, TRX2, MTRX, COXPD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 3, 610505, Otozomal Resesif; COXPD3 (Fatal Mitokondriyal Hastalık, Kombine Oksidatif Fosforilasyon Defektli Tip 3'e Bağlı) (TSFM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 3, 610505, Autosomal recessive; COXPD3 (Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3) (TSFM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSFM	TSFM, COXPD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 3, 610505, Otozomal Resesif; COXPD3 (Fatal Mitokondriyal Hastalık, Kombine Oksidatif Fosforilasyon Defektli Tip 3'e Bağlı) (Prenatal) (TSFM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 3, 610505, Autosomal recessive; COXPD3 (Fatal mitochondrial disease due to combined oxidative phosphorylation deficiency 3) (TSFM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSFM	TSFM, COXPD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 30 616974, Otozomal Resesif; COXPD30 (Kombine Oksidatif Fosforilasyon Defekti Tip 30) (TRMT10C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 30, 616974, Autosomal recessive; COXPD30 (Combined oxidative phosphorylation defect type 30) (TRMT10C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRMT10C	TRMT10C, RG9MTD1, MRPP1, COXPD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 30 616974, Otozomal Resesif; COXPD30 (Kombine Oksidatif Fosforilasyon Defekti Tip 30) (Prenatal) (TRMT10C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 30, 616974, Autosomal recessive; COXPD30 (Combined oxidative phosphorylation defect type 30) (TRMT10C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRMT10C	TRMT10C, RG9MTD1, MRPP1, COXPD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 31, 617228, Otozomal Resesif; COXPD31 (Letal Sol Ventrikülemoncompanction-Nöbet-Hipotoni-Katarakt-Gelişimsel Gecikme Sendromu) (MIPEP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 31, 617228, Autosomal recessive; COXPD31 (Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome) (MIPEP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIPEP	MIPEP, COXPD31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 31, 617228, Otozomal Resesif; COXPD31 (Letal Sol Ventrikülemoncompanction-Nöbet-Hipotoni-Katarakt-Gelişimsel Gecikme Sendromu) (Prenatal) (MIPEP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 31, 617228, Autosomal recessive; COXPD31 (Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome) (MIPEP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MIPEP	MIPEP, COXPD31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 4, 610678, Otozomal Resesif; COXPD4 (Kombine Oksidatif Fosforilasyon Defekti Tip 4) (TUFM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 4, 610678, Autosomal recessive; COXPD4 (Combined oxidative phosphorylation defect type 4) (TUFM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUFM	TUFM, EFTu, COXPD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 4 (COXPD4) [MIM:610678]: A mitochondrial disease resulting in neonatal lactic acidosis, rapidly progressive encephalopathy, severely decreased mitochondrial protein synthesis, and combined deficiency of mtDNA- related mitochondrial respiratory chain complexes. {ECO:0000269|PubMed:17168893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 4, 610678, Otozomal Resesif; COXPD4 (Kombine Oksidatif Fosforilasyon Defekti Tip 4) (Prenatal) (TUFM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 4, 610678, Autosomal recessive; COXPD4 (Combined oxidative phosphorylation defect type 4) (TUFM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUFM	TUFM, EFTu, COXPD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 4 (COXPD4) [MIM:610678]: A mitochondrial disease resulting in neonatal lactic acidosis, rapidly progressive encephalopathy, severely decreased mitochondrial protein synthesis, and combined deficiency of mtDNA- related mitochondrial respiratory chain complexes. {ECO:0000269|PubMed:17168893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 5, 611719, Otozomal Resesif; COXPD5 (Hipotoni-Laktik Asidemi-Hiperamonemi) (MRPS22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 5, 611719, Autosomal recessive; COXPD5 (Hypotonia with lactic acidemia and hyperammonemia) (MRPS22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRPS22	MRPS22, C3orf5, COXPD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 5 (COXPD5) [MIM:611719]: A mitochondrial disease resulting in severe metabolic acidosis, edema, hypertrophic cardiomyopathy, tubulopathy, and hypotonia. {ECO:0000269|PubMed:17873122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 5, 611719, Otozomal Resesif; COXPD5 (Hipotoni-Laktik Asidemi-Hiperamonemi) (Prenatal) (MRPS22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 5, 611719, Autosomal recessive; COXPD5 (Hypotonia with lactic acidemia and hyperammonemia) (MRPS22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MRPS22	MRPS22, C3orf5, COXPD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 5 (COXPD5) [MIM:611719]: A mitochondrial disease resulting in severe metabolic acidosis, edema, hypertrophic cardiomyopathy, tubulopathy, and hypotonia. {ECO:0000269|PubMed:17873122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 6, 300816, X'e Bağlı Resesif; COXPD6 (Ağır X'e Bağlı Mitokondriyal Ensefalomyopati) (AIFM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 6, 300816, X-linked recessive; COXPD6 (Severe X-linked mitochondrial encephalomyopathy) (AIFM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIFM1	AIFM1, PDCD8, AIF, COXPD6, COWCK, CMTX4, DFNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 6 (COXPD6) [MIM:300816]: A mitochondrial disease resulting in a neurodegenerative disorder characterized by psychomotor delay, hypotonia, areflexia, muscle weakness and wasting. Some patients manifest prenatal ventriculomegaly and severe postnatal encephalomyopathy. {ECO:0000269|PubMed:20362274, ECO:0000269|PubMed:22019070}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 6, 300816, X'e Bağlı Resesif; COXPD6 (Ağır X'e Bağlı Mitokondriyal Ensefalomyopati) (Prenatal) (AIFM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 6, 300816, X-linked recessive; COXPD6 (Severe X-linked mitochondrial encephalomyopathy) (AIFM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AIFM1	AIFM1, PDCD8, AIF, COXPD6, COWCK, CMTX4, DFNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 6 (COXPD6) [MIM:300816]: A mitochondrial disease resulting in a neurodegenerative disorder characterized by psychomotor delay, hypotonia, areflexia, muscle weakness and wasting. Some patients manifest prenatal ventriculomegaly and severe postnatal encephalomyopathy. {ECO:0000269|PubMed:20362274, ECO:0000269|PubMed:22019070}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 7, 613559, Otozomal Resesif; COXPD7 (Kombine Oksidatif Fosforilasyon Defekti Tip 7) (C12orf65 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 7, 613559, Autosomal recessive; COXPD7 (Combined oxidative phosphorylation defect type 7) (C12orf65 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C12orf65	C12orf65, COXPD7, SPG55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 7 (COXPD7) [MIM:613559]: A mitochondrial disease resulting in encephalomyopathy. Clinical manifestations include psychomotor delay and regression, ataxia, optic atrophy, nystagmus and muscle atrophy and weakness. {ECO:0000269|PubMed:20598281}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Kombine Oksidatif Fosforilasyon Eksikliği 7, 613559, Otozomal Resesif; COXPD7 (Kombine Oksidatif Fosforilasyon Defekti Tip 7) (Prenatal) (C12orf65 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 7, 613559, Autosomal recessive; COXPD7 (Combined oxidative phosphorylation defect type 7) (C12orf65 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C12orf65	C12orf65, COXPD7, SPG55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 7 (COXPD7) [MIM:613559]: A mitochondrial disease resulting in encephalomyopathy. Clinical manifestations include psychomotor delay and regression, ataxia, optic atrophy, nystagmus and muscle atrophy and weakness. [ECO:0000269]PubMed:20598281). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 8, 614096, Otozomal Resesif; COXPD8 (Kombine Oksidatif Fosforilasyon Defekti Tip 8) (AARS2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 8, 614096, Autosomal recessive; COXPD8 (Combined oxidative phosphorylation defect type 8) (AARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AARS2	AARS2, KIAA1270, MTALARS, COXPD8, LKENP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 8 (COXPD8) [MIM:614096]: A mitochondrial disease characterized by a lethal infantile hypertrophic cardiomyopathy, generalized muscle dysfunction and some neurologic involvement. The liver is not affected. (ECO:0000269	PubMed:21549344). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 8, 614096, Otozomal Resesif; COXPD8 (Kombine Oksidatif Fosforilasyon Defekti Tip 8) (Prenatal) (AARS2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 8, 614096, Autosomal recessive; COXPD8 (Combined oxidative phosphorylation defect type 8) (AARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AARS2	AARS2, KIAA1270, MTALARS, COXPD8, LKENP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 8 (COXPD8) [MIM:614096]: A mitochondrial disease characterized by a lethal infantile hypertrophic cardiomyopathy, generalized muscle dysfunction and some neurologic involvement. The liver is not affected. (ECO:0000269	PubMed:21549344). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 9, 614582, Otozomal Resesif; COXPD9 (Kombine Oksidatif Fosforilasyon Defekti Tip 9) (MRPL3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Combined oxidative phosphorylation deficiency 9, 614582, Autosomal recessive; COXPD9 (Combined oxidative phosphorylation defect type 9) (MRPL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRPL3	MRPL3, MRL3, COXPD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 9 (COXPD9) [MIM:614582]: A mitochondrial disease characterized by failure to thrive, poor feeding, hypertrophic cardiomyopathy, hepatomegaly, and psychomotor retardation. Death in infancy has been observed in some cases. [ECO:0000269	PubMed:21786366). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Kombine Oksidatif Fosforilasyon Eksikliği 9, 614582, Otozomal Resesif; COXPD9 (Kombine Oksidatif Fosforilasyon Defekti Tip 9) (Prenatal) (MRPL3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Combined oxidative phosphorylation deficiency 9, 614582, Autosomal recessive; COXPD9 (Combined oxidative phosphorylation defect type 9) (MRPL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MRPL3	MRPL3, MRL3, COXPD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 9 (COXPD9) [MIM:614582]: A mitochondrial disease characterized by failure to thrive, poor feeding, hypertrophic cardiomyopathy, hepatomegaly, and psychomotor retardation. Death in infancy has been observed in some cases. [ECO:0000269	PubMed:21786366). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kombine SAP Eksikliği- 611721, Otozomal Resesif (Ensefalopati, Prosapozin Eksikliğine Bağlı) (PSAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Combined SAP deficiency, 611721, Autosomal recessive (Encephalopathy due to prosaposin deficiency) (PSAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kombine SAP Eksikliği- 611721, Otozomal Resesif (Ensefalopati, Prosapozin Eksikliğine Bağlı) (Prenatal) (PSAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Combined SAP deficiency, 611721, Autosomal recessive (Encephalopathy due to prosaposin deficiency) (PSAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
COMMAD Sendromu, 617306, Otozomal Resesif (MITF geni) (Tüm Gen Dizi Analizi) (Postnatal)	COMMAD syndrome, 617306, Autosomal recessive (MITF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMAD Sendromu, 617306, Otozomal Resesif (MLPA)	COMMAD syndrome, 617306, Autosomal recessive (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
COMMAD Sendromu, 617306, Otozomal Resesif (Prenatal) (MITF geni) (Tüm Gen Dizi Analizi) (Prenatal)	COMMAD syndrome, 617306, Autosomal recessive (MITF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
COMMAD Sendromu, 617306, Otozomal Resesif (Prenatal) (MLPA)	COMMAD syndrome, 617306, Autosomal recessive (Prenatal) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kompleman 4, Parsiyel Eksikliği, 120790, Otozomal Dominant (C1 Inhibitörü Eksikliği) (SERPING1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Complement component 4, partial deficiency of, 120790, Autosomal dominant (C1 inhibitor deficiency) (SERPING1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPING1	C1NH, HAE1, HAE2, SERPING1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kompleman 4, Parsiyel Eksikliği, 120790, Otozomal Dominant (C1 Inhibitörü Eksikliği) (MLPA)	Complement component 4, partial deficiency of, 120790, Autosomal dominant (C1 inhibitor deficiencyi) (MLPA)	SERPING1	C1NH, HAE1, HAE2, SERPING1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kompleman 4, Parsiyel Eksikliği, 120790, Otozomal Dominant (C1 Inhibitörü Eksikliği) (Prenatal) (SERPING1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Complement component 4, partial deficiency of, 120790, Autosomal dominant (C1 inhibitor deficiency) (SERPING1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPING1	C1NH, HAE1, HAE2, SERPING1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kompleman 4, Parsiyel Eksikliği, 120790, Otozomal Dominant (C1 Inhibitörü Eksikliği) (Prenatal) (MLPA)	Complement component 4, partial deficiency of, 120790, Autosomal dominant (C1 inhibitor deficiencyi) (Prenatal) (MLPA)	SERPING1	C1NH, HAE1, HAE2, SERPING1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kompleman Faktör B Eksikliği, 615561; CFBD (CFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Complement factor B deficiency, 615561; CFBD (CFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFB	CFB, BF, GBG, AHUS4, ARMD14, CFBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kompleman Faktör B Eksikliği, 615561; CFBD (CFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	Complement factor B deficiency, 615561; CFBD (CFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFB	CFB, BF, GBG, AHUS4, ARMD14, CFBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kompleman Faktör D Eksikliği, 613912, Otozomal Resesif; CFDD (Tekrarlayan Neisseria Enfeksiyonları, Faktör D Eksikliğine Bağlı) (CFD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Complement factor D deficiency, 613912, Autosomal recessive; CFDD (Recurrent Neisseria infections due to factor D deficiency) (CFD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFD	CFD, ADN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement factor D deficiency (CFDD) [MIM:613912]: An immunologic disorder characterized by increased susceptibility to bacterial infections, particularly Neisseria infections, due to a defect in the alternative complement pathway. (ECO:0000269	PubMed:16527897). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kompleman Faktör D Eksikliği, 613912, Otozomal Resesif; CFDD (Tekrarlayan Neisseria Enfeksiyonları, Faktör D Eksikliğine Bağlı) (Prenatal) (CFD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Complement factor D deficiency, 613912, Autosomal recessive; CFDD (Recurrent Neisseria infections due to factor D deficiency) (CFD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFD	CFD, ADN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement factor D deficiency (CFDD) [MIM:613912]: An immunologic disorder characterized by increased susceptibility to bacterial infections, particularly Neisseria infections, due to a defect in the alternative complement pathway. (ECO:0000269	PubMed:16527897). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Komplement Faktörü H Eksikliği, 609814, Otozomal Resesif, Otozomal Dominant; CFHD (İmmün Yetmezlik-Faktör H Anomasi) (CFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Complement factor H deficiency, 609814, Autosomal recessive, Autosomal dominant; CFHD (Immunodeficiency with factor H anomaly) (CFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Komplement Faktörü H Eksikliği, 609814, Otozomal Resesif, Otozomal Dominant; CFHD (İmmün Yetmezlik-Faktör H Anomasi) (MLPA)	Complement factor H deficiency, 609814, Autosomal recessive, Autosomal dominant; CFHD (Immunodeficiency with factor H anomaly) (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Komplement Faktörü H Eksikliği, 609814, Otozomal Resesif, Otozomal Dominant; CFHD (İmmün Yetmezlik-Faktör H Anomasi) (Prenatal) (CFH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Complement factor H deficiency, 609814, Autosomal recessive, Autosomal dominant; CFHD (Immunodeficiency with factor H anomaly) (CFH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Komplement Faktörü H Eksikliği, 609814, Otozomal Resesif, Otozomal Dominant; CFHD (İmmün Yetmezlik-Faktör H Anomasi) (Prenatal) (MLPA)	Complement factor H deficiency, 609814, Autosomal recessive, Autosomal dominant; CFHD (Immunodeficiency with factor H anomaly) (Prenatal) (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Komplement Faktör I Eksikliği, 610984, Otozomal Resesif; CFID (İmmün Yetmezlik-Faktör I Anoması) (CFI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Complement factor I deficiency, 610984, Autosomal recessive; CFID (Immunodeficiency with factor I anomaly) (CFI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFI	CFI, FI, AHUS3, ARMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement factor I deficiency (CFI deficiency) [MIM:610984]: Autosomal recessive condition associated with a propensity to pyogenic infections. (ECO:0000269|PubMed:12562389, ECO:0000269|PubMed:17018561, ECO:0000269|PubMed:8613545). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Komplement Faktör I Eksikliği, 610984, Otozomal Resesif; CFID (İmmün Yetmezlik-Faktör I Anoması) (Prenatal) (CFI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Complement factor I deficiency, 610984, Autosomal recessive; CFID (Immunodeficiency with factor I anomaly) (CFI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFI	CFI, FI, AHUS3, ARMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement factor I deficiency (CFI deficiency) [MIM:610984]: Autosomal recessive condition associated with a propensity to pyogenic infections. (ECO:0000269|PubMed:12562389, ECO:0000269|PubMed:17018561, ECO:0000269|PubMed:8613545). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kon Distrofisi 4, 613093, Otozomal Resesif; COD4 (Progresif Kon Distrofisi) (PDE6C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone dystrophy 4, 613093, Autosomal recessive; COD4 (Progressive cone dystrophy) (PDE6C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6C	PDE6C, PDEA2, COD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone dystrophy 4 (COD4) [MIM:613093]: An early-onset cone dystrophy. Cone dystrophies are retinal dystrophies characterized by progressive degeneration of the cone photoreceptors with preservation of rod function, as indicated by electroretinogram. However, some rod involvement may be present in some cone dystrophies, particularly at late stage. Affected individuals suffer from photophobia, loss of visual acuity, color vision and central visual field. Another sign is the absence of macular lesions for many years. Cone dystrophies are distinguished from the cone-rod dystrophies in which some loss of peripheral vision also occurs. [ECO:0000269	PubMed:19615668). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Kon Distrofisi -3,602093, Otozomal Dominant; COD3 (Progresif Kon Distrofisi) (GUCA1A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cone dystrophy-3, 602093, Autosomal dominant; COD3 (Cone rod dystrophy) (GUCA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCA1A	GUCA1A, GCAP, COD3, CORD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi (UNC119 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy (UNC119 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNC119	UNC119, HRG4, IMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 10, 610283, Otozomal Resesif; CORD10 (Progresif Kon Distrofisi) (SEMA4A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 10, 610283, Autosomal recessive; CORD10 (Cone rod dystrophy) (SEMA4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEMA4A	SEMA4A, SEMB, RP35, CORD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 11, 610381, Otozomal Dominant; CORD11 (Progresif Kon Distrofisi) (RAX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 11, 610381, Autosomal dominant; CORD11 (Cone rod dystrophy) (RAX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAX2	RAX2, RAXL1, QRX, CORD11, ARMD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 11 (CORD11) [MIM:610381]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269	PubMed:15028672). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 12, 612657; CORD12 (Progresif Kon Distrofisi) (PROM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 12, 612657; CORD12 (Cone rod dystrophy) (PROM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROM1	PROM1, PROML1, AC133, RP41, CORD12, CD133, MCDR2, STGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 13, 608194; CORD13 (Progresif Kon Distrofisi) (RPGRIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 13, 608194; CORD13 (Cone rod dystrophy) (RPGRIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGRIP1	RPGRIP1, LCA6, CORD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 13 (CORD13) [MIM:608194]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269|PubMed:10958648, ECO:0000269|PubMed:12920076). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kon-Rod Distrofisi 14, 602093, Otozomal Dominant (Progresif Kon Distrofisi) (GUCA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 14, 602093, Autosomal dominant (Cone rod dystrophy) (GUCA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCA1A	GUCA1A, GCAP, COD3, CORD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kon-Rod Distrofisi 15, 613660, Otozomal Resesif, CORD15 (Retinitis Pigmentoza) (CDHR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 15, 613660, Autosomal recessive; CORD15 (Retinitis pigmentosa) (CDHR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDHR1	CDHR1, PCDH21, PRCAD, CORD15, RP65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 15 (CORD15) [MIM:613660]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269[PubMed:20805371). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 16, 614500, Otozomal Resesif, CORD16 (Retinitis Pigmentoza) (C8orf37 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 16, 614500, Autosomal recessive; CORD16 (Retinitis pigmentosa) (C8orf37 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C8orf37	C8orf37, CORD16, RP64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 64 (RP64) [MIM:614500]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. [ECO:0000269[PubMed:22177090). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 18, 615374, Otozomal Resesif, CORD18 (Progresif Kon Distrofisi) (RAB28 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 18, 615374, Autosomal recessive; CORD18 (Cone rod dystrophy) (RAB28 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB28	RAB28, CORD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 19, 615860, Otozomal Resesif, CORD19 (Progresif Kon Distrofisi) (TTLL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 19, 615860, Autosomal recessive; CORD19 (Cone rod dystrophy) (TTLL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTLL5	TTLL5, STAMP, KIAA0998, CORD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 19 (CORD19) [MIM:615860]: A form of cone-rod dystrophy, an inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. [ECO:0000269[PubMed:24791901). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 20, 615973, Otozomal Resesif, CORD20 (Progresif Kon Distrofisi) (POC1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 20, 615973, Autosomal recessive; CORD20 (Cone rod dystrophy) (POC1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POC1B	POC1B, PIX1, CORD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 20 (CORD20) [MIM:615973]: A form of cone-rod dystrophy, an inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269[PubMed:24945461, ECO:0000269[PubMed:25018096, ECO:0000269[PubMed:25044745). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 21, 616502, Otozomal Resesif, CORD21 (Progresif Kon Distrofisi) (DRAM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 21, 616502, Autosomal recessive; CORD21 (Cone rod dystrophy) (DRAM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRAM2	DRAM2, TMEM77, CORD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 3, 604116; CORD3 (Progresif Kon Distrofisi) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 3, 604116; CORD3 (Cone rod dystrophy) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 5, 600977, Otozomal Dominant; CORD5 (Progresif Kon Distrofisi) (PITPNM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 5, 600977, Autosomal dominant; CORD5 (Cone rod dystrophy) (PITPNM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITPNM3	PITPNM3, NIR1, CORD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi 6, 601777, Otozomal Dominant; CORD6 (Progresif Kon Distrofisi) (GUCY2D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 6, 601777, Autosomal dominant; CORD6 (Cone rod dystrophy) (GUCY2D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCY2D	GUCY2D, GUC2D, LCA1, CORD6, RCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 6 (CORD6) [MIM:601777]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269[PubMed:12552567, ECO:0000269[PubMed:15111605, ECO:0000269[PubMed:18323321, ECO:0000269[PubMed:18487367, ECO:0000269[PubMed:20517349, ECO:0000269[PubMed:21552474, ECO:0000269[PubMed:22194653, ECO:0000269[PubMed:23734073, ECO:0000269[PubMed:24480840, ECO:0000269[PubMed:9618177, ECO:0000269[PubMed:9683616). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 7, 603649; CORD7 (Progresif Kon Distrofisi) (RIMS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 7, 603649; CORD7 (Cone rod dystrophy) (RIMS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIMS1	RIMS1, RIM1, RIM, KIAA0340, CORD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 7 (CORD7) [MIM:603649]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269[PubMed:12659814). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kon-Rod Distrofisi 9, 612775 (Progresif Kon Distrofisi) (ADAM9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy 9, 612775 (Cone rod dystrophy) (ADAM9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAM9	ADAM9, MDC9, MCMP, CORD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi ve İşitme Kaybı, 617236, Otozomal Resesif, CRDH4, (CEP78 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy and hearing loss, 617236, Autosomal recessive; CRDH4, (CEP78 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP78	CEP78, CRDH4,	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi, 604393, Otozomal Resesif (Progresif Kon Distrofisi) (AIPL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy, 604393, Autosomal recessive (Cone rod dystrophy) (AIPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIPL1	AIPL1, LCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi, 604393, Otozomal Resesif (Progresif Kon Distrofisi) (AIPL1	Cone-rod dystrophy, 604393, Autosomal	AIPL1	AIPL1, LCA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi, X'e Bağlı, 1, 304020, X'e Bağlı, CORDX1 (Progresif Kon Distrofisi) (RPGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy, X-linked, 1, 304020, X-linked; CORDX1 (Cone rod dystrophy) (RPGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGR	RPGR, RP3, CRD, RP15, COD1, CORDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Distrofisi, 300476, X'e Bağlı, 3, X'e Bağlı, CORDX3 (Progresif Kon Distrofisi) (CACNA1F geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod dystrophy, X-linked, 3, 300476, X-linked recessive; CORDX3 (Cone rod dystrophy) (CACNA1F gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1F	CACNA1F, CSNB2, CORDX3, CSNB2A, AIED, OA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kon-Rod Retina Distrofisisi-2, 120970, Otozomal Dominant; CORD2 (Progresif Kon Distrofisi) (CRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod retinal dystrophy-2, 120970, Autosomal dominant; CORD2 (Cone rod dystrophy) (CRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRX	CRX, CORD2, CRD, LCA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 2 (CORD2) [MIM:120970]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269[PubMed:9390563, ECO:0000269[PubMed:9427255, ECO:0000269[PubMed:9792858). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kon-Rod Retina Distrofisisi-2, 120970, Otozomal Dominant; CORD2 (Progresif Kon Distrofisi) (MLPA)	Cone-rod retinal dystrophy-2, 120970, Autosomal dominant; CORD2 (Cone rod dystrophy) (MLPA)	CRX	CRX, CORD2, CRD, LCA7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cone-rod dystrophy 2 (CORD2) [MIM:120970]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. (ECO:0000269[PubMed:9390563, ECO:0000269[PubMed:9427255, ECO:0000269[PubMed:9792858). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kon-Rod Sinaptik Bozukluğu, Konjenital Progresif Olmayan, 610427, Otozomal Resesif; CRSD (Kongenital Statisyoner Gece Körlüğü) (CABP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cone-rod synaptic disorder, congenital nonprogressive, 610427, Autosomal recessive; CRSD (Congenital stationary night blindness) (CABP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CABP4	CABP4, CRSD, CSNB2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod synaptic disorder, congenital non-progressive (CRSD) [MIM:610427]: A non-progressive retinal disorder characterized by stable low vision, nystagmus, photophobia, a normal or near-normal fundus appearance, and no night blindness. (ECO:0000269[PubMed:16960802). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital adrenal hiperplazi, sitokrom P450 oksidoredüktaz eksikliği (P450 oksidoredüktaz geni (POR)) (MLPA)	Congenital adrenal hyperplasia, cytochrome P450 oxidoreductase deficiency (P450 oxidoreductase gene (POR)) (MLPA)	P450 oksidoredüktaz geni (POR)	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Konjenital adrenal hiperplazi, sitokrom P450 oksidoredüktaz eksikliği (P450 oksidoredüktaz geni (POR)) (MLPA) (Prenatal)	Congenital adrenal hyperplasia, cytochrome P450 oxidoreductase deficiency (P450 oxidoreductase gene (POR)) (MLPA) (Prenatal)	P450 oksidoredüktaz geni (POR)	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal ve Üriner Sistem Konjenital Anomalileri 2, 143400, Otozomal Dominant (Konjenital Hidronefroz) (TBX18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital anomalies of kidney and urinary tract 2, 143400, Autosomal dominant (Congenital hydronephrosis) (TBX18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX18	TBX18, CAKUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital anomalies of kidney and urinary tract 2 (CAKUT2) [MIM:143400]: A disorder encompassing a broad spectrum of renal and urinary tract malformations that include renal agenesis, kidney hypodysplasia, multicystic kidney dysplasia, duplex collecting system, posterior urethral valves and ureter abnormalities. Congenital anomalies of kidney and urinary tract are the commonest cause of chronic kidney disease in children. (ECO:0000269[PubMed:26235987). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal ve Üriner Sistem Konjenital Anomalileri 2, 143400, Otozomal Dominant (Konjenital Hidronefroz) (Prenatal) (TBX18 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital anomalies of kidney and urinary tract 2, 143400, Autosomal dominant (Congenital hydronephrosis) (TBX18 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX18	TBX18, CAKUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital anomalies of kidney and urinary tract 2 (CAKUT2) [MIM:143400]: A disorder encompassing a broad spectrum of renal and urinary tract malformations that include renal agenesis, kidney hypodysplasia, multicystic kidney dysplasia, duplex collecting system, posterior urethral valves and ureter abnormalities. Congenital anomalies of kidney and urinary tract are the commonest cause of chronic kidney disease in children. (ECO:0000269[PubMed:26235987). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal ve İdrar Yollarının Konjenital Anomalileri, Yatkınlık, 610805, Otozomal Dominant (DSTYK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital anomalies of kidney and urinary tract, susceptibility to, 610805, Autosomal dominant (DSTYK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSTYK	DSTYK, KIAA0472, RIP5, DUSTYPK, CAKUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Konjenital Bilateral Vas Deferens Yokluğu, 277180, Otozomal Resesif (Konjenital Bilateral Vaz Deferens Yokluğu) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital bilateral absence of vas deferens, 277180, Autosomal recessive (Congenital bilateral absence of vas deferens) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Bilateral Vas Deferens Yokluğu, 277180, Otozomal Resesif (Konjenital Bilateral Vaz Deferens Yokluğu) (MLPA)	Congenital bilateral absence of vas deferens, 277180, Autosomal recessive (Congenital bilateral absence of vas deferens) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Konjenital Katarakt, Fasiyal Dismorfizm ve Nöropati, 604168, Otozomal Resesif; CCFDN (Konjenital Katarakt-Fasiyal Dismorfizm-Nöropati Sendromu) (CTDP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital cataracts, facial dysmorphism, and neuropathy, 604168, Autosomal recessive; CCFDN (Congenital cataracts-facial dysmorphism-neuropathy syndrome) (CTDP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTDP1	CTDP1, FCP1, CCFDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital cataracts, facial dysmorphism, and neuropathy (CCFDN) [MIM:604168]: An autosomal recessive developmental disorder characterized by a complex clinical phenotype with seemingly unrelated features involving multiple organs and systems. Developmental abnormalities include congenital cataracts and microcomeae, hypomyelination of the peripheral nervous system, impaired physical growth, delayed early motor and intellectual development, facial dysmorphism and hypogonadism. Central nervous system involvement, with cerebral and spinal cord atrophy, may be the result of disrupted development with superimposed degenerative changes. Affected individuals are prone to severe rhabdomyolysis after viral infections and to serious complications related to general anesthesia (such as pulmonary edema and epileptic seizures). {ECO:0000269	PubMed:14517542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Katarakt, Fasiyal Dismorfizm ve Nöropati, 604168, Otozomal Resesif; CCFDN (Konjenital Katarakt-Fasiyal Dismorfizm-Nöropati Sendromu) (Prenatal) (CTDP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital cataracts, facial dysmorphism, and neuropathy, 604168, Autosomal recessive; CCFDN (Congenital cataracts-facial dysmorphism-neuropathy syndrome) (CTDP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTDP1	CTDP1, FCP1, CCFDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital cataracts, facial dysmorphism, and neuropathy (CCFDN) [MIM:604168]: An autosomal recessive developmental disorder characterized by a complex clinical phenotype with seemingly unrelated features involving multiple organs and systems. Developmental abnormalities include congenital cataracts and microcomeae, hypomyelination of the peripheral nervous system, impaired physical growth, delayed early motor and intellectual development, facial dysmorphism and hypogonadism. Central nervous system involvement, with cerebral and spinal cord atrophy, may be the result of disrupted development with superimposed degenerative changes. Affected individuals are prone to severe rhabdomyolysis after viral infections and to serious complications related to general anesthesia (such as pulmonary edema and epileptic seizures). {ECO:0000269	PubMed:14517542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Katarakt, İşitme Kaybı ve Nörodejeneratif Hastalıklar, 614482, Otozomal Resesif; CCHLND (Konjenital Katarakt-İşitme Kaybı-Ağır Gelişimsel Gecikme Sendromu) (SLC33A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital cataracts, hearing loss, and neurodegeneration, 614482, Autosomal recessive; CCHLND (Congenital cataract-hearing loss-severe developmental delay syndrome) (SLC33A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC33A1	SLC33A1, ACATN, AT1, SPG42, CCHLND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital cataracts, hearing loss, and neurodegeneration (CCHLND) [MIM:614482]: An autosomal recessive disorder characterized by congenital cataracts, severe psychomotor retardation, and hearing loss associated with decreased serum ceruloplasmin and copper. Brain MRI shows cerebral and cerebellar atrophy and hypomyelination. {ECO:0000269	PubMed:22243965}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Katarakt, İşitme Kaybı ve Nörodejeneratif Hastalıklar, 614482, Otozomal Resesif; CCHLND (Konjenital Katarakt-İşitme Kaybı-Ağır Gelişimsel Gecikme Sendromu) (Prenatal) (SLC33A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital cataracts, hearing loss, and neurodegeneration, 614482, Autosomal recessive; CCHLND (Congenital cataract-hearing loss-severe developmental delay syndrome) (SLC33A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC33A1	SLC33A1, ACATN, AT1, SPG42, CCHLND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital cataracts, hearing loss, and neurodegeneration (CCHLND) [MIM:614482]: An autosomal recessive disorder characterized by congenital cataracts, severe psychomotor retardation, and hearing loss associated with decreased serum ceruloplasmin and copper. Brain MRI shows cerebral and cerebellar atrophy and hypomyelination. {ECO:0000269	PubMed:22243965}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ekstremitelerin ve Yüzün Konjenital Kontraktürleri, Hipotoni ve Gelişimsel Gecikme, 616266, Otozomal Dominant; CLIFAHDD (Dijitotalar Dismorfizmi) (NALCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266, Autosomal dominant; CLIFAHDD (Digitotalar dysmorphism) (NALCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NALCN	NALCN, IHPRF1, CLIFAHDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekstremitelerin ve Yüzün Konjenital Kontraktürleri, Hipotoni ve Gelişimsel Gecikme, 616266, Otozomal Dominant; CLIFAHDD (Dijitotalar Dismorfizmi) (Prenatal) (NALCN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital contractures of the limbs and face, hypotonia, and developmental delay, 616266, Autosomal dominant; CLIFAHDD (Digitotalar dysmorphism) (NALCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NALCN	NALCN, IHPRF1, CLIFAHDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Deglikosilasyon Defekti, 615273, Otozomal Resesif; CDDG (Alaleinia-Koreoatetozis-Karaciğer Fonksiyon Bozukluğu Sendromu) (NGLY1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of deglycosylation, 615273, Autosomal recessive; CDDG (Alacrima-choreoathetosis-liver dysfunction syndrome) (NGLY1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NGLY1	NGLY1, PNG1, CDDG, CDG1V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of deglycosylation (CDDG) [MIM:615273]: A multisystem disorder characterized by developmental delay, hypotonia, abnormal involuntary movements and alacrima or poor tear production. Other features include microcephaly, intractable seizures, abnormal eye movements and evidence of liver dysfunction, probably due to cytoplasmic accumulation of storage material in vacuoles. {ECO:0000269|PubMed:22581936, ECO:0000269|PubMed:24651605}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Deglikosilasyon Defekti, 615273, Otozomal Resesif; CDDG (Alaleinia-Koreoatetozis-Karaciğer Fonksiyon Bozukluğu Sendromu) (Prenatal) (NGLY1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of deglycosylation, 615273, Autosomal recessive; CDDG (Alacrima-choreoathetosis-liver dysfunction syndrome) (NGLY1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NGLY1	NGLY1, PNG1, CDDG, CDG1V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of deglycosylation (CDDG) [MIM:615273]: A multisystem disorder characterized by developmental delay, hypotonia, abnormal involuntary movements and alacrima or poor tear production. Other features include microcephaly, intractable seizures, abnormal eye movements and evidence of liver dysfunction, probably due to cytoplasmic accumulation of storage material in vacuoles. {ECO:0000269|PubMed:22581936, ECO:0000269|PubMed:24651605}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1aa, 617082, Otozomal Resesif (NUS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1aa, 617082, Autosomal recessive (NUS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUS1	NUS1, NGBR, C6orf68, CDG1AA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in NUS1 are the cause of a congenital disorder of glycosylation (CDG) characterized by reduced protein glycosylation and altered dolichol profiles in the urine and serum. Affected individuals manifest profound psychomotor retardation, refractory epilepsy, congenital scoliosis, hearing deficit, and visual impairment with macular lesions. {ECO:0000269	PubMed:25066056}.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1aa, 617082, Otozomal Resesif (Prenatal) (NUS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1aa, 617082, Autosomal recessive (NUS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUS1	NUS1, NGBR, C6orf68, CDG1AA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Defects in NUS1 are the cause of a congenital disorder of glycosylation (CDG) characterized by reduced protein glycosylation and altered dolichol profiles in the urine and serum. Affected individuals manifest profound psychomotor retardation, refractory epilepsy, congenital scoliosis, hearing deficit, and visual impairment with macular lesions. {ECO:0000269	PubMed:25066056}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1a, 212065, Otozomal Resesif; CDG1A (PMM2-CDG) (PMM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ia, 212065, Autosomal recessive; CDG1A (PMM2-CDG) (PMM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMM2	PMM2, CDG1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1A (CDG1A) [MIM:212065]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Congenital disorder of glycosylation type 1A is an autosomal recessive disorder characterized by a severe encephalopathy with axial hypotonia, abnormal eye movement, and pronounced psychomotor retardation, as well as peripheral neuropathy, cerebellar hypoplasia, and retinitis pigmentosa. Patients show a peculiar distribution of subcutaneous fat, nipple retraction, and hypogonadism. {ECO:0000269|PubMed:10066032, ECO:0000269|PubMed:10571956, ECO:0000269|PubMed:10602363, ECO:0000269|PubMed:10801058, ECO:0000269|PubMed:11058895, ECO:0000269|PubMed:11058896, ECO:0000269|PubMed:11350185, ECO:0000269|PubMed:12357336, ECO:0000269|PubMed:15844218, ECO:0000269|PubMed:17307006, ECO:0000269|PubMed:9140401, ECO:0000269|PubMed:9497260, ECO:0000269|PubMed:9781039}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1a, 212065, Otozomal Resesif; CDG1A (PMM2-CDG) (Prenatal) (PMM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ia, 212065, Autosomal recessive; CDG1A (PMM2-CDG) (PMM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PMM2	PMM2, CDG1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1A (CDG1A) [MIM:212065]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Congenital disorder of glycosylation type 1A is an autosomal recessive disorder characterized by a severe encephalopathy with axial hypotonia, abnormal eye movement, and pronounced psychomotor retardation, as well as peripheral neuropathy, cerebellar hypoplasia, and retinitis pigmentosa. Patients show a peculiar distribution of subcutaneous fat, nipple retraction, and hypogonadism. {ECO:0000269|PubMed:10066032, ECO:0000269|PubMed:10571956, ECO:0000269|PubMed:10602363, ECO:0000269|PubMed:10801058, ECO:0000269|PubMed:11058895, ECO:0000269|PubMed:11058896, ECO:0000269|PubMed:11350185, ECO:0000269|PubMed:12357336, ECO:0000269|PubMed:15844218, ECO:0000269|PubMed:17307006, ECO:0000269|PubMed:9140401, ECO:0000269|PubMed:9497260, ECO:0000269|PubMed:9781039}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1b, 602579, Otozomal Resesif; CDG1B (MPI-CDG) (MPI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ib, 602579, Autosomal recessive; CDG1B (MPI-CDG) (MPI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPI	MPI, PMI1, CDG1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1b, 602579, Otozomal Resesif; CDG1B (MPI-CDG) (Prenatal) (MPI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ib, 602579, Autosomal recessive; CDG1B (MPI-CDG) (MPI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPI	MPI, PMI1, CDG1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip Ic, 603147, Otozomal Resesif; CDG1C (ALG6-CDG) (ALG6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ic, 603147, Autosomal recessive; CDG1C (ALG6-CDG) (ALG6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG6	ALG6, CDG1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1C (CDG1C) [MIM:603147]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:10359825, ECO:0000269|PubMed:10914684, ECO:0000269|PubMed:10924277, ECO:0000269|PubMed:11106564, ECO:0000269|PubMed:11134235, ECO:0000269|PubMed:12357336, ECO:0000269|PubMed:14517965}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ic, 603147, Otozomal Resesif; CDG1C (ALG6-CDG) (Prenatal) (ALG6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ic, 603147, Autosomal recessive; CDG1C (ALG6-CDG) (ALG6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG6	ALG6, CDG1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1C (CDG1C) [MIM:603147]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:10359825, ECO:0000269|PubMed:10914684, ECO:0000269|PubMed:10924277, ECO:0000269|PubMed:11106564, ECO:0000269|PubMed:11134235, ECO:0000269|PubMed:12357336, ECO:0000269|PubMed:14517965}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Id, 601110, Otozomal Resesif; CDG1D (ALG3-CDG) (ALG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Id, 601110, Autosomal recessive; CDG1D (ALG3-CDG) (ALG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG3	ALG3, NOT56L, CDGS4, CDG1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1D (CDG1D) [MIM:601110]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:10581255, ECO:0000269|PubMed:15840742}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Id, 601110, Otozomal Resesif; CDG1D (ALG3-CDG) (Prenatal) (ALG3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Id, 601110, Autosomal recessive; CDG1D (ALG3-CDG) (ALG3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG3	ALG3, NOT56L, CDGS4, CDG1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1D (CDG1D) [MIM:601110]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:10581255, ECO:0000269|PubMed:15840742}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ie, 608799, Otozomal Resesif; CDG1E (DPM1-CDG) (DPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ie, 608799, Autosomal recessive; CDG1E (DPM1-CDG) (DPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPM1	DPM1, MPDS, CDG1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1E (CDG1E) [MIM:608799]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Some CDG1E patients have features consistent with a dystroglycanopathy and congenital muscular dystrophy, including O-mannosylation defect, camptodactyly, elevated creatine kinase, motor delay and dystrophic changes on muscel biopsy. {ECO:0000269|PubMed:10642597, ECO:0000269|PubMed:10642602, ECO:0000269|PubMed:15669674, ECO:0000269|PubMed:23856421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ie, 608799, Otozomal Resesif; CDG1E (DPM1-CDG) (Prenatal) (DPM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ie, 608799, Autosomal recessive; CDG1E (DPM1-CDG) (DPM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPM1	DPM1, MPDS, CDG1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1E (CDG1E) [MIM:608799]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Some CDG1E patients have features consistent with a dystroglycanopathy and congenital muscular dystrophy, including O-mannosylation defect, camptodactyly, elevated creatine kinase, motor delay and dystrophic changes on muscel biopsy. {ECO:0000269|PubMed:10642597, ECO:0000269|PubMed:10642602, ECO:0000269|PubMed:15669674, ECO:0000269|PubMed:23856421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip If, 609180, Otozomal Resesif; CDG1F (MPDU1-CDG) (MPDU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type If, 609180, Autosomal recessive; CDG1F (MPDU1-CDG) (MPDU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPDU1	MPDU1, SL15, CDGIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1F (CDG1F) [MIM:609180]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:11733556, ECO:0000269|PubMed:11733564}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikosilasyon Defekti, Tip If, 609180, Otozomal Resesif; CDG1F (MPDU1-CDG) (Prenatal) (MPDU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type If, 609180, Autosomal recessive; CDG1F (MPDU1-CDG) (MPDU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPDU1	MPDU1, SL15, CDGIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1F (CDG1F) [MIM:609180]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:11733556, ECO:0000269|PubMed:11733564}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ig, 607143 Tip; CDG1G (ALG12-CDG) (ALG12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ig, 607143; CDG1G (ALG12-CDG) (ALG12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG12	ALG12, CDG1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Lg, 607143 Tip; CDG1G (ALG12-CDG) (Prenatal) (ALG12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ig, 607143; CDG1G (ALG12-CDG) (ALG12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG12	ALG12, CDG1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ih, 608104 Tip; CDG1H (ALG8-CDG) (ALG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ih, 608104; CDG1H (ALG8-CDG) (ALG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG8	ALG8, CDG1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1H (CDG1H) [MIM:608104]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:15235028}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip Ih, 608104 Tip; CDG1H (ALG8-CDG) (Prenatal) (ALG8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ih, 608104; CDG1H (ALG8-CDG) (ALG8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG8	ALG8, CDG1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1H (CDG1H) [MIM:608104]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:15235028}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip 2, 607906, Otozomal Resesif; CDG1I (ALG2-CDG) (ALG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ii, 607906, Autosomal recessive; CDG1I (ALG2-CDG) (ALG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG2	ALG2, CDG1I, CMST3, CMS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1I (CDG1I) [MIM:607906]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12684507). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2, 607906, Otozomal Resesif; CDG1I (ALG2-CDG) (Prenatal) (ALG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ii, 607906, Autosomal recessive; CDG1I (ALG2-CDG) (ALG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG2	ALG2, CDG1I, CMST3, CMS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1I (CDG1I) [MIM:607906]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12684507). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2a, 212066, Otozomal Resesif; CDG2A (MGAT2-CDG) (MGAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIa, 212066, Autosomal recessive; CDG2A (MGAT2-CDG) (MGAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MGAT2	MGAT2, CDGS2, CDG2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2A (CDG2A) [MIM:212066]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269|PubMed:11228641, ECO:0000269|PubMed:8808595). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2a, 212066, Otozomal Resesif; CDG2A (MGAT2-CDG) (Prenatal) (MGAT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIa, 212066, Autosomal recessive; CDG2A (MGAT2-CDG) (MGAT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MGAT2	MGAT2, CDGS2, CDG2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2A (CDG2A) [MIM:212066]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269|PubMed:11228641, ECO:0000269|PubMed:8808595). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip IIb, 606056, Otozomal Resesif; CDG2B (GCS1-CDG) (MOGS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIb, 606056, Autosomal recessive; CDG2B (GCS1-CDG) (MOGS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MOGS	MOGS, GCS1, CDG2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Type IIb congenital disorder of glycosylation (CDGIIb) [MIM:606056]: Characterized by marked generalized hypotonia and hypomotility of the neonate, dysmorphic features, including a prominent occiput, short palpebral fissures, retrognathia, high arched palate, generalized edema, and hypoplastic genitalia. Symptoms of the infant included hepatomegaly, hypoventilation, feeding problems and seizures. The clinical course was progressive and the infant did not survive more than a few months. (ECO:0000269	PubMed:10788335). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip IIb, 606056, Otozomal Resesif; CDG2B (GCS1-CDG) (Prenatal) (MOGS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIb, 606056, Autosomal recessive; CDG2B (GCS1-CDG) (MOGS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MOGS	MOGS, GCS1, CDG2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Type IIb congenital disorder of glycosylation (CDGIIb) [MIM:606056]: Characterized by marked generalized hypotonia and hypomotility of the neonate, dysmorphic features, including a prominent occiput, short palpebral fissures, retrognathia, high arched palate, generalized edema, and hypoplastic genitalia. Symptoms of the infant included hepatomegaly, hypoventilation, feeding problems and seizures. The clinical course was progressive and the infant did not survive more than a few months. (ECO:0000269	PubMed:10788335). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2c, 266265, Otozomal Resesif; CDG2C (Lökosit Adezyon Eksikliği) (SLC35C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIc, 266265, Autosomal recessive; CDG2C (Leukocyte adhesion deficiency) (SLC35C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC35C1	SLC35C1, FUCT1, CDG2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2C (CDG2C) [MIM:266265]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. The clinical features of CDG2C include mental retardation, short stature, facial stigmata, and recurrent bacterial peripheral infections with persistently elevated peripheral leukocytes. Biochemically, CDG2C is characterized by a lack of fucosylated glycoconjugates, including selectin ligands. (ECO:0000269|PubMed:11326279, ECO:0000269|PubMed:11326280). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2c, 266265, Otozomal Resesif; CDG2C (Lökosit Adezyon Eksikliği) (Prenatal) (SLC35C1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIc, 266265, Autosomal recessive; CDG2C (Leukocyte adhesion deficiency) (SLC35C1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC35C1	SLC35C1, FUCT1, CDG2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2C (CDG2C) [MIM:266265]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. The clinical features of CDG2C include mental retardation, short stature, facial stigmata, and recurrent bacterial peripheral infections with persistently elevated peripheral leukocytes. Biochemically, CDG2C is characterized by a lack of fucosylated glycoconjugates, including selectin ligands. (ECO:0000269|PubMed:11326279, ECO:0000269|PubMed:11326280). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2d, 607091, Otozomal Resesif; CDG2D (B4GALT1-CDG) (B4GALT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IId, 607091, Autosomal recessive; CDG2D (B4GALT1-CDG) (B4GALT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B4GALT1	B4GALT1, GGTB2, GT1, GTB, CDG2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2D (CDG2D) [MIM:607091]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:11901181). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2d, 607091, Otozomal Resesif; CDG2D (B4GALT1-CDG) (Prenatal) (B4GALT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IId, 607091, Autosomal recessive; CDG2D (B4GALT1-CDG) (B4GALT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B4GALT1	B4GALT1, GGTB2, GT1, GTB, CDG2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2D (CDG2D) [MIM:607091]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:11901181). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2e, 608779; CDG2E (COG7-CDG) (COG7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIe, 608779; CDG2E (COG7-CDG) (COG7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG7	COG7, CDG2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2E (CDG2E) [MIM:608779]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:15107842). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2e, 608779; CDG2E (COG7-CDG) (Prenatal) (COG7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIe, 608779; CDG2E (COG7-CDG) (COG7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG7	COG7, CDG2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2E (CDG2E) [MIM:608779]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:15107842). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip 2f, 603585, Otozomal Resesif; CDG2F (SLC35A1-CDG) (SLC35A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIf, 603585, Autosomal recessive; CDG2F (SLC35A1-CDG) (SLC35A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC35A1	SLC35A1, CST, CDG2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2F (CDG2F) [MIM:603585]: CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. They are characterized by under-glycosylated serum proteins. These multisystem disorders present with a wide variety of clinical features, such as disorders of the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:15576474). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2f, 603585, Otozomal Resesif; CDG2F (SLC35A1-CDG) (Prenatal) (SLC35A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIf, 603585, Autosomal recessive; CDG2F (SLC35A1-CDG) (SLC35A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC35A1	SLC35A1, CST, CDG2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2F (CDG2F) [MIM:603585]: CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. They are characterized by under-glycosylated serum proteins. These multisystem disorders present with a wide variety of clinical features, such as disorders of the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:15576474). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2g, 611209, CDG2G (COG1-CDG) (COG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIg, 611209, CDG2G (COG1-CDG) (COG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG1	COG1, LDLB, KIAA1381, CDG2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2G (CDG2G) [MIM:611209]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2G include failure to thrive, generalized hypotonia, growth retardation and mild psychomotor retardation. CDG2G is biochemically characterized by a defect in O-glycosylation as well as N-glycosylation. (ECO:0000269	PubMed:16537452). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2g, 611209, CDG2G (COG1-CDG) (Prenatal) (COG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIg, 611209, CDG2G (COG1-CDG) (COG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG1	COG1, LDLB, KIAA1381, CDG2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2G (CDG2G) [MIM:611209]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2G include failure to thrive, generalized hypotonia, growth retardation and mild psychomotor retardation. CDG2G is biochemically characterized by a defect in O-glycosylation as well as N-glycosylation. (ECO:0000269	PubMed:16537452). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2h, 611182, CDG2H (COG8-CDG) (COG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIh, 611182, CDG2H (COG8-CDG) (COG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG8	COG8, DOR1, CDG2H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2H (CDG2H) [MIM:611182]: CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. They are characterized by under-glycosylated serum proteins. These multisystem disorders present with a wide variety of clinical features, such as disorders of the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:17331980). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2h, 611182, CDG2H (COG8-CDG) (Prenatal) (COG8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIh, 611182, CDG2H (COG8-CDG) (COG8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG8	COG8, DOR1, CDG2H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2H (CDG2H) [MIM:611182]: CDGs are a family of severe inherited diseases caused by a defect in protein N-glycosylation. They are characterized by under-glycosylated serum proteins. These multisystem disorders present with a wide variety of clinical features, such as disorders of the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:17331980). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2i, 613612, CDG2I (COG5-CDG) (COG5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIi, 613612, CDG2I (COG5-CDG) (COG5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG5	COG5, GOLTC1, GTC90, CDG2I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2I (CDG2I) [MIM:613612]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Congenital disorder of glycosylation type 2I is characterized by mild neurological impairments. (ECO:0000269	PubMed:19690088). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2i, 613612, CDG2I (COG5-CDG) (Prenatal) (COG5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIi, 613612, CDG2I (COG5-CDG) (COG5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG5	COG5, GOLTC1, GTC90, CDG2I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2I (CDG2I) [MIM:613612]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Congenital disorder of glycosylation type 2I is characterized by mild neurological impairments. (ECO:0000269	PubMed:19690088). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2j, 613489, Otozomal Resesif; CDG2J (COG4-CDG) (COG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIj, 613489, Autosomal recessive; CDG2J (COG4-CDG) (COG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG4	COG4, COD1, CDG2J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2J (CDG2J) [MIM:613489]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:19494034). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2j, 613489, Otozomal Resesif; CDG2J (COG4-CDG) (Prenatal) (COG4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIj, 613489, Autosomal recessive; CDG2J (COG4-CDG) (COG4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG4	COG4, COD1, CDG2J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2J (CDG2J) [MIM:613489]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:19494034). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2k, 614727, Otozomal Resesif; CDG2K (TMEM165-CDG) (TMEM165 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIk, 614727, Autosomal recessive; CDG2K (TMEM165-CDG) (TMEM165 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM165	TMEM165, FT27, CDG2K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2k, 614727, Otozomal Resesif; CDG2K (TMEM165-CDG) (Prenatal) (TMEM165 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIk, 614727, Autosomal recessive; CDG2K (TMEM165-CDG) (TMEM165 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM165	TMEM165, FT27, CDG2K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2l, 614576, Otozomal Resesif; CDG2L (COG6-ÇGD) (COG6 geni (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIl, 614576, Autosomal recessive; CDG2L (COG6-CGD) (COG6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG6	COG6, COD2, KIAA1134, CDG2L, SHNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2L (CDG2L) [MIM:614576]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2L include neonatal intractable focal seizures, vomiting, loss of consciousness, intracranial bleeding due to vitamin K deficiency, and death in infancy. (ECO:0000269	PubMed:20605848). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip 2l, 614576, Otozomal Resesif; CDG2L (COG6-CGD) (Prenatal) (COG6 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIl, 614576, Autosomal recessive; CDG2L (COG6-CGD) (COG6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG6	COG6, COD2, KIAA1134, CDG2L, SHNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2L (CDG2L) [MIM:614576]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2L include neonatal intractable focal seizures, vomiting, loss of consciousness, intracranial bleeding due to vitamin K deficiency, and death in infancy. {ECO:0000269|PubMed:20605848}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2m, 300896, X'e Bağlı Dominant, Somatik Mozaik; CDG2M (SLC35A2-CDG) (SLC35A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIm, 300896, X-linked dominant, Somatic mosaicism; CDG2M (SLC35A2-CDG) (SLC35A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC35A2	SLC35A2, UGALT, UGTL, UGT2, CDGX, CDG2M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2M (CDG2M) [MIM:300896]: A disorder characterized by developmental delay, hypotonia, ocular anomalies, and brain malformations. Othere more variable clinical features included seizures, hypsarrhythmia, poor feeding, microcephaly, recurrent infections, dysmorphic features, shortened limbs, and coagulation defects. Congenital disorders of glycosylation are caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins and a wide variety of clinical features. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:23561849, ECO:0000269|PubMed:24115232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2m, 300896, X'e Bağlı Dominant, Somatik Mozaik; CDG2M (SLC35A2-CDG) (Prenatal) (SLC35A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIm, 300896, X-linked dominant, Somatic mosaicism; CDG2M (SLC35A2-CDG) (SLC35A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC35A2	SLC35A2, UGALT, UGTL, UGT2, CDGX, CDG2M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2M (CDG2M) [MIM:300896]: A disorder characterized by developmental delay, hypotonia, ocular anomalies, and brain malformations. Othere more variable clinical features included seizures, hypsarrhythmia, poor feeding, microcephaly, recurrent infections, dysmorphic features, shortened limbs, and coagulation defects. Congenital disorders of glycosylation are caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins and a wide variety of clinical features. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:23561849, ECO:0000269|PubMed:24115232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2n, 616721, Otozomal Resesif; CDG2N (SLC39A8-CDG) (SLC39A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIn, 616721, Autosomal recessive; CDG2N (SLC39A8-CDG) (SLC39A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC39A8	SLC39A8, BIGM103, CDG2N	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2n, 616721, Otozomal Resesif; CDG2N (SLC39A8-CDG) (Prenatal) (SLC39A8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIn, 616721, Autosomal recessive; CDG2N (SLC39A8-CDG) (SLC39A8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC39A8	SLC39A8, BIGM103, CDG2N	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2o, 616828, Otozomal Resesif; CDG2O (CCDC115-CDG) (CCDC115 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIo, 616828, Autosomal recessive; CDG2O (CCDC115-CDG) (CCDC115 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC115	CCDC115, CCP1, CDG2O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2o, 616828, Otozomal Resesif; CDG2O (CCDC115-CDG) (Prenatal) (CCDC115 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIo, 616828, Autosomal recessive; CDG2O (CCDC115-CDG) (CCDC115 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC115	CCDC115, CCP1, CDG2O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2p, Otozomal Resesif; 616829; CDG2P (TMEM199-CDG) (TMEM199 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type IIp, 616829, Autosomal recessive; CDG2P (TMEM199-CDG) (TMEM199 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM199	TMEM199, VMA12, VPH2, C17orf32, CDG2P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 2p, Otozomal Resesif; 616829; CDG2P (TMEM199-CDG) (Prenatal) (TMEM199 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type IIp, 616829, Autosomal recessive; CDG2P (TMEM199-CDG) (TMEM199 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM199	TMEM199, VMA12, VPH2, C17orf32, CDG2P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1j, 608093, Otozomal Resesif (DPAGT1-CDG) (DPAGT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ij, 608093, Autosomal recessive; CDG1J (DPAGT1-CDG) (DPAGT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPAGT1	DPAGT1, DPAGT2, DGPT, CDG1J, CMST42, CMS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1J (CDG1J) [MIM:608093]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:12872255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1j, 608093, Otozomal Resesif (DPAGT1-CDG) (Prenatal) (DPAGT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ij, 608093, Autosomal recessive; CDG1J (DPAGT1-CDG) (DPAGT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPAGT1	DPAGT1, DPAGT2, DGPT, CDG1J, CMST42, CMS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1J (CDG1J) [MIM:608093]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:12872255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1k, 608540, Otozomal Resesif; CDG1K (ALG1-CDG) (ALG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ik, 608540, Autosomal recessive; CDG1K (ALG1-CDG) (ALG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG1	ALG1, HMAT1, HMT1, CDG1K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1K (CDG1K) [MIM:608540]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:14709599, ECO:0000269|PubMed:14973778, ECO:0000269|PubMed:14973782}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1k, 608540, Otozomal Resesif; CDG1K (ALG1-CDG) (Prenatal) (ALG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ik, 608540, Autosomal recessive; CDG1K (ALG1-CDG) (ALG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG1	ALG1, HMAT1, HMT1, CDG1K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1K (CDG1K) [MIM:608540]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:14709599, ECO:0000269|PubMed:14973778, ECO:0000269|PubMed:14973782}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1L, 608776; CDG1L (ALG9-CDG) (ALG9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Il, 608776; CDG1L (ALG9-CDG) (ALG9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG9	ALG9, DIBD1, CDG1L, GIKANIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1L (CDG1L) [MIM:608776]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:15148656, ECO:0000269|PubMed:15945070}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1L, 608776; CDG1L (ALG9-CDG) (Prenatal) (ALG9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Il, 608776; CDG1L (ALG9-CDG) (ALG9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG9	ALG9, DIBD1, CDG1L, GIKANIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1L (CDG1L) [MIM:608776]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:15148656, ECO:0000269|PubMed:15945070}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1m 610768, Otozomal Resesif; CDG1M (DK1-CDG) (DOLK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Im, 610768, Autosomal recessive; CDG1M (DK1-CDG) (DOLK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOLK	TMEM15, DK1, SEC59, KIAA1094, CDG1M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1m 610768, Otozomal Resesif; CDG1M (DK1-CDG) (Prenatal) (DOLK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Im, 610768, Autosomal recessive; CDG1M (DK1-CDG) (DOLK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOLK	TMEM15, DK1, SEC59, KIAA1094, CDG1M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip 1n, 612015, Otozomal Resesif; CDG1N (RFT1-CDG) (RFT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1n, 612015, Autosomal recessive; CDG1N (RFT1-CDG) (RFT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RFT1	RFT1, CDG1N	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1N (CDG1N) [MIM:612015]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:18313027, ECO:0000269|PubMed:19701946}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1n, 612015, Otozomal Resesif; CDG1N (RFT1-CDG) (Prenatal) (RFT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1n, 612015, Autosomal recessive; CDG1N (RFT1-CDG) (RFT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFT1	RFT1, CDG1N	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1N (CDG1N) [MIM:612015]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:18313027, ECO:0000269|PubMed:19701946}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1o, 612937, CDG1O (DPM3-CDG) (DPM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1o, 612937, CDG1O (DPM3-CDG) (DPM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPM3	DPM3, CDG1O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1O (CDG1O) [MIM:612937]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. CDG1O patients have increased serum creatine kinase, dystrophic changes on muscle biopsy, and reduced O-mannosylation of alpha-dystroglycan. {ECO:0000269|PubMed:19576565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1o, 612937, CDG1O (DPM3-CDG) (Prenatal) (DPM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1o, 612937, CDG1O (DPM3-CDG) (DPM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPM3	DPM3, CDG1O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1O (CDG1O) [MIM:612937]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. CDG1O patients have increased serum creatine kinase, dystrophic changes on muscle biopsy, and reduced O-mannosylation of alpha-dystroglycan. {ECO:0000269|PubMed:19576565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1p, 613661, Otozomal Resesif (ALG11-CDG) (ALG11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1p, 613661, Autosomal recessive (ALG11-CDG) (ALG11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG11	ALG11, KIAA1266, CDG1P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1P (CDG1P) [MIM:613661]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:20080937, ECO:0000269|PubMed:22213132}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1p, 613661, Otozomal Resesif (ALG11-CDG) (Prenatal) (ALG11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1p, 613661, Autosomal recessive (ALG11-CDG) (ALG11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG11	ALG11, KIAA1266, CDG1P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1P (CDG1P) [MIM:613661]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:20080937, ECO:0000269|PubMed:22213132}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1q, 612379, Otozomal Resesif; CDG1Q (El-Gazali-Dattani Sendromu) (SRD5A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1q, 612379, Autosomal recessive; CDG1Q (Al-Gazali-Dattani syndrome) (SRD5A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRD5A3	SRD5A3, SRD5A2L, CDG1Q, KRIZI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1q, 612379, Otozomal Resesif; CDG1Q (El-Gazali-Dattani Sendromu) (Prenatal) (SRD5A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1q, 612379, Autosomal recessive; CDG1Q (Al-Gazali-Dattani syndrome) (SRD5A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRD5A3	SRD5A3, SRD5A2L, CDG1Q, KRIZI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1r, 614507, Otozomal Resesif; CDG1R (DDOST-CDG) (DDOST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1r, 614507, Autosomal recessive; CDG1R (DDOST-CDG) (DDOST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDOST	DDOST, OST, OST48, CDG1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1R (CDG1R) [MIM:614507]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:22305527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1r, 614507, Otozomal Resesif; CDG1R (DDOST-CDG) (Prenatal) (DDOST geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1r, 614507, Autosomal recessive; CDG1R (DDOST-CDG) (DDOST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDOST	DDOST, OST, OST48, CDG1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1R (CDG1R) [MIM:614507]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:22305527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1S, 300884, X'e Bağlı Dominant (ALG13-CDG) (ALG13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1s, 300884, X-linked dominant (ALG13-CDG) (ALG13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG13	ALG13, GLT28D1, CDG1S, EIEE36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1S (CDG1S) [MIM:300884]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:22492991}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1S, 300884, X'e Bağlı Dominant (ALG13-CDG) (Prenatal) (ALG13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type 1s, 300884, X-linked dominant (ALG13-CDG) (ALG13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG13	ALG13, GLT28D1, CDG1S, EIEE36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1S (CDG1S) [MIM:300884]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:22492991}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1t, 614921, Otozomal Resesif; CDG1T (PGM1-CDG) (PGM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type 1t, 614921, Autosomal recessive; CDG1T(PGM1-CDG) (PGM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGM1	PGM1, GSD14, CDG1T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1T (CDG1T) [MIM:614921]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:19625727, ECO:0000269|PubMed:22492991, ECO:0000269|PubMed:22976764, ECO:0000269|PubMed:24499211, ECO:0000269|PubMed:25288802}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Konjenital Glikozilasyon Defekti, Tip 1t, 614921, Otozomal Resesif; CDG1T (PGM1-CDG) (Prenatal) (PGM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type It, 614921, Autosomal recessive; CDG1T(PGM1-CDG) (PGM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGM1	PGM1, GSD14, CDG1T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1T (CDG1T) [MIM:614921]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269|PubMed:19625727, ECO:0000269|PubMed:22482991, ECO:0000269|PubMed:22976764, ECO:0000269|PubMed:24499211, ECO:0000269|PubMed:25288802}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1u, 615042, Otozomal Resesif; CDG1U (Konjenital Müsküler Distrofi-Entelektüel Gerilik ve Ağır Epilepsi) (DPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Iu, 615042, Autosomal recessive; CDG1U (Congenital muscular dystrophy with intellectual disability and severe epilepsy) (DPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPM2	DPM2, CDG1U	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1U (CDG1U) [MIM:615042]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Some CDG1U patients have dystrophic changes seen on muscle biopsy and reduced O-mannosyl glycans on alpha-dystroglycan. {ECO:0000269	PubMed:23109149}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1u, 615042, Otozomal Resesif; CDG1U (Konjenital Müsküler Distrofi-Entelektüel Gerilik ve Ağır Epilepsi) (Prenatal) (DPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Iu, 615042, Autosomal recessive; CDG1U (Congenital muscular dystrophy with intellectual disability and severe epilepsy) (DPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPM2	DPM2, CDG1U	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1U (CDG1U) [MIM:615042]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Some CDG1U patients have dystrophic changes seen on muscle biopsy and reduced O-mannosyl glycans on alpha-dystroglycan. {ECO:0000269	PubMed:23109149}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1w, 615596, Otozomal Resesif; CDG1W (STT3A-CDG) (STT3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Iw, 615596, Autosomal recessive; CDG1W (STT3A-CDG) (STT3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STT3A	STT3A, ITM1, TMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1w, 615596, Otozomal Resesif; CDG1W (STT3A-CDG) (Prenatal) (STT3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Iw, 615596, Autosomal recessive; CDG1W (STT3A-CDG) (STT3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STT3A	STT3A, ITM1, TMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1x, 615597, Otozomal Resesif; CDG1X (STT3B-CDG) (STT3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Ix, 615597, Autosomal recessive; CDG1X (STT3B-CDG) (STT3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STT3B	STT3B, SIMP, CDG1X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1x, 615597, Otozomal Resesif; CDG1X (STT3B-CDG) (Prenatal) (STT3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Ix, 615597, Autosomal recessive; CDG1X (STT3B-CDG) (STT3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STT3B	STT3B, SIMP, CDG1X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1y, 300934, X'e Bağlı; CDG1Y (SSR4-CDG) (SSR4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital disorder of glycosylation, type Iy, 300934, X-linked recessive; CDG1Y (SSR4-CDG) (SSR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SSR4	SSR4, TRAPD, CDG1Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1Y (CDG1Y) [MIM:300934]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269	PubMed:24218363}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Glikozilasyon Defekti, Tip 1y, 300934, X'e Bağlı; CDG1Y (SSR4-CDG) (Prenatal) (SSR4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital disorder of glycosylation, type Iy, 300934, X-linked recessive; CDG1Y (SSR4-CDG) (SSR4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SSR4	SSR4, TRAPD, CDG1Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1Y (CDG1Y) [MIM:300934]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269	PubMed:24218363}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları ve Ektodermal Displazi, 617364, Otozomal Dominant; CHDED (PRKD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects and ectodermal dysplasia, 617364, Autosomal dominant; CHDED (PRKD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKD1	PRKD1, PRKCM, PKD, CHDED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Kalp Kusurları ve Ektodermal Displazi, 617364, Otozomal Dominant; CHDED (Prenatal) (PRKD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects and ectodermal dysplasia, 617364, Autosomal dominant; CHDED (PRKD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKD1	PRKD1, PRKCM, PKD, CHDED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları, Dismorfik Yüz Bulguları ve Entelektüel Gelişim Bozukluğu, 617360, Otozomal Dominant (CDK13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360, Autosomal dominant (CDK13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDK13	CDK13, CDC2L5, CHED, CHDFIDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Kalp Kusurları, Dismorfik Yüz Bulguları ve Entelektüel Gelişim Bozukluğu, 617360, Otozomal Dominant (Prenatal) (CDK13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects, dysmorphic facial features, and intellectual developmental disorder, 617360, Autosomal dominant (CDK13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDK13	CDK13, CDC2L5, CHED, CHDFIDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları, Dil Hamartomları ve Polisindaktili, 217085, Otozomal Resesif; CHDTHP (Kalp Defekti-Dil Hamartoması--Polisindaktili Sendromu) (WDPCP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085, Autosomal recessive; CHDTHP (Heart defect-tongue hamartoma-polysyndactyly syndrome) (WDPCP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDPCP	WDPCP, C2orf86, BBS15, CHDTHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital heart defects, hamartomas of tongue, and polysyndactyly (CHDTHP) [MIM:217085]: A disease characterized by a constellation of anomalies including tongue hamartomas, polysyndactyly, and congenital heart defects such as atrioventricular canal and coarctation of the aorta. {ECO:0000269	PubMed:25427950}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Kalp Kusurları, Dil Hamartomları ve Polisindaktili, 217085, Otozomal Resesif; CHDTHP (Kalp Defekti-Dil Hamartoması--Polisindaktili Sendromu) (Prenatal) (WDPCP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085, Autosomal recessive; CHDTHP (Heart defect-tongue hamartoma-polysyndactyly syndrome) (WDPCP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDPCP	WDPCP, C2orf86, BBS15, CHDTHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital heart defects, hamartomas of tongue, and polysyndactyly (CHDTHP) [MIM:217085]: A disease characterized by a constellation of anomalies including tongue hamartomas, polysyndactyly, and congenital heart defects such as atrioventricular canal and coarctation of the aorta. {ECO:0000269	PubMed:25427950}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları, Çeşitli Tipler, 4, 615779, Otozomal Dominant; CHTD4 (Atriyoventriküler Kanal Defekti) (NR2F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects, multiple types, 4, 615779, Autosomal dominant; CHTD4 (Atrioventricular canal defect) (NR2F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR2F2	NR2F2, TFCOUP2, ARP1, CHTD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Kalp Kusurları, Çeşitli Tipler, 4, 615779, Otozomal Dominant; CHTD4 (Atriyoventriküler Kanal Defekti) (Prenatal) (NR2F2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects, multiple types, 4, 615779, Autosomal dominant; CHTD4 (Atrioventricular canal defect) (NR2F2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NR2F2	NR2F2, TFCOUP2, ARP1, CHTD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları, Sendromik Olmayan, 1, X'e Bağlı, 306955, X'e Bağlı Resesif (Heterotaksi) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects, nonsyndromic, 1, X-linked, 306955, X-linked recessive (Heterotaxia) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Kalp Kusurları, Sendromik Olmayan, 1, X'e Bağlı, 306955, X'e Bağlı Resesif (Heterotaksi) (Prenatal) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects, nonsyndromic, 1, X-linked, 306955, X-linked recessive (Heterotaxia) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kalp Kusurları, Sendromik Olmayan, 2, 614980, Otozomal Dominant (TAB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital heart defects, nonsyndromic, 2, 614980, Autosomal dominant (TAB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAB2	TAB2, MAP3K7IP2, KIAA0733, CHTD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital heart defects, multiple types, 2 (CHTD2) [MIM:614980]: A disease characterized by congenital developmental abnormalities involving structures of the heart. CHTD2 patients have left ventricular outflow tract obstruction, subaortic stenosis, residual aortic regurgitation, atrial fibrillation, bicuspid aortic valve and aortic dilation. {ECO:0000269	PubMed:20493459}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving TAB2 has been found in a family with congenital heart disease. Translocation t(7;10)(p21;q25).	EDTA Blood Sample

Konjenital Kalp Kusurları, Sendromik Olmayan, 2, 614980, Otozomal Dominant (Prenatal) (TAB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital heart defects, nonsyndromic, 2, 614980, Autosomal dominant (TAB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAB2	TAB2, MAP3K7IP2, KIAA0733, CHTD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital heart defects, multiple types, 2 (CHTD2) [MIM:614980]: A disease characterized by congenital developmental abnormalities involving structures of the heart. CHTD2 patients have left ventricular outflow tract obstruction, subaortic stenosis, residual aortic regurgitation, atrial fibrillation, bicuspid aortic valve and aortic dilation. {ECO:0000269	PubMed:20493459}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving TAB2 has been found in a family with congenital heart disease. Translocation t(2;6)(p21;q25).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Hemidisplazi-İktiyoziform Eritrodermi ve Ekstremite Sorunları (CHILD Sendromu) (NSDHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS (CHILD syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Hemidisplazi-İktiyoziform Eritrodermi ve Ekstremite Sorunları (CHILD Sendromu) (Prenatal) (NSDHL geni) (Tüm Gen Dizi Analizi) (Prenatal)	CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS (CHILD syndrome) (NSDHL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSDHL	NSDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Lipomatöz Aşırı Büyüme, Vasküler Malformasyonlar ve Epidermal Nevüsler (CLOVES Sendromu) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI (CLOVES syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Lipomatöz Aşırı Büyüme, Vasküler Malformasyonlar ve Epidermal Nevüsler (CLOVES Sendromu) (Prenatal) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Prenatal)	CONGENITAL LIPOMATOUS OVERGROWTH, VASCULAR MALFORMATIONS, AND EPIDERMAL NEVI (CLOVES syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital müsküler distrofi tip 1A (konjenital merosin eksikliği) (Mix 1) (6q22.33) (MLPA)/ Konjenital müsküler distrofi tip 1A (konjenital merosin eksikliği) (Mix 2) (6q22.33) (MLPA)	Congenital muscular dystrophy type 1A (congenital merosin deficiency) (Mix 1) (6q22.33) (MLPA) / Congenital muscular dystrophy type 1A (congenital merosin deficiency) (Mix 2) (6q22.33) (MLPA)	6q22.33	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Konjenital müsküler distrofi tip 1A (konjenital merosin eksikliği) (Miks 1) (6q22.33) (MLPA)/ Konjenital müsküler distrofi tip 1A (konjenital merosin eksikliği) (Miks 2) (6q22.33) (MLPA) (Prenatal)	Congenital muscular dystrophy type 1A (congenital merosin deficiency) (Mix 1) (6q22.33) (MLPA) / Congenital muscular dystrophy type 1A (congenital merosin deficiency) (Mix 2) (6q22.33) (MLPA) (Prenatal)	6q22.33	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Miyopati, Kas İğciklerinde Artış 218040, Otozomal Dominant, Sporadik Vakalar (Costello Sendromu) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital myopathy with excess of muscle spindles, 218040, Autosomal dominant, Isolated cases (Costello syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Miyopati, Kas İğciklerinde Artış 218040, Otozomal Dominant, Sporadik Vakalar (Costello Sendromu) (MLPA)	Congenital myopathy with excess of muscle spindles, 218040, Autosomal dominant, Isolated cases (Costello syndrome) (MLPA)	HRAS	HRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Konjenital Miyopati, Kas İğciklerinde Artış 218040, Otozomal Dominant, Sporadik Vakalar (Costello Sendromu) (Prenatal) (HRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital myopathy with excess of muscle spindles, 218040, Autosomal dominant, Isolated cases (Costello syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Miyopati, Kas İğciklerinde Artış 218040, Otozomal Dominant, Sporadik Vakalar (Costello Sendromu) (Prenatal) (MLPA)	Congenital myopathy with excess of muscle spindles, 218040, Autosomal dominant, Isolated cases (Costello syndrome) (Prenatal) (MLPA)	HRAS	HRAS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 300048, X'e Bağlı Resesif (Konjenital Kısa Bağırsak Sendromu) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital short bowel syndrome, 300048, X-linked recessive (Congenital short bowel syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 300048, X'e Bağlı Resesif (Konjenital Kısa Bağırsak Sendromu) (MLPA)	Congenital short bowel syndrome, 300048, X-linked recessive (Congenital short bowel syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 300048, X'e Bağlı Resesif (Konjenital Kısa Bağırsak Sendromu) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital short bowel syndrome, 300048, X-linked recessive (Congenital short bowel syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 300048, X'e Bağlı Resesif (Konjenital Kısa Bağırsak Sendromu) (Prenatal) (MLPA)	Congenital short bowel syndrome, 300048, X-linked recessive (Congenital short bowel syndrome) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 615237, Otozomal Resesif (Konjenital Kısa Bağırsak Sendromu) (CLMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congenital short bowel syndrome, 615237, Autosomal recessive; CSBS (Congenital short bowel syndrome) (CLMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLMP	CLMP, ASAM, ACAM, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital short bowel syndrome (CSBS) [MIM:615237]: A disease characterized by a shortened small intestine, intestinal malrotation, and malabsorption. The mean length of the small intestine in CSBS patients is approximately 50 cm, compared with a normal length at birth of 190-280 cm. Patients with CSBS may develop severe malnutrition as a result of the hugely reduced absorptive surface of the small intestine. Infants require parenteral nutrition for survival. However, parenteral nutrition itself causes life-threatening complications such as sepsis and liver failure which are associated with a high rate of mortality early in life. {ECO:0000269	PubMed:22155368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konjenital Kısa Barsak Sendromu, 615237, Otozomal Resesif (Konjenital Kısa Bağırsak Sendromu) (Prenatal) (CLMP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Congenital short bowel syndrome, 615237, Autosomal recessive; CSBS (Congenital short bowel syndrome) (CLMP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLMP	CLMP, ASAM, ACAM, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital short bowel syndrome (CSBS) [MIM:615237]: A disease characterized by a shortened small intestine, intestinal malrotation, and malabsorption. The mean length of the small intestine in CSBS patients is approximately 50 cm, compared with a normal length at birth of 190-280 cm. Patients with CSBS may develop severe malnutrition as a result of the hugely reduced absorptive surface of the small intestine. Infants require parenteral nutrition for survival. However, parenteral nutrition itself causes life-threatening complications such as sepsis and liver failure which are associated with a high rate of mortality early in life. {ECO:0000269	PubMed:22155368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjestif Kalp Yetmezliği ve Beta Blokör Yanıtı, Modifiye Edici (ADRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congestive heart failure and beta-blocker response, modifier of (ADRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB1	ADRB1, ADRB1R, RHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjestif Kalp Yetmezliği ve Beta Blokör Yanıtı, Modifiye Edici (ADRA2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Congestive heart failure and beta-blocker response, modifier of (ADRA2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRA2C	ADRA2C, ADRA2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konotrunkal Anomali Yüz Sendromu, 217095 (Konotrunkal Kalp Malformasyonları) (TBX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Conotruncal anomaly face syndrome, 217095 (Conotruncal heart malformations) (TBX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konotrunkal Anomali Yüz Sendromu, 217095 (Konotrunkal Kalp Malformasyonları) (Prenatal) (TBX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Conotruncal anomaly face syndrome, 217095 (Conotruncal heart malformations) (TBX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konotrunkal Kalp Malformasyonları, 217095; CTHM (Konotrunkal Kalp Malformasyonları) (NKX2-6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Conotruncal heart malformations, 217095; CTHM (Conotruncal heart malformations) (NKX2-6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-6	NKX2-6, CSX2, CTHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Conotruncal heart malformations (CTHM) [MIM:217095]: A group of congenital heart defects involving the outflow tracts. Examples include truncus arteriosus communis, double-outlet right ventricle and transposition of great arteries. Truncus arteriosus communis is characterized by a single outflow tract instead of a separate aorta and pulmonary artery. In transposition of the great arteries, the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. In double outlet of the right ventricle, both the pulmonary artery and aorta arise from the right ventricle. {ECO:0000269|PubMed:15649947, ECO:0000269|PubMed:24421281, ECO:0000269|PubMed:25195019, ECO:0000269|PubMed:25380965}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Konotrunkal Kalp Malformasyonları, 217095; CTHM (Konotrunkal Kalp Malformasyonları) (Prenatal) (NKX2-6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Conotruncal heart malformations, 217095; CTHM (Conotruncal heart malformations) (NKX2-6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-6	NKX2-6, CSX2, CTHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Conotruncal heart malformations (CTHM) [MIM:217095]: A group of congenital heart defects involving the outflow tracts. Examples include truncus arteriosus communis, double-outlet right ventricle and transposition of great arteries. Truncus arteriosus communis is characterized by a single outflow tract instead of a separate aorta and pulmonary artery. In transposition of the great arteries, the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. In double outlet of the right ventricle, both the pulmonary artery and aorta arise from the right ventricle. {ECO:0000269|PubMed:15649947, ECO:0000269|PubMed:24421281, ECO:0000269|PubMed:25195019, ECO:0000269|PubMed:25380965}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konotrunkal Kalp Malformasyonları, Değişken, 217095 (Konotrunkal Kalp Malformasyonları) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Conotruncal heart malformations, variable, 217095 (Conotruncal heart malformations) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Konotruntkal Kalp Malformasyonlar, Değişiken, 217095 (Konotruntkal Kalp Malformasyonlar) (Prenatal) (NKX2-5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Conotruncal heart malformations, variable, 217095 (Conotruncal heart malformations) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kontraktürel Araknodaktili, Konjenital, 121050, Otozomal Dominant; DA9 (Konjenital Kontraktürel Araknodaktili) (FBN2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Contractural arachnodactyly, congenital, 121050, Autosomal dominant; DA9 (Congenital contractural arachnodactyly) (FBN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN2	FBN2, CCA, EOMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kontraktürel Araknodaktili, Konjenital, 121050, Otozomal Dominant; DA9 (Konjenital Kontraktürel Araknodaktili) (Prenatal) (FBN2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Contractural arachnodactyly, congenital, 121050, Autosomal dominant; DA9 (Congenital contractural arachnodactyly) (FBN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBN2	FBN2, CCA, EOMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Paroksismal Koreoatetozlu Konvülsiyonlar, Ailesel İnfantil, 602066, Otozomal Dominant; ICCA (İnfantil Konvülsiyon ve Koreoatetozis) (PRRT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Convulsions, familial infantile, with paroxysmal choreoathetosis, 602066, Autosomal dominant; ICCA (Infantile convulsions and choreoathetosis) (PRRT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRRT2	PRRT2, PKC, DYT10, EKD1, BFIS2, BFIC2, ICCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KOAH, Akciğer Fonksiyonlarında Azalma Hızı, 606963 (MMP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	COPD, rate of decline of lung function in, 606963 (MMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP1	MMP1, CLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koproporfiri, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (CPOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coproporphyria, 121300, Autosomal dominant; HCP (Hereditary coproporphyria) (CPOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPOX	CPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koproporfiri, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (MLPA)	Coproporphyria, 121300, Autosomal dominant; HCP (Hereditary coproporphyria) (MLPA)	CPOX	CPOX	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koproporfiri, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (Prenatal) (CPOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Coproporphyria, 121300, Autosomal dominant; HCP (Hereditary coproporphyria) (CPOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPOX	CPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koproporfiri, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (Prenatal) (MLPA)	Coproporphyria, 121300, Autosomal dominant; HCP (Hereditary coproporphyria) (Prenatal) (MLPA)	CPOX	CPOX	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kornea Plana 2, Otozomal Resesif, 217300, Otozomal Resesif; CNA2 (Konjenital Kornea Plana) (KERA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornea plana 2, autosomal recessive, 217300, Autosomal recessive; CNA2 (Congenital cornea plana) (KERA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KERA	KERA, CNA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: The autosomal recessive cornea plana 2 (CNA2) [MIM:217300]: In CNA2, the forward convex curvature is flattened, leading to a decrease in refraction, reduced visual activity, extreme hyperopia (usually plus 10 d or more), hazy corneal limbus, opacities in the corneal parenchyma, and marked arcus senilis (often detected at an early age). CNA2 is a rare disorder with a worldwide distribution, but a high prevalence in the Finnish population. {ECO:0000269|PubMed:10802664, ECO:0000269|PubMed:11726611}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kornea Bulutlanma, Otozomal Resesif (APOA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal clouding, autosomal recessive (APOA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA1	APOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi ve Perseptif Sağırlık; CDPD (Korneal Distrofi-Perseptif Sağırlık Sendromu) (SLC4A11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CORNEAL DYSTROPHY AND PERCEPTIVE DEAFNESS; CDPD (Corneal dystrophy-perceptive deafness syndrome) (SLC4A11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A11	SLC4A11, BTR1, NABC1, CHED, CDPD, FECD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy and perceptive deafness (CDPD) [MIM:217400]: An ocular disease characterized by the association of corneal clouding with progressive perceptive hearing loss. {ECO:0000269|PubMed:17230209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kornea Distrofisi, Avellino Tipi, 607541, Otozomal Dominant; CDA (Granül Kornea Distrofisi Tip 2) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Avellino type, 607541, Autosomal dominant; CDA (Granular corneal dystrophy type II) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Konjenital Stromal 610048, Otozomal Dominant; CSCD (Konjenital Stromal Kornea Distrofisi) (DCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, congenital stromal, 610048, Autosomal dominant; CSCD (Congenital stromal corneal dystrophy) (DCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCN	DCN, CSCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, congenital stromal (CSCD) [MIM:610048]: A corneal dystrophy characterized by congenital corneal opacification consisting of a large number of flakes and spots throughout all layers of the stroma. It results in progressive, painless visual loss. Corneal erosions and photophobia are absent. {ECO:0000269|PubMed:15671264}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kornea Distrofisi, Bazal Membran, 121820, Otozomal Dominant; EBMD (Mikrokistik Kornea Distrofisi) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, epithelial basement membrane, 121820, Autosomal dominant; EBMD (Microcystic corneal dystrophy) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, epithelial basement membrane (EBMD) [MIM:121820]: A bilateral anterior corneal dystrophy characterized by grayish epithelial fingerprint lines, geographic map-like lines, and dots (or microcysts) on slit-lamp examination. Pathologic studies show abnormal, redundant basement membrane and intraepithelial lacunae filled with cellular debris. {ECO:0000269|PubMed:16652336}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kornea Distrofisi, Fleck (Fleck Kornea Distrofisi) (PIKFYVE geni) (Tüm Gen Dizi Analizi) (Postnatal)	CORNEAL DYSTROPHY, FLECK (Fleck corneal dystrophy) (PIKFYVE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIKFYVE	PIKFYVE, PIP5K3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, fleck (CFD) [MIM:121850]: A form of stromal corneal dystrophy characterized by numerous small white flecks scattered in all levels of the stroma, with configurations varying from semicircular to wreath-like, curvilinear, or punctate. Although CFD may occasionally cause mild photophobia, patients are typically asymptomatic and have normal vision. {ECO:0000269|PubMed:15902656}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kornea Distrofisi, Fuchs Endotelyal, 1, 136800, Otozomal Dominant (Fuchs Endotelyal Kornea Distrofisi) (COL8A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Fuchs endothelial, 1, 136800, Autosomal dominant (Fuchs endothelial corneal dystrophy) (COL8A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL8A2	COL8A2, FECD1, PPCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, Fuchs endothelial, 1 (FECD1) [MIM:136800]: A corneal disease caused by loss of endothelium of the central cornea. It is characterized by focal wart-like guttata that arise from Descemet membrane and develop in the central cornea, epithelial blisters, reduced vision and pain. Descemet membrane is thickened by abnormal collagenous deposition. {ECO:0000269|PubMed:11689489}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kornea Distrofisi, Fuchs Endotelyal, 3, 613267, Otozomal Dominant; FECD3 (Fuchs Endotelyal Kornea Distrofisi) (TCF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Fuchs endothelial, 3, 613267, Autosomal dominant; FECD3 (Fuchs endothelial corneal dystrophy) (TCF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCF4	TCF4, SEF2, ITF2, PTHS, FECD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kornea Distrofisi, Fuchs Endotelyal, 4, 613268; FECD4 (Fuchs Endotelyal Kornea Distrofisi) (SLC4A11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Fuchs endothelial, 4, 613268; FECD4 (Fuchs endothelial corneal dystrophy) (SLC4A11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A11	SLC4A11, BTR1, NABC1, CHED, CDPD, FECD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Fuchs Endotelyal, 6, 613270; FECD6 (Fuchs Endotelyal Kornea Distrofisi) (ZEB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Fuchs endothelial, 6, 613270; FECD6 (Fuchs endothelial corneal dystrophy) (ZEB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZEB1	ZEB1, TCF8, NIL2A, PPCD3, FECD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, Fuchs endothelial, 6 (FECD6) [MIM:613270]: A corneal disease caused by loss of endothelium of the central cornea. It is characterized by focal wart-like guttata that arise from Descemet membrane and develop in the central cornea, epithelial blisters, reduced vision and pain. Descemet membrane is thickened by abnormal collagenous deposition. {ECO:0000269|PubMed:20036349, ECO:0000269|PubMed:23599324}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kornea Distrofisi, Fuchs Endotelyal, 8, 615523, Otozomal Dominant; FECD8 (Fuchs Endotelyal Kornea Distrofisi) (AGBL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Fuchs endothelial, 8, 615523, Autosomal dominant; FECD8 (Fuchs endothelial corneal dystrophy) (AGBL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGBL1	AGBL1, CCP4, FECD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Jelatinimsi Damla Benzeri, 204870, Otozomal Resesif; GDLD (Jelatinöz Damla Benzeri Kornea Distrofisi) (TACSTD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, gelatinous drop-like, 204870, Autosomal recessive; GDLD (Gelatinous drop-like corneal dystrophy) (TACSTD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TACSTD2	TACSTD2, TROP2, M1S1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, gelatinous drop-like (GDLD) [MIM:204870]: A form of lattice corneal dystrophy, a class of inherited stromal amyloidoses characterized by pathognomonic branching lattice figures in the cornea. GDLD is an autosomal recessive disorder characterized by severe corneal amyloidosis leading to blindness. Clinical manifestations, which appear in the first decade of life, include blurred vision, photophobia, and foreign-body sensation. By the third decade, raised, yellowish-gray, gelatinous masses severely impair visual acuity. {ECO:0000269|PubMed:10192395}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kornea Distrofisi, Groenouw Tipi I, 121900, Otozomal Dominant; CDGG1 (Granül Kornea Distrofisi Tip 1) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Groenouw type I, 121900, Autosomal dominant; CDGG1 (Granular corneal dystrophy type I) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Kafes Tipi I, 122200, Otozomal Dominant; LCD1 (Lattice Kornea Distrofi Tip 1) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, lattice type I, 122200, Autosomal dominant; LCD1 (Lattice corneal dystrophy type I) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Kornea Distrofisi, Kafes Tipi 3A, 608471, Otozomal Dominant; CDL3A (Lattice Kornea Distrofi Tip 1) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, lattice type IIIA, 608471, Autosomal dominant; CDL3A (Lattice corneal dystrophy type I) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Posterior Polimorf 2, 609140; PPCD2 (Posterior Polimorfoz Kornea Distrofisi) (COL8A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, posterior polymorphous 2, 609140; PPCD2 (Posterior polymorphous corneal dystrophy) (COL8A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL8A2	COL8A2, FECD1, PPCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Otozomal Dominant Polimorf Atka, 1, 122000; PPCD1 (Posterior Polimorfoz Kornea Distrofisi) (OVOL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, posterior polymorphous, 1, 122000, Autosomal dominant; PPCD1 (Posterior polymorphous corneal dystrophy) (OVOL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OVOL2	OVOL2, ZNF339, PPCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Posterior Polimorf, 3, 609141; PPCD3 (Posterior Polimorfoz Kornea Distrofisi) (ZEB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, posterior polymorphous, 3, 609141; PPCD3 (Posterior polymorphous corneal dystrophy) (ZEB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZEB1	ZEB1, TCF8, NIL2A, PPCD3, FECD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Reis-Bucklers Tipi, 608470; CDRB (Reis-Bücklers Kornea Distrofisi) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Reis-Bucklers type, 608470; CDRB (Reis-Bücklers corneal dystrophy) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Schnyder Tipi, 121800, Otozomal Dominant; SCCD (Schnyder Kornea Distrofisi) (UBIAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Schnyder type, 121800, Autosomal dominant; SCCD (Schnyder corneal dystrophy) (UBIAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBIAD1	UBIAD1, TERE1, SCCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Distrofisi, Thiel-Behnke Tipi, 602082, Otozomal Dominant; CDTB (Thiel-Behnke Kornea Distrofisi) (TGFBI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal dystrophy, Thiel-Behnke type, 602082, Autosomal dominant; CDTB (Thiel-Behnke corneal dystrophy) (TGFBI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBI	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2, EBMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kornea Endotel Distrofi ve Perseptif Sağırlık, 217400, Otozomal Resesif (Korneal Distrofi-Perseptif Sağırlık Sendromu) (SLC4A11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal endothelial dystrophy and perceptive deafness, 217400, Autosomal recessive (Corneal dystrophy-perceptive deafness syndrome) (SLC4A11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A11	SLC4A11, BTR1, NABC1, CHED, CDPD, FECD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy and perceptive deafness (CDPD) [MIM:217400]: An ocular disease characterized by the association of corneal clouding with progressive perceptive hearing loss. {ECO:0000269|PubMed:17220209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Korneal Endotelial Distrofi, Otozomal Resesif, 217700, Otozomal Resesif; CHED (Konjenital Kalıtsal Endotelyal Distrofi Tip 2) (SLC4A11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal endothelial dystrophy, autosomal recessive, 217700, Autosomal recessive; CHED (Congenital hereditary endothelial dystrophy type II) (SLC4A11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A11	SLC4A11, BTR1, NABC1, CHED, CDPD, FECD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy and perceptive deafness (CDPD) [MIM:217400]: An ocular disease characterized by the association of corneal clouding with progressive perceptive hearing loss. {ECO:0000269|PubMed:17220209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kornea Leke Distrofi, 121850, Otozomal Dominant (Fleck Kornea Distrofisi) (PIKFYVE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corneal fleck dystrophy, 121850, Autosomal dominant (Fleck corneal dystrophy) (PIKFYVE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIKFYVE	PIKFYVE, PIP5K3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corneal dystrophy, fleck (CFD) [MIM:121850]: A form of stromal corneal dystrophy characterized by numerous small white flecks scattered in all levels of the stroma, with configurations varying from semicircular to wreath-like, curvilinear, or punctate. Although CFD may occasionally cause mild photophobia, patients are typically asymptomatic and have normal vision. {ECO:0000269|PubMed:15902656}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 1, 122470, Otozomal Dominant; CDLS1 (Cornelia De Lange Sendromu) (NIPBL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornelia de Lange syndrome 1, 122470, Autosomal dominant; CDLS1 (Cornelia de Lange syndrome) (NIPBL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NIPBL	NIPBL, CDLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cornelia de Lange syndrome 1 (CDLS1) [MIM:122470]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:15146185, ECO:0000269|PubMed:15146186, ECO:0000269|PubMed:15318302, ECO:0000269|PubMed:20124326, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:21934712, ECO:0000269|PubMed:23254390, ECO:0000269|PubMed:25447906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 1, 122470, Otozomal Dominant; CDLS1 (Cornelia De Lange Sendromu) (MLPA)	Cornelia de Lange syndrome 1, 122470, Autosomal dominant; CDLS1 (Cornelia de Lange syndrome) (MLPA)	NIPBL	NIPBL, CDLS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cornelia de Lange syndrome 1 (CDLS1) [MIM:122470]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:15146185, ECO:0000269|PubMed:15146186, ECO:0000269|PubMed:15318302, ECO:0000269|PubMed:20124326, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:21934712, ECO:0000269|PubMed:23254390, ECO:0000269|PubMed:25447906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 1, 122470, Otozomal Dominant; CDLS1 (Cornelia De Lange Sendromu) (Prenatal) (NIPBL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cornelia de Lange syndrome 1, 122470, Autosomal dominant; CDLS1 (Cornelia de Lange syndrome) (NIPBL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NIPBL	NIPBL, CDLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 1 (CDLS1) [MIM:122470]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:15146185, ECO:0000269|PubMed:15146186, ECO:0000269|PubMed:15318302, ECO:0000269|PubMed:20124326, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:21934712, ECO:0000269|PubMed:23254390, ECO:0000269|PubMed:25447906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cornelia De Lange Sendromu 1, 122470, Otozomal Dominant; CDLS1 (Cornelia De Lange Sendromu) (Prenatal) (MLPA)	Cornelia de Lange syndrome 1, 122470, Autosomal dominant; CDLS1 (Cornelia de Lange syndrome) (Prenatal) (MLPA)	NIPBL	NIPBL, CDLS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 1 (CDLS1) [MIM:122470]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:15146185, ECO:0000269|PubMed:15146186, ECO:0000269|PubMed:15318302, ECO:0000269|PubMed:20124326, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:21934712, ECO:0000269|PubMed:23254390, ECO:0000269|PubMed:25447906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Cornelia De Lange Sendromu 2, 300590, X'e Bağlı Dominant; CDLS2 (Cornelia De Lange Sendromu) (SMC1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornelia de Lange syndrome 2, 300590, X-linked dominant; CDLS2 (Cornelia de Lange syndrome) (SMC1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMC1A	SMC1A, SMC1L1, SMC1, DXS423E, KIAA0178, CDLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cornelia de Lange syndrome 2 (CDLS2) [MIM:300590]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:16604071, ECO:0000269|PubMed:17221863, ECO:0000269|PubMed:17273969, ECO:0000269|PubMed:19701948, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:20635401}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 2, 300590, X'e Bağlı Dominant; CDLS2 (Cornelia De Lange Sendromu) (Prenatal) (SMC1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cornelia de Lange syndrome 2, 300590, X-linked dominant; CDLS2 (Cornelia de Lange syndrome) (SMC1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMC1A	SMC1A, SMC1L1, SMC1, DXS423E, KIAA0178, CDLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 2 (CDLS2) [MIM:300590]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269|PubMed:16604071, ECO:0000269|PubMed:17221863, ECO:0000269|PubMed:17273969, ECO:0000269|PubMed:19701948, ECO:0000269|PubMed:20358602, ECO:0000269|PubMed:20635401}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cornelia De Lange Sendromu 3, 610759, Otozomal Dominant; CDLS3 (Cornelia De Lange Sendromu) (SMC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornelia de Lange syndrome 3, 610759, Autosomal dominant; CDLS3 (Cornelia de Lange syndrome) (SMC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMC3	SMC3, CSPG6, HCAP, BAM, CDLS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cornelia de Lange syndrome 3 (CDLS3) [MIM:610759]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. Cornelia de Lange syndrome type 3 is a mild form with absence of major structural anomalies. The phenotype in some instances approaches that of apparently non- syndromic mental retardation. {ECO:0000269	PubMed:17273969}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 3, 610759, Otozomal Dominant; CDLS3 (Cornelia De Lange Sendromu) (SMC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cornelia de Lange syndrome 3, 610759, Autosomal dominant; CDLS3 (Cornelia de Lange syndrome) (SMC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMC3	SMC3, CSPG6, HCAP, BAM, CDLS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 3 (CDLS3) [MIM:610759]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. Characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. Cornelia de Lange syndrome type 3 is a mild form with absence of major structural anomalies. The phenotype in some instances approaches that of apparently non- syndromic mental retardation. {ECO:0000269	PubMed:17273969}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cornelia De Lange Sendromu 4, 614701, Otozomal Dominant; CDLS4 (Cornelia De Lange Sendromu) (RAD21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornelia de Lange syndrome 4, 614701, Autosomal dominant; CDLS4 (Cornelia de Lange syndrome) (RAD21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD21	RAD21, SCC1, NXP1, KIAA0078, CDLS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cornelia de Lange syndrome 4 (CDLS4) [MIM:614701]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. It is characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269	PubMed:22633399}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cornelia De Lange Sendromu 4, 614701, Otozomal Dominant; CDLS4 (Cornelia De Lange Sendromu) (Prenatal) (RAD21 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cornelia de Lange syndrome 4, 614701, Autosomal dominant; CDLS4 (Cornelia de Lange syndrome) (RAD21 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAD21	RAD21, SCC1, NXP1, KIAA0078, CDLS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 4 (CDLS4) [MIM:614701]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. It is characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269	PubMed:22633399}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cornelia De Lange Sendromu 5, 300882, X'e Bağlı Dominant; CDLS5 (Cornelia De Lange Sendromu) (HDAC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cornelia de Lange syndrome 5, 300882, X-linked dominant; CDLS5 (Cornelia de Lange syndrome) (HDAC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HDAC8	HDAC8, MRXS6, CDLS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cornelia de Lange syndrome 5 (CDLS5) [MIM:300882]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. It is characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269	PubMed:22885700}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Cornelia De Lange Sendromu 5, 300882, X'e Bağlı Dominant; CDLS5 (Cornelia De Lange Sendromu) (Prenatal) (HDAC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cornelia de Lange syndrome 5, 300882, X-linked dominant; CDLS5 (Cornelia de Lange syndrome) (HDAC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HDAC8	HDAC8, MRXS6, CDLS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cornelia de Lange syndrome 5 (CDLS5) [MIM:300882]: A form of Cornelia de Lange syndrome, a clinically heterogeneous developmental disorder associated with malformations affecting multiple systems. It is characterized by facial dysmorphisms, abnormal hands and feet, growth delay, cognitive retardation, hirsutism, gastroesophageal dysfunction and cardiac, ophthalmologic and genitourinary anomalies. {ECO:0000269	PubMed:22885700}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ailesel Hiperkolesterolemiye Bağlı Koroner Arter Hastalığı, Korunma, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (ABCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease in familial hypercholesterolemia, protection against, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (ABCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA1	ABCA1, ABC1, HDLDT1, TGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Otozomal Dominant, 1, 608320, Otozomal Dominant; ADCAD1 (MEF2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, autosomal dominant, 1, 608320, Autosomal dominant; ADCAD1 (MEF2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEF2A	MEF2A, ADCAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coronary artery disease, autosomal dominant, 1 (ADCAD1) [MIM:608320]: A common heart disease characterized by reduced or absent blood flow in one or more of the arteries that encircle and supply the heart. Its most important complication is acute myocardial infarction. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koroner Arter Hastalığı, Otozomal Dominant, 2, 610947, Otozomal Dominant (LRP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, autosomal dominant, 2, 610947, Autosomal dominant (LRP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP6	LRP6, ADCAD2, STHAG7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coronary artery disease, autosomal dominant, 2 (ADCAD2) [MIM:610947]: A common heart disease characterized by reduced or absent blood flow in one or more of the arteries that encircle and supply the heart. Its most important complication is acute myocardial infarction. {ECO:0000269	PubMed:17332414}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koroner Arter Hastalığı, Modifiye Edici (CCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, modifier of (CCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL2	CCL2, SCYA2, MCP1, MCAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Direnç, 607339 (CX3CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, resistance to, 607339 (CX3CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CX3CR1	CX3CR1, GPR13, V28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Şiddetli Yatkınlık, 617347 (Hiperlipoproteinemi Tip 3) (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, severe, susceptibility to, 617347 (Hyperlipoproteinemia type 3) (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Yatkınlık (KL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, susceptibility to (KL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KL	KL, KLOTHO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Yatkınlık (PON2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, susceptibility to (PON2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PON2	PON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Yatkınlık (PON1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, susceptibility to (PON1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PON1	PON1, PON, ESA, MVCD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Hastalığı, Yatkınlık (IRS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery disease, susceptibility to (IRS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRS1	IRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Arter Spazmı 1, Yatkınlık (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery spasm 1, susceptibility to (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Variation in NOS3 seem to be associated with susceptibility to coronary spasm. {ECO:0000269|PubMed:11740345, ECO:0000269|PubMed:9737795.;	EDTA Blood Sample
Koroner Arter Spazmı 2, Yatkınlık (PON1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary artery spasm 2, susceptibility to (PON1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PON1	PON1, PON, ESA, MVCD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroner Kalp Hastalığı, Yatkınlık, 5, 608901 (KALRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary heart disease, susceptibility to, 5, 608901 (KALRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KALRN	KALRN, HAPIP, DUO, CHDS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coronary heart disease 5 (CHDS5) [MIM:608901]: A multifactorial disease characterized by an imbalance between myocardial functional requirements and the capacity of the coronary vessels to supply sufficient blood flow. Decreased capacity of the coronary vessels is often associated with thickening and loss of elasticity of the coronary arteries. {ECO:0000269	PubMed:17357071}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Koroner Kalp Hastalığı, Yatkınlık, 6, 614466 (MMP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary heart disease, susceptibility to, 6, 614466 (MMP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP3	MMP3, STMY1, CHDS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coronary heart disease 6 (CHDS6) [MIM:614466]: A multifactorial disease characterized by an imbalance between myocardial functional requirements and the capacity of the coronary vessels to supply sufficient blood flow. Decreased capacity of the coronary vessels is often associated with thickening and loss of elasticity of the coronary arteries. {ECO:0000269|PubMed:12477941, ECO:0000269|PubMed:8662692}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A polymorphism in the MMP3 promoter region is associated with the risk of coronary heart disease and myocardial infarction, due to lower MMP3 proteolytic activity and higher extracellular matrix deposition in atherosclerotic lesions.;	EDTA Blood Sample
Koroner Kalp Hastalığı, Yatkınlık, 7, 610938 (CD36 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coronary heart disease, susceptibility to, 7, 610938 (CD36 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD36	CD36, CHDS7, BDPLT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Korpus Kallozum AGENEZİSİ-Anormal Gentilalya (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Gentialya Sendromu (Proud Sendromu)) (ARX gen) (Tüm Gen Dizi Analizi) (Postnatal)	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENTALIA (Microcephaly-corpus callosum agenesis-abnormal gentialia syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Korpus Kallozum AGENEZİSİ-Anormal Gentilalya (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Gentialya Sendromu (Proud Sendromu)) (MLPA)	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENITALIA (Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Korpus Kallozum AGENEZİSİ-Anormal Gentilalya (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Gentialya Sendromu (Proud Sendromu)) (Prenatal) (ARX gen) (Tüm Gen Dizi Analizi) (Prenatal)	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENTALIA (Microcephaly-corpus callosum agenesis-abnormal gentialia syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korpus Kallozum AGENEZİSİ-Anormal Gentilalya (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Gentialya Sendromu (Proud Sendromu)) (Prenatal) (MLPA)	CORPUS CALLOSUM, AGENESIS OF, WITH ABNORMAL GENTALIA (Microcephaly-corpus callosum agenesis-abnormal gentialia syndrome) (Prenatal) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korpus Kallosumun Agenezisi, Yüz Anomalileri ve Serebellar Ataksi, 616819, Otozomal Resesif (Ağır Mental Retardasyon-Korpus Kallozum Agenezisi-Fasiyal Dismorfizm-Serebellar Ataksi Sendromu) (FRMD4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819, Autosomal recessive (Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome) (FRMD4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRMD4A	FRMD4A, KIAA1294, CCAFCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Korpus Kallosumun Agenezisi, Yüz Anomalileri ve Serebellar Ataksi, 616819, Otozomal Resesif (Ağır Mental Retardasyon-Korpus Kallozum Agenezisi-Fasiyal Dismorfizm-Serebellar Ataksi Sendromu) (Prenatal) (FRMD4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Corpus callosum, agenesis of, with facial anomalies and cerebellar ataxia, 616819, Autosomal recessive (Severe intellectual disability-corpus callosum agenesis-facial dysmorphism-cerebellar ataxia syndrome) (FRMD4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FRMD4A	FRMD4A, KIAA1294, CCAFCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korpus Kallosum, Agenezis,Mental Retardasyon, Oküler Kolobom ve Mikrognati İle Birlikte, 300472, X'e Bağlı Resesif (Korpus Kallozum Agenezisi-Entelektüel Yetersizlik-Kolobom-Mikrognati Sendromu) (IGBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472, X-linked recessive (Agenesis of the corpus callosum-intellectual disability-coloboma-micrognathia syndrome) (IGBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGBP1	IGBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 28 (MRXS28) [MIM:300472]: A mental retardation syndrome characterized by agenesis of the corpus callosum, coloboma of the iris and optic nerve, severe retrognathia, and intellectual deficit. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:14556245}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Korpus Kallosum, Agenezis,Mental Retardasyon, Oküler Kolobom ve Mikrognati İle Birlikte, 300472, X'e Bağlı Resesif (Korpus Kallozum Agenezisi-Entelektüel Yetersizlik-Kolobom-Mikrognati Sendromu) (Prenatal) (IGBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472, X-linked recessive (Agenesis of the corpus callosum-intellectual disability-coloboma-micrognathia syndrome) (IGBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGBP1	IGBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 28 (MRXS28) [MIM:300472]: A mental retardation syndrome characterized by agenesis of the corpus callosum, coloboma of the iris and optic nerve, severe retrognathia, and intellectual deficit. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:14556245}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korpus Kallozum, Parsiyel Agenezisi, 304100, X'e Bağlı Resesif (X'e Bağlı Komplike Korpus Kallozum Disgenezisi) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corpus callosum, partial agenesis of, 304100, X-linked recessive (X-linked complicated corpus callosum dysgenesis) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Korpus Kallozum, Parsiyel Agenezisi, 304100, X'e Bağlı Resesif (X'e Bağlı Komplike Korpus Kallozum Disgenezisi) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Corpus callosum, partial agenesis of, 304100, X-linked recessive (X-linked complicated corpus callosum dysgenesis) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi-Fokal Epilepsi Sendromu, 610042; COFES (Kortikal Displazi-Fokal Epilepsi Sendromu) (CNTNAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia-focal epilepsy syndrome, 610042; COFES (Cortical dysplasia-focal epilepsy syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kortikal Displazi-Fokal Epilepsi Sendromu, 610042; COFES (Kortikal Displazi-Fokal Epilepsi Sendromu) (Prenatal) (CNTNAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia-focal epilepsy syndrome, 610042; COFES (Cortical dysplasia-focal epilepsy syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 1, 614039, Otozomal Dominant; CDCBM1 (Kortikal Disgenezi-Pontoserebellar Hipoplazi, TUBB3 Mutasyona Bağlı) (TUBB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 1, 614039, Autosomal dominant; CDCBM1 (Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation) (TUBB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB3	TUBB3, TUBB4, CFEOM3A, CDCBM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 1, 614039, Otozomal Dominant; CDCBM1 (Kortikal Disgenezi-Pontoserebellar Hipoplazi, TUBB3 Mutasyona Bağlı) (Prenatal) (TUBB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 1, 614039, Autosomal dominant; CDCBM1 (Cortical dysgenesis with pontocerebellar hypoplasia due to TUBB3 mutation) (TUBB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB3	TUBB3, TUBB4, CFEOM3A, CDCBM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 2, 615282, Otozomal Dominant; CDCBM2 (KIF5C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 2, 615282, Autosomal dominant; CDCBM2 (KIF5C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF5C	KIF5C, NKHC2, CDCBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 2 (CDCBM2) [MIM:615282]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include intrauterine growth retardation, fetal akinesia, seizures, microcephaly, lack of psychomotor development, and arthrogryposis. Brain imaging shows malformations of cortical development, including polymicrogyria, gyral simplification, and thin corpus callosum. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 2, 615282, Otozomal Dominant; CDCBM2 (Prenatal) (KIF5C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 2, 615282, Autosomal dominant; CDCBM2 (KIF5C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF5C	KIF5C, NKHC2, CDCBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 2 (CDCBM2) [MIM:615282]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include intrauterine growth retardation, fetal akinesia, seizures, microcephaly, lack of psychomotor development, and arthrogryposis. Brain imaging shows malformations of cortical development, including polymicrogyria, gyral simplification, and thin corpus callosum. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 3, 615411, Otozomal Dominant; CDCBM3 (KIF2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 3, 615411, Autosomal dominant; CDCBM3 (KIF2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF2A	KIF2A, CDCBM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 3 (CDCBM3) [MIM:615411]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include early-onset epilepsy, and various malformations of cortical development such as agyria, posterior or frontal pachygyria, subcortical band heterotopia, and thin corpus callosum. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 3, 615411, Otozomal Dominant; CDCBM3 (Prenatal) (KIF2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 3, 615411, Autosomal dominant; CDCBM3 (KIF2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF2A	KIF2A, CDCBM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 3 (CDCBM3) [MIM:615411]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include early-onset epilepsy, and various malformations of cortical development such as agyria, posterior or frontal pachygyria, subcortical band heterotopia, and thin corpus callosum. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 6, 615771, Otozomal Dominant; CDCBM4 (TUBG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 4, 615412, Autosomal dominant; CDCBM4 (TUBG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBG1	TUBG1, CDCBM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 4 (CDCBM4) [MIM:615412]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include early-onset seizures, microcephaly, spastic tetraplegia, and various malformations of cortical development, such as agyria, posterior or frontal pachygyria, thick cortex, and subcortical band heterotopia and thin corpus callosum in some patients. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 6, 615771, Otozomal Dominant, CDCBM4 (Prenatal) (TUBG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 4, 615412, Autosomal dominant; CDCBM4 (TUBG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBG1	TUBG1, CDCBM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 4 (CDCBM4) [MIM:615412]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include early-onset seizures, microcephaly, spastic tetraplegia, and various malformations of cortical development, such as agyria, posterior or frontal pachygyria, thick cortex, and subcortical band heterotopia and thin corpus callosum in some patients. {ECO:0000269	PubMed:23603762}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 5, 615763, Otozomal Dominant; CDCBM5 (TUBB2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 5, 615763, Autosomal dominant; CDCBM5 (TUBB2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB2A	TUBB2A, CDCBM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 5 (CDCBM5) [MIM:615763]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include seizures, global developmental delay, and various brain malformations such as a diffuse simplified gyral pattern with reduced volume of white matter, globular basal ganglia, thin and dysmorphic corpus callosum, mild brainstem hypoplasia with a flat pons, mild cerebellar vermis hypoplasia, and mildly enlarged posterior fossa. {ECO:0000269	PubMed:24702957}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 5, 615763, Otozomal Dominant; CDCBM5 (TUBB2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 5, 615763, Autosomal dominant; CDCBM5 (TUBB2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB2A	TUBB2A, CDCBM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 5 (CDCBM5) [MIM:615763]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Clinical features include seizures, global developmental delay, and various brain malformations such as a diffuse simplified gyral pattern with reduced volume of white matter, globular basal ganglia, thin and dysmorphic corpus callosum, mild brainstem hypoplasia with a flat pons, mild cerebellar vermis hypoplasia, and mildly enlarged posterior fossa. {ECO:0000269	PubMed:24702957}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 6, 615771, Otozomal Dominant; CDCBM6 (TUBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical dysplasia, complex, with other brain malformations 6, 615771, Autosomal dominant; CDCBM6 (TUBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB	TUBB, TUBB5, M40, CDCBM6, CSCSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 6 (CDCBM6) [MIM:615771]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Affected individuals have microcephaly, ataxia, and severe delayed psychomotor development. Brain imaging shows variable malformations of cortical development, including white matter streaks, dysmorphic basal ganglia, corpus callosum abnormalities, brainstem and cerebellar hypoplasia, cortical dysplasia, polymicrogyria. {ECO:0000269	PubMed:23246003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikal Displazi, Kompleks, Diğer Beyin Malformasyonları 6, 615771, Otozomal Dominant; CDCBM6 (Prenatal) (TUBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical dysplasia, complex, with other brain malformations 6, 615771, Autosomal dominant; CDCBM6 (TUBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB	TUBB, TUBB5, M40, CDCBM6, CSCSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 6 (CDCBM6) [MIM:615771]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Affected individuals have microcephaly, ataxia, and severe delayed psychomotor development. Brain imaging shows variable malformations of cortical development, including white matter streaks, dysmorphic basal ganglia, corpus callosum abnormalities, brainstem and cerebellar hypoplasia, cortical dysplasia, polymicrogyria. {ECO:0000269	PubMed:23246003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikal Malformasyonlar, Oksipital, 614115, Otozomal Resesif; OCCM (Oksipital Pakigiri ve Polimikrogiri) (LAMC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortical malformations, occipital, 614115, Autosomal recessive; OCCM (Occipital pachygyria and polymicrogyria) (LAMC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMC3	LAMC3, OCCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical malformations occipital (OCCM) [MIM:614115]: A disease in which affected individuals develop seizures, sometimes associated with transient visual changes. Brain MRI shows both pachygyria and polymicrogyria restricted to the lateral occipital lobes. {ECO:0000269	PubMed:21572413}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikal Malformasyonlar, Oksipital, 614115, Otozomal Resesif; OCCM (Oksipital Pakigiri ve Polimikrogiri) (Prenatal) (LAMC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cortical malformations, occipital, 614115, Autosomal recessive; OCCM (Occipital pachygyria and polymicrogyria) (LAMC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMC3	LAMC3, OCCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical malformations occipital (OCCM) [MIM:614115]: A disease in which affected individuals develop seizures, sometimes associated with transient visual changes. Brain MRI shows both pachygyria and polymicrogyria restricted to the lateral occipital lobes. {ECO:0000269	PubMed:21572413}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kortikosteroid Bağlayıcı Globulin Eksikliği, 611489, Otozomal Resesif, Otozomal Dominant (Kortikosteroid Bağlayıcı Globulin Eksikliği) (SERPINA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Corticosteroid-binding globulin deficiency, 611489, Autosomal recessive, Autosomal dominant (Corticosteroid-binding globulin deficiency) (SERPINA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA6	CBG, SERPINA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Corticosteroid-binding globulin deficiency (CBG deficiency) [MIM:611489]: Extremely rare hereditary disorder characterized by reduced corticosteroid-binding capacity with normal or low plasma corticosteroid-binding globulin concentration, and normal or low basal cortisol levels associated with hypo/hypertension and muscle fatigue. {ECO:0000269|PubMed:10634411, ECO:0000269|PubMed:1504007, ECO:0000269|PubMed:17245537, ECO:0000269|PubMed:8212073}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kortikosteron Metiloksidaz Tip 1 Eksikliği (Aldosteronizm) (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CORTICOSTERONE METHYLOXIDASE TYPE I DEFICIENCY (Familial hypoaldosteronism) (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.	EDTA Blood Sample
Kortikosteron Metiloksidaz Tip 2 Eksikliği (Aldosteronizm) (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CORTICOSTERONE METHYLOXIDASE TYPE II DEFICIENCY (Familial hypoaldosteronism) (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.	EDTA Blood Sample
Kortizon Redüktaz Eksikliği 1, 604931, Otozomal Resesif; CORTRD1 (Hiperandrojenizm, Kortizon Redüktaz Eksikliğine Bağlı) (H6PD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortisone reductase deficiency 1, 604931, Autosomal recessive; CORTRD1 (Hyperandrogenism due to cortisone reductase deficiency) (H6PD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	H6PD	H6PD, GDH, G6PDH, CORTRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kortizon Redüktaz Eksikliği 2, 614662, Otozomal Dominant; CORTRD2 (Hiperandrojenizm, Kortizon Redüktaz Eksikliğine Bağlı) (Hiperandrojenizm, Kortizon Redüktaz Eksikliğine Bağlı) (HSD11B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cortisone reductase deficiency 2, 614662, Autosomal dominant; CORTRD2 (Hyperandrogenism due to cortisone reductase deficiency) (Hyperandrogenism due to cortisone reductase deficiency) (HSD11B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD11B1	HSD11B1, HSD11, HSD11L, CORTRD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortisone reductase deficiency (CRD) [MIM:604931]: In CRD, activation of cortisone to cortisol does not occur, resulting in adrenocorticotropin-mediated androgen excess and a phenotype resembling polycystic ovary syndrome (PCOS). {ECO:0000269	PubMed:11858176}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Costello Sendromu, 218040, Otozomal Dominant, Sporadik Vakalar; CSTLO (Costello Sendromu) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Costello syndrome, 218040, Autosomal dominant, Isolated cases; CSTLO (Costello syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Costello Sendromu, 218040, Otozomal Dominant, Sporadik Vakalar; CSTLO (Costello Sendromu) (MLPA)	Costello syndrome, 218040, Autosomal dominant, Isolated cases; CSTLO (Costello syndrome) (MLPA)	HRAS	HRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Costello Sendromu, 218040, Otozomal Dominant, Sporadik Vakalar; CSTLO (Costello Sendromu) (HRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Costello syndrome, 218040, Autosomal dominant, Isolated cases; CSTLO (Costello syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Costello Sendromu, 218040, Otozomal Dominant, Sporadik Vakalar; CSTLO (Costello Sendromu) (Prenatal) (MLPA)	Costello syndrome, 218040, Autosomal dominant, Isolated cases; CSTLO (Costello syndrome) (Prenatal) (MLPA)	HRAS	HRAS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Coumarin Direnci, 122700, Otozomal Dominant (CYP2A6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Coumarin resistance, 122700, Autosomal dominant (CYP2A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Coumarin Direnci, 122700, Otozomal Dominant (MLPA)	Coumarin resistance, 122700, Autosomal dominant (MLPA)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Cousin Sendromu, 260660, Otozomal Resesif (Pelviskapular Displazi) (TBX15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cousin syndrome, 260660, Autosomal recessive (Pelviscapular dysplasia) (TBX15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX15	TBX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cousin syndrome (COUSS) [MIM:260660]: Defined as pelviscapular dysplasia with epiphyseal abnormalities, congenital dwarfism and facial dysmorphy (frontal bossing, hypertelorism, narrow palpebral fissures, deep set globes, strabismus, low-set posteriory rotated and unusually formed external ears, dysplasia of conchae, small chin, short neck with redundant skin folds, and a low hairline). Intelligence may vary from normal to moderately impaired. Radiographic features comprise aplasia of the body of the scapula, hypoplasia of the iliac bone, humeroradial synostosis, dislocation of the femoral heads, and moderate brachydactyly. {ECO:0000269	PubMed:19068278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Cousin Sendromu, 260660, Otozomal Resesif (Pelviskapular Displazi) (Prenatal) (TBX15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cousin syndrome, 260660, Autosomal recessive (Pelviscapular dysplasia) (TBX15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX15	TBX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cousin syndrome (COUSS) [MIM:260660]: Defined as pelviscapular dysplasia with epiphyseal abnormalities, congenital dwarfism and facial dysmorphy (frontal bossing, hypertelorism, narrow palpebral fissures, deep set globes, strabismus, low-set posteriory rotated and unusually formed external ears, dysplasia of conchae, small chin, short neck with redundant skin folds, and a low hairline). Intelligence may vary from normal to moderately impaired. Radiographic features comprise aplasia of the body of the scapula, hypoplasia of the iliac bone, humeroradial synostosis, dislocation of the femoral heads, and moderate brachydactyly. {ECO:0000269	PubMed:19068278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cowchock Sendromu, 310490, X'e Bağlı resesif; COWCK (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 4) (AIFM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cowchock syndrome, 310490, X-linked recessive; COWCK (X-linked Charcot-Marie-Tooth disease type 4) (AIFM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIFM1	AIFM1, PDCD8, AIF, COXPD6, COWCK, CMTX4, DFNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cowchock Sendromu, 310490, X'e Bağlı resesif; COWCK (X'e Bağlı Charcot-Marie-Tooth Hastalığı Tip 4) (Prenatal) (AIFM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cowchock syndrome, 310490, X-linked recessive; COWCK (X-linked Charcot-Marie-Tooth disease type 4) (AIFM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AIFM1	AIFM1, PDCD8, AIF, COXPD6, COWCK, CMTX4, DFNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cowden Sendromu 1, 158350, Otozomal Dominant; CWS1 (Cowden Sendromu) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 1, 158350, Autosomal dominant; CWS1 (Cowden syndrome) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Cowden Sendromu 1, 158350, Otozomal Dominant; CWS1 (Cowden Sendromu) (MLPA)	Cowden syndrome 1, 158350, Autosomal dominant; CWS1 (Cowden syndrome) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Cowden Sendromu 2, 612359, Otozomal Dominant; CWS2 (Cowden Sendromu) (SDHB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 2, 612359, Autosomal dominant; CWS2 (Cowden syndrome) (SDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHB	SDHB, SDH2, SDHP, PGL4, CWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cowden Sendromu 3, 615106; CWS3 (Cowden Sendromu) (SDHD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 3, 615106; CWS3 (Cowden syndrome) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cowden Sendromu 4, 615107; CWS4 (Cowden Sendromu) (KLLN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 4, 615107; CWS4 (Cowden syndrome) (KLLN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLLN	KLLN, CWS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cowden syndrome 4 (CWS4) [MIM:615107]: A form of Cowden syndrome, a hamartomatous polyposis syndrome with age-related penetrance. Cowden syndrome is characterized by hamartomatous lesions affecting derivatives of ectodermal, mesodermal and endodermal layers, macrocephaly, facial trichilemmomas (benign tumors of the hair follicle infundibulum), acral keratoses, papillomatous papules, and elevated risk for development of several types of malignancy, particularly breast carcinoma in women and thyroid carcinoma in both men and women. Colon cancer and renal cell carcinoma have also been reported. Hamartomas can be found in virtually every organ, but most commonly in the skin, gastrointestinal tract, breast and thyroid. {ECO:0000269|PubMed:21177507}. Note=The gene represented in this entry is involved in disease pathogenesis. Germline KLLN methylation is common among patients with Cowden syndrome or Cowden-like syndrome and is associated with increased risks of breast and renal cancer over PTEN mutation-positive individuals {PubMed:21177507}. {ECO:0000269|PubMed:21177507};.	EDTA Blood Sample
Cowden Sendromu 5, 615108; CWS5 (Cowden Sendromu) (PIK3CA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 5, 615108; CWS5 (Cowden syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cowden Sendromu 6, 615109; CWS6 (Cowden Sendromu) (AKT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 6, 615109; CWS6 (Cowden syndrome) (AKT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Cowden Sendromu 7, 616858, Otozomal Dominant; CWS7 (Cowden Sendromu) (SEC23B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Cowden syndrome 7, 616858, Autosomal dominant; CWS7 (Cowden syndrome) (SEC23B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC23B	SEC23B, CDAN2, HEMPAS, CWS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPT Eksikliği, Hepatik, Tip 1A, 255120, Otozomal Resesif (Karnitin Palmitoil Transferaz 1A Eksikliği) (CPT1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	CPT deficiency, hepatic, type IA, 255120, Autosomal recessive (Carnitine palmitoyl transferase 1A deficiency) (CPT1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT1A	CPT1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carnitine palmitoyltransferase 1A deficiency (CPT1AD) [MIM:255120]: Rare autosomal recessive metabolic disorder of long- chain fatty acid oxidation characterized by severe episodes of hypoketotic hypoglycemia usually occurring after fasting or illness. Onset is in infancy or early childhood. {ECO:0000269|PubMed:11350182, ECO:0000269|PubMed:11441142, ECO:0000269|PubMed:12189492, ECO:0000269|PubMed:14517221, ECO:0000269|PubMed:15110323, ECO:0000269|PubMed:15669684, ECO:0000269|PubMed:9691089}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
CPT Eksikliği, Hepatik, Tip 1A, 255120, Otozomal Resesif (Karnitin Palmitoil Transferaz 1A Eksikliği) (Prenatal) (CPT1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	CPT deficiency, hepatic, type IA, 255120, Autosomal recessive (Carnitine palmitoyl transferase 1A deficiency) (CPT1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT1A	CPT1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carnitine palmitoyltransferase 1A deficiency (CPT1AD) [MIM:255120]: Rare autosomal recessive metabolic disorder of long- chain fatty acid oxidation characterized by severe episodes of hypoketotic hypoglycemia usually occurring after fasting or illness. Onset is in infancy or early childhood. {ECO:0000269|PubMed:11350182, ECO:0000269|PubMed:11441142, ECO:0000269|PubMed:12189492, ECO:0000269|PubMed:14517221, ECO:0000269|PubMed:15110323, ECO:0000269|PubMed:15669684, ECO:0000269|PubMed:9691089}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CPT 2 Eksikliği, İnfantil, 600649, Otozomal Resesif (Karnitin Palmitoil Transferaz 2 Eksikliği) (CPT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CPT II deficiency, infantile, 600649, Autosomal recessive (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
CPT 2 Eksikliği, İnfantil, 600649, Otozomal Resesif (Karnitin Palmitoil Transferaz 2 Eksikliği) (Prenatal) (CPT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CPT II deficiency, infantile, 600649, Autosomal recessive (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CPT 2 Eksikliği, Letal Neonatal, 608836, Otozomal Resesif (Karnitin Palmitoil Transferaz 2 Eksikliği) (CPT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CPT II deficiency, lethal neonatal, 608836, Autosomal recessive (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
CPT 2 Eksikliği, Letal Neonatal, 608836, Otozomal Resesif (Karnitin Palmitoil Transferaz 2 Eksikliği) (Prenatal) (CPT2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	CPT II deficiency, lethal neonatal, 608836, Autosomal recessive (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CPT 2 Eksikliği, Miyopatik, Stresle İndüklenen, 255110, Otozomal Resesif, Otozomal Dominant (Karnitin Palmitoil Transferaz 2 Eksikliği) (CPT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CPT II deficiency, myopathic, stress-induced, 255110, Autosomal recessive, Autosomal dominant (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
CPT 2 Eksikliği, Miyopatik, Stresle İndüklenen, 255110, Otozomal Resesif, Otozomal Dominant (Karnitin Palmitoil Transferaz 2 Eksikliği) (Prenatal) (CPT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CPT II deficiency, myopathic, stress-induced, 255110, Autosomal recessive, Autosomal dominant (Carnitine palmitoyltransferase II deficiency) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carnitine palmitoyltransferase 2 deficiency late-onset (CPT2D) [MIM:255110]: Autosomal recessive disorder characterized by recurrent myoglobinuria, episodes of muscle pain, stiffness, and rhabdomyolysis. These symptoms are triggered by prolonged exercise, fasting or viral infection and patients are usually young adults. In addition to this classical, late-onset, muscular type, a hepatic or hepatocardiomuscular form has been reported in infants. Clinical pictures in these children or neonates include hypoketotic hypoglycemia, liver dysfunction, cardiomyopathy and sudden death. {ECO:0000269|PubMed:10090476, ECO:0000269|PubMed:11477613, ECO:0000269|PubMed:14605500, ECO:0000269|PubMed:14615409, ECO:0000269|PubMed:1528846, ECO:0000269|PubMed:15489334, ECO:0000269|PubMed:15622536, ECO:0000269|PubMed:7711730, ECO:0000269|PubMed:8358442, ECO:0000269|PubMed:8651281, ECO:0000269|PubMed:9600456, ECO:0000269|PubMed:9758712, ECO:0000269|Ref.13, ECO:0000269|Ref.17}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CR1 Eksikliği (CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CR1 deficiency (CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR1	CR1, C3BR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CR1 Eksikliği (Prenatal) (CR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	CR1 deficiency (CR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CR1	CR1, C3BR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniodiafizyal Displazi, Otozomal Dominant, 122860, Otozomal Dominant; CDD (Kraniyodiafizyal Displazi) (SOST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniodiaphyseal dysplasia, autosomal dominant, 122860, Autosomal dominant; CDD (Craniodiaphyseal dysplasia) (SOST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of craniofacial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269|PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	EDTA Blood Sample
Kraniodiafizyal Displazi, Otozomal Dominant, 122860, Otozomal Dominant; CDD (Kraniyodiafizyal Displazi) (Prenatal) (SOST geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniodiaphyseal dysplasia, autosomal dominant, 122860, Autosomal dominant; CDD (Craniodiaphyseal dysplasia) (SOST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of craniofacial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269|PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kranioektodermal Displazi 1, 218330, Otozomal Resesif; CED1 (Kranioektodermal Displazi 4) (IFT122 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cranioectodermal dysplasia 1, 218330, Autosomal recessive; CED1 (Cranioectodermal dysplasia) (IFT122 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT122	IFT122, WDR10, CED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kranioektodermal Displazi 1, 218330, Otozomal Resesif; CED1 (Kranioektodermal Displazi 4) (Prenatal) (IFT122 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cranioectodermal dysplasia 1, 218330, Autosomal recessive; CED1 (Cranioectodermal dysplasia) (IFT122 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT122	IFT122, WDR10, CED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kranioektodermal Displazi 2, 613610, Otozomal Resesif; CED2 (Kraniyoektodermal Displazi 4) (WDR35 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cranioectodermal dysplasia 2, 613610, Autosomal recessive; CED2 (Cranioectodermal dysplasia) (WDR35 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR35	WDR35, NAOFEN, KIAA1336, CED2, SRTD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cranioectodermal dysplasia 2 (CED2) [MIM:613610]: A disorder characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include short stature, dolichocephaly, craniosynostosis, narrow thorax with pectus excavatum, short limbs, brachydactyly, joint laxity, narrow palpebral fissures, telecanthus with hypertelorism, low-set simple ears, everted lower lip, and short neck. Teeth abnormalities include widely spaced, hypoplastic and fused teeth. [ECO:0000269	PubMed:20817137]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kranioektodermal Displazi 2, 613610, Otozomal Resesif; CED2 (Kraniyoektodermal Displazi 4) (Prenatal) (WDR35 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cranioectodermal dysplasia 2, 613610, Autosomal recessive; CED2 (Cranioectodermal dysplasia) (WDR35 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR35	WDR35, NAOFEN, KIAA1336, CED2, SRTD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cranioectodermal dysplasia 2 (CED2) [MIM:613610]: A disorder characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include short stature, dolichocephaly, craniosynostosis, narrow thorax with pectus excavatum, short limbs, brachydactyly, joint laxity, narrow palpebral fissures, telecanthus with hypertelorism, low-set simple ears, everted lower lip, and short neck. Teeth abnormalities include widely spaced, hypoplastic and fused teeth. [ECO:0000269	PubMed:20817137]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kranioektodermal Displazi 3, 614099, Otozomal Resesif; CED3 (Kraniyoektodermal Displazi 4) (IFT43 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cranioectodermal dysplasia 3, 614099, Autosomal recessive; CED3 (Cranioectodermal dysplasia) (IFT43 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT43	IFT43, C14orf179, CED3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cranioectodermal dysplasia 3 (CED3) [MIM:614099]: A disorder primarily characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include craniosynostosis, narrow rib cage, short limbs, brachydactyly, hypoplastic and widely spaced teeth, sparse hair, skin laxity and abnormal nails. Nephronophthisis leading to progressive renal failure, hepatic fibrosis, heart defects, and retinitis pigmentosa have also been described. (ECO:0000269	PubMed:21378380). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kranioektodermal Displazi 3, 614099, Otozomal Resesif; CED3 (Kraniyoektodermal Displazi 4) (Prenatal) (IFT43 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cranioectodermal dysplasia 3, 614099, Autosomal recessive; CED3 (Cranioectodermal dysplasia) (IFT43 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT43	IFT43, C14orf179, CED3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cranioectodermal dysplasia 3 (CED3) [MIM:614099]: A disorder primarily characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include craniosynostosis, narrow rib cage, short limbs, brachydactyly, hypoplastic and widely spaced teeth, sparse hair, skin laxity and abnormal nails. Nephronophthisis leading to progressive renal failure, hepatic fibrosis, heart defects, and retinitis pigmentosa have also been described. (ECO:0000269	PubMed:21378380). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kranioektodermal Displazi 4, 614378, Otozomal Resesif; CED4 (Kraniyoektodermal Displazi 4) (WDR19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cranioectodermal dysplasia 4, 614378, Autosomal recessive; CED4 (Cranioectodermal dysplasia) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kranioektodermal Displazi 4, 614378, Otozomal Resesif; CED4 (Kraniyoektodermal Displazi 4) (Prenatal) (WDR19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cranioectodermal dysplasia 4, 614378, Autosomal recessive; CED4 (Cranioectodermal dysplasia) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal Anomaliler ve Ön Segment Disgenezisi Sendromu, 614195; CAASDS (VSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195; CAASDS (VSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VSX1	VSX1, RINX, KTCN1, CAASDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniofasiyal Anomaliler ve Ön Segment Disgenezisi Sendromu, 614195; CAASDS (Prenatal) (VSX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniofacial anomalies and anterior segment dysgenesis syndrome, 614195; CAASDS (VSX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VSX1	VSX1, RINX, KTCN1, CAASDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal boşluklular (FGFRs, TWIST, MSX2, ALX4, RUNX2) (MLPA)	.	FGFRs, TWIST, MSX2, ALX4, RUNX2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Kraniofasial boşluklular (FGFRs, TWIST, MSX2, ALX4, RUNX2) (MLPA) (Prenatal)	Craniofacial disorders (FGFRs, TWIST, MSX2, ALX4, RUNX2) (MLPA) (Prenatal)	FGFRs, TWIST, MSX2, ALX4, RUNX2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal Dismorfizm İskelet Anomalileri ve Mental Retardasyon Sendromu, 213980, Otozomal Resesif; CFSMR (Serebra-Fasio-Torasik Displazi) (TMCO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980, Autosomal recessive; CFSMR (Cerebro-facio-thoracic dysplasia) (TMCO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMCO1	TMCO1, CFSMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniofasiyal Dismorfizm İskelet Anomalileri ve Mental Retardasyon Sendromu, 213980, Otozomal Resesif; CFSMR (Serebra-Fasio-Torasik Displazi) (Prenatal) (TMCO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980, Autosomal recessive; CFSMR (Cerebro-facio-thoracic dysplasia) (TMCO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMCO1	TMCO1, CFSMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal-Sağırlık-El Sendromu, 122880, Otozomal Dominant; CDHS (Kraniyofasiyal - Sağırlık-El Sendromu) (PAX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniofacial-deafness-hand syndrome, 122880, Autosomal dominant; CDHS (Craniofacial-deafness-hand syndrome) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. (ECO:0000269	PubMed:8664898). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kraniofasiyal-Sağırlık-El Sendromu, 122880, Otozomal Dominant; CDHS (Kraniyofasiyal - Sağırlık-El Sendromu) (MLPA)	Craniofacial-deafness-hand syndrome, 122880, Autosomal dominant; CDHS (Craniofacial-deafness-hand syndrome) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. (ECO:0000269	PubMed:8664898). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kraniofasiyal-Sağırlık-El Sendromu, 122880, Otozomal Dominant; CDHS (Kraniyofasiyal - Sağırlık-El Sendromu) (Prenatal) (PAX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniofacial-deafness-hand syndrome, 122880, Autosomal dominant; CDHS (Craniofacial-deafness-hand syndrome) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. (ECO:0000269	PubMed:8664898). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal-Sağırlık-El Sendromu, 122880, Otozomal Dominant; CDHS (Kraniyofasiyal - Sağırlık-El Sendromu) (Prenatal) (MLPA)	Craniofacial-deafness-hand syndrome, 122880, Autosomal dominant; CDHS (Craniofacial-deafness-hand syndrome) (Prenatal) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. (ECO:0000269	PubMed:8664898). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofasiyal-İskelet-Dermatolojik Displazi, 101600, Otozomal Dominant (Pfeiffer Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniofacial-skeletal-dermatologic dysplasia, 101600, Autosomal dominant (Pfeiffer syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Kraniofasiyal-İskelet-Dermatolojik Displazi, 101600, Otozomal Dominant (Pfeiffer Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniofacial-skeletal-dermatologic dysplasia, 101600, Autosomal dominant (Pfeiffer syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniofrontonazal Displazi, 304110, X'e Bağlı Dominant; CFNS (Kraniyofrontonazal Displazi) (EFNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniofrontonasal dysplasia, 304110, X-linked dominant; CFNS (Craniofrontonasal dysplasia) (EFNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFNB1	EFNB1, EPLG2, CFNS, CFND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniofrontonasal syndrome (CFNS) [MIM:304110]: X-linked inherited syndrome characterized by hypertelorism, coronal synostosis with brachycephaly, downslanting palpebral fissures, clefting of the nasal tip, joint anomalies, longitudinally grooved fingernails and other digital anomalies. (ECO:0000269|PubMed:15124102, ECO:0000269|PubMed:15166289, ECO:0000269|PubMed:15959873). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kraniofrontonazal Displazi, 304110, X'e Bağlı Dominant; CFNS (Kraniyofrontonazal Displazi) (Prenatal) (EFNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniofrontonasal dysplasia, 304110, X-linked dominant; CFNS (Craniofrontonasal dysplasia) (EFNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EFNB1	EFNB1, EPLG2, CFNS, CFND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniofrontonasal syndrome (CFNS) [MIM:304110]: X-linked inherited syndrome characterized by hypertelorism, coronal synostosis with brachycephaly, downslanting palpebral fissures, clefting of the nasal tip, joint anomalies, longitudinally grooved fingernails and other digital anomalies. (ECO:0000269|PubMed:15124102, ECO:0000269|PubMed:15166289, ECO:0000269|PubMed:15959873). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniolentikülosutural Displazi, 607812, Otozomal Resesif; CLSD (Kraniolentikülosutural Displazi) (SEC23A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniolenticulosutural dysplasia, 607812, Autosomal recessive; CLSD (Craniolenticulosutural dysplasia) (SEC23A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC23A	SEC23A, CLSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniolenticulosutural dysplasia (CLSD) [MIM:607812]: Autosomal recessive syndrome characterized by late-closing fontanels, sutural cataracts, facial dysmorphisms and skeletal defects. (ECO:0000269	PubMed:16980979). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kraniolentikülosutural Displazi, 607812, Otozomal Resesif; CLSD (Kraniolentikülosutural Displazi) (Prenatal) (SEC23A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniolenticulosutural dysplasia, 607812, Autosomal recessive; CLSD (Craniolenticulosutural dysplasia) (SEC23A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEC23A	SEC23A, CLSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniolenticulosutural dysplasia (CLSD) [MIM:607812]: Autosomal recessive syndrome characterized by late-closing fontanels, sutural cataracts, facial dysmorphisms and skeletal defects. (ECO:0000269	PubMed:16980979). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniometafizyal Displazi, 123000, Otozomal Dominant; CMDD (Kraniyometafizyal Displazi, Otozomal Resesif) (ANKH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniometaphyseal dysplasia, 123000, Autosomal dominant; CMDD (Craniometaphyseal dysplasia) (ANKH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKH	ANKH, HANK, ANK, CMDJ, CCAL2, CPPDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniometaphyseal dysplasia, autosomal dominant (CMDD) [MIM:123000]: An osteochondrodysplasia characterized by hyperostosis and sclerosis of the craniofacial bones associated with abnormal modeling of the metaphyses. Sclerosis of the skull may lead to asymmetry of the mandible, as well as to cranial nerve compression, that may finally result in hearing loss and facial palsy. (ECO:0000269|PubMed:11326272, ECO:0000269|PubMed:11326338). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kraniometafizyal Displazi, 123000, Otozomal Dominant; CMDD (Kraniyometafizyal Displazi, Otozomal Resesif) (Prenatal) (ANKH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniometaphyseal dysplasia, 123000, Autosomal dominant; CMDD (Craniometaphyseal dysplasia) (ANKH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANKH	ANKH, HANK, ANK, CMDJ, CCAL2, CPPDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniometaphyseal dysplasia, autosomal dominant (CMDD) [MIM:123000]: An osteochondrodysplasia characterized by hyperostosis and sclerosis of the craniofacial bones associated with abnormal modeling of the metaphyses. Sclerosis of the skull may lead to asymmetry of the mandible, as well as to cranial nerve compression, that may finally result in hearing loss and facial palsy. (ECO:0000269|PubMed:11326272, ECO:0000269|PubMed:11326338). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kraniometafiziyal Displazi, Otozomal Resesif, 218400, Otozomal Resesif, CMDR (Kraniyometafiziyal Displazi, Otozomal Resesif) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniometaphyseal dysplasia, autosomal recessive, 218400, Autosomal recessive; CMDR (Craniometaphyseal dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODCD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniometafiziyal Displazi, Otozomal Resesif, 218400, Otozomal Resesif; CMDR (Kraniyometafiziyal Displazi, Otozomal Resesif) (Prenatal) (GJA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniometaphyseal dysplasia, autosomal recessive, 218400, Autosomal recessive; CMDR (Craniometaphyseal dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODCD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kranioosteoartropati, 259100, Otozomal Resesif (Kraniyo-Osteoartropati) (HPGD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cranioosteoarthropathy, 259100, Autosomal recessive (Cranio-osteoarthropathy) (HPGD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPGD	HPGD, PGDH1, PHOAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kranioosteoartropati, 259100, Otozomal Resesif (Kraniyo-Osteoartropati) (Prenatal) (HPGD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cranioosteoarthropathy, 259100, Autosomal recessive (Cranio-osteoarthropathy) (HPGD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPGD	HPGD, PGDH1, PHOAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 1, 123100, Otozomal Dominant; CRS1 (İzole Oksisefali) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 1, 123100, Autosomal dominant; CRS1 (Isolated oxycephaly) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kraniosinostozis 1, 123100, Otozomal Dominant; CRS1 (İzole Oksisefali) (MLPA)	Craniosynostosis 1, 123100, Autosomal dominant; CRS1 (Isolated oxycephaly) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kraniosinostozis 1, 123100, Otozomal Dominant; CRS1 (İzole Oksisefali) (Prenatal) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 1, 123100, Autosomal dominant; CRS1 (Isolated oxycephaly) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 1, 123100, Otozomal Dominant; CRS1 (İzole Oksisefali) (Prenatal) (MLPA)	Craniosynostosis 1, 123100, Autosomal dominant; CRS1 (Isolated oxycephaly) (Prenatal) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 2, 604757, Otozomal Dominant; CRS2 (Kraniosynostozis, Boston Tipi, 2) (MSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 2, 604757, Autosomal dominant; CRS2 (Craniosynostosis, Boston type) (MSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX2	MSX2, CRS2, HOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 2, 604757, Otozomal Dominant; CRS2 (Kraniosynostozis, Boston Tipi, 2) (MLPA)	Craniosynostosis 2, 604757, Autosomal dominant; CRS2 (Craniosynostosis, Boston type) (MLPA)	MSX2	MSX2, CRS2, HOX8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 2, 604757, Otozomal Dominant; CRS2 (Kraniosynostozis, Boston Tipi, 2) (Prenatal) (MSX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 2, 604757, Autosomal dominant; CRS2 (Craniosynostosis, Boston type) (MSX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSX2	MSX2, CRS2, HOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 2, 604757, Otozomal Dominant; CRS2 (Kraniosynostozis, Boston Tipi, 2) (Prenatal) (MLPA)	Craniosynostosis 2, 604757, Autosomal dominant; CRS2 (Craniosynostosis, Boston type) (Prenatal) (MLPA)	MSX2	MSX2, CRS2, HOX8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 3, 615314, Otozomal Dominant; CRS3 (İzole Brakisefali) (TCF12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 3, 615314, Autosomal dominant; CRS3 (Isolated brachycephaly) (TCF12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCF12	TCF12, HTF4, CRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 3 (CRS3) [MIM:615314]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:23354436, ECO:0000269|PubMed:25271085}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kraniosinostozis 3, 615314, Otozomal Dominant; CRS3 (İzole Brakisefali) (Prenatal) (TCF12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 3, 615314, Autosomal dominant; CRS3 (Isolated brachycephaly) (TCF12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCF12	TCF12, HTF4, CRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 3 (CRS3) [MIM:615314]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:23354436, ECO:0000269|PubMed:25271085}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 4, 600775, Otozomal Dominant; CRS4 (İzole Yonca Yaprağı Skalp Sendromu) (ERF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 4, 600775, Autosomal dominant; CRS4 (Isolated cloverleaf skull syndrome) (ERF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERF	ERF, PE2, CRS4, CHYTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 4 (CRS4) [MIM:600775]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:23354439}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kraniosinostozis 4, 600775, Otozomal Dominant; CRS4 (İzole Yonca Yaprağı Skalp Sendromu) (Prenatal) (ERF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 4, 600775, Autosomal dominant; CRS4 (Isolated cloverleaf skull syndrome) (ERF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERF	ERF, PE2, CRS4, CHYTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 4 (CRS4) [MIM:600775]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:23354439}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 5, Yatkınlık, 615529, Otozomal Dominant; CRS5 (İzole Skafosefali) (ALX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 5, susceptibility to, 615529, Autosomal dominant; CRS5 (Isolated scaphocephaly) (ALX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 5, Yatkınlık, 615529, Otozomal Dominant; CRS5 (İzole Skafosefali) (MLPA)	Craniosynostosis 5, susceptibility to, 615529, Autosomal dominant; CRS5 (Isolated scaphocephaly) (MLPA)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 5, Yatkınlık, 615529, Otozomal Dominant; CRS5 (İzole Skafosefali) (Prenatal) (ALX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 5, susceptibility to, 615529, Autosomal dominant; CRS5 (Isolated scaphocephaly) (ALX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 5, Yatkınlık, 615529, Otozomal Dominant; CRS5 (İzole Skafosefali) (Prenatal) (MLPA)	Craniosynostosis 5, susceptibility to, 615529, Autosomal dominant; CRS5 (Isolated scaphocephaly) (Prenatal) (MLPA)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 6, 616602, Otozomal Dominant; CRS6 (İzole Oksisefali) (ZIC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis 6, 616602, Autosomal dominant; CRS6 (Isolated oxycephaly) (ZIC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZIC1	ZIC1, CRS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 6, 616602, Otozomal Dominant; CRS6 (İzole Oksisefali) (MLPA)	Craniosynostosis 6, 616602, Autosomal dominant; CRS6 (Isolated oxycephaly) (MLPA)	ZIC1	ZIC1, CRS6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kraniosinostozis 6, 616602, Otozomal Dominant; CRS6 (İzole Oksisefali) (Prenatal) (ZIC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis 6, 616602, Autosomal dominant; CRS6 (Isolated oxycephaly) (ZIC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZIC1	ZIC1, CRS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis 6, 616602, Otozomal Dominant; CRS6 (İzole Oksisefali) (Prenatal) (MLPA)	Craniosynostosis 6, 616602, Autosomal dominant; CRS6 (Isolated oxycephaly) (Prenatal) (MLPA)	ZIC1	ZIC1, CRS6	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis ve Dış Anomalleri, 614188, Otozomal Resesif; CRSDA (Kraniyosinostozis ve Dış Anomalleri) (IL11RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis and dental anomalies, 614188, Autosomal recessive; CRSDA (Craniosynostosis and dental anomalies) (IL11RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL11RA	IL11RA, CRSDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniosinostozis ve Dış Anomalleri, 614188, Otozomal Resesif; CRSDA (Kraniyosinostozis ve Dış Anomalleri) (Prenatal) (IL11RA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis and dental anomalies, 614188, Autosomal recessive; CRSDA (Craniosynostosis and dental anomalies) (IL11RA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL11RA	IL11RA, CRSDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis, Radiohumeral Füzyon, Diğer İskelet ve Kraniofasiyal Anomaliler, 614416 (Letal Oksipital Ensefalosel-İskelet Displazisi Sendromu) (CYP26B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (Lethal occipital encephalocele-skeletal dysplasia syndrome) (CYP26B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP26B1	CYP26B1, CYP26A2, P450RAI2, RHFCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Radiohumeral fusions with other skeletal and craniofacial anomalies (RHFCA) [MIM:614416]: A disease characterized by craniofacial malformations, occipital encephalocele, radiohumeral fusions, oligodactyly, advanced osseous maturation, and calvarial mineralization defects. {ECO:0000269	PubMed:22019272}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kraniosinostozis, Radiohumeral Füzyon, Diğer İskelet ve Kraniofasiyal Anomaliler, 614416 (Letal Oksipital Ensefalosel-İskelet Displazisi Sendromu) (Prenatal) (CYP26B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 (Lethal occipital encephalocele-skeletal dysplasia syndrome) (CYP26B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP26B1	CYP26B1, CYP26A2, P450RAI2, RHFCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Radiohumeral fusions with other skeletal and craniofacial anomalies (RHFCA) [MIM:614416]: A disease characterized by craniofacial malformations, occipital encephalocele, radiohumeral fusions, oligodactyly, advanced osseous maturation, and calvarial mineralization defects. {ECO:0000269	PubMed:22019272}, Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kraniosinostozis, Nonspesifik (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Craniosynostosis, nonspecific (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kraniosinostozis, Nonspesifik (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Craniosynostosis, nonspecific (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CRASH Sendromu, 303350, X'e Bağlı (MASA Sendromu) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	CRASH syndrome, 303350, X-linked recessive (MASA syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRASH Sendromu, 303350, X'e Bağlı (MASA Sendromu) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	CRASH syndrome, 303350, X-linked recessive (MASA syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kreatin Fosfokinaz, Artmış Serum Seviyesi, 123320, Otozomal Dominant (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Creatine phosphokinase, elevated serum, 123320, Autosomal dominant (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kreatin Fosfokinaz, Artmış Serum Seviyesi, 123320, Otozomal Dominant (Dalgalanan Kas Hastalığı (MLPA)	Creatine phosphokinase, elevated serum, 123320, Autosomal dominant (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Creutzfeldt-Jakob Hastalığı, 123400, Otozomal Dominant (Sporadik Creutzfeldt-Jakob Hastalığı) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Creutzfeldt-Jakob disease, 123400, Autosomal dominant (Sporadic Creutzfeldt-Jakob disease) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Creutzfeldt-Jakob Hastalığı, 123400, Otozomal Dominant (Sporadik Creutzfeldt-Jakob Hastalığı) (Prenatal) (PRNP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Creutzfeldt-Jakob disease, 123400, Autosomal dominant (Sporadic Creutzfeldt-Jakob disease) (PRNP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Creutzfeldt-Jakob Hastalığı, Varyant, Direnç, 123400, Otozomal Dominant; CJD (Sporadik Creutzfeldt-Jakob Hastalığı) (HLA-DQB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Creutzfeldt-Jakob disease, variant, resistance to, 123400, Autosomal dominant; CJD (Sporadic Creutzfeldt-Jakob disease) (HLA-DQB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DQB1	HLA-DQB1, CELIAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Creutzfeldt-Jakob Hastalığı, Varyant, Direnç, 123400, Otozomal Dominant; CJD (Sporadik Creutzfeldt-Jakob Hastalığı) (Prenatal) (HLA-DQB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Creutzfeldt-Jakob disease, variant, resistance to, 123400, Autosomal dominant; CJD (Sporadic Creutzfeldt-Jakob disease) (HLA-DQB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HLA-DQB1	HLA-DQB1, CELIAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Crigler-Najjar Sendromu, Tip 1, 218800, Otozomal Resesif (Crigler-Najjar Sendromu) (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Crigler-Najjar syndrome, type I, 218800, Autosomal recessive (Crigler-Najjar syndrome) (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Crigler-Najjar Sendromu, Tip 1, 218800, Otozomal Resesif (Crigler-Najjar Sendromu) (Prenatal) (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Crigler-Najjar syndrome, type I, 218800, Autosomal recessive (Crigler-Najjar syndrome) (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Crigler-Najjar Sendromu, Tip 2, 606785, Otozomal Resesif (Crigler-Najjar Sendromu) (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Crigler-Najjar syndrome, type II, 606785, Autosomal recessive (Crigler-Najjar syndrome) (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Crigler-Najjar Sendromu, Tip 2, 606785, Otozomal Resesif (Crigler-Najjar Sendromu) (Prenatal) (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Crigler-Najjar syndrome, type II, 606785, Autosomal recessive (Crigler-Najjar syndrome) (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Crohn Hastalığı ile İlişkili Büyüme Gerilği, 266600, Multifaktöriyel (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Crohn disease-associated growth failure, 266600, Multifactorial (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Crouzon Sendromu, Acanthosis Nigricans, 612247, Otozomal Dominant; CAN (Crouzon Sendromu-Akantozis Nigrikans Sendromu) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Crouzon syndrome with acanthosis nigricans, 612247, Autosomal dominant; CAN (Crouzon syndrome-acanthosis nigricans syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Crouzon Sendromu, Acanthosis Nigricans, 612247, Otozomal Dominant; CAN (Crouzon Sendromu-Akantozis Nigrikans Sendromu) (MLPA)	Crouzon syndrome with acanthosis nigricans, 612247, Autosomal dominant; CAN (Crouzon syndrome-acanthosis nigricans syndrome) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Crouzon Sendromu, Acanthosis Nigricans, 612247, Otozomal Dominant; CAN (Crouzon Sendromu-Akantozis Nigrikans Sendromu) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Crouzon syndrome with acanthosis nigricans, 612247, Autosomal dominant; CAN (Crouzon syndrome-acanthosis nigricans syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Crouzon Sendromu, Acanthosis Nigricans, 612247, Otozomal Dominant; CAN (Crouzon Sendromu-Akantozis Nigrikans Sendromu) (Prenatal) (MLPA)	Crouzon syndrome with acanthosis nigricans, 612247, Autosomal dominant; CAN (Crouzon syndrome-acanthosis nigricans syndrome) (Prenatal)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Crouzon Sendromu, 123500, Otozomal Dominant (Crouzon Hastalığı) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Crouzon syndrome, 123500, Autosomal dominant (Crouzon disease) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Crouzon Sendromu, 123500, Otozomal Dominant (Crouzon Hastalığı) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Crouzon syndrome, 123500, Autosomal dominant (Crouzon disease) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kriyohidrositoz, 185020, Otozomal Dominant (Kalıtsal Kriyohidrositoz-Normal Stomatin) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cryohydrocytosis, 185020, Autosomal dominant (Hereditary cryohydrocytosis with normal stomatin) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kriptorşidizm, Unilateral Veya Bilateral, 219050, Otozomal Dominant (INSL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CRYPTORCHIDISM, UNILATERAL OR BILATERAL, 219050, Autosomal dominant (INSL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INSL3	INSL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Culler-Jones Sendromu, 615849, Otozomal Dominant; CJS (Postaksiyal Polidaktili-Ön Hipofiz Anomalileri-Fasiyal Dismorfizm Sendromu (Culler Jones Sendromu)) (GLI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Culler-Jones syndrome, 615849, Autosomal dominant; CJS (Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome) (GLI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI2	GLI2, HPE9, CJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Culler-Jones Sendromu, 615849, Otozomal Dominant; CJS (Postaksiyal Polidaktili-Ön Hipofiz Anomalileri-Fasiyal Dismorfizm Sendromu (Culler Jones Sendromu)) (Prenatal) (GLI2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Culler-Jones syndrome, 615849, Autosomal dominant; CJS (Postaxial polydactyly-anterior pituitary anomalies-facial dysmorphism syndrome) (GLI2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLI2	GLI2, HPE9, CJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Currarino Sendromu, 176450, Otozomal Dominant (Currarino Triadı) (MNX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Currarino syndrome, 176450, Autosomal dominant (Currarino triad) (MNX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MNX1	MNX1, HLXB9, HOXHB9, SCRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Currarino Sendromu, 176450, Otozomal Dominant (Currarino Triadı) (Prenatal) (MNX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Currarino syndrome, 176450, Autosomal dominant (Currarino triad) (MNX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MNX1	MNX1, HLXB9, HOXHB9, SCRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Curry-Jones Sendromu, Somatik Mozaik, 601707; CRJS (Curry Jones Sendromu) (SMO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Curry-Jones syndrome, somatic mosaic, 601707; CRJS (Curry-Jones syndrome) (SMO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMO	SMOH, SMO, CRJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Curry-Jones Sendromu, Somatik Mozaik, 601707; CRJS (Curry Jones Sendromu) (Prenatal) (SMO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Curry-Jones syndrome, somatic mosaic, 601707; CRJS (Curry-Jones syndrome) (SMO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMO	SMOH, SMO, CRJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cushing Sendromu, ACTH Bağımsız Adrenal Somatik, 615830 (PRKACA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cushing syndrome, ACTH-independent adrenal, somatic, 615830 (PRKACA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKACA	PRKACA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Kutanöz Telanjiyektazi ve Kanser Sendromu, Ailesel, 614564, Otozomal Dominant; FCTCS (Ailesel Kutanöz Telanjiektazi ve Orofaringeal Kanser Yatkınlık Sendromu) (ATR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutaneous telangiectasia and cancer syndrome, familial, 614564, Autosomal dominant; FCTCS (Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome) (ATR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATR	ATR, FRP1, SCKL1, FCTCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kutanöz Telanjiyektazi ve Kanser Sendromu, Ailesel, 614564, Otozomal Dominant; FCTCS (Ailesel Kutanöz Telanjiektazi ve Orofaringeal Kanser Yatkınlık Sendromu) (MLPA)	Cutaneous telangiectasia and cancer syndrome, familial, 614564, Autosomal dominant; FCTCS (Familial cutaneous telangiectasia and oropharyngeal predisposition cancer syndrome) (MLPA)	ATR	ATR, FRP1, SCKL1, FCTCS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kutis Laksa, AD, 123700, Otozomal Dominant; ADCL1 (Autosomal Dominant Kutis Laksa) (ELN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, AD, 123700, Autosomal dominant; ADCL1 (Autosomal dominant cutis laxa) (ELN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELN	ELN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal dominant, 1 (ADCL1) [MIM:123700]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. Additional variable clinical features are gastrointestinal diverticula, hernia, and genital prolapse. Rare manifestations are pulmonary artery stenosis, aortic aneurysm, bronchiectasis, and emphysema. {ECO:0000269	PubMed:9873040}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kutis Laksa, AD, 123700, Otozomal Dominant; ADCL1 (Autosomal Dominant Kutis Laksa) (Prenatal) (ELN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, AD, 123700, Autosomal dominant; ADCL1 (Autosomal dominant cutis laxa) (ELN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELN	ELN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal dominant, 1 (ADCL1) [MIM:123700]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. Additional variable clinical features are gastrointestinal diverticula, hernia, and genital prolapse. Rare manifestations are pulmonary artery stenosis, aortic aneurysm, bronchiectasis, and emphysema. {ECO:0000269	PubMed:9873040}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Dominant 2, 614434; ADCL2 (Otozomal Dominant Kutis Laksa) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal dominant 2, 614434; ADCL2 (Autosomal dominant cutis laxa) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kutis Laksa, Otozomal Dominant 2, 614434; ADCL2 (Otozomal Dominant Kutis Laksa) (Prenatal) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal dominant 2, 614434; ADCL2 (Autosomal dominant cutis laxa) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Dominant 3, 616603, Otozomal Dominant; ADCL3 (Autosomal Dominant Kutis Laksa) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal dominant 3, 616603, Autosomal dominant; ADCL3 (Autosomal dominant cutis laxa) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SPG9A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, athetoid movements and hypermffeoia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. {ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18479038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kutis Laksa, Otozomal Dominant 3, 616603, Otozomal Dominant; ADCL3 (Autosomal Dominant Kutis Laksa) (Prenatal) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal dominant 3, 616603, Autosomal dominant; ADCL3 (Autosomal dominant cutis laxa) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SPG9A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, athetoid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. (ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18478038). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1A, 219100, Otozomal Resesif; ARCL1A (Autozomal Resesif Kutis Laksa Tip 1) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IA, 219100, Autosomal recessive; ARCL1A (Autosomal recessive cutis laxa type 1) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 1A (ARCL1A) [MIM:219100]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. Type I autosomal recessive cutis laxa is a specific, life-threatening disorder with organ involvement, lung ateléctasis and emphysema, diverticula of the gastrointestinal and genitourinary systems, and vascular anomalies. Associated cranial anomalies, late closure of the fontanel, joint laxity, hip dislocation, and inguinal hernia have been observed but are uncommon. (ECO:0000269|PubMed:12189163, ECO:0000269|PubMed:16652333, ECO:0000269|PubMed:16691202, ECO:0000269|PubMed:17035250, ECO:0000269|PubMed:18185537). Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations affecting this gene can modify the phenotype of diseases caused by ELN mutations. (ECO:0000269|PubMed:19194475).	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1A, 219100, Otozomal Resesif; ARCL1A (Autosomal Resesif Kutis Laksa Tip 1) (Prenatal) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IA, 219100, Autosomal recessive; ARCL1A (Autosomal recessive cutis laxa type 1) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 1A (ARCL1A) [MIM:219100]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. Type I autosomal recessive cutis laxa is a specific, life-threatening disorder with organ involvement, lung ateléctasis and emphysema, diverticula of the gastrointestinal and genitourinary systems, and vascular anomalies. Associated cranial anomalies, late closure of the fontanel, joint laxity, hip dislocation, and inguinal hernia have been observed but are uncommon. (ECO:0000269|PubMed:12189163, ECO:0000269|PubMed:16652333, ECO:0000269|PubMed:16691202, ECO:0000269|PubMed:17035250, ECO:0000269|PubMed:18185537). Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations affecting this gene can modify the phenotype of diseases caused by ELN mutations. (ECO:0000269|PubMed:19194475).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1B, 614437, Otozomal Resesif; ARCL1B (Autosomal Resesif Kutis Laksa Tip 2) (EFEMP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IB, 614437, Autosomal recessive; ARCL1B (Autosomal recessive cutis laxa type 1) (EFEMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFEMP2	EFEMP2, FBLN4, UPH1, ARCL1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 1B (ARCL1B) [MIM:614437]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. ARCL1B features include emphysema, lethal pulmonary artery occlusion, aortic aneurysm, cardiopulmonary insufficiency, birth fractures, arachnodactyly, and fragility of blood vessels. (ECO:0000269|PubMed:16685658, ECO:0000269|PubMed:17937443, ECO:0000269|PubMed:19664000). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1B, 614437, Otozomal Resesif; ARCL1B (Autosomal Resesif Kutis Laksa Tip 2) (Prenatal) (EFEMP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IB, 614437, Autosomal recessive; ARCL1B (Autosomal recessive cutis laxa type 1) (EFEMP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EFEMP2	EFEMP2, FBLN4, UPH1, ARCL1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 1B (ARCL1B) [MIM:614437]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. ARCL1B features include emphysema, lethal pulmonary artery occlusion, aortic aneurysm, cardiopulmonary insufficiency, birth fractures, arachnodactyly, and fragility of blood vessels. (ECO:0000269|PubMed:16685658, ECO:0000269|PubMed:17937443, ECO:0000269|PubMed:19664000). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1C, 613177, Otozomal Resesif; ARCL1C (Kutis Laksa-Ağır Pulmoner, Gastrointestinal ve Üriner Anomaliler) (LTBP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IC, 613177, Autosomal recessive; ARCL1C (Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies) (LTBP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTBP4	LTBP4, LTBP4S, LTBP4L, ARCL1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Urban-Rifkin-Davis syndrome (URDS) [MIM:613177]: A syndrome characterized by disrupted pulmonary, gastrointestinal, urinary, musculoskeletal, craniofacial and dermal development. Clinical features include cutis laxa, mild cardiovascular lesions, respiratory distress with cystic and atelectatic changes in the lungs, and diverticulosis, tortuosity and stenosis at various levels of the intestinal tract. Craniofacial features include microretrognathia, flat midface, receding forehead and wide fontanelles. (ECO:0000269	PubMed:19836019). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 1C, 613177, Otozomal Resesif; ARCL1C (Kutis Laksa-Ağır Pulmoner, Gastrointestinal ve Üriner Anomaliler) (Prenatal) (LTBP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IC, 613177, Autosomal recessive; ARCL1C (Cutis laxa with severe pulmonary, gastrointestinal and urinary anomalies) (LTBP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LTBP4	LTBP4, LTBP4S, LTBP4L, ARCL1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Urban-Rifkin-Davis syndrome (URDS) [MIM:613177]: A syndrome characterized by disrupted pulmonary, gastrointestinal, urinary, musculoskeletal, craniofacial and dermal development. Clinical features include cutis laxa, mild cardiovascular lesions, respiratory distress with cystic and atelectatic changes in the lungs, and diverticulosis, tortuosity and stenosis at various levels of the intestinal tract. Craniofacial features include microretrognathia, flat midface, receding forehead and wide fontanelles. (ECO:0000269	PubMed:19836019). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 2A, 219200, Otozomal Resesif; ARCL2A (Otozomal Resesif Kutis Laksa Tip 2, Klasik Tip) (ATP6V0A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IIA, 219200, Autosomal recessive; ARCL2A (Autosomal recessive cutis laxa type 2, classic type) (ATP6V0A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V0A2	ATP6V0A2, WSS, ARCL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 2A (ARCL2A) [MIM:219200]: A disorder characterized by an excessive congenital skin wrinkling, a large fontanelle with delayed closure, a typical facial appearance with downslanting palpebral fissures, a general connective tissue weakness, and varying degrees of growth and developmental delay and neurological abnormalities. Some affected individuals develop seizures and mental deterioration later in life, whereas the skin phenotype tends to become milder with age. At the molecular level, an abnormal glycosylation of serum proteins is observed in many cases. (ECO:0000269	PubMed:18157129). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 2A, 219200, Otozomal Resesif; ARCL2A (Otozomal Resesif Kutis Laksa Tip 2, Klasik Tip) (Prenatal) (ATP6V0A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IIA, 219200, Autosomal recessive; ARCL2A (Autosomal recessive cutis laxa type 2, classic type) (ATP6V0A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP6V0A2	ATP6V0A2, WSS, ARCL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 2A (ARCL2A) [MIM:219200]: A disorder characterized by an excessive congenital skin wrinkling, a large fontanelle with delayed closure, a typical facial appearance with downslanting palpebral fissures, a general connective tissue weakness, and varying degrees of growth and developmental delay and neurological abnormalities. Some affected individuals develop seizures and mental deterioration later in life, whereas the skin phenotype tends to become milder with age. At the molecular level, an abnormal glycosylation of serum proteins is observed in many cases. (ECO:0000269	PubMed:18157129). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 2B, 612940, Otozomal Resesif; ARCL2B (Otozomal Resesif Cutis Laxa Tip 2B) (PYCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IIB, 612940, Autosomal recessive; ARCL2B (Autosomal recessive cutis laxa type 2B) (PYCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYCR1	PYCR1, PRO3, ARCL2B, ARCL3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 2B (ARCL2B) [MIM:612940]: A disorder characterized by an excessive congenital skin wrinkling, a large fontanelle with delayed closure, a typical facial appearance with downslanting palpebral fissures, a general connective tissue weakness, and varying degrees of growth and developmental delay and neurological abnormalities. Patients do not manifest metabolic abnormalities. (ECO:0000269|PubMed:19576563, ECO:0000269|PubMed:19649221). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 2B, 612940, Otozomal Resesif; ARCL2B (Otozomal Resesif Cutis Laxa Tip 2B) (Prenatal) (PYCR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IIB, 612940, Autosomal recessive; ARCL2B (Autosomal recessive cutis laxa type 2B) (PYCR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYCR1	PYCR1, PRO3, ARCL2B, ARCL3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 2B (ARCL2B) [MIM:612940]: A disorder characterized by an excessive congenital skin wrinkling, a large fontanelle with delayed closure, a typical facial appearance with downslanting palpebral fissures, a general connective tissue weakness, and varying degrees of growth and developmental delay and neurological abnormalities. Patients do not manifest metabolic abnormalities. (ECO:0000269|PubMed:19576563, ECO:0000269|PubMed:19649221). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 3A, 219150, Otozomal Resesif, Sporadik Vakalar; ARCL3A (De Barsy Sendromu) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IIIA, 219150, Autosomal recessive, Isolated cases; ARCL3A (De Barsy syndrome) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SPG9A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, athetoid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. (ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18478038). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kutis Laksa, Otozomal Resesif, Tip 3A, 219150, Otozomal Resesif, Sporadik Vakalar; ARCL3A (De Barsy Sendromu) (Prenatal) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IIIA, 219150, Autosomal recessive, Isolated cases; ARCL3A (De Barsy syndrome) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SP029A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, athetoid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. {ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18478038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 3B, 614438; ARCL3B (De Barsy Sendromu) (PYCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cutis laxa, autosomal recessive, type IIIB, 614438; ARCL3B (De Barsy syndrome) (PYCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYCR1	PYCR1, PRO3, ARCL2B, ARCL3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kutis Laksa, Otozomal Resesif, Tip 3B, 614438; ARCL3B (De Barsy Sendromu) (Prenatal) (PYCR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cutis laxa, autosomal recessive, type IIIB, 614438; ARCL3B (De Barsy syndrome) (PYCR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYCR1	PYCR1, PRO3, ARCL2B, ARCL3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Siyanoz, Geçici Neonatal, 613977, Otozomal Dominant; TNCY (Hemoglobinopati Toms River) (HBG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cyanosis, transient neonatal, 613977, Autosomal dominant; TNCY (Hemoglobinopathy Toms River) (HBG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBG2	HBG2, TNCY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cyanosis transient neonatal (TNCY) [MIM:613977]: A disorder characterized by cyanosis in the fetus and neonate, due to a defect in the fetal hemoglobin chain which has reduced affinity for oxygen. Some patients develop anemia resulting from increased destruction of red cells containing abnormal or unstable hemoglobin. The cyanosis resolves spontaneously by 5 to 6 months of age or earlier, as the adult beta-globin chain is produced and replaces the fetal gamma-globin chain. {ECO:0000269|PubMed:19065339, ECO:0000269|PubMed:21561349, ECO:0000269|PubMed:24502349, ECO:0000269|PubMed:2470017, ECO:0000269|PubMed:2483933, ECO:0000269|PubMed:7741137}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Silindromatozis, Allesel, 132700, Otozomal Dominant (Allesel Silindromatozis) (CYLD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cylindromatosis, familial, 132700, Autosomal dominant (Familial cylindromatosis) (CYLD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYLD	CYLD, CDMT, EAC, MFT1, KIAA0849, BRSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cylindromatosis, familial (FCYL) [MIM:132700]: A disorder characterized by multiple skin tumors that develop from skin appendages, such as hair follicles and sweat glands. Affected individuals typically develop large numbers of tumors called cylindromas that arise predominantly in hairy parts of the body with approximately 90% on the head and neck. In severely affected individuals, cylindromas may combine into a confluent mass which may ulcerate or become infected (turban tumor syndrome). Individuals with familial cylindromatosis occasionally develop other types of tumors including spiradenomas that begin in sweat glands, and trichoepithelomas arising from hair follicles. {ECO:0000269|PubMed:12190880, ECO:0000269|PubMed:16922728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Silindromatozis, Allesel, 132700, Otozomal Dominant (Allesel Silindromatozis) (Prenatal) (CYLD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cylindromatosis, familial, 132700, Autosomal dominant (Familial cylindromatosis) (CYLD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYLD	CYLD, CDMT, EAC, MFT1, KIAA0849, BRSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cylindromatosis, familial (FCYL) [MIM:132700]: A disorder characterized by multiple skin tumors that develop from skin appendages, such as hair follicles and sweat glands. Affected individuals typically develop large numbers of tumors called cylindromas that arise predominantly in hairy parts of the body with approximately 90% on the head and neck. In severely affected individuals, cylindromas may combine into a confluent mass which may ulcerate or become infected (turban tumor syndrome). Individuals with familial cylindromatosis occasionally develop other types of tumors including spiradenomas that begin in sweat glands, and trichoepithelomas arising from hair follicles. {ECO:0000269|PubMed:12190880, ECO:0000269|PubMed:16922728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistatiyoninüri, 219500, Otozomal Resesif (Sistatiyoninüri) (CTH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystathioninuria, 219500, Autosomal recessive (Cystathioninuria) (CTH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTH	CTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystathioninuria (CSTNU) [MIM:219500]: Autosomal recessive phenotype characterized by abnormal accumulation of plasma cystathionine, leading to increased urinary excretion. {ECO:0000269|PubMed:12574942}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kistik Fibroz Akciğer Hastalığı, Modifiye Edici, 219700, Otozomal Resesif (TGFB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystic fibrosis lung disease, modifier of, 219700, Autosomal recessive (TGFB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFB1	TGFB1, DPD1, CED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kistik Fibrozis, 219700, Otozomal Resesif (Kistik Fibroz) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystic fibrosis, 219700, Autosomal recessive (Cystic fibrosis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Kistik Fibrozis, 219700, Otozomal Resesif (Kistik Fibroz) (MLPA)	Cystic fibrosis, 219700, Autosomal recessive (Cystic fibrosis) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Kistik Fibrozis, 219700, Otozomal Resesif (Kistik Fibroz) (Prenatal) (CFTR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystic fibrosis, 219700, Autosomal recessive (Cystic fibrosis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kistik Fibrozis, 219700, Otozomal Resesif (Kistik Fibroz) (Prenatal) (MLPA)	Cystic fibrosis, 219700, Autosomal recessive (Cystic fibrosis) (Prenatal) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kistik Fibroz Cf (Kistik Fibroz) (FCGR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	CYSTIC FIBROSIS; CF (Cystic fibrosis) (FCGR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2A	FCGR2A, IGFR2, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sistinoz, Erişkin Nonnefropatik (Sistinozis) (CTNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	CYSTINOSIS, ADULT NONNEPHROPATHIC (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinoz, Erişkin Nonnefropatik (Sistinozis) (MLPA)	CYSTINOSIS, ADULT NONNEPHROPATHIC (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Atipik Nefropatik, 219800, Otozomal Resesif; CTNS (Sistinozis) (CTNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinosis, atypical nephropathic, 219800, Autosomal recessive; CTNS (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Atipik Nefropatik, 219800, Otozomal Resesif; CTNS (Sistinozis) (MLPA)	Cystinosis, atypical nephropathic, 219800, Autosomal recessive; CTNS (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Atipik Nefropatik, 219800, Otozomal Resesif; CTNS (Sistinozis) (Prenatal) (CTNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinosis, atypical nephropathic, 219800, Autosomal recessive; CTNS (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Atipik Nefropatik, 219800, Otozomal Resesif; CTNS (Sistinozis) (MLPA)	Cystinosis, atypical nephropathic, 219800, Autosomal recessive; CTNS (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Geç Başlangıçlı Jüvenil Veya Adolesan Nefropati, 219900, Otozomal Resesif (Sistinozis) (CTNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinosis, late-onset juvenile or adolescent nephropathic, 219900, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Geç Başlangıçlı Jüvenil Veya Adolesan Nefropati, 219900, Otozomal Resesif (Sistinozis) (MLPA)	Cystinosis, late-onset juvenile or adolescent nephropathic, 219900, Autosomal recessive (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Geç Başlangıçlı Jüvenil Veya Adolesan Nefropati, 219900, Otozomal Resesif (Sistinozis) (Prenatal) (CTNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinosis, late-onset juvenile or adolescent nephropathic, 219900, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Geç Başlangıçlı Jüvenil Veya Adolesan Nefropati, 219900, Otozomal Resesif (Sistinozis) (Prenatal) (MLPA)	Cystinosis, late-onset juvenile or adolescent nephropathic, 219900, Autosomal recessive (Cystinosis) (Prenatal) (MLPA)	CTNS	CTNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Nefropatik, 219800, Otozomal Resesif (Sistinozis) (CTNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinosis, nephropathic, 219800, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Nefropatik, 219800, Otozomal Resesif (Sistinozis) (MLPA)	Cystinosis, nephropathic, 219800, Autosomal recessive (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269|PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Sistinozis, Nefropatik, 219800, Otozomal Resesif (Sistinozis) (Prenatal) (CTNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinosis, nephropathic, 219800, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Nefropatik, 219800, Otozomal Resesif (Sistinozis) (Prenatal) (MLPA)	Cystinosis, nephropathic, 219800, Autosomal recessive (Cystinosis) (Prenatal) (MLPA)	CTNS	CTNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Okiler Nonnefropatik, 219750, Otozomal Resesif (Sistinozis) (CTNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinosis, ocular nonnephropathic, 219750, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Okiler Nonnefropatik, 219750, Otozomal Resesif (Sistinozis) (MLPA)	Cystinosis, ocular nonnephropathic, 219750, Autosomal recessive (Cystinosis) (MLPA)	CTNS	CTNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sistinozis, Okiler Nonnefropatik, 219750, Otozomal Resesif (Sistinozis) (Prenatal) (CTNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinosis, ocular nonnephropathic, 219750, Autosomal recessive (Cystinosis) (CTNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTNS	CTNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinozis, Okiler Nonnefropatik, 219750, Otozomal Resesif (Sistinozis) (Prenatal) (MLPA)	Cystinosis, ocular nonnephropathic, 219750, Autosomal recessive (Cystinosis) (Prenatal) (MLPA)	CTNS	CTNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystinosis, adult, non-nephropathic type (CTNSANN) [MIM:219750]: A form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benign course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. {ECO:0000269	PubMed:10625078}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinüri paneli (SLC3A1, 2p12, SLC7A9) (MLPA)	Cystinuria panel (SLC3A1, 2p12, SLC7A9) (MLPA)	SLC3A1, 2p12, SLC7A9	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sistinüri paneli (SLC3A1, 2p12, SLC7A9) (MLPA) (Prenatal)	Cystinuria panel (SLC3A1, 2p12, SLC7A9) (MLPA) (Prenatal)	SLC3A1, 2p12, SLC7A9	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (SLC7A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (SLC7A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC7A9	SLC7A9, CSNU3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (SLC3A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (SLC3A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC3A1	SLC3A1, ATR1, D2H, NBAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (MLPA)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (MLPA)	SLC7A9	SLC7A9, CSNU3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (MLPA)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (MLPA)	SLC3A1	SLC3A1, ATR1, D2H, NBAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (SLC7A9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (SLC7A9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC7A9	SLC7A9, CSNU3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (SLC3A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (SLC3A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC3A1	SLC3A1, ATR1, D2H, NBAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (MLPA)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (Prenatal) (MLPA)	SLC7A9	SLC7A9, CSNU3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sistinüri, 220100, Otozomal Resesif, Otozomal Dominant (Sistinüri) (MLPA)	Cystinuria, 220100, Autosomal recessive, Autosomal dominant (Cystinuria) (Prenatal) (MLPA)	SLC3A1	SLC3A1, ATR1, D2H, NBAT	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sitokrom P-450 paneli (CYP2D6, CYP2C9, CYP2C19, CYP1B1, CYP3A4, CYP3A5, CYP2E1, CYP1A1, CYP1A2, CYP2A6, CYP2B6, GSTP1, GSTT1, GSTM1) (MLPA)	Cytochrome P-450 (CYP2D6, CYP2C9, CYP2C19, CYP1B1, CYP3A4, CYP3A5, CYP2E1, CYP1A1, CYP1A2, CYP2A6, CYP2B6, GSTP1, GSTT1, GSTM1)	CYP2D6, CYP2C9, CYP2C19, CYP1B1, CYP3A4, CYP3A5, CYP2E1, CYP1A1, CYP1A2, CYP2A6, CYP2B6, GSTP1, GSTT1, GSTM1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sitrokrom P450, Alt Ailesi I, Polipeptid 2; Cyp1a2 (CYP1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CYTOCHROME P450, SUBFAMILY I, POLYPEPTIDE 2; CYP1A2 (CYP1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP1A2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sitrokrom P450, Alt Ailesi I, Polipeptid 2; Cyp1a2 (MLPA)	CYTOCHROME P450, SUBFAMILY I, POLYPEPTIDE 2; CYP1A2 (MLPA)	CYP1A2		MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sitrokrom P450, Alt Ailesi Ic, Polipeptid 8; Cyp2c8 (CYP2C8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	CYTOCHROME P450, SUBFAMILY IC, POLYPEPTIDE 8; CYP2C8 (CYP2C8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C8	CYP2C8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çek Displazi, 609162, Otozomal Dominant (Çek Displazi, Metatarsal Tip) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Czech dysplasia, 609162, Autosomal dominant (Czech dysplasia, metatarsal type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çek Displazi, 609162, Otozomal Dominant (Çek Displazi, Metatarsal Tip) (MLPA)	Czech dysplasia, 609162, Autosomal dominant (Czech dysplasia, metatarsal type) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Çek Displazi, 609162, Otozomal Dominant (Çek Displazi, Metatarsal Tip) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Czech dysplasia, 609162, Autosomal dominant (Czech dysplasia, metatarsal type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çek Displazi, 609162, Otozomal Dominant (Çek Displazi, Metatarsal Tip) (Prenatal) (MLPA)	Czech dysplasia, 609162, Autosomal dominant (Czech dysplasia, metatarsal type) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
D-2-Hidroksiglutarik Asidüri 2, 613657; D2HGA2 (D-2-Hidroksi Glutarik Asidüri) (IDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	D-2-hydroxyglutaric aciduria 2, 613657; D2HGA2 (D-2-hydroxyglutaric aciduria) (IDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDH2	IDH2, IDPM, D2HGA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
D-2-Hidroksiglutarik Asidüri 2, 613657; D2HGA2 (D-2-Hidroksi Glutarik Asidüri) (Prenatal) (IDH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	D-2-hydroxyglutaric aciduria 2, 613657; D2HGA2 (D-2-hydroxyglutaric aciduria) (IDH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDH2	IDH2, IDPM, D2HGA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
D-2-Hidroksiglutarik Asidüri, 600721, Otozomal Resesif; D2HGA1 (D-2-Hidroksi Glutarik Asidüri) (D2HGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	D-2-hydroxyglutaric aciduria, 600721, Autosomal recessive; D2HGA1 (D-2-hydroxyglutaric aciduria) (D2HGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	D2HGDH	D2HGDH, D2HGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: D-2-hydroxyglutaric aciduria 1 (D2HGA1) [MIM:600721]: A rare recessive neurometabolic disorder causing developmental delay, epilepsy, hypotonia, and dysmorphic features. Both a mild and a severe phenotype exist. The severe phenotype is homogeneous and is characterized by early infantile-onset epileptic encephalopathy and cardiomyopathy. The mild phenotype has a more variable clinical presentation. Diagnosis is based on the presence of an excess of D-2-hydroxyglutaric acid in the urine. {ECO:0000269|PubMed:15609246, ECO:0000269|PubMed:16037974, ECO:0000269|PubMed:16081310}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
D-2-Hidroksiglutarik Asidüri, 600721, Otozomal Resesif; D2HGA1 (D-2-Hidroksi Glutarik Asidüri) (MLPA)	D-2-hydroxyglutaric aciduria, 600721, Autosomal recessive; D2HGA1 (D-2-hydroxyglutaric aciduria) (MLPA)	D2HGDH	D2HGDH, D2HGD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: D-2-hydroxyglutaric aciduria 1 (D2HGA1) [MIM:600721]: A rare recessive neurometabolic disorder causing developmental delay, epilepsy, hypotonia, and dysmorphic features. Both a mild and a severe phenotype exist. The severe phenotype is homogeneous and is characterized by early infantile-onset epileptic encephalopathy and cardiomyopathy. The mild phenotype has a more variable clinical presentation. Diagnosis is based on the presence of an excess of D-2-hydroxyglutaric acid in the urine. {ECO:0000269|PubMed:15609246, ECO:0000269|PubMed:16037974, ECO:0000269|PubMed:16081310}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
D-2-Hidroksiglutarik Asidüri, 600721, Otozomal Resesif; D2HGA1 (D-2-Hidroksi Glutarik Asidüri) (Prenatal) (D2HGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	D-2-hydroxyglutaric aciduria, 600721, Autosomal recessive; D2HGA1 (D-2-hydroxyglutaric aciduria) (D2HGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	D2HGDH	D2HGDH, D2HGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: D-2-hydroxyglutaric aciduria 1 (D2HGA1) [MIM:600721]: A rare recessive neurometabolic disorder causing developmental delay, epilepsy, hypotonia, and dysmorphic features. Both a mild and a severe phenotype exist. The severe phenotype is homogeneous and is characterized by early infantile-onset epileptic encephalopathy and cardiomyopathy. The mild phenotype has a more variable clinical presentation. Diagnosis is based on the presence of an excess of D-2-hydroxyglutaric acid in the urine. {ECO:0000269|PubMed:15609246, ECO:0000269|PubMed:16037974, ECO:0000269|PubMed:16081310}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

D-2-Hidroksiglutarik Asidüri, 600721, Otozomal Resesif; D2HGA1 (D-2-Hidroksi Glutarik Asidüri) (Prenatal) (MLPA)	D-2-hydroxyglutaric aciduria, 600721, Autosomal recessive; D2HGA1 (D-2-hydroxyglutaric aciduria) (Prenatal) (MLPA)	D2HGDH	D2HGDH, D2HGD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: D-2-hydroxyglutaric aciduria 1 (D2HGA1) [MIM:600721]: A rare recessive neurometabolic disorder causing developmental delay, epilepsy, hypotonia, and dysmorphic features. Both a mild and a severe phenotype exist. The severe phenotype is homogeneous and is characterized by early infantile-onset epileptic encephalopathy and cardiomyopathy. The mild phenotype has a more variable clinical presentation. Diagnosis is based on the presence of an excess of D-2-hydroxyglutaric acid in the urine. {ECO:0000269|PubMed:15609246, ECO:0000269|PubMed:16037974, ECO:0000269|PubMed:16081310}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
D-İki İşlevli Protein Eksikliği, 261515, Otozomal Resesif (İki Fonksiyonlu Enzim Eksikliği) (HSD17B4 gen) (Tüm Gen Dizi Analizi) (Prenatal)	D-bifunctional protein deficiency, 261515, Autosomal recessive (Bifunctional enzyme deficiency) (HSD17B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B4	HSD17B4, PRLTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	.	Postnatal	.	EDTA Blood Sample
D-İki İşlevli Protein Eksikliği, 261515, Otozomal Resesif (İki Fonksiyonlu Enzim Eksikliği) (Prenatal) (HSD17B4 gen) (Tüm Gen Dizi Analizi) (Prenatal)	D-bifunctional protein deficiency, 261515, Autosomal recessive (Bifunctional enzyme deficiency) (HSD17B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B4	HSD17B4, PRLTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
D-Gliserik Asidüri, 220120, Otozomal Resesif (D-Gliserik Asidüri) (GLYCTK gen) (Tüm Gen Dizi Analizi) (Postnatal)	D-glyceric aciduria, 220120, Autosomal recessive (D-glyceric aciduria) (GLYCTK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLYCTK	GLYCTK, GLYCTK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
D-Gliserik Asidüri, 220120, Otozomal Resesif (D-Gliserik Asidüri) (Prenatal) (GLYCTK gen) (Tüm Gen Dizi Analizi) (Prenatal)	D-glyceric aciduria, 220120, Autosomal recessive (D-glyceric aciduria) (GLYCTK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLYCTK	GLYCTK, GLYCTK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dandy-Walker Malformasyonu (DWM) paneli (ZIC1, ZIC4 3p2, VLDLR 9p24) (MLPA)	Dandy-Walker Malformation (DWM) panel (ZIC1, ZIC4 3p2, VLDLR 9p24) (MLPA)	ZIC1, ZIC4 3p2, VLDLR 9p24	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	.	EDTA Blood Sample
Dandy-Walker Malformasyonu (DWM) paneli (ZIC1, ZIC4 3p2, VLDLR 9p24) (MLPA) (Prenatal)	Dandy-Walker Malformation (DWM) panel (ZIC1, ZIC4 3p2, VLDLR 9p24) (MLPA) (Prenatal)	ZIC1, ZIC4 3p2, VLDLR 9p24	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dandy-Walker Sendromu, 220200, Sporadik Vakalar (İzole Dandy-Walker Malformasyonu) (2200)	Dandy-Walker syndrome, 220200, Isolated cases (Isolated Dandy-Walker malformation) (440)	DWS	DWS, C3OEL-q23q24, DEL3q23q24	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Dandy-Walker Sendromu, 220200, Sporadik Vakalar (İzole Dandy-Walker Malformasyonu) (Prenatal)	Dandy-Walker syndrome, 220200, Isolated cases (Isolated Dandy-Walker malformation) (Prenatal)	DWS	DWS, C3OEL-q23q24, DEL3q23q24	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Danon Hastalığı, 300257, X'e Bağlı Dominant (Glikojen Depo Hastalığı, LAMP-2 Eksikliğine Bağlı (Danon Hastalığı)) (LAMP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Danon disease, 300257, X-linked dominant (Glycogen storage disease due to LAMP-2 deficiency) (LAMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMP2	LAMP2, LAMPB, LGP110	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Danon disease (DAND) [MIM:300257]: DAND is a lysosomal glycogen storage disease characterized by the clinical triad of cardiomyopathy, vacuolar myopathy and mental retardation. It is often associated with an accumulation of glycogen in muscle and lysosomes. {ECO:0000269|PubMed:15673802, ECO:0000269|PubMed:15907287}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Danon Hastalığı, 300257, X'e Bağlı Dominant (Glikojen Depo Hastalığı, LAMP-2 Eksikliğine Bağlı (Danon Hastalığı)) (Prenatal) (LAMP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Danon disease, 300257, X-linked dominant (Glycogen storage disease due to LAMP-2 deficiency) (LAMP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMP2	LAMP2, LAMPB, LGP110	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Danon disease (DAND) [MIM:300257]: DAND is a lysosomal glycogen storage disease characterized by the clinical triad of cardiomyopathy, vacuolar myopathy and mental retardation. It is often associated with an accumulation of glycogen in muscle and lysosomes. {ECO:0000269|PubMed:15673802, ECO:0000269|PubMed:15907287}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Darier Hastalığı, 124200, Otozomal Dominant; DAR (Darier Hastalığı) (ATP2A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Darier disease, 124200, Autosomal dominant; DAR (Darier disease) (ATP2A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2A2	ATP2A2, ATP2B, DAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Darier Hastalığı, 124200, Otozomal Dominant; DAR (Darier Hastalığı) (Prenatal) (ATP2A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Darier disease, 124200, Autosomal dominant; DAR (Darier disease) (ATP2A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP2A2	ATP2A2, ATP2B, DAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
De la Chapelle Displazi, 256050, Otozomal Resesif (Atelosteogenesis Tip II) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	De la Chapelle dysplasia, 256050, Autosomal recessive (Atelosteogenesis type II) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 4 (EDM4) [MIM:226900]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. Multiple epiphyseal dysplasia type 4 is a recessively inherited form, characterized by early childhood-onset hip dysplasia and recurrent patella dislocation. Short stature is not frequent. {ECO:0000269|PubMed:12966518, ECO:0000269|PubMed:21927596}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
De la Chapelle Displazi, 256050, Otozomal Resesif (Atelosteogenesis Tip II) (Prenatal) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	De la Chapelle dysplasia, 256050, Autosomal recessive (Atelosteogenesis type II) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 4 (EDM4) [MIM:226900]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. Multiple epiphyseal dysplasia type 4 is a recessively inherited form, characterized by early childhood-onset hip dysplasia and recurrent patella dislocation. Short stature is not frequent. {ECO:0000269|PubMed:12966518, ECO:0000269|PubMed:21927596}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
De Sanctis-Cacchione Sendromu, 278800, Otozomal Resesif (De Sanctis-Cacchione Sendromu) (ERCC6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	De Sanctis-Cacchione syndrome, 278800, Autosomal recessive (De Sanctis-Cacchione syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
De Sanctis-Cacchione Sendromu, 278800, Otozomal Resesif (De Sanctis-Cacchione Sendromu) (Prenatal) (ERCC6 gen) (Tüm Gen Dizi Analizi) (Prenatal)	De Sanctis-Cacchione syndrome, 278800, Autosomal recessive (De Sanctis-Cacchione syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İşitme Kaybı, Digenic GJB2/ GJB6 (Tüm Gen - Dizi Analizi) (GJB2, GJB6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness (GJB2, GJB6 genes) (All coding exons - Sequence analysis) (GJB2, GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2, GJB6	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
İşitme Kaybı, Digenic GJB2/ GJB6 + Mitokondriyal A1555G (Tüm Gen - Dizi Analizi) (GJB2, GJB6, mtDNA 12S rRNA (MTRNR1) gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness (GJB2, GJB6 genes) (All coding exons - Sequence analysis) and (MTRNR1 gene A1555G mutation) (GJB2, GJB6, mtDNA 12S rRNA (MTRNR1) gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2, GJB6, mtDNA 12S rRNA (MTRNR1)	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık ve Erkek İnfertilitesi, 611102, Otozomal Resesif (Sağırlık-İnfertilite Sendromu) (2200)	Deafness and male infertility, 611102, Autosomal recessive (Deafness-infertility syndrome) (440)	.	DEL15q15.3, C15DELq15.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Sağırlık ve Miyopi, 221200, Otozomal Resesif; DFNMYP (Yüksek Miyopi-Sensorinöral Sağırlık Sendromu) (SLITRK6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness and myopia, 221200, Autosomal recessive; DFNMYP (High myopia-sensorineural deafness syndrome) (SLITRK6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLITRK6	SLITRK6, DFNMYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 1, 124900, Otozomal Dominant; DFNA1 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (DIAPH1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 1, 124900, Autosomal dominant; DFNA1 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (DIAPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIAPH1	DIAPH1, DFNA1, LFHL1, SCBMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 1 (DFNA1) [MIM:124900]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:9360932}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 10, 601316, Otozomal Dominant; DFNA10 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (EYA4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 10, 601316, Autosomal dominant; DFNA10 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (EYA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA4	EYA4, DFNA10, CMD1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 11, 601317, Otozomal Dominant; DFNA11 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MYO7A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 11, 601317, Autosomal dominant; DFNA11 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MYO7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO7A	MYO7A, USH1B, DFNB2, DFNA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 13, 601868, Otozomal Dominant (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (COL11A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 13, 601868, Autosomal dominant (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık Otozomal Dominant 13, Dfna13 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (COL11A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL DOMINANT 13; DFNA13 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 15, 602459, Otozomal Dominant; DFNA15 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (POU4F3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 15, 602459, Autosomal dominant; DFNA15 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (POU4F3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POU4F3	POU4F3, BRN3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 17, 603622, Otozomal Dominant; DFNA17 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MYH9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 17, 603622, Autosomal dominant; DFNA17 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample

Sağırlık, Otozomal Dominant 20/26, 604717, Otozomal Dominant; DFNA20 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (ACTG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 20/26, 604717, Autosomal dominant; DFNA20 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (ACTG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTG1	ACTG1, DFNA20, DFNA26, BRWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 20 (DFNA20) [MIM:604717]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:19805526, ECO:0000269|PubMed:14684684, ECO:0000269|PubMed:16773128, ECO:0000269|PubMed:18804074, ECO:0000269|PubMed:19477959}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 22 606346, Otozomal Dominant; DFNA22 (Progresif İşitme Kaybı-Hipertrofik Kardiyomiyopati Sendromu) (MYO6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 22, 606346, Autosomal dominant; DFNA22 (Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome) (MYO6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO6	MYO6, DFNA22, DFNB37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 22 (DFNA22) [MIM:606346]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNA22 is progressive and postlingual, with onset during childhood. By the age of approximately 50 years, affected individuals invariably have profound sensorineural deafness. {ECO:0000269	PubMed:11468689}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 23, 605192, Otozomal Dominant; DFNA23 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (SIX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 23, 605192, Autosomal dominant; DFNA23 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (SIX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX1	SIX1, BOS3, DFNA23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 23 (DFNA23) [MIM:605192]: A form of non-syndromic deafness characterized by prelingual, bilateral, symmetric hearing loss with a conductive component present in some but not all patients. {ECO:0000269	PubMed:15141091}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 25, 605583, Otozomal Dominant; DFNA25 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (SLC17A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 25, 605583, Autosomal dominant; DFNA25 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (SLC17A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A8	SLC17A8, VGLUT3, DFNA25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Sağırlık, Otozomal Dominant 28 608641, Otozomal Dominant; DFNA28 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (GRHL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 28, 608641, Autosomal dominant; DFNA28 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (GRHL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRHL2	GRHL2, TFCP2L3, DFNA28, ECTDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 28 (DFNA28) [MIM:608641]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNA28 is characterized by mild to moderate hearing loss across most frequencies that progresses to severe loss in the higher frequencies by the fifth decade. {ECO:0000269	PubMed:12993799}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 2A, 600101, Otozomal Dominant; DFNA2A (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (KCNQ4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 2A, 600101, Autosomal dominant; DFNA2A (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (KCNQ4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ4	KCNQ4, DFNA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 2A (DFNA2A) [MIM:600101]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:10025409, ECO:0000269|PubMed:10399879, ECO:0000269|PubMed:10571947, ECO:0000269|PubMed:10925378, ECO:0000269|PubMed:21242547}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Sağırlık, Otozomal Dominant 2B, 612644, Otozomal Dominant (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (GJB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 2B, 612644, Autosomal dominant (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 2B, 612644, Otozomal Dominant (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MLPA)	Deafness, autosomal dominant 2B, 612644, Autosomal dominant (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MLPA)	GJB3	GJB3, CX31, DFNA2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 2b; Dfna2b (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (GJB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL DOMINANT 2B, DFNA2B (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 36, 606705, Otozomal Dominant; DFNA36 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (TMC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 36, 606705, Autosomal dominant; DFNA36 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (TMC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMC1	TMC1, DFNB7, DFNB11, DFNA36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Dentinogenez İle Otozomal Dominant 39, 605594, Otozomal Dominant (Dentinogenezis İmperfekta Type 2) (DSPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 39, with dentinogenesis, 605594, Autosomal dominant (Dentinogenesis imperfecta type 2) (DSPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSPP	DSPP, DPP, DGI1, DFNA39, DTDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 3A, 601544, Otozomal Dominant; DFNA3A (DFNA) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 3A, 601544, Autosomal dominant; DFNA3A (DFNA) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 3B, 612643, Otozomal Dominant; DFNA3B (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (GJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 3B, 612643, Autosomal dominant; DFNA3B (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 40, 616357, Otozomal Dominant; DFNA40 (DFNA) (CRYM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 40, 616357, Autosomal dominant; DFNA40 (DFNA) (CRYM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYM	CRYM, DFNA40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 41, 608224, Otozomal Dominant; DFNA41 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (P2RX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 41, 608224, Autosomal dominant; DFNA41 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (P2RX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P2RX2	P2RX2, P2X2, DFNA41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 41 (DFNA41) [MIM:608224]: A form of non-syndromic deafness characterized by onset of progressive sensorineural hearing loss usually in the second decade. The hearing loss is severe and ultimately affects all frequencies. Exposure to noise exacerbates the hearing loss, particularly at high frequencies. {ECO:0000269	PubMed:23345450, ECO:0000269	PubMed:24211385}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant 44, 607453, Otozomal Dominant; DFNA44 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (CCDC50 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 44, 607453, Autosomal dominant; DFNA44 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (CCDC50 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC50	CCDC50, C3orf6, DFNA44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 44 (DFNA44) [MIM:607453]: A form of non-syndromic deafness characterized by initially moderate hearing loss that affects mainly low to mid frequencies. Later, it progresses to involve all the frequencies and leads to a profound hearing loss by the 6th decade. {ECO:0000269	PubMed:17503326}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Sağırlık, Otozomal Dominant 4A, 600652, Otozomal Dominant; DFNA4A (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MYH14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 4A, 600652, Autosomal dominant; DFNA4A (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MYH14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH14	MYH14, KIAA2034, DFNA4A, PNMHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 4A (DFNA4A) [MIM:600652]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:15015131, ECO:0000269	PubMed:16222661}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 4B, 614614, Otozomal Dominant; DFNA4B (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (CEACAM16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 4B, 614614, Autosomal dominant; DFNA4B (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (CEACAM16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEACAM16	CEACAM16, CEAL2, DFNA4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 4B (DFNA4B) [MIM:614614]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:21368133, ECO:0000269	PubMed:25589040}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant 5, 600994, Otozomal Dominant; DFNA5 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (DFNA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 5, 600994, Autosomal dominant (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (DFNA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DFNA5	DFNA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Is a tumor suppressor gene with an important role in colorectal cancer (CRC). {ECO:0000303	PubMed:18223688}.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant 50, 613074, Otozomal Dominant; DFNA50 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MIR96 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 50, 613074, Autosomal dominant; DFNA50 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MIR96 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIR96	MIR96, MIRN96, DFNA50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 51, 613558, Otozomal Dominant (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (2200)	Deafness, autosomal dominant 51, 613558, Autosomal dominant (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (44/2)	.	DFNA51, C9ORF21.11, DUP9q21.11	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Sağırlık, Otozomal Dominant 56, 615629, Otozomal Dominant; DFNA56 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (TNC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 56, 615629, Autosomal dominant; DFNA56 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (TNC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNC	TNC, HXB, DFNA56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 56 (DFNA56) [MIM:615629]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNA56 is characterized by progressive hearing impairment with post-lingual onset. {ECO:0000269	PubMed:23936043}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant 6/14/38, 600965, Otozomal Dominant; DFNA6 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (WFS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 6/14/38, 600965, Autosomal dominant; DFNA6 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (WFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 6/14/38, 600965, Otozomal Dominant; DFNA6 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (MLPA)	Deafness, autosomal dominant 6/14/38, 600965, Autosomal dominant; DFNA6 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (MLPA)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 64, 614152, Otozomal Dominant; DFNA64 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (DIABLO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 64, 614152, Autosomal dominant; DFNA64 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (DIABLO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIABLO	SMAC, DIABLO, DFNA64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 64 (DFNA64) [MIM:614152]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:21722859}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant 65, 616044, Otozomal Dominant; DFNA65 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 65, 616044, Autosomal dominant; DFNA65 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sağırlık, Otozomal Dominant 66, 616969, Otozomal Dominant; DFNA66 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (CD164 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 66, 616969, Autosomal dominant; DFNA66 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (CD164 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD164	CD164, DFNA66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 67, 616340, Otozomal Dominant; DFNA67 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (OSBPL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 67, 616340, Autosomal dominant; DFNA67 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (OSBPL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OSBPL2	OSBPL2, ORP2, KIAA0772, DNFA67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 68, 616707, Otozomal Dominant; DFNA68 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (HOMER2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 68, 616707, Autosomal dominant; DFNA68 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (HOMER2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOMER2	HOMER2, HOMER2B, HOMER2A, DFNA68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 69, Tek Taraflı Veya Asimetrik, 616697, Otozomal Dominant; DFNA69 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (KITLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 69, unilateral or asymmetric, 616697, Autosomal dominant; DFNA69 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (KITLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KITLG	KITLG, MGF, SF, SCF, SHEP7, FPHH, DCUA, DFNA69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 70, 616968, Otozomal Dominant; DFNA70 (DFNA) (MCM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 70, 616968, Autosomal dominant; DFNA70 (DFNA) (MCM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCM2	MCM2, CDCL1, DFNA70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 8/12, 601543, Otozomal Dominant; DFNA12 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (TECTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 8/12, 601543, Autosomal dominant; DFNA12 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (TECTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TECTA	TECTA, DFNA8, DFNA12, DFNB21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 12 (DFNA12) [MIM:601543]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:10196713, ECO:0000269|PubMed:10987647, ECO:0000269|PubMed:12162770, ECO:0000269|PubMed:15319541, ECO:0000269|PubMed:16718611, ECO:0000269|PubMed:17651817, ECO:0000269|PubMed:20947814, ECO:0000269|PubMed:21520338, ECO:0000269|PubMed:9590290}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Dominant 9, 601369, Otozomal Dominant; DFNA9 (Otozomal Dominant Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNA) (COCH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant 9, 601369, Autosomal dominant; DFNA9 (Autosomal dominant non-syndromic sensorineural deafness type DFNA) (COCH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COCH	COCH, DFNA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 9 (DFNA9) [MIM:601369]: A form of non-syndromic hearing loss characterized by onset in the fourth or fifth decade of life and initially involves the high frequencies. Hearing loss is progressive and usually complete by the sixth decade. In addition to cochlear involvement, DFNA9 patients also exhibit a spectrum of vestibular dysfunctions. Penetrance of the vestibular symptoms is often incomplete, and some patients are minimally affected, whereas others suffer from severe balance disturbances and episodes of vertigo. Affected individuals have mucopolysaccharide depositions in the channels of the cochlear and vestibular nerves. These depositions apparently cause strangulation and degeneration of dendritic fibers. {ECO:0000269|PubMed:10400989, ECO:0000269|PubMed:11295836, ECO:0000269|PubMed:14512963, ECO:0000269|PubMed:16835021, ECO:0000269|PubMed:17561763, ECO:0000269|PubMed:18312449, ECO:0000269|PubMed:22610276, ECO:0000269|PubMed:22931125, ECO:0000269|PubMed:23983205, ECO:0000269|PubMed:9806553, ECO:0000269|PubMed:9931344}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Dominant, Periferik Nöropati (GJB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal dominant, with peripheral neuropathy (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Dominant, Periferik Nöropati (MLPA)	Deafness, autosomal dominant, with peripheral neuropathy (MLPA)	GJB3	GJB3, CX31, DFNA2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 101, 615837, Otozomal Resesif; DFNB101 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GRXCR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 101, 615837, Autosomal recessive; DFNB101 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GRXCR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRXCR2	GRXCR2, DFNB101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 101 (DFNB101) [MIM:615837]: A form of non-syndromic deafness characterized by bilateral, moderate to severe hearing loss. Vestibular function is unaffected. {ECO:0000269	PubMed:24619944}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 102, 615974, Otozomal Resesif; DFNB102 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (EPS8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 102, 615974, Autosomal recessive; DFNB102 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (EPS8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPS8	EPS8, DFNB102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 102 (DFNB102) [MIM:615974]: A form of non-syndromic deafness characterized by profound hearing loss affecting all frequencies. Vestibular function is unaffected. {ECO:0000269	PubMed:24741995}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 103, 616042, Otozomal Resesif; DFNB103 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CLIC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 103, 616042, Autosomal recessive; DFNB103 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CLIC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLIC5	CLIC5, DFNB103	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 103 (DFNB103) [MIM:616042]: A form of sensorineural deafness with onset in early childhood. Hearing impairment progresses from mild to severe or even profound before the second decade, and is accompanied by vestibular areflexia. {ECO:0000269	PubMed:24781754}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 104, 616515, Otozomal Resesif; DFNB104 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (RIPOR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 104, 616515, Autosomal recessive; DFNB104 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (RIPOR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIPOR2	FAM65B, C6orf32, KIAA0386, PL48, DFNB104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Sağırlık, Otozomal Resesif 105, 616958, Otozomal Resesif; DFNB105 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CDC14A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 105, 616958, Autosomal recessive; DFNB105 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CDC14A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC14A	CDC14A, DFNB105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 12, 601386, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CDH23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 12, 601386, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CDH23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH23	CDH23, USH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 12, Modifiye Edici, 601386, Otozomal Resesif (DFNB) (ATP2B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 12, modifier of, 601386, Autosomal recessive (DFNB) (ATP2B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2B2	ATP2B2, PMCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 12, Dfnb12 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CDH23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL RECESSIVE 12; DFNB12 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CDH23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH23	CDH23, USH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 15, 601869, Otozomal Resesif; DFNB15 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GIPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 15, 601869, Autosomal recessive; DFNB15 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GIPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GIPC3	GIPC3, DFNB15, DFNB72, DFNB95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 15 (DFNB15) [MIM:601869]: A form of non-syndromic sensorineural hearing loss with prelingual onset. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:21336233, ECO:0000269|PubMed:21660509}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 16, 603720, Otozomal Resesif; DFNB16 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (STRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 16, 603720, Autosomal recessive; DFNB16 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (STRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STRC	STRC, DFNB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 16 (DFNB16) [MIM:603720]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:11687802}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 18A, 602092, Otozomal Resesif; DFNB18A (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (USH1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 18A, 602092, Autosomal recessive; DFNB18A (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (USH1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USH1C	USH1C, DFNB18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 18B, 614945, Otozomal Resesif; DFNB18B (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (OTOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 18B, 614945, Autosomal recessive; DFNB18B (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (OTOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTOG	OTOG, OTGN, DFNB18B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 18B (DFNB18B) [MIM:614945]: A form of non-syndromic deafness characterized by a moderate hearing impairment, which can be associated with vestibular dysfunction, and a flat to shallow 'U' or slightly downsloping shaped audiograms. {ECO:0000269	PubMed:23172587}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 1A, 220290, Otozomal Resesif, Digenic Dominant (DFNB) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 1A, 220290, Autosomal recessive, Digenic dominant (DFNB) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 1a; Dfnb1a (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GJB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL RECESSIVE 1A; DFNB1A (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 1B, 612645, Otozomal Resesif; DFNB1B (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 1B, 612645, Autosomal recessive; DFNB1B (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 2, 600060, Otozomal Resesif; DFNB2 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MYO7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 2, 600060, Autosomal recessive; DFNB2 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MYO7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO7A	MYO7A, USH1B, DFNB2, DFNA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 21, 603629, Otozomal Resesif; DFNB21 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TECTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 21, 603629, Autosomal recessive; DFNB21 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TECTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TECTA	TECTA, DFNA8, DFNA12, DFNB21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sağırlık, Otozomal Resesif 22, 607039, Otozomal Resesif; DFNB22 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (OTOA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 22, 607039, Autosomal recessive; DFNB22 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (OTOA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTOA	OTOA, DFNB22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 22 (DFNB22) [MIM:607039]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:11972037}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 23, 609533, Otozomal Resesif; DFNB23 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (PCDH15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 23, 609533, Autosomal recessive; DFNB23 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (PCDH15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCDH15	PCDH15, DFNB23, USH1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 24, 611022, Otozomal Resesif; DFNB24 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (RDX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 24, 611022, Autosomal recessive; DFNB24 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (RDX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RDX	RDX, DFNB24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 24 (DFNB24) [MIM:611022]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:17226784}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 25, 613285, Otozomal Resesif; DFNB25 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GRXCR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 25, 613285, Autosomal recessive; DFNB25 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GRXCR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRXCR1	GRXCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 28 609823, Otozomal Resesif; DFNB28 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TRIOBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 28, 609823, Autosomal recessive; DFNB28 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TRIOBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIOBP	TRIOBP, KIAA1662	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 28 (DFNB28) [MIM:609823]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:16385457, ECO:0000269|PubMed:16385458}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 29 614035, Otozomal Resesif; DFNB29 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CLDN14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 29, 614035, Autosomal recessive; DFNB29 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CLDN14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLDN14	CLDN14, DFNB29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 3, 600316, Otozomal Resesif; DFNB3 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MYO15A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 3, 600316, Autosomal recessive; DFNB3 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MYO15A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO15A	MYO15A, DFNB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 30, 607101, Otozomal Resesif; DFNB30 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MYO3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 30, 607101, Autosomal recessive; DFNB30 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MYO3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO3A	MYO3A, DFNB30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 31 607084, Otozomal Resesif; DFNB31 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (WHRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 31, 607084, Autosomal recessive; DFNB31 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (WHRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WHRN	WHRN, CIP98, KIAA1526, DFNB31, USH2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 35, 608565, Otozomal Resesif; DFNB35 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ESRRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 35, 608565, Autosomal recessive; DFNB35 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ESRRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESRRB	ESRRB, ESRL2, DFNB35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 35 (DFNB35) [MIM:608565]: A form of non-syndromic deafness characterized by non-progressive, prelingual hearing loss. {ECO:0000269	PubMed:18179891}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 36, 609006, Otozomal Resesif; DFNB36 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ESPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 36, 609006, Autosomal recessive; DFNB36 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ESPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESPN	ESPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 36, with or without vestibular involvement (DFNB36) [MIM:609006]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB36 is characterized by prelingual, profound hearing loss, and vestibular areflexia in some patients. {ECO:0000269|PubMed:15286153, ECO:0000269|PubMed:15930085}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 37, 607821, Otozomal Resesif; DFNB37 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MYO6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 37, 607821, Autosomal recessive; DFNB37 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MYO6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO6	MYO6, DFNA22, DFNB37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 39, 608265, Otozomal Resesif; DFNB39 (DFNB) (HGF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 39, 608265, Autosomal recessive; DFNB39 (DFNB) (HGF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HGF	HGF, DFNB39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 39 (DFNB39) [MIM:608265]: A form of profound prelingual sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:19576567}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Genişlemiş Vestibüler Kanal, Otozomal Resesif 4, 600791, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (SLC26A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (SLC26A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A4	SLC26A4, PDS, DFNB4, EVA, TDH2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 4 (DFNB4) [MIM:600791]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB4 is associated with an enlarged vestibular aqueduct. {ECO:0000269|PubMed:10190331, ECO:0000269|PubMed:10700480, ECO:0000269|PubMed:11748854, ECO:0000269|PubMed:12676893, ECO:0000269|PubMed:14508505, ECO:0000269|PubMed:14679580, ECO:0000269|PubMed:19204907, ECO:0000269|PubMed:20198392, ECO:0000269|PubMed:20597900, ECO:0000269|PubMed:9500541}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Genişlemiş Vestibüler Kanal, Otozomal Resesif 4, 600791, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MLPA)	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct, 600791, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MLPA)	SLC26A4	SLC26A4, PDS, DFNB4, EVA, TDH2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Deafness, autosomal recessive, 4 (DFNB4) [MIM:600791]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB4 is associated with an enlarged vestibular aqueduct. {ECO:0000269|PubMed:10190331, ECO:0000269|PubMed:10700480, ECO:0000269|PubMed:11748854, ECO:0000269|PubMed:12676893, ECO:0000269|PubMed:14508505, ECO:0000269|PubMed:14679580, ECO:0000269|PubMed:19204907, ECO:0000269|PubMed:20198392, ECO:0000269|PubMed:20597900, ECO:0000269|PubMed:9500541}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 42, 609646, Otozomal Resesif; DFNB42 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ILDR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 42, 609646, Autosomal recessive; DFNB42 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ILDR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ILDR1	ILDR1, DFNB42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 44, 610154, Otozomal Resesif; DFNB44 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ADCY1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 44, 610154, Autosomal recessive; DFNB44 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ADCY1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADCY1	ADCY1, DFNB44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 44 (DFNB44) [MIM:610154]: A form of non-syndromic deafness characterized by prelingual profound hearing loss affecting all frequencies. {ECO:0000269	PubMed:24482543}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 48, 609439, Otozomal Resesif; DFNB48 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CIB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 48, 609439, Autosomal recessive; DFNB48 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CIB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIB2	CIB2, KIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1J (USH1J) [MIM:614869]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1), Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. {ECO:0000269	PubMed:23023331}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 49, 610153, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MARVELD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 49, 610153, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MARVELD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARVELD2	MARVELD2, MARVD2, TRIC, DFNB49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 49 (DFNB49) [MIM:610153]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:17186462}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 49; Dfnb49 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MARVELD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL RECESSIVE 49; DFNB49 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MARVELD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARVELD2	MARVELD2, MARVD2, TRIC, DFNB49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 49 (DFNB49) [MIM:610153]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:17186462}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sağırlık, Otozomal Resesif 53, 609706, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 53, 609706, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 53; Dfnb53 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL RECESSIVE 53; DFNB53 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sağırlık, Otozomal Resesif 59, 610220, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (DFNB59 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 59, 610220, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (DFNB59 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DFNB59	PJVK, DFNB59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 6, 600971, Otozomal Resesif; DFNB6 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TMIE gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 6, 600971, Autosomal recessive; DFNB6 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TMIE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMIE	TMIE, DFNB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 61, 613865, Otozomal Resesif; DFNB61 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (SLC26A5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 61, 613865, Autosomal recessive; DFNB61 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (SLC26A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A5	SLC26A5, PRES, DFNB61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 61 (DFNB61) [MIM:613865]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:12719379}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 63, 611451, Otozomal Resesif; DFNB63 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (LRTOMT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 63, 611451, Autosomal recessive; DFNB63 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (LRTOMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRTOMT	LRTOMT, LRTOMT1, LRTOMT2, DFNB63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 63 (DFNB63) [MIM:611451]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:18794526, ECO:0000269	PubMed:18953341}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 66, 610212, Otozomal Resesif; DFNB66 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (DCDC2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 66, 610212, Autosomal recessive; DFNB66 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (DCDC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCDC2	DCDC2, RU2, KIAA1154, NPHP19, DFNB66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 66 (DFNB66) [MIM:610212]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:25601850}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 67, 610265, Otozomal Resesif; DFNB67 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (LHFPL5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 67, 610265, Autosomal recessive; DFNB67 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (LHFPL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHFPL5	LHFPL5, TMHS, DFNB67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 67 (DFNB67) [MIM:610265]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:16459341, ECO:0000269	PubMed:16752389}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 68, 610419, Otozomal Resesif; DFNB68 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (S1PR2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 68, 610419, Autosomal recessive; DFNB68 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (S1PR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	S1PR2	S1PR2, EDG5, DFNB68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 7, 600974, Otozomal Resesif; DFNB7 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TMC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 7, 600974, Autosomal recessive; DFNB7 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TMC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMC1	TMC1, DFNB7, DFNB11, DFNA36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 7 (DFNB7) [MIM:600974]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:11850618}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 70, 614934, Otozomal Resesif; DFNB70 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (PNPT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 70, 614934, Autosomal recessive; DFNB70 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (PNPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPT1	PNPT1, OLD35, COXPD13, DFNB70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 74, 613718, Otozomal Resesif; DFNB74 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MSRB3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 74, 613718, Autosomal recessive; DFNB74 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MSRB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSRB3	MSRB3, DFNB74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 76, 615540, Otozomal Resesif; DFNB76 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (SYNE4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 76, 615540, Autosomal recessive; DFNB76 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (SYNE4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNE4	SYNE4, NESP4, C19orf46, DFNB76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 76 (DFNB76) [MIM:615540]: A form of non-syndromic sensorineural deafness, a disorder resulting from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB76 affected individuals have onset of progressive high frequency hearing impairment between birth and 6 years of age. The hearing loss is severe at high frequencies by adulthood. {ECO:0000269	PubMed:23348741}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 77, 613079, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (LOXHD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 77, 613079, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (LOXHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LOXHD1	LOXHD1, DFNB77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 77 (DFNB77) [MIM:613079]: A form of non-syndromic deafness characterized by preserved low- frequency hearing, and a trend toward mild to moderate mid- frequency and high-frequency hearing loss during childhood and adolescence. Hearing loss progresses to become moderate to severe at mid and high frequencies during adulthood. {ECO:0000269	PubMed:19732867}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 79, 613307, Otozomal Resesif; DFNB79 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TPRN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 79, 613307, Autosomal recessive; DFNB79 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TPRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPRN	TPRN, C9orf75, DFNB79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 8/10, 601072, Otozomal Resesif; DFNB8 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TMPRSS3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 8/10, 601072, Autosomal recessive; DFNB8 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TMPRSS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMPRSS3	TMPRSS3, ECHOS1, DFNB8, DFNB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 8 (DFNB8) [MIM:601072]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:11424022, ECO:0000269|PubMed:11462234, ECO:0000269|PubMed:11907649, ECO:0000269|PubMed:12393794, ECO:0000269|PubMed:16021470}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 84A, 613391, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (PTPRQ gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 84A, 613391, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (PTPRQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRQ	PTPRQ, PTPGMC1, DFNB84A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 84A (DFNB84A) [MIM:613391]: A form of non-syndromic deafness characterized by progressive, sensorineural hearing loss and vestibular dysfunction. {ECO:0000269	PubMed:20346435, ECO:0000269	PubMed:20472657}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 84a; Dfnb84a (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (PTPRQ gen) (Tüm Gen Dizi Analizi) (Postnatal)	DEAFNESS, AUTOSOMAL RECESSIVE 84A; DFNB84A (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (PTPRQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRQ	PTPRQ, PTPGMC1, DFNB84A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 84A (DFNB84A) [MIM:613391]: A form of non-syndromic deafness characterized by progressive, sensorineural hearing loss and vestibular dysfunction. {ECO:0000269	PubMed:20346435, ECO:0000269	PubMed:20472657}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Otozomal Resesif 84B, 614944, Otozomal Resesif; DFNB84B (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (OTOGL gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 84B, 614944, Autosomal recessive; DFNB84B (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (OTOGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTOGL	OTOGL, C12orf64, DFNB84B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 86, 614617, Otozomal Resesif; DFNB86 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TBC1D24 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 86, 614617, Autosomal recessive; DFNB86 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 88, 615429, Otozomal Resesif; DFNB88 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ELMOD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 88, 615429, Autosomal recessive; DFNB88 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ELMOD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELMOD3	ELMOD3, RBED1, DFNB88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 89, 613916, Otozomal Resesif; DFNB89 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (KARS gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 89, 613916, Autosomal recessive; DFNB89 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (KARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KARS	KARS, CMTRIB, DFNB89	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 9, 601071, Otozomal Resesif; DFNB9 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (OTOF gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 9, 601071, Autosomal recessive; DFNB9 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (OTOF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTOF	OTOF, DFNB9, NSRD9, AUNB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 9 (DFNB9) [MIM:601071]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:10192385, ECO:0000269|PubMed:12127154, ECO:0000269|PubMed:16097006, ECO:0000269|PubMed:16283880, ECO:0000269|PubMed:16371502}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 91, 613453, Otozomal Resesif; DFNB91 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (SERPINB6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 91, 613453, Autosomal recessive; DFNB91 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (SERPINB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINB6	SERPINB6, PI6, PTI, SPI3, DFNB91	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 93, 614899, Otozomal Resesif; DFNB93 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (CABP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 93, 614899, Autosomal recessive; DFNB93 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (CABP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CABP2	CABP2, DFNB93	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Otozomal Resesif 97, 616705, Otozomal Resesif; DFNB97 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MET gen) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 97, 616705, Autosomal recessive; DFNB97 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MET	MET, DFNB97, OSFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sağırlık, Otozomal Resesif 98, 614861, Otozomal Resesif; DFNB98 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (TSPEAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, autosomal recessive 98, 614861, Autosomal recessive; DFNB98 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (TSPEAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSPEAR	TSPEAR, C21orf29, DFNB98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 98 (DFNB98) [MIM:614861]: A form of non-syndromic sensorineural hearing loss with prelingual onset. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:22678063}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Konjenital Kalp Defekti, ve Posterior Embryotokson (Alagille Sendromu) (JAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, congenital heart defects, and posterior embryotoxon (Alagille syndrome) (JAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAG1	JAG1, AGS1, AHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Konjenital Kalp Defekti, ve Posterior Embryotokson (Alagille Sendromu) (MLPA)	Deafness, congenital heart defects, and posterior embryotoxon (Alagille syndrome) (MLPA)	JAG1	JAG1, AGS1, AHD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sağırlık, Konjenital, İç Kulak Agenezisi, Mikrotİ ve Mikrodontİ, 610706, Otozomal Resesif (Labirent Aplazisi- Mikrotİ-Mikrodontİ-Sağırlık) (FGF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706, Autosomal recessive (Deafness with labyrinthine aplasia, microtia, and microdontia) (FGF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF3	FGF3, INT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness with labyrinthine aplasia, microtia and microdontia (LAMM) [MIM:610706]: Unique autosomal recessive syndrome characterized by type I microtia, microdontia, and profound congenital deafness associated with a complete absence of inner ear structures (Michel aplasia). {ECO:0000269|PubMed:17236138, ECO:0000269|PubMed:18435799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, Konjenital, Onikodistrofİ, Otozomal Dominant, 124480, Otozomal Dominant; DOOD (Sağırlık-Onikodistrofİ Sendromu) (ATP6V1B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, congenital, with onychodystrophy, autosomal dominant, 124480, Autosomal dominant; DOOD (Deafness–onychodystrophy syndrome) (ATP6V1B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V1B2	ATP6B1B2, ATP6B2, VPP3, DOOD, ZLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Dijenik GJB2 / GJB6, 220290, Otozomal Resesif, Digenic Dominant (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, digenic GJB2/GJB6, 220290, Autosomal recessive, Digenic dominant (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Dijenik, GJB2 / GJB3, 220290, Otozomal Resesif, Dijenik Dominant (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (GJB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, digenic, GJB2/GJB3, 220290, Autosomal recessive, Digenic dominant (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Dijenik, GJB2 / GJB3, 220290, Otozomal Resesif, Dijenik Dominant (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (MLPA)	Deafness, digenic, GJB2/GJB3, 220290, Autosomal recessive, Digenic dominant (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (MLPA)	GJB3	GJB3, CX31, DFNA2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sağırlık, Distoni ve Serebral Hipomyelinizasyon, 300475, X'e Bağlı (CADDS) (BCAP31 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, dystonia, and cerebral hypomyelination, 300475, X-linked recessive (CADDS) (BCAP31 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAP31	BCAP31, BAP31, DXS1357E, DDCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, dystonia, and cerebral hypomyelination (DDCH) [MIM:300475]: An X-linked recessive mental retardation syndrome characterized by almost no psychomotor development, dysmorphic facial features, sensorineural deafness, dystonia, pyramidal signs, and hypomyelination on brain imaging. {ECO:0000269	PubMed:24011989}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağırlık, Distoni ve Serebral Hipomyelinizasyon, 300475, X'e Bağlı (CADDS) (Prenatal) (BCAP31 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Deafness, dystonia, and cerebral hypomyelination, 300475, X-linked recessive (CADDS) (BCAP31 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCAP31	BCAP31, BAP31, DXS1357E, DDCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Deafness, dystonia, and cerebral hypomyelination (DDCH) [MIM:300475]: An X-linked recessive mental retardation syndrome characterized by almost no psychomotor development, dysmorphic facial features, sensorineural deafness, dystonia, pyramidal signs, and hypomyelination on brain imaging. {ECO:0000269	PubMed:24011989}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sağırlık, Mitokondriyal, Modifiye Edici, 580000, Mitokondriyal (Mitokondriyal Sendromik Olmayan Sensorinöral Sağırlık, Aminoglikozid Maruziyetine Bağlı) (TRMU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, mitochondrial, modifier of, 580000, Mitochondrial (Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure) (TRMU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRMU	TRMU, MTO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, Nörosensor, Vestibüler Tutulum Olmaksızın, Otozomal Dominant (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (ESPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, neurosensory, without vestibular involvement, autosomal dominant (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (ESPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESPN	ESPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, X'e Bağlı 1, 304500, X'e Bağlı; DFNX1 (X'e Bağlı Sendromik Olmayan Sensorinöral İşitme Kaybıtip DFN) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, X-linked 1, 304500, X-linked; DFNX1 (X-linked non-syndromic sensorineural deafness type DFN) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, X'e Bağlı 2, 304400, X'e Bağlı Resesif; DFNX2 (X'e Bağlı Miks Tip Sağırlık - Perilenfatik Guşer) (POU3F4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, X-linked 2, 304400, X-linked recessive; DFNX2 (X-linked mixed deafness with perilymphatic gusher) (POU3F4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POU3F4	POU3F4, DFN3, DFNX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, X'e Bağlı 4, 300066, X'e Bağlı Dominant; DFNX4 (X'e Bağlı Sendromik Olmayan Sensorinöral İşitme Kaybıtip DFN) (SMPX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, X-linked 4, 300066, X-linked dominant; DFNX4 (X-linked non-syndromic sensorineural deafness type DFN) (SMPX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMPX	SMPX, DFNX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, X-linked, 4 (DFNX4) [MIM:300066]: A non-syndromic form of sensorineural, progressive hearing loss with postlingual onset. In affected males, the auditory impairment affects initially high-frequency hearing. It later evolves to become severe to profound and affects all frequencies. Carrier females manifest moderate hearing impairment in the high frequencies. {ECO:0000269|PubMed:21549338, ECO:0000269|PubMed:21549342, ECO:0000269|PubMed:22911656}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağırlık, X'e Bağlı 5, 300614, X'e Bağlı Resesif; DFNX5 (X'e Bağlı Kalıtsal Duysal ve Otonom Nöropati-Sağırlık) (AIFM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deafness, X-linked 5, 300614, X-linked recessive; DFNX5 (X-linked hereditary sensory and autonomic neuropathy with deafness) (AIFM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIFM1	AIFM1, PDCD8, AIF, COXPD6, COWCK, CMTX4, DFNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sağırlık, X'e Bağlı, 6, 300914, X'e Bağlı; DFNX6 (X'e Bağlı Sendromik Olmayan Sensorinöral İşitme Kaybıtip DFN) (2200)	Deafness, X-linked 6, 300914, X-linked recessive; DFNX6 (X-linked non-syndromic sensorineural deafness type DFN) (440)	COL4A6	COL4A6, DELXq22.3, CXDELq22.3, DFNX6	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	DISEASE: Deafness, X-linked, 6 (DFNX6) [MIM:300914]: A non-syndromic form of sensorineural hearing loss with prelingual onset. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269	PubMed:23714752}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample and Heparinized blood sample
DEB testi (Fankoni anemisi)	DEB test (Fanconi anemia)	.	.	Kromozom analizi/ Karyotype analysis	Heparinli Kan (2-4 ml)		15	45	.	.	21	Postnatal	.	.	Heparinized blood sample
Debrisokuin Sensitivity, 608902, Otozomal Resesif (CYP2D6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Debrisoquine sensitivity, 608902, Autosomal recessive (CYP2D6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2D6	CYP2D6, CPD6, P450DB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Debrisokuin Sensitivity, 608902, Otozomal Resesif (MLPA)	Debrisoquine sensitivity, 608902, Autosomal recessive (MLPA)	CYP2D6	CYP2D6, CPD6, P450DB1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Derin Ven Tromboz, Karşı Koruma, 300807 (F9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Deep venous thrombosis, protection against, 300807 (F9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F9	F9, HEMB, THPHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia, X-linked, due to factor IX defect (THPH6) [MIM:300807]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269	PubMed:19846852}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Derin Ven Tromboz, Karşı Koruma, 300807 (MLPA)	Deep venous thrombosis, protection against, 300807 (MLPA)	F9	F9, HEMB, THPHB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Thrombophilia, X-linked, due to factor IX defect (THPH6) [MIM:300807]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269	PubMed:19846852}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dehidrate Kalıtsal Stomatositozis 2, 616689, Otozomal Dominant; DHS2 (Dehidrate Kalıtsal Stomatositoz) (KCNN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dehydrated hereditary stomatocytosis 2, 616689, Autosomal dominant; DHS2 (Dehydrated hereditary stomatocytosis) (KCNN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNN4	KCNN4, KCA4, SK4, DHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:26148990, ECO:0000269|PubMed:26178367, ECO:0000269|PubMed:26198474}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dehidrate Kalıtsal Stomatositozis 2, 616689, Otozomal Dominant; DHS2 (Dehidrate Kalıtsal Stomatositoz) (Prenatal) (KCNN4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dehydrated hereditary stomatocytosis 2, 616689, Autosomal dominant; DHS2 (Dehydrated hereditary stomatocytosis) (KCNN4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNN4	KCNN4, KCA4, SK4, DHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:26148990, ECO:0000269|PubMed:26178367, ECO:0000269|PubMed:26198474}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Dehidrate Kalıtsal Stomatositozis, ± Psödohiperkalemi ve / Veya Perinatal Ödem, 194380, Otozomal Dominant; DHS1 (Dehidrate Kalıtsal Stomatositoz) (PIEZO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380, Autosomal dominant; DHS1 (Dehydrated hereditary stomatocytosis) (PIEZO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO1	PIEZO1, FAM38A, MIB, DHS, LMPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:22529292, ECO:0000269|PubMed:23479567, ECO:0000269|PubMed:23487776, ECO:0000269|PubMed:23581886, ECO:0000269|PubMed:23695678, ECO:0000269|PubMed:23973043}. Note=The disease is caused by mutations affecting the gene represented in this entry. All disease-causing mutations characterized so far produce a gain-of- function phenotype, mutated channels exhibiting increased cation transport in erythroid cells, that could be due to slower channel inactivation rate compared to the wild-type protein.,	EDTA Blood Sample
Dehidrate Kalıtsal Stomatositozis, ± Psödohiperkalemi ve / Veya Perinatal Ödem, 194380, Otozomal Dominant; DHS1 (Dehidrate Kalıtsal Stomatositoz) (Prenatal) (PIEZO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380, Autosomal dominant; DHS1 (Dehydrated hereditary stomatocytosis) (PIEZO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO1	PIEZO1, FAM38A, MIB, DHS, LMPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:22529292, ECO:0000269|PubMed:23479567, ECO:0000269|PubMed:23487776, ECO:0000269|PubMed:23581886, ECO:0000269|PubMed:23695678, ECO:0000269|PubMed:23973043}. Note=The disease is caused by mutations affecting the gene represented in this entry. All disease-causing mutations characterized so far produce a gain-of- function phenotype, mutated channels exhibiting increased cation transport in erythroid cells, that could be due to slower channel inactivation rate compared to the wild-type protein.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Deiyodinaz İyodotironin, Tip I, Dio1 (DIO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DEIODINASE, IODOTHYRONINE, TYPE I; DIO1 (DIO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIO1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (PRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (PRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRX	PRX, CMT4F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (EGR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (EGR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGR2	EGR2, KROX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MLPA)	PRX	PRX, CMT4F	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MLPA)	EGR2	EGR2, KROX20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (PRX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (PRX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRX	PRX, CMT4F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (PMP22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269|PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (MPZ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (MPZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (EGR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (EGR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EGR2	EGR2, KROX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (Prenatal) (MLPA)	PRX	PRX, CMT4F	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (Prenatal) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (Prenatal) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:11133365}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dejerine-Sottas Hastalığı, 145900, Otozomal Resesif, Otozomal Dominant (Dejerine-Sottas Sendromu) (Prenatal) (MLPA)	Dejerine-Sottas disease, 145900, Autosomal recessive, Autosomal dominant (Dejerine-Sottas syndrome) (Prenatal) (MLPA)	EGR2	EGR2, KROX20	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Delta-Beta Talasemi, 141749, Otozomal Dominant (Delta-Beta-Talasemi) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Delta-beta thalassemia, 141749, Autosomal dominant (Delta-beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Delta-Beta Talasemi, 141749, Otozomal Dominant (Delta-Beta-Talasemi) (MLPA)	Delta-beta thalassemia, 141749, Autosomal dominant (Delta-beta-thalassemia) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Delta-Beta Talasemi, 141749, Otozomal Dominant (Delta-Beta-Talasemi) (Prenatal) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Delta-beta thalassemia, 141749, Autosomal dominant (Delta-beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Delta-Beta Talasemi, 141749, Otozomal Dominant (Delta-Beta-Talasemi) (Prenatal) (MLPA)	Delta-beta thalassemia, 141749, Autosomal dominant (Delta-beta-thalassemia) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Demans, Ailesel İngiliz 176500, Otozomal Dominant (ITM2B Amiloidoz) (ITM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, familial British, 176500, Autosomal dominant (ITM2B amyloidosis) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. {ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362}. Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. {ECO:0000269|PubMed:10781099}.	EDTA Blood Sample
Demans, Ailesel Danimarka Tipi, 117300, Otozomal Dominant (ITM2B Amiloidoz) (ITM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, familial Danish, 117300, Autosomal dominant (ITM2B amyloidosis) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. {ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362}. Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. {ECO:0000269|PubMed:10781099}.	EDTA Blood Sample
Demans, Ailesel, Nonspesifik, 600795, Otozomal Dominant, FTD3 (Frontotemporal Demans Davranışsal Varyant) (CHMP2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, familial, nonspecific, 600795, Autosomal dominant, FTD3 (Behavioral variant of frontotemporal dementia) (CHMP2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHMP2B	CHMP2B, DMT1, VPS2B, ALS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frontotemporal dementia, chromosome 3-linked (FTD3) [MIM:600795]: Characterized by an onset of dementia in the late 50's initially characterized by behavioral and personality changes including apathy, restlessness, disinhibition and hyperorality, progressing to stereotyped behaviors, non-fluent aphasia, mutism and dystonia, with a marked lack of insight. The brains of individuals with FTD3 have no distinctive neuropathological features. They show global cortical and central atrophy, but no beta-amyloid deposits. {ECO:0000269	PubMed:16041373}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Demans, Frontotemporal, 600274, Otozomal Dominant (Frontotemporal Demans Davranışsal Varyant) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, frontotemporal, 600274, Autosomal dominant(Behavioral variant of frontotemporal dementia) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frontotemporal dementia (FTD) [MIM:600274]: A form of dementia characterized by pathologic finding of frontotemporal lobar degeneration, presenile dementia with behavioral changes, deterioration of cognitive capacities and loss of memory. In some cases, parkinsonian symptoms are prominent. Neuropathological changes include frontotemporal atrophy often associated with atrophy of the basal ganglia, substantia nigra, amygdala. In most cases, protein tau deposits are found in glial cells and/or neurons. {ECO:0000269	PubMed:11094121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Demans, Frontotemporal, ± Parkinsonizm, 600274, Otozomal Dominant (Frontotemporal Demans) (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, frontotemporal, with or without parkinsonism, 600274, Autosomal dominant (Frontotemporal dementia) (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Demans, Frontotemporal, ± Parkinsonizm, 600274, Otozomal Dominant (Frontotemporal Demans) (MLPA)	Dementia, frontotemporal, with or without parkinsonism, 600274, Autosomal dominant (Frontotemporal dementia) (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Demans, Lewy Cismi, 127750, Otozomal Dominant (SNCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, Lewy body, 127750, Autosomal dominant (SNCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNCA	SNCA, NACP, PARK1, PARK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dementia Lewy body (DLB) [MIM:127750]: A neurodegenerative disorder characterized by mental impairment leading to dementia, parkinsonism, fluctuating cognitive function, visual hallucinations, falls, syncopal episodes, and sensitivity to neuroleptic medication. Brainstem or cortical intraneuronal accumulations of aggregated proteins (Lewy bodies) are the only essential pathologic features. Patients may also have hippocampal and neocortical senile plaques, sometimes in sufficient number to fulfill the diagnostic criteria for Alzheimer disease. {ECO:0000269	PubMed:14755719}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Demans, Lewy Cismi, 127750, Otozomal Dominant: DLB (SNCB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, Lewy body, 127750, Autosomal dominant: DLB (SNCB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNCB	SNCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Demans, Vaskiler, Yatkınlık (TNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dementia, vascular, susceptibility to (TNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNF	TNF, TNFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dengue Humması, Karşı Koruma, 614371 (Dang Ateşi) (CD209 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dengue fever, protection against, 614371 (Dengue fever) (CD209 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD209	CD209, CDSIGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Dent Hastalığı 2, 300555, X'e Bağlı (Dent Hastalığı) (OCRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dent disease 2, 300555, X-linked recessive (Dent disease) (OCRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCRL	OCRL, LOCR, OCRL1, NPHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dent disease 2 (DD2) [MIM:300555]: A renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Characteristic abnormalities include low-molecular-weight proteinuria and other features of Fanconi syndrome, such as glycosuria, aminoaciduria, and phosphaturia, but typically do not include proximal renal tubular acidosis. Progressive renal failure is common, as are nephrocalcinosis and kidney stones. {ECO:0000269|PubMed:15627218, ECO:0000269|PubMed:17384968, ECO:0000269|PubMed:21031565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dent Hastalığı 2, 300555, X'e Bağlı (Dent Hastalığı) (Prenatal) (OCRL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dent disease 2, 300555, X-linked recessive (Dent disease) (OCRL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OCRL	OCRL, LOCR, OCRL1, NPHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dent disease 2 (DD2) [MIM:300555]: A renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Characteristic abnormalities include low-molecular-weight proteinuria and other features of Fanconi syndrome, such as glycosuria, aminoaciduria, and phosphaturia, but typically do not include proximal renal tubular acidosis. Progressive renal failure is common, as are nephrocalcinosis and kidney stones. {ECO:0000269|PubMed:15627218, ECO:0000269|PubMed:17384968, ECO:0000269|PubMed:21031565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dent Hastalığı, 300009, X'e Bağlı (Dent Hastalığı) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dent disease, 300009, X-linked recessive (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. {ECO:0000269|PubMed:15086899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8558248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dent Hastalığı, 300009, X'e Bağlı (Dent Hastalığı) (Prenatal) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dent diseaase, 300009, X-linked recessive (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. {ECO:0000269|PubMed:15086899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8558248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dış Anomaliler ve Kısa Boy, 601216, Otozomal Resesif; DASS (Brakidilmi-Amelogenezis Imperfekta Sendromu) (LTBP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dental anomalies and short stature, 601216, Autosomal recessive; DASS (Brachydilmia-amelogenesis imperfecta syndrome) (LTBP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTBP3	LTBP3, LTBP2, DASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dental anomalies and short stature (DASS) [MIM:601216]: A disorder characterized by hypoplastic amelogenesis imperfecta, significant short stature, brachydilmia-like anomalies including platyspondyly with short pedicles, narrow intervertebral and interpedicular distances, rectangular-shaped vertebrae with posterior scalloping and herniation of the nuclei, and broad femoral necks. Dental anomalies include widely spaced, small, yellow teeth, oligodontia, and severely reduced to absent enamel. {ECO:0000269|PubMed:19344874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dentin Displazi, Tip 1, Mikrodonti ve Diğerde Şekil Düzensizliği, 125400, Otozomal Resesif; DTDP1 (Dentin Displazi) (SMOC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400, Autosomal recessive; DTDP1 (Dentin dysplasia) (SMOC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMOC2	SMOC2, SMAP2, DTDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dentin dysplasia 1 (DTDP1) [MIM:125400]: A dental defect in which both primary and secondary dentitions are affected. The clinical crowns of both permanent and deciduous teeth are of normal shape, form and color in most cases, although they may be slightly opalescent and blue or brown. Teeth may be very mobile and exfoliate spontaneously because of inadequate root formation. On radiographs, the roots are short and may be more pointed than normal. Pulp chambers are usually absent except for a chevron- shaped remnant in the crown. Root canals are usually absent. {ECO:0000269|PubMed:22152679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dentin Displazi, Tip 2, 125420, Otozomal Dominant; DTDP2 (Dentin Displazi) (DSPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dentin dysplasia, type II, 125420, Autosomal dominant; DTDP2 (Dentin dysplasia) (DSPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSPP	DSPP, DPP, DGI1, DFNA39, DTDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dentin dysplasia 2 (DTDP2) [MIM:125420]: A dental defect in which the deciduous teeth are opalescent. The permanent teeth are of normal shape, form, and color in most cases. The root length is normal. On radiographs, the pulp chambers of permanent teeth are obliterated, have a thistle-tube deformity and contain pulp stones. {ECO:0000269|PubMed:12354781, ECO:0000269|PubMed:18521831}. Note=The disease is caused by mutations affecting the gene represented in this entry. DSPP defects causing dentin abnormalities act in a dominant negative manner and include missense, splice-site, frameshift mutations. 5' frameshift mutations cause dentin dysplasia while frameshift mutations at the 3' end cause the more severe dentinogenesis imperfecta phenotype (PubMed:18521831, PubMed:22392858). {ECO:0000269|PubMed:18521831, ECO:0000269|PubMed:22392858}.;	EDTA Blood Sample
Dentinogenezis Imperfekta, Shields Tip 2, 125490, Otozomal Dominant (Dentinogenezis Imperfekta Type 2) (DSPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dentinogenesis imperfecta, Shields type II, 125490, Autosomal dominant (Dentinogenesis imperfecta type 2) (DSPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSPP	DSPP, DPP, DGI1, DFNA39, DTDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Dentinogenezis Imperfekta, Shields Tip 3, 125500, Otozomal Dominant (Dentinogenezis Imperfekta Type 3) (DSPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dentinogenesis imperfecta, Shields type III, 125500, Autosomal dominant (Dentinogenesis imperfecta type 3) (DSPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSPP	DSPP, DPP, DGI1, DFNA39, DTDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Denys-Drash Sendromu, 194080, Otozomal Dominant, Somatik Mutasyon; DDS (Denys-Drash Sendromu) (WT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Denys-Drash syndrome, 194080, Autosomal dominant, Somatic mutation; DDS (Denys-Drash syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Denys-Drash Sendromu, 194080, Otozomal Dominant, Somatik Mutasyon; DDS (Denys-Drash Sendromu) (MLPA)	Denys-Drash syndrome, 194080, Autosomal dominant, Somatic mutation; DDS (Denys-Drash syndrome) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)/ Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		20	60	.	.	30	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Denys-Drash Sendromu, 194080, Otozomal Dominant, Somatik Mutasyon; DDS (Denys-Drash Sendromu) (Prenatal) (WT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Denys-Drash syndrome, 194080, Autosomal dominant, Somatic mutation; DDS (Denys-Drash syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Denys-Drash Sendromu, 194080, Otozomal Dominant, Somatik Mutasyon; DDS (Denys-Drash Sendromu) (MLPA)	Denys-Drash syndrome, 194080, Autosomal dominant, Somatic mutation; DDS (Denys-Drash syndrome) (Prenatal) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Atopik Dermatit, Yatkınlık 2, 605803 (FLG geni) (Tüm Gen Dizi Analizi) (Postnatal) K.E.	Dermatitis, atopic, susceptibility to, 2, 605803 (FLG gene) (Sequence Analysis-All Coding Exons) (Postnatal) K.E.	FLG	FLG, ATOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dermatofibrosarkoma Protuberans, 607907; DFSP (Dermatofibrosarkoma Protuberans) (PDGFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dermatofibrosarcoma protuberans, 607907; DFSP (Dermatofibrosarcoma protuberans) (PDGFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFB	PDGFB, SIS, IBGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving PDGFB is found in dermatofibrosarcoma protuberans. Translocation t(17;22)(q22;q13) with PDGFB. {ECO:0000269|PubMed:12660034}.;	EDTA Blood Sample
Dermatofibrosarkoma Protuberans, 607907; DFSP (Dermatofibrosarkoma Protuberans) (Prenatal) (PDGFB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dermatofibrosarcoma protuberans, 607907; DFSP (Dermatofibrosarcoma protuberans) (PDGFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDGFB	PDGFB, SIS, IBGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving PDGFB is found in dermatofibrosarcoma protuberans. Translocation t(17;22)(q22;q13) with PDGFB. {ECO:0000269|PubMed:12660034}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dermatopati Pigmentosa Retikülaris, 125595, Otozomal Dominant; DPR (Dermatopatia Pigmentosa Retikularis) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dermatopathia pigmentosa reticularis, 125595, Autosomal dominant; DPR (Dermatopathia pigmentosa reticularis) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dermatopati Pigmentosa Retikülaris, 125595, Otozomal Dominant; DPR (Dermatopatia Pigmentosa Retikularis) (MLPA)	Dermatopathia pigmentosa reticularis, 125595, Autosomal dominant; DPR (Dermatopathia pigmentosa reticularis) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Desanto-Shinawi Sendromu, 616708, Otozomal Dominant (Fasiyal Dismorfizm-Gelişimsel Gecikme-Davranışsal Anormallikler Sendromu, 10p11.21p12.31 Mikrodelesyonuna Bağlı) (WAC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Desanto-Shinawi syndrome, 616708, Autosomal dominant (Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion) (WAC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WAC	WAC, KIAA1844, DESSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Desanto-Shinawi Sendromu, 616708, Otozomal Dominant (Fasiyal Dismorfizm-Gelişimsel Gecikme-Davranışsal Anormallikler Sendromu, 10p11.21p12.31 Mikrodelesyonuna Bağlı) (Prenatal) (WAC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Desanto-Shinawi syndrome, 616708, Autosomal dominant (Facial dysmorphism-developmental delay-behavioral abnormalities syndrome due to 10p11.21p12.31 microdeletion) (WAC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WAC	WAC, KIAA1844, DESSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Desbuquois Displazi 1, 251450, Otozomal Resesif, DBQD1 (Desbuquois Displazi) (CANT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Desbuquois dysplasia 1, 251450, Autosomal recessive; DBQD1 (Desbuquois syndrome) (CANT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CANT1	CANT1, SCAN1, DBQD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Desbuquois dysplasia 1 (DBQD1) [MIM:251450]: A chondrodysplasia characterized by severe prenatal and postnatal growth retardation (less than -5 SD), joint laxity, short extremities, progressive scoliosis, round face, midface hypoplasia, prominent bulging eyes. The main radiologic features are short long bones with metaphyseal splay, a 'Swedish key' appearance of the proximal femur (exaggerated trochanter), and advance carpal and tarsal bone age. Two forms of Desbuquois dysplasia are distinguished on the basis of the presence or absence of characteristic hand anomalies: an extra ossification center distal to the second metacarpal, delta phalanx, bifid distal thumb phalanx, and phalangeal dislocations. {ECO:0000269|PubMed:19853239, ECO:0000269|PubMed:20425819, ECO:0000269|PubMed:21037275, ECO:0000269|PubMed:21412251, ECO:0000269|PubMed:21654728, ECO:0000269|PubMed:22539336}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Desbuquois Displazi 1, 251450, Otozomal Resesif, DBQD1 (Desbuquois Displazi) (Prenatal) (CANT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Desbuquois dysplasia 1, 251450, Autosomal recessive; DBQD1 (Desbuquois syndrome) (CANT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CANT1	CANT1, SCAN1, DBQD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Desbuquois dysplasia 1 (DBQD1) [MIM:251450]: A chondrodysplasia characterized by severe prenatal and postnatal growth retardation (less than -5 SD), joint laxity, short extremities, progressive scoliosis, round face, midface hypoplasia, prominent bulging eyes. The main radiologic features are short long bones with metaphyseal splay, a 'Swedish key' appearance of the proximal femur (exaggerated trochanter), and advance carpal and tarsal bone age. Two forms of Desbuquois dysplasia are distinguished on the basis of the presence or absence of characteristic hand anomalies: an extra ossification center distal to the second metacarpal, delta phalanx, bifid distal thumb phalanx, and phalangeal dislocations. {ECO:0000269|PubMed:19853239, ECO:0000269|PubMed:20425819, ECO:0000269|PubMed:21037275, ECO:0000269|PubMed:21412251, ECO:0000269|PubMed:21654728, ECO:0000269|PubMed:22539336}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Desbuquois Displazi 2, 615777, Otozomal Resesif, DBQD2 (Desbuquois Displazi) (XYLT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Desbuquois dysplasia 2, 615777, Autosomal recessive; DBQD2 (Desbuquois dysplasia syndrome) (XYLT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XYLT1	XYLT1, XT1, DBQD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Desbuquois Displazi 2, 615777, Otozomal Resesif, DBQD2 (Desbuquois Displazi) (Prenatal) (XYLT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Desbuquois dysplasia 2, 615777, Autosomal recessive; DBQD2 (Desbuquois dysplasia syndrome) (XYLT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XYLT1	XYLT1, XT1, DBQD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Desmoid Hastalığı, Kalıtsal, 135290, Otozomal Dominant (Desmoid Tümör) (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Desmoid disease, hereditary, 135290, Autosomal dominant (Desmoid tumor) (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Desmoid Hastalığı, Kalıtsal, 135290, Otozomal Dominant (Desmoid Tümör) (MLPA)	Desmoid disease, hereditary, 135290, Autosomal dominant (Desmoid tumor) (MLPA)	APC	APC, GS, FPC, BTPS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Desmosterolosis, 602398, Otozomal Resesif (Desmosterolosis) (DHCR24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Desmosterolosis, 602398, Autosomal recessive (Desmosterolosis) (DHCR24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHCR24	DHCR24, KIAA0018	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gelişme Geriliği, Boy Kısalığı, Dismorfik Özellikler ve Seyrek Saç, 616901, Otozomal Resesif (Kraniofasiyel Displazi-Boy Kısalığı-Ektodermal Anomaliler-Mental Retardasyon Sendromu) (DPH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901, Autosomal recessive (Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome) (DPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPH1	DPH1, DPH2L1, OVCA1, DEDSSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gelişme Geriliği, Boy Kısalığı, Dismorfik Özellikler ve Seyrek Saç, 616901, Otozomal Resesif (Kraniofasiyel Displazi-Boy Kısalığı-Ektodermal Anomaliler-Mental Retardasyon Sendromu) (Prenatal) (DPH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Developmental delay with short stature, dysmorphic features, and sparse hair, 616901, Autosomal recessive (Craniofacial dysplasia-short stature-ectodermal anomalies-intellectual disability syndrome) (DPH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPH1	DPH1, DPH2L1, OVCA1, DEDSSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyabetes Mellitus + Sağırlık, Maternal Kalıtım (Tüm Mitokondriyal Genom - MTTL1, MTTE, MTTK genleri)	Diabetes and Deafness, Maternally Inherited (Whole Mitochondrial Genome Analysis including MTTL1, MTTE, MTTK genes)	Mitokondrial Genom	.	Tüm Mitokondrial Genom Dizi Analizi/ Whome Mitochondrial Genome Sequence Analysis	EDTA'lı Kan (2-4 ml)/ Cilt-Kas-Sokil Doku Biyopsisi (Transport besi yeri içinde)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Skin-Muscle-Soft Tissue Biopsy (Transport Media)
Diyabet İnsipidus, Nefrojenik, 125800, Otozomal Resesif, Otozomal Dominant (Nefrojenik Diabetes İnsipidus) (AQP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes insipidus, nephrogenic, 125800, Autosomal recessive, Autosomal dominant (Nephrogenic diabetes insipidus) (AQP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP2	AQP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes insipidus, nephrogenic, autosomal (AND) [MIM:125800]: A disorder caused by the inability of the renal collecting ducts to absorb water in response to arginine vasopressin. Characterized by excessive water drinking (polydipsia), excessive urine excretion (polyuria), persistent hypotonic urine, and hypokalemia. Inheritance can be autosomal dominant or recessive. {ECO:0000269|PubMed:12050236, ECO:0000269|PubMed:12191971, ECO:0000269|PubMed:15509592, ECO:0000269|PubMed:16120822, ECO:0000269|PubMed:16361827, ECO:0000269|PubMed:16845277, ECO:0000269|PubMed:19585583, ECO:0000269|PubMed:24944815, ECO:0000269|PubMed:7524315, ECO:0000269|PubMed:8882880, ECO:0000269|PubMed:9048343, ECO:0000269|PubMed:9302264, ECO:0000269|PubMed:9402087, ECO:0000269|PubMed:9550615, ECO:0000269|PubMed:9649557, ECO:0000269|PubMed:9745427}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabet İnsipidus, Nefrojenik, 304800, X'e Bağlı (Nefrojenik Diabetes İnsipidus) (AVPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes insipidus, nephrogenic, 304800, X-linked recessive (Nephrogenic diabetes insipidus) (AVPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AVPR2	AVPR2, DIR, DI1, ADHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes insipidus, nephrogenic, X-linked (XNDI) [MIM:304800]: A disorder caused by the inability of the renal collecting ducts to absorb water in response to arginine vasopressin. Characterized by excessive water drinking (polydipsia), excessive urine excretion (polyuria), persistent hypotonic urine, and hypokalemia. {ECO:0000269|PubMed:10694923, ECO:0000269|PubMed:10770218, ECO:0000269|PubMed:11232028, ECO:0000269|PubMed:11916004, ECO:0000269|PubMed:1303257, ECO:0000269|PubMed:1303271, ECO:0000269|PubMed:1358229, ECO:0000269|PubMed:16845277, ECO:0000269|PubMed:7560098, ECO:0000269|PubMed:7833930, ECO:0000269|PubMed:7984150, ECO:0000269|PubMed:7987330, ECO:0000269|PubMed:7999078, ECO:0000269|PubMed:8037205, ECO:0000269|PubMed:8045948, ECO:0000269|PubMed:8078903, ECO:0000269|PubMed:8267567, ECO:0000269|PubMed:8479490, ECO:0000269|PubMed:8514744, ECO:0000269|PubMed:9402087, ECO:0000269|PubMed:9452109, ECO:0000269|PubMed:9711877}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabet İnsipidus, Nörohipofizyal, 125700, Otozomal Dominant (Santral Diabetes İnsipitus) (AVP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes insipidus, neurohypophyseal, 125700, Autosomal dominant (Central diabetes insipidus) (AVP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AVP	AVP, AVRP, VP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes insipidus, neurohypophyseal (NDI) [MIM:125700]: A disease characterized by persistent thirst, polydipsia and polyuria. Affected individuals are apparently normal at birth, but characteristically develop symptoms of vasopressin deficiency during childhood. {ECO:0000269|PubMed:10369876, ECO:0000269|PubMed:10487710, ECO:0000269|PubMed:10677561, ECO:0000269|PubMed:11017955, ECO:0000269|PubMed:11150885, ECO:0000269|PubMed:11161827, ECO:0000269|PubMed:11443218, ECO:0000269|PubMed:11748489, ECO:0000269|PubMed:11980620, ECO:0000269|PubMed:12012274, ECO:0000269|PubMed:12107248, ECO:0000269|PubMed:12359138, ECO:0000269|PubMed:12519420, ECO:0000269|PubMed:12931042, ECO:0000269|PubMed:14510916, ECO:0000269|PubMed:14673472, ECO:0000269|PubMed:15538939, ECO:0000269|PubMed:1740104, ECO:0000269|PubMed:7714110, ECO:0000269|PubMed:8045958, ECO:0000269|PubMed:8103767, ECO:0000269|PubMed:8370682, ECO:0000269|PubMed:8514868, ECO:0000269|PubMed:8554046, ECO:0000269|PubMed:9360520, ECO:0000269|PubMed:9580132, ECO:0000269|PubMed:9814750}. Note=The disease is caused	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 12, 601388 (CTLA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 12, 601388 (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Diyabetes Mellitus, İnsülin Bağımlı, 2, 125852, Otozomal Dominant (INS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 2, 125852, Autosomal dominant (INS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INS	INS, MODY10, IDDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 2 (IDDM2) [MIM:125852]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:18192540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 20, 612520 (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 20, 612520 (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 20, 612520 (MLPA)	Diabetes mellitus, insulin-dependent, 20, 612520 (MLPA)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 13, 601318 (CCRS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 22, 612522 (CCRS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCRS	CCRS, CMKBR5, CCCKR5, IDDM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 22 (IDDM22) [MIM:612522]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:19073967}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 222100, Otozomal Resesif (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 222100, Autosomal recessive (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:1470163, ECO:0000269|PubMed:1607076, ECO:0000269|PubMed:7657032}. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 222100, Otozomal Resesif (MLPA)	Diabetes mellitus, insulin-dependent, 222100, Autosomal recessive (MLPA)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:1470163, ECO:0000269|PubMed:1607076, ECO:0000269|PubMed:7657032}. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, 5, 600320 (SUMO4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-dependent, 5, 600320 (SUMO4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUMO4	SUMO4, IDDM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Direnci, Akantozis Nigricans, 610549 (İnsülin Direnci Sendromu Tip A) (INSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549 (insulin-resistance syndrome type A) (INSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INSR	INSR, HHF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:1470163, ECO:0000269|PubMed:1607076, ECO:0000269|PubMed:7657032}. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Diyabetes Mellitus, Ketozis Eğilimli, Yatkınlık, 612227, Otozomal Dominant, KPD (PAX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, ketosis-prone, susceptibility to, 612227, Autosomal dominant; KPD (PAX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX4	PAX4, MODY9, KPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:11723072}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Konjenital Hipotiroidizm İle Diyabetes Mellitus, Neonatal, 610199, Otozomal Resesif; NDH (Neonatal Diyabet-Konjenital Hipotiroidizm-Konjenital Glokom-Hepatik Fibroz-Polikistik Böbrek Sendromu) (GLIS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199, Autosomal recessive; NDH (Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome) (GLIS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLIS3	GLIS3, ZNF515, NDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konjenital Hipotiroidizm İle Diyabetes Mellitus, Neonatal, 610199, Otozomal Resesif; NDH (Neonatal Diyabet-Konjenital Hipotiroidizm-Konjenital Glokom-Hepatik Fibroz-Polikistik Böbrek Sendromu) (Prenatal) (GLIS3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diabetes mellitus, neonatal, with congenital hypothyroidism, 610199, Autosomal recessive; NDH (Neonatal diabetes-congenital hypothyroidism-congenital glaucoma-hepatic fibrosis-polycystic kidneys syndrome) (GLIS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLIS3	GLIS3, ZNF515, NDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİS) BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, Yatkınlık, 125853, Otozomal Dominant (ENPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, non-insulin-dependent, susceptibility to, 125853, Autosomal dominant (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENPP1	ENPP1, PDNP1, NPPS, M6S1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:16186408}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan 1, 601283 (CAPN10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent 1, 601283 (CAPN10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAPN10	CAPN10, NIDDM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (SLC2A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (SLC2A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A2	SLC2A2, GLUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:16186408}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (MAPK8IP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (MAPK8IP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPK8IP1	MAPK8IP1, IB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:10700186}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (LIPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (LIPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPC	LIPC, HL, LIPH, HDLCQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:16186408}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (IRS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (IRS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRS2	IRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (IRS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (IRS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRS1	IRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:10206679, ECO:0000269|PubMed:12843189, ECO:0000269|PubMed:8723689}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (HNF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (HNF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (GCGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (GCGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCGR	GCGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (ABCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC8	ABCC8, SUR, PHH1, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:16186408}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (MLPA)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (MLPA)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (MODY) (NEUROD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (MODY) (NEUROD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEUROD1	NEUROD1, NIDDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 6 (MODY6) [MIM:606394]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269	PubMed:10545951}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (MODY) (HNF4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (MODY) (HNF4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 1 (MODY1) [MIM:125850]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269|PubMed:17407387, ECO:0000269|PubMed:9243109, ECO:0000269|PubMed:9313765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 125853, Otozomal Dominant (MODY) (MLPA)	Diabetes mellitus, noninsulin-dependent, 125853, Autosomal dominant (MODY) (MLPA)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Maturity-onset diabetes of the young 1 (MODY1) [MIM:125850]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269|PubMed:17407387, ECO:0000269|PubMed:9243109, ECO:0000269|PubMed:9313765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 2, 125853, Otozomal Dominant (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 2, 125853, Autosomal dominant (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 2, 125853, Otozomal Dominant (MLPA)	Diabetes mellitus, noninsulin-dependent, 2, 125853, Autosomal dominant (MLPA)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, 5, 616087 (TBC1D4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, 5, 616087 (TBC1D4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D4	TBC1D4, AS160, KIAA0603, NIDDM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent, 5 (NIDDM5) [MIM:616087]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:25043022}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, İle İlişki, 125853, Otozomal Dominant (Erken Başlangıçlı Sendromik Olmayan Katarakt) (WFS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, association with, 125853, Autosomal dominant (Early-onset non-syndromic cataract) (WFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, İle İlişki, 125853, Otozomal Dominant (Erken Başlangıçlı Sendromik Olmayan Katarakt) (MLPA)	Diabetes mellitus, noninsulin-dependent, association with, 125853, Autosomal dominant (Early-onset non-syndromic cataract) (MLPA)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, Geç Başlangıçlı, 125853, Otozomal Dominant (GCK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, late onset, 125853, Autosomal dominant (GCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCK	GCK, HHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan, Geç Başlangıçlı, 125853, Otozomal Dominant (MLPA)	Diabetes mellitus, noninsulin-dependent, late onset, 125853, Autosomal dominant (MLPA)	GCK	GCK, HHF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan Yatkınlık, 125853, Otozomal Dominant (SLC30A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853, Autosomal dominant (SLC30A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC30A8	SLC30A8, ZNT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan Yatkınlık, 125853, Otozomal Dominant (RETN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853, Autosomal dominant (RETN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RETN	RETN, RSTN, FIZZ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan Yatkınlık, 125853, Otozomal Dominant (IGF2BP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853, Autosomal dominant (IGF2BP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGF2BP2	IGF2BP2, IMP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan Yatkınlık, 125853, Otozomal Dominant (HMGA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853, Autosomal dominant (HMGA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGA1	HMGA1, HMGIY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı Olmayan Yatkınlık, 125853, Otozomal Dominant (CDKAL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853, Autosomal dominant (CDKAL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKAL1	CDKAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Kalıcı Neonatal, 606176, Otozomal Dominant (DEND Sendromu) (INS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, permanent neonatal, 606176, Autosomal dominant (DEND syndrome) (INS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INS	INS, MODY10, IDDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Kalıcı Neonatal, 606176, Otozomal Dominant (DEND Sendromu) (ABCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, permanent neonatal, 606176, Autosomal dominant (DEND syndrome) (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC8	ABCC8, SUR, PHH, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Kalıcı Neonatal, 606176, Otozomal Dominant (Kalıcı Neonatal Diabetes Mellitus) (GCK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, permanent neonatal, 606176, Autosomal dominant (Permanent neonatal diabetes mellitus) (GCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCK	GCK, HHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Kalıcı Neonatal, 606176, Otozomal Dominant (Kalıcı Neonatal Diabetes Mellitus) (MLPA)	Diabetes mellitus, permanent neonatal, 606176, Autosomal dominant (Permanent neonatal diabetes mellitus) (MLPA)	GCK	GCK, HHF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Kalıcı Neonatal, Prein (DEND Sendromu) (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DIABETES MELLITUS, PERMANENT NEONATAL, PNDM (DEND syndrome) (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Geçici Neonatal 2, 610374 (Geçici Neonatal Diabetes Mellitus) (ABCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, transient neonatal 2, 610374 (Transient neonatal diabetes mellitus) (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC8	ABCC8, SUR, PHH, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Geçici Neonatal, 1, 601410 (Geçici Neonatal Diabetes Mellitus) (ZFP57 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, transient neonatal, 1, 601410 (Transient neonatal diabetes mellitus) (ZFP57 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFP57	ZFP57, TNDM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Transient neonatal diabetes mellitus 1 (TNDM1) [MIM:601410]: Neonatal diabetes is a form of diabetes mellitus defined by the onset of mild-to-severe hyperglycemia within the first months of life. In about half of the neonates, diabetes is transient and resolves at a median age of 3 months, whereas the rest have a permanent form of diabetes. {ECO:0000269	PubMed:18622393}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, Geçici Neonatal, 3, 610582, Otozomal Dominant (Geçici Neonatal Diabetes Mellitus) (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, transient neonatal, 3, 610582, Autosomal dominant (Transient neonatal diabetes mellitus) (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Geçici Neonatal, 601410 (Geçici Neonatal Diabetes Mellitus) (PLAGL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, transient neonatal, 601410 (Transient neonatal diabetes mellitus) (PLAGL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAGL1	PLAGL1, ZAC, LOT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Tip 1, Yatkınlık, 222100, Otozomal Resesif (OAS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type 1, susceptibility to, 222100, Autosomal recessive (OAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OAS1	OAS1, OIAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Tip 2, 125853, Otozomal Dominant (PAX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type 2, 125853, Autosomal dominant (PAX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX4	PAX4, MODY9, KPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:11723072}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, Tip 2, Yatkınlık, 125853, Otozomal Dominant (TCF7L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type 2, susceptibility to, 125853, Autosomal dominant (TCF7L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCF7L2	TCF7L2, TCF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:16415884, ECO:0000269|PubMed:24390345}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, Tip 2, Yatkınlık, 125853, Otozomal Dominant (MTNR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type 2, susceptibility to, 125853, Autosomal dominant (MTNR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTNR1B	MTNR1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Tip 2, Yatkınlık, 125853, Otozomal Dominant (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type 2, susceptibility to, 125853, Autosomal dominant (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabetes Mellitus, Tip 1, Yatkınlık, 222100, Otozomal Resesif (FOXP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type I, susceptibility to, 222100, Autosomal recessive (FOXP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXP3	FOXP3, IPEX, AIID, XPID, PIDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Diyabetes Mellitus, Tip 2, 125853, Otozomal Dominant (AKT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type II, 125853, Autosomal dominant (AKT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT2	AKT2, HHGHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:15186380, ECO:0000269|PubMed:19164855}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, Tip 2, Yatkınlık, 125853, Otozomal Dominant (PDX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes mellitus, type II, susceptibility to, 125853, Autosomal dominant (PDX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDX1	PDX1, IPF1, MODY4, PAGEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetes Mellitus, İnsülin Bağımlı, Yatkınlık, 10, 601942 (IL2RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, mellitus, insulin-dependent, susceptibility to, 10, 601942 (IL2RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL2RA	IL2RA, CD25, IL2R, IDDM10, IMD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 10 (IDDM10) [MIM:601942]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:17676041}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabet, Kalıcı Neonatal, ± Nörolojik Bulgular, 606176, Otozomal Dominant (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, permanent neonatal, with or without neurologic features, 606176, Autosomal dominant (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabet, Yatkınlık 222100, Otozomal Resesif (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, susceptibility to, 222100, Autosomal recessive (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabet, Tip 1, Yatkınlık, 222100, Otozomal Resesif (PTPN22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, type 1, susceptibility to, 222100, Autosomal recessive (PTPN22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN22	PTPN22, PEP, PTPN8, LYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabet, Tip 1, Yatkınlık, 222100, Otozomal Resesif (ITPR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, type 1, susceptibility to, 222100, Autosomal recessive (ITPR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR3	ITPR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabet, Tip 2, 125853, Otozomal Dominant (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, type 2, 125853, Autosomal dominant (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CIMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyabet, Tip 2, Yatkınlık, 125853, Otozomal Dominant (GPD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diabetes, type 2, susceptibility to, 125853, Autosomal dominant (GPD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPD2	GPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diamond Blackfan Anemi 15, Mandibulofasiyal Disostozis, 606164, Otozomal Dominant; DBA15 (Blackfan-Diamond Anemisi) (RPS28 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164, Autosomal dominant; DBA15 (Blackfan-Diamond anemia) (RPS28 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS28	RPS28, DBA15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 15, with mandibulofacial dysostosis (DBA15) [MIM:606164]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:24942156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond Blackfan Anemi 15, Mandibulofasiyal Disostozis, 606164, Otozomal Dominant; DBA15 (Blackfan-Diamond Anemisi) (Prenatal) (RPS28 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164, Autosomal dominant; DBA15 (Blackfan-Diamond anemia) (RPS28 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS28	RPS28, DBA15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 15, with mandibulofacial dysostosis (DBA15) [MIM:606164]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:24942156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 1, 105650, Otozomal Dominant; DBA1 (Blackfan-Diamond Anemisi) (RPS19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 1, 105650, Autosomal dominant; DBA1 (Blackfan-Diamond anemia) (RPS19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS19	RPS19, DBA, DBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diamond-Blackfan Anemi 1, 105650, Otozomal Dominant; DBA1 (Blackfan-Diamond Anemisi) (Prenatal) (RPS19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 1, 105650, Autosomal dominant; DBA1 (Blackfan-Diamond anemia) (RPS19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS19	RPS19, DBA, DBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 10, 613309, Otozomal Dominant; DBA10 (Blackfan-Diamond Anemisi) (RPS26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 10, 613309, Autosomal dominant; DBA10 (Blackfan-Diamond anemia) (RPS26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS26	RPS26, DBA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 10 (DBA10) [MIM:613309]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:20116044}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 10, 613309, Otozomal Dominant; DBA10 (Blackfan-Diamond Anemisi) (Prenatal) (RPS26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 10, 613309, Autosomal dominant; DBA10 (Blackfan-Diamond anemia) (RPS26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS26	RPS26, DBA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 10 (DBA10) [MIM:613309]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:20116044}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond Blackfan Anemi 11, 614900, Otozomal Dominant; DBA11 (Blackfan-Diamond Anemisi) (RPL26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 11, 614900, Autosomal dominant; DBA11 (Blackfan-Diamond anemia) (RPL26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL26	RPL26, DBA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 11 (DBA11) [MIM:614900]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:22431104}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond Blackfan Anemi 11, 614900, Otozomal Dominant; DBA11 (Blackfan-Diamond Anemisi) (Prenatal) (RPL26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 11, 614900, Autosomal dominant; DBA11 (Blackfan-Diamond anemia) (RPL26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL26	RPL26, DBA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 11 (DBA11) [MIM:614900]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:22431104}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond Blackfan Anemi 12, 615550, Otozomal Dominant; DBA12 (Blackfan-Diamond Anemisi) (RPL15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 12, 615550, Autosomal dominant; DBA12 (Blackfan-Diamond anemia) (RPL15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL15	RPL15, DBA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 12 (DBA12) [MIM:615550]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:23812780}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond Blackfan Anemi 12, 615550, Otozomal Dominant; DBA12 (Blackfan-Diamond Anemisi) (Prenatal) (RPL15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 12, 615550, Autosomal dominant; DBA12 (Blackfan-Diamond anemia) (RPL15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL15	RPL15, DBA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 12 (DBA12) [MIM:615550]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:23812780}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 13, 615909, Otozomal Dominant; DBA13 (Blackfan-Diamond Anemisi) (RPS29 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 13, 615909, Autosomal dominant; DBA13 (Blackfan-Diamond anemia) (RPS29 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS29	RPS29, DBA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 13 (DBA13) [MIM:615909]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:24829207}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Diamond-Blackfan Anemi 13, 615909, Otozomal Dominant; DBA13 (Blackfan-Diamond Anemisi) (Prenatal) (RPS29 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 13, 615909, Autosomal dominant; DBA13 (Blackfan-Diamond anemia) (RPS29 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS29	RPS29, DBA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 13 (DBA13) [MIM:615909]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:24829207}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 14-Mandibulofasiyal Disostoz, 300946, X'e Bağlı Resesif; DBA14 (Blackfan-Diamond Anemisi) (TSR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946, X-linked recessive; DBA14 (Blackfan-Diamond anemia) (TSR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSR2	TSR2, WGG1, DBA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 14, with mandibulofacial dysostosis (DBA14) [MIM:300946]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:24942156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 14-Mandibulofasiyal Disostoz, 300946, X'e Bağlı Resesif; DBA14 (Blackfan-Diamond Anemisi) (Prenatal) (TSR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 14 with mandibulofacial dysostosis, 300946, X-linked recessive; DBA14 (Blackfan-Diamond anemia) (TSR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSR2	TSR2, WGG1, DBA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 14, with mandibulofacial dysostosis (DBA14) [MIM:300946]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:24942156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 3, 610629, Otozomal Dominant; DBA3 (Blackfan-Diamond Anemisi) (RPS24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-blackfan anemia 3, 610629, Autosomal dominant; DBA3 (Blackfan-Diamond anemia) (RPS24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS24	RPS24, DBA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diamond-Blackfan Anemi 3, 610629, Otozomal Dominant; DBA3 (Blackfan-Diamond Anemisi) (Prenatal) (RPS24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-blackfan anemia 3, 610629, Autosomal dominant; DBA3 (Blackfan-Diamond anemia) (RPS24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS24	RPS24, DBA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 4, 612527, Otozomal Dominant; DBA4 (Blackfan-Diamond Anemisi) (RPS17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 4, 612527, Autosomal dominant; DBA4 (Blackfan-Diamond anemia) (RPS17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS17	RPS17, RPS17L1, RPS17L2, DBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 4 (DBA4) [MIM:612527]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of developing leukemia. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:17647292, ECO:0000269|PubMed:19061985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 4, 612527, Otozomal Dominant; DBA4 (Blackfan-Diamond Anemisi) (Prenatal) (RPS17 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 4, 612527, Autosomal dominant; DBA4 (Blackfan-Diamond anemia) (RPS17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS17	RPS17, RPS17L1, RPS17L2, DBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 4 (DBA4) [MIM:612527]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of developing leukemia. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:17647292, ECO:0000269|PubMed:19061985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 5, 612528, Otozomal Dominant; DBA5 (Blackfan-Diamond Anemisi) (RPL35A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 5, 612528, Autosomal dominant; DBA5 (Blackfan-Diamond anemia) (RPL35A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL35A	RPL35A, DBA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 5 (DBA5) [MIM:612528]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:18535205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 5, 612528, Otozomal Dominant; DBA5 (Blackfan-Diamond Anemisi) (Prenatal) (RPL35A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 5, 612528, Autosomal dominant; DBA5 (Blackfan-Diamond anemia) (RPL35A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL35A	RPL35A, DBA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 5 (DBA5) [MIM:612528]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:18535205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 6, 612561, Otozomal Dominant; DBA6 (Blackfan-Diamond Anemisi) (RPL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 6, 612561, Autosomal dominant; DBA6 (Blackfan-Diamond anemia) (RPL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL5	RPL5, DBA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 6 (DBA6) [MIM:612561]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:19061985, ECO:0000269|PubMed:19191325}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 6, 612561, Otozomal Dominant; DBA6 (Blackfan-Diamond Anemisi) (Prenatal) (RPL5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 6, 612561, Autosomal dominant; DBA6 (Blackfan-Diamond anemia) (RPL5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL5	RPL5, DBA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 6 (DBA6) [MIM:612561]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:19061985, ECO:0000269|PubMed:19191325}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 7, 612562, Otozomal Dominant; DBA7 (Blackfan-Diamond Anemisi) (RPL11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 7, 612562, Autosomal dominant; DBA7 (Blackfan-Diamond anemia) (RPL11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL11	RPL11, DBA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 7 (DBA7) [MIM:612562]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:19061985, ECO:0000269|PubMed:19191325}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 7, 612562, Otozomal Dominant; DBA7 (Blackfan-Diamond Anemisi) (Prenatal) (RPL11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 7, 612562, Autosomal dominant; DBA7 (Blackfan-Diamond anemia) (RPL11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPL11	RPL11, DBA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 7 (DBA7) [MIM:612562]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269|PubMed:19061985, ECO:0000269|PubMed:19191325}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diamond-Blackfan Anemi 8, 612563, Otozomal Dominant; DBA8 (Blackfan-Diamond Anemisi) (RPS7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 8, 612563, Autosomal dominant; DBA8 (Blackfan-Diamond anemia) (RPS7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS7	RPS7, DBA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 8 (DBA8) [MIM:612563]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:19061985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diamond-Blackfan Anemi 8, 612563, Otozomal Dominant; DBA8 (Blackfan-Diamond Anemisi) (Prenatal) (RPS7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 8, 612563, Autosomal dominant; DBA8 (Blackfan-Diamond anemia) (RPS7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS7	RPS7, DBA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 8 (DBA8) [MIM:612563]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:19061985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Diamond-Blackfan Anemi 9, 613308, Otozomal Dominant; DBA9 (Blackfan-Diamond Anemisi) (RPS10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diamond-Blackfan anemia 9, 613308, Autosomal dominant; DBA9 (Blackfan-Diamond anemia) (RPS10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS10	RPS10, DBA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diamond-Blackfan anemia 9 (DBA9) [MIM:613308]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:20116044}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diamond-Blackfan Anemi 9, 613308, Otozomal Dominant; DBA9 (Blackfan-Diamond Anemisi) (Prenatal) (RPS10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diamond-Blackfan anemia 9, 613308, Autosomal dominant; DBA9 (Blackfan-Diamond anemia) (RPS10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS10	RPS10, DBA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diamond-Blackfan anemia 9 (DBA9) [MIM:613308]: A form of Diamond-Blackfan anemia, a congenital non-regenerative hypoplastic anemia that usually presents early in infancy. Diamond-Blackfan anemia is characterized by a moderate to severe macrocytic anemia, erythroblastopenia, and an increased risk of malignancy. 30 to 40% of Diamond-Blackfan anemia patients present with short stature and congenital anomalies, the most frequent being craniofacial (Pierre-Robin syndrome and cleft palate), thumb and urogenital anomalies. {ECO:0000269	PubMed:20116044}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diafonospondilodisostozis, 608022, Otozomal Resesif (Diafanospondilodisostozis) (BMPER geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diaphanospondylodysostosis, 608022, Autosomal recessive (Diaphanospondylodysostosis) (BMPER gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPER	BMPER, CV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diafonospondilodisostozis, 608022, Otozomal Resesif (Diafanospondilodisostozis) (Prenatal) (BMPER geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diaphanospondylodysostosis, 608022, Autosomal recessive (Diaphanospondylodysostosis) (BMPER gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMPER	BMPER, CV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyafragmatik Herni 3, 610187; DIH3 (Konjenital Diafragma Hernisi) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diaphragmatic hernia 3, 610187; DIH3 (Congenital diaphragmatic hernia) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diyafragmatik Herni 3, 610187; DIH3 (Konjenital Diafragma Hernisi) (Prenatal) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diaphragmatic hernia 3, 610187; DIH3 (Congenital diaphragmatic hernia) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Malign Fibröz Histiyositoma İle Diyafizyel Medüller Stenoz, 112250, Otozomal Dominant; DMSMFH (Dafiz-Medüller Stenozu-Kemik Maligniteisi Sendromu) (MTAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diaphyseal medullary stenosis with malignant fibrous histiocytoma, 112250, Autosomal dominant; DMSMFH (Diaphyseal medullary stenosis-bone malignancy syndrome) (MTAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTAP	MTAP, DMSMFH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Loss of MTAP activity may play a role in human cancer. MTAP loss has been reported in a number of cancers, including osteosarcoma, malignant melanoma and gastric cancer.,	EDTA Blood Sample
Diyare 1, Klorür Salgılama, Konjenital, 214700, Otozomal Resesif; DIAR1 (Konjenital Klorür Diyaresi) (SLC26A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 1, secretory chloride, congenital, 214700, Autosomal recessive; DIAR1 (Congenital chloride diarrhea) (SLC26A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A3	SLC26A3, DRA, CLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 1, secretory chloride, congenital (DIAR1) [MIM:214700]: A disease characterized by voluminous watery stools containing an excess of chloride. The children with this disease are often premature. {ECO:0000269|PubMed:11524734, ECO:0000269|PubMed:19861545, ECO:0000269|PubMed:21150650, ECO:0000269|PubMed:21394828, ECO:0000269|PubMed:8896562, ECO:0000269|PubMed:9554749, ECO:0000269|PubMed:9718329}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diyare 1, Klorür Salgılama, Konjenital, 214700, Otozomal Resesif; DIAR1 (Konjenital Klorür Diyaresi) (Prenatal) (SLC26A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 1, secretory chloride, congenital, 214700, Autosomal recessive; DIAR1 (Congenital chloride diarrhea) (SLC26A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC26A3	SLC26A3, DRA, CLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 1, secretory chloride, congenital (DIAR1) [MIM:214700]: A disease characterized by voluminous watery stools containing an excess of chloride. The children with this disease are often premature. {ECO:0000269|PubMed:11524734, ECO:0000269|PubMed:19861545, ECO:0000269|PubMed:21150650, ECO:0000269|PubMed:21394828, ECO:0000269|PubMed:8896562, ECO:0000269|PubMed:9554749, ECO:0000269|PubMed:9718329}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 2-Mikrovillus Atrofisi; Diar2 (Mikrovillus İnklüzyon Hastalığı) (MYO5B geni) (Tüm Gen Dizi Analizi) (Postnatal)	DIARRHEA 2, WITH MICROVILLUS ATROPHY; DIAR2 (Microvillus inclusion disease) (MYO5B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO5B	MYO5B, KIAA1119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 2, with microvillus atrophy (DIAR2) [MIM:251850]: A disease characterized by onset of intractable life-threatening watery diarrhea during infancy. Two forms are recognized: early-onset microvillus inclusion disease with diarrhea beginning in the neonatal period, and late-onset, with first symptoms appearing after 3 or 4 months of life. {ECO:0000269|PubMed:18724368, ECO:0000269|PubMed:19006234, ECO:0000269|PubMed:20186687, ECO:0000269|PubMed:21206382, ECO:0000269|PubMed:24138727, ECO:0000269|PubMed:24892806}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diyare 2-Mikrovillus Atrofisi; Diar2 (Mikrovillus İnklüzyon Hastalığı) (Prenatal) (MYO5B geni) (Tüm Gen Dizi Analizi) (Prenatal)	DIARRHEA 2, WITH MICROVILLUS ATROPHY; DIAR2 (Microvillus inclusion disease) (MYO5B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYO5B	MYO5B, KIAA1119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 2, with microvillus atrophy (DIAR2) [MIM:251850]: A disease characterized by onset of intractable life-threatening watery diarrhea during infancy. Two forms are recognized: early-onset microvillus inclusion disease with diarrhea beginning in the neonatal period, and late-onset, with first symptoms appearing after 3 or 4 months of life. {ECO:0000269|PubMed:18724368, ECO:0000269|PubMed:19006234, ECO:0000269|PubMed:20186687, ECO:0000269|PubMed:21206382, ECO:0000269|PubMed:24138727, ECO:0000269|PubMed:24892806}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 3, Sodyum Salgılayan, Konjenital, Sendromik, 270420, Otozomal Resesif; DIAR3 (Konjenital Sodyum Diyaresi) (SPINT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 3, secretory sodium, congenital, syndromic, 270420, Autosomal recessive; DIAR3 (Congenital sodium diarrhea) (SPINT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINT2	SPINT2, HAI2, DIAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 3, secretory sodium, congenital, with or without other congenital anomalies (DIAR3) [MIM:270420]: A disease characterized by life-threatening secretory diarrhea, severe metabolic acidosis and hyponatremia. Hyponatremia is secondary to extraordinarily high fecal sodium loss, with low or normal excretion of urinary sodium, in the absence of infectious, autoimmune and endocrine causes. {ECO:0000269|PubMed:19185281}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diyare 3, Sodyum Salgılayan, Konjenital, Sendromik, 270420, Otozomal Resesif; DIAR3 (Konjenital Sodyum Diyaresi) (Prenatal) (SPINT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 3, secretory sodium, congenital, syndromic, 270420, Autosomal recessive; DIAR3 (Congenital sodium diarrhea) (SPINT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPINT2	SPINT2, HAI2, DIAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 3, secretory sodium, congenital, with or without other congenital anomalies (DIAR3) [MIM:270420]: A disease characterized by life-threatening secretory diarrhea, severe metabolic acidosis and hyponatremia. Hyponatremia is secondary to extraordinarily high fecal sodium loss, with low or normal excretion of urinary sodium, in the absence of infectious, autoimmune and endocrine causes. {ECO:0000269|PubMed:19185281}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 4, Malabsorptif, Konjenital, 610370, Otozomal Resesif; DIAR4 (Konjenital Malabsorbtif Diyare, Enteroendokrin Hücre Azlığına Bağlı) (NEUROG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 4, malabsorptive, congenital, 610370, Autosomal recessive; DIAR4 (Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells) (NEUROG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEUROG3	NEUROG3, NGN3, ATOH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 4, malabsorptive, congenital (DIAR4) [MIM:610370]: A disease characterized by severe, life-threatening watery diarrhea associated with generalized malabsorption and a paucity of enteroendocrine cells. {ECO:0000269|PubMed:16855267}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diyare 4, Malabsorptif, Konjenital, 610370, Otozomal Resesif; DIAR4 (Konjenital Malabsorbtif Diyare, Enteroendokrin Hücre Azlığına Bağlı) (Prenatal) (NEUROG3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 4, malabsorptive, congenital, 610370, Autosomal recessive; DIAR4 (Congenital malabsorptive diarrhea due to paucity of enteroendocrine cells) (NEUROG3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEUROG3	NEUROG3, NGN3, ATOH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 4, malabsorptive, congenital (DIAR4) [MIM:610370]: A disease characterized by severe, life-threatening watery diarrhea associated with generalized malabsorption and a paucity of enteroendocrine cells. {ECO:0000269|PubMed:16855267}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 5, Tafting Enteropati, Konjenital, 613217, Otozomal Resesif; DIAR5 (Intestinal Epitelial Displazi) (EPCAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 5, with tufting enteropathy, congenital, 613217, Autosomal recessive; DIAR5 (Intestinal epithelial dysplasia) (EPCAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPCAM	EPCAM, ACSTD1, TROP1, M4S1, MIC18, DIAR5, HNPCC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 5, with tufting enteropathy, congenital (DIAR5) [MIM:613217]: An intractable diarrhea of infancy characterized by villous atrophy and absence of inflammation, with intestinal epithelial cell dysplasia manifesting as focal epithelial tufts in the duodenum and jejunum. {ECO:0000269|PubMed:18572020}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyare 5, Tafting Enteropati, Konjenital, 613217, Otozomal Resesif; DIAR5 (Intestinal Epitelial Displazi) (Prenatal) (EPCAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 5, with tufting enteropathy, congenital, 613217, Autosomal recessive; DIAR5 (Intestinal epithelial dysplasia) (EPCAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPCAM	EPCAM, ACSTD1, TROP1, M4S1, MIC18, DIAR5, HNPCC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 5, with tufting enteropathy, congenital (DIAR5) [MIM:613217]: An intractable diarrhea of infancy characterized by villous atrophy and absence of inflammation, with intestinal epithelial cell dysplasia manifesting as focal epithelial tufts in the duodenum and jejunum. {ECO:0000269|PubMed:18572020}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 6, 614616, Otozomal Dominant; DIAR6 (Kronik Diyare, Guanilat Siklaz 2C Aşırı Aktivitesine Bağlı) (GUCY2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 6, 614616, Autosomal dominant; DIAR6 (Chronic diarrhea due to guanylate cyclase 2C overactivity) (GUCY2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCY2C	GUCY2C, GUC2C, DIAR6, MECIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 6 (DIAR6) [MIM:614616]: A relatively mild, early-onset chronic diarrhea that may be associated with increased susceptibility to inflammatory bowel disease, small bowel obstruction, and esophagitis. {ECO:0000269|PubMed:22436048}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyare 6, 614616, Otozomal Dominant; DIAR6 (Kronik Diyare, Guanilat Siklaz 2C Aşırı Aktivitesine Bağlı) (Prenatal) (GUCY2C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 6, 614616, Autosomal dominant; DIAR6 (Chronic diarrhea due to guanylate cyclase 2C overactivity) (GUCY2C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GUCY2C	GUCY2C, GUC2C, DIAR6, MECIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 6 (DIAR6) [MIM:614616]: A relatively mild, early-onset chronic diarrhea that may be associated with increased susceptibility to inflammatory bowel disease, small bowel obstruction, and esophagitis. {ECO:0000269|PubMed:22436048}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 7, 615863, Otozomal Resesif; DIAR7 (Konjenital Kronik Diyare-Protein Kaybettiren Enteropati) (DGAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 7, 615863, Autosomal recessive; DIAR7 (Congenital chronic diarrhea with protein-losing enteropathy) (DGAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGAT1	DGAT1, ARGP1, DIAR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 7 (DIAR7) [MIM:615863]: A life-threatening disease characterized by severe, intractable, watery diarrhea. {ECO:0000269|PubMed:23114594}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diyare 7, 615863, Otozomal Resesif; DIAR7 (Konjenital Kronik Diyare-Protein Kaybettiren Enteropati) (Prenatal) (DGAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 7, 615863, Autosomal recessive; DIAR7 (Congenital chronic diarrhea with protein-losing enteropathy) (DGAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGAT1	DGAT1, ARGP1, DIAR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 7 (DIAR7) [MIM:615863]: A life-threatening disease characterized by severe, intractable, watery diarrhea. {ECO:0000269|PubMed:23114594}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyare 8, Klorür Salgılama, Konjenital, 616868, Otozomal Resesif; DIAR8 (Konjenital Sodyum Diyaresi) (SLC9A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diarrhea 8, secretory sodium, congenital, 616868, Autosomal recessive; DIAR8 (Congenital sodium diarrhea) (SLC9A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC9A3	SLC9A3, NHE3, DIAR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Diyare 8, Klorür Salgılama, Konjenital, 616868, Otozomal Resesif; DIAR8 (Konjenital Sodyum Diyaresi) (Prenatal) (SLC9A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Diarrhea 8, secretory sodium, congenital, 616868, Autosomal recessive; DIAR8 (Congenital sodium diarrhea) (SLC9A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC9A3	SLC9A3, NHE3, DIAR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dias-Logan Sendromu, 617101, Otozomal Dominant (BCL11A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dias-Logan syndrome, 617101, Autosomal dominant (BCL11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL11A	BCL11A, CTIP1, EVI9, KIAA1809, DILOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dias-Logan Sendromu, 617101, Otozomal Dominant (Prenatal) (BCL11A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dias-Logan syndrome, 617101, Autosomal dominant (BCL11A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCL11A	BCL11A, CTIP1, EVI9, KIAA1809, DILOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diyastrofik Displazi, Geniş Kemikler-Platispondilik Varyant, 222600, Otozomal Resesif; DTD (Diastropik Displazi) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diastrophic dysplasia, broad bone-platyspondylic variant, 222600, Autosomal recessive; DTD (Diastrophic dwarfism) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A2	SLC26A2, DTD, DTDST, DTDST, DTD, DTDST, DTD, DTDST, DTD

Dimetilglisin Dehidrogenaz Eksikliği, 605850, Otozomal Resesif; DMGDHD (Dimetilglisin Dehidrogenaz Eksikliği) (DMGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dimethylglycine dehydrogenase deficiency, 605850, Autosomal recessive; DMGDHD (Dimethylglycine dehydrogenase deficiency) (DMGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMGDH	DMGDH, DMGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: DMGDH deficiency (DMGDHD) [MIM:605850]: Disorder characterized by fish odor, muscle fatigue with increased serum creatine kinase. Biochemically it is characterized by an increase of N,N-dimethylglycine (DMG) in serum and urine. (ECO:0000269	PubMed:11231903). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dimetilglisin Dehidrogenaz Eksikliği, 605850, Otozomal Resesif; DMGDHD (Dimetilglisin Dehidrogenaz Eksikliği) (Prenatal) (DMGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dimethylglycine dehydrogenase deficiency, 605850, Autosomal recessive; DMGDHD (Dimethylglycine dehydrogenase deficiency) (DMGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DMGDH	DMGDH, DMGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: DMGDH deficiency (DMGDHD) [MIM:605850]: Disorder characterized by fish odor, muscle fatigue with increased serum creatine kinase. Biochemically it is characterized by an increase of N,N-dimethylglycine (DMG) in serum and urine. (ECO:0000269	PubMed:11231903). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Difenilhidantoin Toksisitesi (EPHX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diphenylhydantoin toxicity (EPHX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHX1	EPHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Difteri, Yatkınlık (HBEGF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Diphtheria, susceptibility to (HBEGF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBEGF	HBEGF, DTR, DTSF, HEGFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sitokrom P450 Oksidoredüktaz Nedeniyle Düzensiz Steroidogenezis, 613571 (Konjenital Adrenal Hiperplazi, Sitokrom P450 Oksidoredüktaz Eksikliği) (POR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency) (POR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POR	POR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. (ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sitokrom P450 Oksidoredüktaz Nedeniyle Düzensiz Steroidogenezis, 613571 (Konjenital Adrenal Hiperplazi, Sitokrom P450 Oksidoredüktaz Eksikliği) (MLPA)	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (Congenital adrenal hyperplasia due to cytochrome P450 oxidoreductase deficiency) (MLPA)	POR	POR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Antley-Bixler syndrome, with genital anomalies and disordered steroidogenesis (ABS1) [MIM:201750]: A disease characterized by the association of Antley-Bixler syndrome with steroidogenesis defects and abnormal genitalia. Antley-Bixler syndrome is characterized by craniosynostosis, radiohumeral synostosis present from the perinatal period, midface hypoplasia, choanal stenosis or atresia, femoral bowing and multiple joint contractures. (ECO:0000269|PubMed:14758361, ECO:0000269|PubMed:15264278, ECO:0000269|PubMed:15483095). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
DNA Ligaz 1 Eksikliği (LIG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DNA ligase I deficiency (LIG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIG1	LIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNA Ligaz 1 Eksikliği (Prenatal) (LIG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	DNA ligase I deficiency (LIG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIG1	LIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
DNA Sperm Integrity Testing (DNA Sperm Hasar Testi) (TUNEL testi)	DNA sperm integrity testing (TUNEL test)	.	.	TUNEL	Sperm (Transport besi yeri içinde)		5	30	.	.	15	Postnatal	.	Sperm (Transport besi yeri içinde)
DNA Topoizomeraz I, Kamptotesine Dirençli (TOP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DNA topoisomerase I, camptothecin-resistant (TOP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOP1	TOP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving TOP1 is found in a form of therapy-related myelodysplastic syndrome. Translocation t(11;20)(q15;q11) with NUP98. (ECO:0000269	PubMed:10556215).:	EDTA Blood Sample
DNA Topoizomeraz 2, Amsakrin İnhibisyonuna Direnç, (TOP2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	DNA topoisomerase II, resistance to inhibition of, by amsacrine (TOP2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOP2A	TOP2A, TOP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Donnai-Barrow Sendromu, 222448, Otozomal Resesif (Donnai-Barrow Sendromu (LRP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Donnai-Barrow syndrome, 222448, Autosomal recessive (Donnai-Barrow syndrome) (LRP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP2	LRP2, DBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Donnai-Barrow Sendromu, 222448, Otozomal Resesif (Donnai-Barrow Sendromu) (Prenatal) (LRP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Donnai-Barrow syndrome, 222448, Autosomal recessive (Donnai-Barrow syndrome) (LRP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP2	LRP2, DBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
DOOR Sendromu, 220500, Otozomal Resesif; DOORS (Sağırlık-Onikodistrofi Sendromu) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DOOR syndrome, 220500, Autosomal recessive; DOORS (Deafness-onychodystrophy syndrome) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome (DOORS) [MIM:220500]: A syndrome characterized by sensorineural deafness, mental retardation, hypoplastic or absent nails, small or absent distal phalanges of hands and feet. Additional features include coarse facies, a large nose with wide nasal bridge, bulbous tip and anteverted nares, a long prominent philtrum and downturned corners of the mouth. Progressive neurological manifestations include seizures from infancy, optic atrophy, and peripheral polyneuropathy. (ECO:0000269	PubMed:24291220). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
DOOR Sendromu, 220500, Otozomal Resesif; DOORS (Sağırlık-Onikodistrofi Sendromu) (Prenatal) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	DOOR syndrome, 220500, Autosomal recessive; DOORS (Deafness-onychodystrophy syndrome) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome (DOORS) [MIM:220500]: A syndrome characterized by sensorineural deafness, mental retardation, hypoplastic or absent nails, small or absent distal phalanges of hands and feet. Additional features include coarse facies, a large nose with wide nasal bridge, bulbous tip and anteverted nares, a long prominent philtrum and downturned corners of the mouth. Progressive neurological manifestations include seizures from infancy, optic atrophy, and peripheral polyneuropathy. (ECO:0000269	PubMed:24291220). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dopa-duyarlı distoni, Otozomal resesif/ Dopa-duyarlı distoni, Otozomal dominant/ Miyoklonus-distoni sendromu (Distoni 11, miyoklonik) (TH 11p15.5, GCH11 4q22, SGCE 7q21) (MLPA)	Dopa-sensitive dystonia, Autosomal recessive / Dopa-sensitive dystonia, Autosomal dominant / Myoclonus-dystonia syndrome (Distoni 11, Myoclonic) (TH 11p15.5, GCH11 4q22, SGCE 7q21) (MLPA)	TH 11p15.5, GCH11 4q22, SGCE 7q21	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Dopa-duyarlı distoni, Otozomal resesif/ Dopa-duyarlı distoni, Otozomal dominant/ Miyoklonus-distoni sendromu (Distoni 11, miyoklonik) (TH 11p15.5, GCH11 4q22, SGCE 7q21) (MLPA) (Prenatal)	Dopa-sensitive dystonia, Autosomal recessive / Dopa-sensitive dystonia, Autosomal dominant / Myoclonus-dystonia syndrome (Distoni 11, Myoclonic) (TH 11p15.5, GCH11 4q22, SGCE 7q21) (MLPA) (Prenatal)	TH 11p15.5, GCH11 4q22, SGCE 7q21	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dopamin Beta-Hidroksilaz Eksikliği, 223360, Otozomal Resesif (Dopamin Beta-Hidroksilaz Eksikliği) (DBH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dopamine beta-hydroxylase deficiency, 223360, Autosomal recessive (Dopamine beta-hydroxylase deficiency) (DBH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DBH	DBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dopamine beta-hydroxylase deficiency (DBH deficiency) [MIM:223360]: Characterized by profound deficits in autonomic and cardiovascular function, but apparently only subtle signs, if any, of central nervous system dysfunction. (ECO:0000269	PubMed:1857564). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dopamin Beta-Hidroksilaz Eksikliği, 223360, Otozomal Resesif (Dopamin Beta-Hidroksilaz Eksikliği) (Prenatal) (DBH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dopamine beta-hydroxylase deficiency, 223360, Autosomal recessive (Dopamine beta-hydroxylase deficiency) (DBH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DBH	DBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dopamine beta-hydroxylase deficiency (DBH deficiency) [MIM:223360]: Characterized by profound deficits in autonomic and cardiovascular function, but apparently only subtle signs, if any, of central nervous system dysfunction. (ECO:0000269	PubMed:1857564). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dopamin Reseptör D2, Azalmış Beyin Yoğunluğu (ANKK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dopamine receptor D2, reduced brain density of (ANKK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKK1	ANKK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Çift Çıkışlı Sağ Ventrikül, 217095 (Çift Çıkışlı Sağ Ventrikül) (GDF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Double-outlet right ventricle, 217095 (Double outlet right ventricle) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. (ECO:0000269	PubMed:17924340). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Çift Çıkışlı Sağ Ventrikül, 217095 (Çift Çıkışlı Sağ Ventrikül) (Prenatal) (GDF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Double-outlet right ventricle, 217095 (Double outlet right ventricle) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. (ECO:0000269	PubMed:17924340). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dowling-Degos Hastalığı 1, 179850, Otozomal Dominant; DDD1 (Dowling-Degos Hastalığı) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dowling-Degos disease 1, 179850, Autosomal dominant; DDD1 (Dowling-Degos disease) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dowling-Degos Hastalığı 1, 179850, Otozomal Dominant; DDD1 (Dowling-Degos Hastalığı) (MLPA)	Dowling-Degos disease 1, 179850, Autosomal dominant; DDD1 (Dowling-Degos disease) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Dowling-Degos Hastalığı 1, 179850, Otozomal Dominant; DDD1 (Dowling-Degos Hastalığı) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dowling-Degos disease 1, 179850, Autosomal dominant; DDD1 (Dowling-Degos disease) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dowling-Degos Hastalığı 1, 179850, Otozomal Dominant; DDD1 (Dowling-Degos Hastalığı) (Prenatal) (MLPA)	Dowling-Degos disease 1, 179850, Autosomal dominant; DDD1 (Dowling-Degos disease) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dowling-Degos Hastalığı 2, 615327, Otozomal Dominant; DDD2 (Dowling-Degos Hastalığı) (POFUT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dowling-Degos disease 2, 615327, Autosomal dominant; DDD2 (Dowling-Degos disease) (POFUT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POFUT1	POFUT1, OFUCT1, KIAA0180, DDD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dowling-Degos disease 2 (DDD2) [MIM:615327]: An autosomal dominant genodermatosis. Affected individuals develop a postpubertal reticulate hyperpigmentation that is progressive and disfiguring, and small hyperkeratotic dark brown papules that affect mainly the flexures and great skin folds. Patients usually show no abnormalities of the hair or nails. (ECO:0000269	PubMed:23684010). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dowling-Degos Hastalığı 2, 615327, Otozomal Dominant; DDD2 (Dowling-Degos Hastalığı) (Prenatal) (POFUT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dowling-Degos disease 2, 615327, Autosomal dominant; DDD2 (Dowling-Degos disease) (POFUT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POFUT1	POFUT1, OFUCT1, KIAA0180, DDD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dowling-Degos disease 2 (DDD2) [MIM:615327]: An autosomal dominant genodermatosis. Affected individuals develop a postpubertal reticulate hyperpigmentation that is progressive and disfiguring, and small hyperkeratotic dark brown papules that affect mainly the flexures and great skin folds. Patients usually show no abnormalities of the hair or nails. (ECO:0000269	PubMed:23684010). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dowling-Degos Hastalığı 4, 615696, Otozomal Dominant; DDD4 (Dowling-Degos Hastalığı) (POGLUT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dowling-Degos disease 4, 615696, Autosomal dominant; DDD4 (Dowling-Degos disease) (POGLUT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POGLUT1	POGLUT1, CLP46, KTELC1, RUMI, C3orf9, DDD4, LGMD2Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dowling-Degos Hastalığı 4, 615696, Otozomal Dominant; DDD4 (Dowling-Degos Hastalığı) (Prenatal) (POGLUT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dowling-Degos disease 4, 615696, Autosomal dominant; DDD4 (Dowling-Degos disease) (POGLUT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POGLUT1	POGLUT1, CLP46, KTELC1, RUMI, C3orf9, DDD4, LGMD2Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Doyne Balpeteği Retina Dejenerasyonu, 126600, Otozomal Dominant; DHRD (Allesel Druzen) (EFEMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Doyne honeycomb degeneration of retina, 126600. Autosomal dominant; DHRD (Familial drusen) (EFEMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFEMP1	EFEMP1, FBNL, DHRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dravet Sendromu, Modifiye Edici, 607208, Otozomal Dominant (Dravet Sendromu) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dravet syndrome, modifier of, 607208, Autosomal dominant (Dravet syndrome) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dravet Sendromu, Modifiye Edici, 607208, Otozomal Dominant (Dravet Sendromu) (Prenatal) (SCN9A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dravet syndrome, modifier of, 607208, Autosomal dominant (Dravet syndrome) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İlaç Bağımlılığı, Yatkınlık, 606581 (FAAH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Drug addiction, susceptibility to, 606581 (FAAH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAAH	FAAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İlaç Bağımlılığı, Yatkınlık, 606581 (Prenatal) (FAAH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Drug addiction, susceptibility to, 606581 (FAAH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAAH	FAAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İlaca Bağlı Karaciğer Hasarı, Flukloksasilin Nedeniyle (Stevens-Johnson Sendromu) (HLA-B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Drug-induced liver injury due to flucloxacillin (Stevens-Johnson syndrome) (HLA-B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-B	HLA-B, SPDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Du Pan Sendromu, 228900, Otozomal Resesif (Fibula Aplazisi-Kompleks Brakidaktili Sendromu (Du Pan Sendromu)) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Du Pan syndrome, 228900, Autosomal recessive (Fibular aplasia-complex brachydactyly syndrome) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Du Pan Sendromu, 228900, Otozomal Resesif (Fibula Aplazisi-Kompleks Brakidaktili Sendromu (Du Pan Sendromu)) (Prenatal) (GDF5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Du Pan syndrome, 228900, Autosomal recessive (Fibular aplasia-complex brachydactyly syndrome) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Duane Retraksiyon Sendromu 2, 604356; DURS2 (Duane Retraksiyon Sendromu) (CHN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Duane retraction syndrome 2, 604356; DURS2 (Duane retraction syndrome) (CHN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHN1	CHN1, CHN, ARHGAP2, RHOGAP2, DURS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Duane retraction syndrome 2 (DURS2) [MIM:604356]: Duane retraction syndrome is a congenital eye movement disorder characterized by a failure of cranial nerve VI (the abducens nerve) to develop normally, resulting in restriction or absence of abduction, adduction, or both, and narrowing of the palpebral fissure and retraction of the globe on attempted adduction. Undiagnosed in children, it can lead to amblyopia, a permanent uncorrectable loss of vision. [ECO:0000269	PubMed:18653847]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Duane Retraksiyon Sendromu 3, 617041, Otozomal Dominant; DURS3 (Duane Retraksiyon Sendromu) (MAFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Duane retraction syndrome 3, 617041, Autosomal dominant; DURS3 (Duane retraction syndrome) (MAFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAFB	MAFB, KRML, MCTO, DURS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Duane Radyal Ray Sendromu, 607323, Otozomal Dominant; DRRS (Akro-Reno-Okuler Sendrom (Duane-Radius Sendromu)) (SALL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Duane-radial ray syndrome, 607323, Autosomal dominant; DRRS (Acro-renal-ocular syndrome) (SALL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SALL4	SALL4, HSAL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Duane Radyal Ray Sendromu, 607323, Otozomal Dominant; DRRS (Akro-Reno-Okuler Sendrom (Duane-Radius Sendromu)) (Prenatal) (SALL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Duane-radial ray syndrome, 607323, Autosomal dominant; DRRS (Acro-renal-ocular syndrome) (SALL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SALL4	SALL4, HSAL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dubin-Johnson Sendromu, 237500, Otozomal Resesif; DJS (Dubin-Johnson Sendromu) (ABCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dubin-Johnson syndrome, 237500, Autosomal recessive; DJS (Dubin-Johnson syndrome) (ABCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC2	ABCC2, CMOAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dubin-Johnson Sendromu, 237500, Otozomal Resesif; DJS (Dubin-Johnson Sendromu) (Prenatal) (ABCC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dubin-Johnson syndrome, 237500, Autosomal recessive; DJS (Dubin-Johnson syndrome) (ABCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCC2	ABCC2, CMOAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Duchenne Müsküler Distrofi, 310200, X'e Bağlı Resesif; DMD (Duchenne Müsküler Distrofi) (DMD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Duchenne muscular dystrophy, 310200, X-linked recessive; DMD (Duchenne muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Duchenne Müsküler Distrofi, 310200, X'e Bağlı Resesif; DMD (Duchenne Müsküler Distrofi) (MLPA)	Duchenne muscular dystrophy, 310200, X-linked recessive; DMD (Duchenne muscular dystrophy) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Duchenne Müsküler Distrofi, 310200, X'e Bağlı Resesif; DMD (Duchenne Müsküler Distrofi) (Prenatal) (DMD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Duchenne muscular dystrophy, 310200, X-linked recessive; DMD (Duchenne muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. [ECO:0000269	PubMed:10573008]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Duchenne Müsküler Distrofi, 310200, X'e Bağlı Resesif; DMD (Duchenne Müsküler Distrofi) (Prenatal) (MLPA)	Duchenne muscular dystrophy, 310200, X-linked recessive; DMD (Duchenne muscular dystrophy) (Prenatal) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. [ECO:0000269	PubMed:10573008]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dursun Sendromu, 612541, Otozomal Resesif (Dursun Sendromu) (G6PC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dursun syndrome, 612541, Autosomal recessive (Dursun syndrome) (G6PC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PC3	G6PC3, UGRP, SCN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dursun syndrome (DURSS) [MIM:612541]: A disease characterized by pulmonary arterial hypertension, cardiac abnormalities including secundum-type atrial septal defect, intermittent neutropenia, lymphopenia, monocytosis and anemia. [ECO:0000269	PubMed:20799326]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Dursun Sendromu, 612541, Otozomal Resesif (Dursun Sendromu) (Prenatal) (G6PC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dursun syndrome, 612541, Autosomal recessive (Dursun syndrome) (G6PC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	G6PC3	G6PC3, UGRP, SCN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dursun syndrome (DURSS) [MIM:612541]: A disease characterized by pulmonary arterial hypertension, cardiac abnormalities including secundum-type atrial septal defect, intermittent neutropenia, lymphopenia, monocytosis and anemia. [ECO:0000269	PubMed:20799326]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dyggve-Melchior-Clausen Hastalığı, 223800, Otozomal Resesif; DMC (Dyggve-Melchior-Clausen Hastalığı) (DYM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyggve-Melchior-Clausen disease, 223800, Autosomal recessive; DMC (Dyggve-Melchior-Clausen disease) (DYM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYM	DYM, FLJ90130, DMC, SMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyggve-Melchior-Clausen syndrome (DMC) [MIM:223800]: A rare autosomal recessive disorder belonging to the group of spondyloepimetaphyseal dysplasias. DMC is characterized by progressive short stature with short trunk dwarfism, microcephaly, protruding sternum, and psychomotor retardation. Radiological features include a platyspondyly with double vertebral humps, an epiphyso-metaphyseal dysplasia and lacy pelvis iliac crests. [ECO:0000269|PubMed:12491225, ECO:0000269|PubMed:12554689]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dyggve-Melchior-Clausen Hastalığı, 223800, Otozomal Resesif; DMC (Dyggve-Melchior-Clausen Hastalığı) (Prenatal) (DYM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyggve-Melchior-Clausen disease, 223800, Autosomal recessive; DMC (Dyggve-Melchior-Clausen disease) (DYM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYM	DYM, FLJ90130, DMC, SMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyggve-Melchior-Clausen syndrome (DMC) [MIM:223800]: A rare autosomal recessive disorder belonging to the group of spondyloepimetaphyseal dysplasias. DMC is characterized by progressive short stature with short trunk dwarfism, microcephaly, protruding sternum, and psychomotor retardation. Radiological features include a platyspondyly with double vertebral humps, an epiphyso-metaphyseal dysplasia and lacy pelvis iliac crests. [ECO:0000269|PubMed:12491225, ECO:0000269|PubMed:12554689]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disotonomi, Allesel, 223900, Otozomal Resesif (Allesel Disotonomi) (ELP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysautonomia, familial, 223900, Autosomal recessive (Familial dysautonomia) (IKBKAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELP1	IKBKAP, IKAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 3 (HSAN3) [MIM:223900]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN3 patients manifest a variety of symptoms such as alacrima, decreased taste, decreased sensitivity to pain and temperature, vasomotor instability, hypoactive or absent deep tendon reflexes, vomiting crises, and gastrointestinal dysfunction. [ECO:0000269|PubMed:11179008, ECO:0000269|PubMed:11179021]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diskromatozis Simetrica Hereditariya, 127400, Otozomal Dominant; DSH (Simetrik Kahsal Diskromatoz) (ADAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyschromatosis symmetrica hereditaria, 127400, Autosomal dominant; DSH (Dyschromatosis symmetrica hereditaria) (ADAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAR	ADAR, DRADA, DSH, DSRAD, IFI4, G1P1, AGS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyschromatosis symmetrica hereditaria (DSH) [MIM:127400]: An autosomal dominant pigmentary genodermatosis characterized by a mixture of hyperpigmented and hypopigmented macules distributed on the face and the dorsal parts of the hands and feet, that appear in infancy or early childhood. [ECO:0000269|PubMed:12916015, ECO:0000269|PubMed:15146470, ECO:0000269|PubMed:15659327]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diskromatozis Universalis Hereditaria 3, 615402, Otozomal Dominant; DUH3 (Diskokatozis Universalis) (ABCB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyschromatosis universalis hereditaria 3, 615402, Autosomal dominant; DUH3 (Dyschromatosis universalis) (ABCB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB6	ABCB6, MTABC3, MCOPCB7, LAN, DUH3, PSHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Tip 1a, 224120, Otozomal Resesif; CDAN1A (Konjenital Diseritropoietik Anemi Tip 1) (CDAN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyserythropoietic anemia, congenital, type Ia, 224120, Autosomal recessive; CDAN1A (Congenital dyserythropoietic anemia type I) (CDAN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDAN1	CDAN1, CDA1, CDAN1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Tip 1a, 224120, Otozomal Resesif; CDAN1A (Konjenital Diseritropoietik Anemi Tip 1) (Prenatal) (CDAN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyserythropoietic anemia, congenital, type Ia, 224120, Autosomal recessive; CDAN1A (Congenital dyserythropoietic anemia type I) (CDAN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDAN1	CDAN1, CDA1, CDAN1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Diseritropoietik Anemi, Konjenital, Tip 1b, 615631, Otozomal Resesif, CDAN1B (Konjenital Diseritropoietik Anemi Tip 1) (C15orf41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyserythropoietic anemia, congenital, type Ib, 615631, Autosomal recessive; CDAN1B (Congenital dyserythropoietic anemia type I) (C15orf41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C15orf41	C15orf41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, congenital dyserythropoietic, 1B (CDAN1B) [MIM:615631]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, macrocytic anemia and secondary hemochromatosis. It is occasionally associated with bone abnormalities, especially of the hands and feet (acrodysostosis), nail hypoplasia, and scoliosis. Ultrastructural features include internuclear chromatin bridges connecting some nearly completely separated erythroblasts and an abnormal appearance (spongy or Swiss-cheese appearance) of the heterochromatin in a high proportion of the erythroblasts. {ECO:0000269	PubMed:23716552}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Tip 1b, 615631, Otozomal Resesif, CDAN1B (Konjenital Diseritropoietik Anemi Tip 1) (Prenatal) (C15orf41 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyserythropoietic anemia, congenital, type Ib, 615631, Autosomal recessive; CDAN1B (Congenital dyserythropoietic anemia type I) (C15orf41 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C15orf41	C15orf41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, congenital dyserythropoietic, 1B (CDAN1B) [MIM:615631]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, macrocytic anemia and secondary hemochromatosis. It is occasionally associated with bone abnormalities, especially of the hands and feet (acrodysostosis), nail hypoplasia, and scoliosis. Ultrastructural features include internuclear chromatin bridges connecting some nearly completely separated erythroblasts and an abnormal appearance (spongy or Swiss-cheese appearance) of the heterochromatin in a high proportion of the erythroblasts. {ECO:0000269	PubMed:23716552}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Otozomal Resesif Tip 2, 224100 (Konjenital Diseritropoietik Anemi Tip 2) (SEC23B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyserythropoietic anemia, congenital, type II, 224100, Autosomal recessive (Congenital dyserythropoietic anemia type II) (SEC23B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC23B	SEC23B, CDAN2, HEMPAS, CWS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, congenital dyserythropoietic, 2 (CDAN2) [MIM:224100]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, normocytic anemia, iron overload, jaundice, and variable splenomegaly. Ultrastructural features include bi- or multinucleated erythroblasts in bone marrow, karyorrhexis, and the presence of Gaucher-like bone marrow histiocytes. The main biochemical feature of the disease is defective glycosylation of some red blood cells membrane proteins. {ECO:0000269|PubMed:19561605, ECO:0000269|PubMed:19621418}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Otozomal Resesif Tip 2, 224100 (Konjenital Diseritropoietik Anemi Tip 2) (Prenatal) (SEC23B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyserythropoietic anemia, congenital, type II, 224100, Autosomal recessive (Congenital dyserythropoietic anemia type II) (SEC23B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEC23B	SEC23B, CDAN2, HEMPAS, CWS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, congenital dyserythropoietic, 2 (CDAN2) [MIM:224100]: An autosomal recessive blood disorder characterized by morphological abnormalities of erythroblasts, ineffective erythropoiesis, normocytic anemia, iron overload, jaundice, and variable splenomegaly. Ultrastructural features include bi- or multinucleated erythroblasts in bone marrow, karyorrhexis, and the presence of Gaucher-like bone marrow histiocytes. The main biochemical feature of the disease is defective glycosylation of some red blood cells membrane proteins. {ECO:0000269|PubMed:19561605, ECO:0000269|PubMed:19621418}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Tip 4, 613673, Otozomal Dominant (Konjenital Diseritropoietik Anemi Tip 4) (KLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyserythropoietic anemia, congenital, type IV, 613673, Autosomal dominant (Congenital dyserythropoietic anemia type IV) (KLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, congenital dyserythropoietic, 4 (CDAN4) [MIM:613673]: A blood disorder characterized by ineffective erythropoiesis and hemolysis resulting in anemia. Circulating erythroblasts and erythroblasts in the bone marrow show various morphologic abnormalities. Affected individuals with CDA4 also have increased levels of fetal hemoglobin. {ECO:0000269|PubMed:21055716, ECO:0000269|PubMed:25585695}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diseritropoietik Anemi, Konjenital, Tip 4, 613673, Otozomal Dominant (Konjenital Diseritropoietik Anemi Tip 4) (Prenatal) (KLF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyserythropoietic anemia, congenital, type IV, 613673, Autosomal dominant (Congenital dyserythropoietic anemia type IV) (KLF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, congenital dyserythropoietic, 4 (CDAN4) [MIM:613673]: A blood disorder characterized by ineffective erythropoiesis and hemolysis resulting in anemia. Circulating erythroblasts and erythroblasts in the bone marrow show various morphologic abnormalities. Affected individuals with CDA4 also have increased levels of fetal hemoglobin. {ECO:0000269|PubMed:21055716, ECO:0000269|PubMed:25585695}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Disfibrinojenemi) (FGB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysfibrinogenemia, congenital, 616004 (Familial dysfibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Disfibrinojenemi) (FGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysfibrinogenemia, congenital, 616004 (Familial dysfibrinogenemia) (FGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Disfibrinojenemi) (Prenatal) (FGB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dysfibrinogenemia, congenital, 616004 (Familial dysfibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Disfibrinojenemi) (Prenatal) (FGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dysfibrinogenemia, congenital, 616004 (Familial dysfibrinogenemia) (FGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Hipodisfibrinojenemi) (FGG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysfibrinogenemia, congenital, 616004 (Familial hypodysfibrinogenemia) (FGG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Disfibrinojenemi, Konjenital, 616004 (Ailesel Hipodisfibrinojenemi) (Prenatal) (FGG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dysfibrinogenemia, congenital, 616004 (Familial hypodysfibrinogenemia) (FGG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 1, 127550, Otozomal Dominant; DKCA1 (Diskeratozis Congenita) (TERC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal dominant 1, 127550, Autosomal dominant; DKCA1 (Dyskeratosis congenita) (TERC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 1, 127550, Otozomal Dominant; DKCA1 (Diskeratozis Congenita) (MLPA)	Dyskeratosis congenita, autosomal dominant 1, 127550, Autosomal dominant; DKCA1 (Dyskeratosis congenita) (MLPA)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 1, 127550, Otozomal Dominant; DKCA1 (Diskeratozis Congenita) (Prenatal) (TERC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal dominant 1, 127550, Autosomal dominant; DKCA1 (Dyskeratosis congenita) (TERC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 1, 127550, Otozomal Dominant; DKCA1 (Diskeratozis Congenita) (Prenatal) (MLPA)	Dyskeratosis congenita, autosomal dominant 1, 127550, Autosomal dominant; DKCA1 (Dyskeratosis congenita) (Prenatal) (MLPA)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 2, 613989, Otozomal Resesif, Otozomal Dominant; DKCA2 (Diskeratozis Congenita) (TERT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal dominant 2, 613989, Autosomal recessive, Autosomal dominant; DKCA2 (Dyskeratosis congenita) (TERT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 2, 613989, Otozomal Resesif, Otozomal Dominant; DKCA2 (Diskeratozis Congenita) (Prenatal) (TERT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal dominant 2, 613989, Autosomal recessive, Autosomal dominant; DKCA2 (Dyskeratosis congenita) (TERT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 3, 613990, Otozomal Dominant; DKCA3 (Diskeratozis Congenita) (TINF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal dominant 3, 613990, Autosomal dominant; DKCA3 (Dyskeratosis congenita) (TINF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TINF2	TINF2, TIN2, DKCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal dominant, 5 (DKCA5) [MIM:268130]: A disease characterized by bone marrow hypoplasia, nail dystrophy, fine sparse hair, fine reticulate skin pigmentation, oral leukoplakia, bilateral exudative retinopathy, cerebellar hypoplasia, and growth retardation. {ECO:0000269	PubMed:18252230}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 3, 613990, Otozomal Dominant; DKCA3 (Diskeratozis Congenita) (Prenatal) (TINF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal dominant 3, 613990, Autosomal dominant; DKCA3 (Dyskeratosis congenita) (TINF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TINF2	TINF2, TIN2, DKCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal dominant, 5 (DKCA5) [MIM:268130]: A disease characterized by bone marrow hypoplasia, nail dystrophy, fine sparse hair, fine reticulate skin pigmentation, oral leukoplakia, bilateral exudative retinopathy, cerebellar hypoplasia, and growth retardation. {ECO:0000269	PubMed:18252230}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Diskeratozis Konjenita, Otozomal Dominant 4, 615190, Otozomal Resesif, Otozomal Dominant (Diskeratozis Congenita) (RTEL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal dominant 4, 615190, Autosomal recessive, Autosomal dominant (Dyskeratosis congenita) (RTEL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTEL1	RTEL1, C20orf41, NHL, KIAA1088, DKCB5, DKCA4, PFBMFT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453664, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). {ECO:0000269|PubMed:23329068}.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 4, 615190, Otozomal Resesif, Otozomal Dominant (Diskeratozis Congenita) (Prenatal) (RTEL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal dominant 4, 615190, Autosomal recessive, Autosomal dominant (Dyskeratosis congenita) (RTEL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RTEL1	RTEL1, C20orf41, NHL, KIAA1088, DKCB5, DKCA4, PFBMFT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453664, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). {ECO:0000269|PubMed:23329068}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 6, 616553, Otozomal Resesif, Otozomal Dominant; DKCA6 (Hoyeraal-Hreidarsson Sendromu) (ACD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal dominant 6, 616553, Autosomal recessive, Autosomal dominant; DKCA6 (hoyeraal-Hreidarsson syndrome) (ACD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACD	ACD, PTOP, PIP1, TINT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Dominant 6, 616553, Otozomal Resesif, Otozomal Dominant; DKCA6 (Hoyeraal-Hreidarsson Sendromu) (Prenatal) (ACD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal dominant 6, 616553, Autosomal recessive, Autosomal dominant; DKCA6 (hoyeraal-Hreidarsson syndrome) (ACD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACD	ACD, PTOP, PIP1, TINT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 1, 224230, Otozomal Resesif, DKCB1 (Diskeratozis Congenita) (NOP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 1, 224230, Autosomal recessive; DKCB1 (Dyskeratosis congenita) (NOP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOP10	NOLA3, NOP10, DKCB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 1 (DKCB1) [MIM:224230]: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:17507419}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 1, 224230, Otozomal Resesif, DKCB1 (Diskeratozis Congenita) (Prenatal) (NOP10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 1, 224230, Autosomal recessive; DKCB1 (Dyskeratosis congenita) (NOP10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOP10	NOLA3, NOP10, DKCB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 1 (DKCB1) [MIM:224230]: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:17507419}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 2, 613987, Otozomal Resesif, DKCB2 (Diskeratozis Congenita) (NHP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 2, 613987, Autosomal recessive; DKCB2 (Dyskeratosis congenita) (NHP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NHP2	NOLA2, NHP2, DKCB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 2 (DKCB2) [MIM:613987]: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:18523010}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 2, 613987, Otozomal Resesif, DKCB2 (Diskeratozis Congenita) (Prenatal) (NHP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 2, 613987, Autosomal recessive; DKCB2 (Dyskeratosis congenita) (NHP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NHP2	NOLA2, NHP2, DKCB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 2 (DKCB2) [MIM:613987]: A rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:18523010}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 3, 613988, Otozomal Resesif, DKCB3 (Diskeratozis Congenita) (WRAP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 3, 613988, Autosomal recessive; DKCB3 (Dyskeratosis congenita) (WRAP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WRAP53	WRAP53, TCAB1, WDR79, DKCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 3, 613988, Otozomal Resesif, DKCB3 (Diskeratozis Congenita) (Prenatal) (WRAP53 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 3, 613988, Autosomal recessive; DKCB3 (Dyskeratosis congenita) (WRAP53 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WRAP53	WRAP53, TCAB1, WDR79, DKCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratoz Konjenita, Otozomal Resesif 4, 613989, Otozomal Resesif, Otozomal Dominant (Diskeratozis Konjenita) (TERT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 4, 613989, Autosomal recessive, Autosomal dominant (Dyskeratosis congenita) (TERT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diskeratoz Konjenita, Otozomal Resesif 4, 613989, Otozomal Resesif, Otozomal Dominant (Diskeratozis Konjenita) (Prenatal) (TERT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 4, 613989, Autosomal recessive, Autosomal dominant (Dyskeratosis congenita) (TERT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 5, 615190, Otozomal Resesif, Otozomal Dominant, DKCB5 (Diskeratozis Congenita) (RTEL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 5, 615190, Autosomal recessive, Autosomal dominant; DKCB5 (Dyskeratosis congenita) (RTEL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTEL1	RTEL1, C20orf41, NHL, KIAA1088, DKCB5, DKCA4, PFBMFT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453664, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). {ECO:0000269|PubMed:23329068}.	EDTA Blood Sample

Diskeratozis Konjenita, Otozomal Resesif 5, 615190, Otozomal Resesif, Otozomal Dominant, DKCB5 (Diskeratozis Congenita) (Prenatal) (RTEL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 5, 615190, Autosomal recessive, Autosomal dominant; DKCB5 (Dyskeratosis congenita) (RTEL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RTEL1	RTEL1, C20orf41, NHL, KIAA1088, DKCB5, DKCA4, PFBMFT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453864, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). {ECO:0000269|PubMed:23329068}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 6, 616353, Otozomal Resesif; DKCB6 (Diskeratozis Congenita) (PARN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, autosomal recessive 6, 616353, Autosomal recessive; DKCB6 (Dyskeratosis congenita) (PARN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PARN	PARN, DAN, DKCB6, PFBMFT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 6 (DKCB6) [MIM:616353]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:25893599}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diskeratozis Konjenita, Otozomal Resesif 6, 616353, Otozomal Resesif; DKCB6 (Diskeratozis Congenita) (Prenatal) (PARN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, autosomal recessive 6, 616353, Autosomal recessive; DKCB6 (Dyskeratosis congenita) (PARN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PARN	PARN, DAN, DKCB6, PFBMFT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 6 (DKCB6) [MIM:616353]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269	PubMed:25893599}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskeratozis Konjenita, X'e Bağlı, 305000, X'e Bağlı; DKCX (Diskeratozis Congenita) (DKC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskeratosis congenita, X-linked, 305000, X-linked recessive; DKCX (Dyskeratosis congenita) (DKC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DKC1	DKC1, DKCX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Hoyeraal-Hreidarsson syndrome (HHS) [MIM:305000]: A clinically severe variant of dyskeratosis congenita that is characterized by multisystem involvement, early onset in utero, and often results in death in childhood. Affected individuals show intrauterine growth retardation, microcephaly, cerebellar hypoplasia, delayed development, and bone marrow failure resulting in immunodeficiency. {ECO:0000269|PubMed:10583221, ECO:0000269|PubMed:12437656, ECO:0000269|PubMed:19734544, ECO:0000269|PubMed:24914498}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diskeratozis Konjenita, X'e Bağlı, 305000, X'e Bağlı; DKCX (Diskeratozis Congenita) (Prenatal) (DKC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskeratosis congenita, X-linked, 305000, X-linked recessive; DKCX (Dyskeratosis congenita) (DKC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DKC1	DKC1, DKCX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hoyeraal-Hreidarsson syndrome (HHS) [MIM:305000]: A clinically severe variant of dyskeratosis congenita that is characterized by multisystem involvement, early onset in utero, and often results in death in childhood. Affected individuals show intrauterine growth retardation, microcephaly, cerebellar hypoplasia, delayed development, and bone marrow failure resulting in immunodeficiency. {ECO:0000269|PubMed:10583221, ECO:0000269|PubMed:12437656, ECO:0000269|PubMed:19734544, ECO:0000269|PubMed:24914498}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Diskinezi, Ailesel, Yüz Miyokimisi, 606703, Otozomal Dominant (Ailesel Diskinezi ve Fasiyal Miyokimi) (ADCY5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskinesia, familial, with facial myokymia, 606703, Autosomal dominant (Familial dyskinesia and facial myokymia) (ADCY5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADCY5	ADCY5, FDFM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Dyskinesia, familial, with facial myokymia (FDFM) [MIM:606703]: A disorder characterized by predominantly perioral and periorbital myokymia, and face, neck and upper limb dystonic/choreic movements. Initially paroxysmal and worsened by stress, the dyskinetic episodes become nearly constant by the end of the third decade of life, but in some individuals, they may diminish in frequency and severity at older ages. {ECO:0000269	PubMed:22782511, ECO:0000269	PubMed:24700542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diskinezi, Bacak ve Orofasiyal, İnfantil Başlangıçlı, 616921, Otozomal Resesif (PDE10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskinesia, limb and orofacial, infantile-onset, 616921, Autosomal recessive (PDE10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE10A	PDE10A, IOLOD, ADSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Diskinezi, Nöbetler ve Entelektüel Gelişim Bozukluğu, 617171, Otozomal Resesif (Mental Retardasyon Epilepsi-Ekstrapiramidal Sendromi) (DEAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyskinesia, seizures, and intellectual developmental disorder, 617171, Autosomal recessive (Intellectual disability-epilepsy-extrapyramidal syndrome) (DEAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DEAF1	DEAF1, SPN, ZMYND5, MRD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Diskinezi, Nöbetler ve Entelektüel Gelişim Bozukluğu, 617171, Otozomal Resesif (Mental Retardasyon Epilepsi-Ekstrapiramidal Sendromi) (Prenatal) (DEAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dyskinesia, seizures, and intellectual developmental disorder, 617171, Autosomal recessive (Intellectual disability-epilepsy-extrapyramidal syndrome) (DEAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DEAF1	DEAF1, SPN, ZMYND5, MRD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disleksi, Yatkınlık, 1, 127700, Otozomal Dominant (DYX1C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyslexia, susceptibility to, 1, 127700, Autosomal dominant (DYX1C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYX1C1	DYX1C1, DYXC1, DYX1, CILD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Disleksi, 2 Yatkınlık, 600202, Otozomal Dominant (KIAA0319 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyslexia, susceptibility to, 2, 600202, Autosomal dominant (KIAA0319 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA0319	KIAA0319, DYX2, DYLX2, DLX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Dyslexia 2 (DYX2) [MIM:600202]: A relatively common, complex cognitive disorder characterized by an impairment of reading performance despite adequate motivational, educational and intellectual opportunities. It is a multifactorial trait, with evidence for familial clustering and heritability. {ECO:0000269	PubMed:16600991}. Note=Disease susceptibility is associated with variations affecting the gene representedin this entry.	EDTA Blood Sample
Displazminojenemi, 217090, Otozomal Resesif (Hipoplazminojenemi) (PLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysplasminogenemia, 217090, Autosomal recessive (Hypoplasminogenemia) (PLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLG	PLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Plasminogen deficiency (PLGD) [MIM:217090]: A disorder characterized by decreased serum plasminogen activity. Two forms of the disorder are distinguished: type 1 deficiency is additionally characterized by decreased plasminogen antigen levels and clinical symptoms, whereas type 2 deficiency, also known as dysplasminogenemia, is characterized by normal, or slightly reduced antigen levels, and absence of clinical manifestations. Plasminogen deficiency type 1 results in markedly impaired extracellular fibrinolysis and chronic mucosal pseudomembranous lesions due to subepithelial fibrin deposition and inflammation. The most common clinical manifestation of type 1 deficiency is ligneous conjunctivitis in which pseudomembranes formation on the palpebral surfaces of the eye progresses to white, yellow-white, or red thick masses with a wood-like consistency that replace the normal mucosa. {ECO:0000269|PubMed:10233898, ECO:0000269|PubMed:1427790, ECO:0000269|PubMed:1986355, ECO:0000269|PubMed:6216475, ECO:0000269|PubMed:6238949, ECO:0000269|PubMed:8302398, ECO:0000269|PubMed:9242524, ECO:0000269|PubMed:9858247}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Disprotrombinemi, 613679, Otozomal Resesif (Konjenital Faktör 2 Eksikliği) (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysprothrombinemia, 613679, Autosomal recessive (Congenital factor II deficiency) (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Dysegmental Displazi, Silverman-Handmaker Tipi, 224410, Otozomal Resesif; DDSH (Disegmental Displazi, Silverman-Handmaker Tipi) (HSPG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dysegmental dysplasia, Silverman-Handmaker type, 224410, Autosomal recessive; DDSH (Dysegmental dysplasia, Silverman-Handmaker type) (HSPG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPG2	HSPG2, PLC, SJS, SJA, SJS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Dysegmental dysplasia Silverman-Handmaker type (DDSH) [MIM:224410]: The dysegmental dysplasias are rare, autosomal recessive skeletal dysplasias with anisospondyly and micromelia. There are two recognized types: the severe, lethal DDSH and the milder Rolland-Desbuquois form. Individuals with DDSH also have a flat face, micrognathia, cleft palate and reduced joint mobility, and frequently have an encephalocoele. The endochondral growth plate is short, the calcospherites (which are spherical calcium- phosphorus crystals produced by hypertrophic chondrocytes) are unfused, and there is mucoid degeneration of the resting cartilage. {ECO:0000269	PubMed:11279527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dysegmental Displazi, Silverman-Handmaker Tipi, 224410, Otozomal Resesif; DDSH (Disegmental Displazi, Silverman-Handmaker Tipi) (Prenatal) (HSPG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dysegmental dysplasia, Silverman-Handmaker type, 224410, Autosomal recessive; DDSH (Dysegmental dysplasia, Silverman-Handmaker type) (HSPG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPG2	HSPG2, PLC, SJS, SJA, SJS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dysegmental dysplasia Silverman-Handmaker type (DDSH) [MIM:224410]: The dysegmental dysplasias are rare, autosomal recessive skeletal dysplasias with anisospondyly and micromelia. There are two recognized types: the severe, lethal DDSH and the milder Rolland-Desbuquois form. Individuals with DDSH also have a flat face, micrognathia, cleft palate and reduced joint mobility, and frequently have an encephalocoele. The endochondral growth plate is short, the calcospherites (which are spherical calcium- phosphorus crystals produced by hypertrophic chondrocytes) are unfused, and there is mucoid degeneration of the resting cartilage. {ECO:0000269	PubMed:11279527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disspondiloenkondromatozis (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dyspondyloenchondromatosis (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	.	EDTA Blood Sample
Disspondiloenkondromatozis (MLPA)	Dysspondyloenchondromatosis (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	.	EDTA Blood Sample

Disspondiloenkondromatozis (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Dysspondyboenchondromatosis (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Disspondiloenkondromatozis (Prenatal) (MLPA)	Dysspondyloenchondromatosis (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Distoni 1, Torsiyon, Otozomal Dominant, DYT1 (Erken Başlangıçlı Jeneralize Ekstremite Başlangıçlı Distoni) (TOR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	DYSTONIA 1, TORSION, AUTOSOMAL DOMINANT; DYT1 (Early-onset generalized limb-onset dystonia) (TOR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOR1A	DYT1, TOR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 1, torsion, autosomal dominant (DYT1) [MIM:128100]: A primary torsion dystonia, and the most common and severe form. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 1 is characterized by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body, in the absence of other neurological symptoms. Typically, symptoms develop first in an arm or leg in middle to late childhood and progress in approximately 30% of patients to other body regions (generalized dystonia) within about five years. 'Torsion' refers to the twisting nature of body movements observed in DYT1, often affecting the trunk. Distribution and severity of symptoms vary widely between affected individuals, ranging from mild focal dystonia to severe generalized dystonia, even within families. {ECO:0000269|PubMed:18477710, ECO:0000269|PubMed:19955557, ECO:0000269|PubMed:9288096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 1, Torsiyon, Otozomal Dominant, DYT1 (Erken Başlangıçlı Jeneralize Ekstremite Başlangıçlı Distoni) (MLPA)	DYSTONIA 1, TORSION, AUTOSOMAL DOMINANT; DYT1 (Early-onset generalized limb-onset dystonia) (MLPA)	TOR1A	DYT1, TOR1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 1, torsion, autosomal dominant (DYT1) [MIM:128100]: A primary torsion dystonia, and the most common and severe form. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 1 is characterized by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body, in the absence of other neurological symptoms. Typically, symptoms develop first in an arm or leg in middle to late childhood and progress in approximately 30% of patients to other body regions (generalized dystonia) within about five years. 'Torsion' refers to the twisting nature of body movements observed in DYT1, often affecting the trunk. Distribution and severity of symptoms vary widely between affected individuals, ranging from mild focal dystonia to severe generalized dystonia, even within families. {ECO:0000269|PubMed:18477710, ECO:0000269|PubMed:19955557, ECO:0000269|PubMed:9288096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 16, 612067, Otozomal Resesif (Distoni 16) (PRKRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 16, 612067, Autosomal recessive (Dystonia 16) (PRKRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKRA	PRKRA, PACT, RAX, DYT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 16 (DYT16) [MIM:612067]: An early-onset dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contraction, often leading to abnormal postures. DYT16 patients have progressive, generalized dystonia with axial muscle involvement, oro-mandibular (sardonic smile) and laryngeal dystonia and, in some cases, parkinsonian features. {ECO:0000269|PubMed:18243799, ECO:0000269|PubMed:18420150}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 16, 612067, Otozomal Resesif (Distoni 16) (MLPA)	Dystonia 16, 612067, Autosomal recessive (Dystonia 16) (MLPA)	PRKRA	PRKRA, PACT, RAX, DYT16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 16 (DYT16) [MIM:612067]: An early-onset dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contraction, often leading to abnormal postures. DYT16 patients have progressive, generalized dystonia with axial muscle involvement, oro-mandibular (sardonic smile) and laryngeal dystonia and, in some cases, parkinsonian features. {ECO:0000269|PubMed:18243799, ECO:0000269|PubMed:18420150}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 2, Torsiyon, Otozomal Resesif, 224500, Otozomal Resesif; DYT2 (Primer Distoni, DYT2 Tipi) (HPCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 2, torsion, autosomal recessive, 224500, Autosomal recessive; DYT2 (Primary dystonia, DYT2 type) (HPCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPCA	HPCA, DYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 2, torsion, autosomal recessive (DYT2) [MIM:224500]: A form of torsion dystonia, a disease defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. 'Torsion' refers to the twisting nature of body movements, often affecting the trunk. DYT2 is a slowly progressive form that first affects distal limbs and later involves the neck, orofacial, and craniocervical regions. {ECO:0000269|PubMed:25799188}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 23, 614860, Otozomal Dominant; DYT23 (Yetişkin Başlangıçlı Servikal Distoni, DYT23 Tipi) (CACNA1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 23, 614860, Autosomal dominant; DYT23 (Adult-onset cervical dystonia, DYT23 type) (CACNA1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1B	CACNA1B, CACNL1A5, DYT23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 23 (DYT23) [MIM:614860]: A form of dystonia, a disorder defined by the presence of sustained involuntary muscle contraction, often leading to abnormal postures. DYT23 is an autosomal dominant dystonia affecting the face, neck, limbs. Some DYT23 patients manifest generalized myoclonus in addition to progressive action-induced multifocal dystonia. {ECO:0000269|PubMed:25296916}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 24, 615034, Otozomal Dominant; DYT24 (Kranioservikal Distoni-Larinks ve Üst Ekstremite Tutulumu (Distoni 24)) (ANO3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 24, 615034, Autosomal dominant; DYT24 (Cranio-cervical dystonia with laryngeal and upper-limb involvement) (ANO3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO3	ANO3, TMEM16C, C11orf25, DYT24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni 25, 615073, Otozomal Dominant (Otozomal Dominant Fokal Distoni, DYT25) (GNAL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 25, 615073, Autosomal dominant (Autosomal dominant focal dystonia, DYT25) (GNAL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAL	GNAL, DYT25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni 26, Miyoklonik, 616398, Otozomal Dominant (Miyoklonus-Distoni Sendromu) (KCTD17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 26, myoclonic, 616398, Autosomal dominant (Myoclonus-dystonia syndrome) (KCTD17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCTD17	KCTD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni 27, 616411, Otozomal Resesif (Primer Distoni, DYT27 Tipi) (COL6A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 27, 616411, Autosomal recessive (Primary dystonia, DYT27 type) (COL6A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A3	COL6A3, DYT27, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Distoni 28, Çocuklukta Başlayan, 617284, Otozomal Dominant (KMT2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 28, childhood-onset, 617284, Autosomal dominant (KMT2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KMT2B	KMT2B, MLL4, KIAA0304, DYT28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni 3, Torsiyon, X'e Bağlı; Dy3 (X'e Bağlı Distoni-Parkinsonizm) (TAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	DYSTONIA 3, TORSION, X-LINKED; DYT3 (X-linked dystonia-parkinsonism) (TAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF1	TAF1, TAF2A, CCG1, BA2R, DYT3, MRXS33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 3, torsion, X-linked (DYT3) [MIM:314250]: A X- linked dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT3 is characterized by severe progressive torsion dystonia followed by parkinsonism. It has a well-defined pathology of extensive neuronal loss and mosaic gliosis in the striatum (caudate nucleus and putamen) which appears to resemble that in Huntington disease. {ECO:0000269|PubMed:12928496, ECO:0000269|PubMed:17273961}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 4, Torsiyon, Otozomal Dominant, 128101, Otozomal Dominant; DYT4 (Primer Distoni, DYT4 Tipi) (TUBB4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 4, torsion, autosomal dominant, 128101, Autosomal dominant; DYT4 (Primary dystonia, DYT4 type) (TUBB4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB4A	TUBB4A, DYT4, HLD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 4, torsion, autosomal dominant (DYT4) [MIM:128101]: A form of torsion dystonia, a disease defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. 'Torsion' refers to the twisting nature of body movements, often affecting the trunk. DYT4 is characterized by onset in the second to third decade of progressive laryngeal dysphonia followed by the involvement of other muscles, such as the neck or limbs. Some patients develop an ataxic gait. {ECO:0000269|PubMed:23424103}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Distoni 6, Torsiyon, 602629, Otozomal Dominant (Primer Distoni, DYT6 Tipi) (THAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 6, torsion, 602629, Autosomal dominant (Primary dystonia, DYT6 type) (THAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THAP1	THAP1, DYT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni 6, Torsiyon, 602629, Otozomal Dominant (Primer Distoni, DYT6 Tipi) (MLPA)	Dystonia 6, torsion, 602629, Autosomal dominant (Primary dystonia, DYT6 type) (MLPA)	THAP1	THAP1, DYT6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Distoni 9, 601042, Otozomal Dominant; DYT9 (Paroksismal Distonik Koreoatetoz-Epizodik Ataksi-Spastisite) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia 9, 601042, Autosomal dominant; DYT9 (Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269|PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni 9, 601042, Otozomal Dominant; DYT9 (Paroksismal Distonik Koreoatetoz-Epizodik Ataksi-Spastisite) (MLPA)	Dystonia 9, 601042, Autosomal dominant; DYT9 (Paroxysmal dystonic choreathetosis with episodic ataxia and spasticity) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269|PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni-1, Modifiye Edici (Erken Başlangıçlı Jeneralize Ekstremite Başlangıçlı Distoni) (TOR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia-1, modifier of (Early-onset generalized limb-onset dystonia) (TOR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOR1A	DYT1, TOR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 1, torsion, autosomal dominant (DYT1) [MIM:128100]: A primary torsion dystonia, and the most common and severe form. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 1 is characterized by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body, in the absence of other neurological symptoms. Typically, symptoms develop first in an arm or leg in middle to late childhood and progress in approximately 30% of patients to other body regions (generalized dystonia) within about five years. 'Torsion' refers to the twisting nature of body movements observed in DYT1, often affecting the trunk. Distribution and severity of symptoms vary widely between affected individuals, ranging from mild focal dystonia to severe generalized dystonia, even within families. {ECO:0000269|PubMed:18477710, ECO:0000269|PubMed:19955557, ECO:0000269|PubMed:9288096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Distoni-1, Modifiye Edici (Erken Başlangıçlı Jeneralize Ekstremite Başlangıçlı Distoni) (MLPA)	Dystonia-1, modifier of (Early-onset generalized limb-onset dystonia) (MLPA)	TOR1A	DYT1, TOR1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 1, torsion, autosomal dominant (DYT1) [MIM:128100]: A primary torsion dystonia, and the most common and severe form. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 1 is characterized by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body, in the absence of other neurological symptoms. Typically, symptoms develop first in an arm or leg in middle to late childhood and progress in approximately 30% of patients to other body regions (generalized dystonia) within about five years. 'Torsion' refers to the twisting nature of body movements observed in DYT1, often affecting the trunk. Distribution and severity of symptoms vary widely between affected individuals, ranging from mild focal dystonia to severe generalized dystonia, even within families. {ECO:0000269|PubMed:18477710, ECO:0000269|PubMed:19955557, ECO:0000269|PubMed:9288096). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni-1, Torsiyon, 128100, Otozomal Dominant (Erken Başlangıçlı Jeneralize Ekstremite Başlangıçlı Distoni) (MLPA)	Dystonia-1, torsion, 128100, Autosomal dominant (Early-onset generalized limb-onset dystonia) (MLPA)	TOR1A	DYT1, TOR1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 1, torsion, autosomal dominant (DYT1) [MIM:128100]: A primary torsion dystonia, and the most common and severe form. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 1 is characterized by involuntary, repetitive, sustained muscle contractions or postures involving one or more sites of the body, in the absence of other neurological symptoms. Typically, symptoms develop first in an arm or leg in middle to late childhood and progress in approximately 30% of patients to other body regions (generalized dystonia) within about five years. 'Torsion' refers to the twisting nature of body movements observed in DYT1, often affecting the trunk. Distribution and severity of symptoms vary widely between affected individuals, ranging from mild focal dystonia to severe generalized dystonia, even within families. {ECO:0000269|PubMed:18477710, ECO:0000269|PubMed:19955557, ECO:0000269|PubMed:9288096). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni-11, Miyoklonik, 159900, Otozomal Dominant; DYT11 (Miyoklonus-Distoni Sendromu) (SGCE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia-11, myoclonic, 159900, Autosomal dominant; DYT11 (Myoclonus-dystonia syndrome) (SGCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCE	SGCE, DYT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 11, myoclonic (DYT11) [MIM:159900]: A myoclonic dystonia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT11 is characterized by involuntary lightning jerks and dystonic movements and postures alleviated by alcohol. Inheritance is autosomal dominant. The age of onset, pattern of body involvement, presence of myoclonus and response to alcohol are all variable. {ECO:0000269|PubMed:11528394, ECO:0000269|PubMed:12402271, ECO:0000269|PubMed:15079037, ECO:0000269|PubMed:15258227, ECO:0000269|PubMed:16227522, ECO:0000269|PubMed:17853490, ECO:0000269|PubMed:18175340, ECO:0000269|PubMed:18362280, ECO:0000269|PubMed:19066193). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni-11, Miyoklonik, 159900, Otozomal Dominant; DYT11 (Miyoklonus-Distoni Sendromu) (MLPA)	Dystonia-11, myoclonic, 159900, Autosomal dominant; DYT11 (Myoclonus-dystonia syndrome) (MLPA)	SGCE	SGCE, DYT11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 11, myoclonic (DYT11) [MIM:159900]: A myoclonic dystonia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT11 is characterized by involuntary lightning jerks and dystonic movements and postures alleviated by alcohol. Inheritance is autosomal dominant. The age of onset, pattern of body involvement, presence of myoclonus and response to alcohol are all variable. {ECO:0000269|PubMed:11528394, ECO:0000269|PubMed:12402271, ECO:0000269|PubMed:15079037, ECO:0000269|PubMed:15258227, ECO:0000269|PubMed:16227522, ECO:0000269|PubMed:17853490, ECO:0000269|PubMed:18175340, ECO:0000269|PubMed:18362280, ECO:0000269|PubMed:19066193). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni-12, 128235, Otozomal Dominant; DYT12 (Hızlı Başlangıçlı Distoni-Parkinsonizmi) (ATP1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia-12, 128235, Autosomal dominant; DYT12 (Rapid-onset dystonia-parkinsonism) (ATP1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 12 (DYT12) [MIM:128235]: An autosomal dominant dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT12 patients develop dystonia and parkinsonism between 15 and 45 years of age. The disease is characterized by an unusually rapid evolution of signs and symptoms. The sudden onset of symptoms over hours to a few weeks, often associated with physical or emotional stress, suggests a trigger initiating a nervous system insult resulting in permanent neurologic disability. {ECO:0000269|PubMed:15260953, ECO:0000269|PubMed:19351654, ECO:0000269|PubMed:19652145). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Distoni-12, 128235, Otozomal Dominant; DYT12 (Hızlı Başlangıçlı Distoni-Parkinsonizmi) (MLPA)	Dystonia-12, 128235, Autosomal dominant; DYT12 (Rapid-onset dystonia-parkinsonism) (MLPA)	ATP1A3	ATP1A3, DYT12, RDP, AHC2, CAPOS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 12 (DYT12) [MIM:128235]: An autosomal dominant dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT12 patients develop dystonia and parkinsonism between 15 and 45 years of age. The disease is characterized by an unusually rapid evolution of signs and symptoms. The sudden onset of symptoms over hours to a few weeks, often associated with physical or emotional stress, suggests a trigger initiating a nervous system insult resulting in permanent neurologic disability. {ECO:0000269|PubMed:15260953, ECO:0000269|PubMed:19351654, ECO:0000269|PubMed:19652145). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Distoni-Parkinsonizm, X'e Bağlı, 314250, X'e Bağlı Resesif (X' e Bağlı Distoni-Parkinsonizmi) (TAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia-Parkinsonism, X-linked, 314250, X-linked recessive (X-linked dystonia-parkinsonism) (TAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF1	TAF1, TAF2A, CCG1, BA2R, DYT3, MRX533	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 3, torsion, X-linked (DYT3) [MIM:314250]: A X- linked dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT3 is characterized by severe progressive torsion dystonia followed by parkinsonism. It has a well-defined pathology of extensive neuronal loss and mosaic gliosis in the striatum (caudate nucleus and putamen) which appears to resemble that in Huntington disease. {ECO:0000269|PubMed:12929496, ECO:0000269|PubMed:17273961). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni, Çocuklukta Başlangıç, Optik Atrofi ve Bazal Ganglyon Anormallikleri, 617282, Otozomal Resesif (MECR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia, childhood-onset, with optic atrophy and basal ganglia abnormalities, 617282, Autosomal recessive (MECR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECR	MECR, NRBF1, DYTOABG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni, Dopa Yanıtlı, Sepiapterin Redüktaz Eksikliğine Bağlı, 612716, Otozomal Dominant, Otozomal Resesif (Dopa-Yanıtlı Distoni, Sepiapterin Redüktaz Eksikliğine Bağlı) (SPR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia, dopa-responsive, due to sepiapterin reductase deficiency, 612716, Autosomal dominant, Autosomal recessive (Dopa-responsive dystonia due to sepiapterin reductase deficiency) (SPR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPR	SPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia, DOPA-responsive, due to sepiapterin reductase deficiency (DRDSPRD) [MIM:612716]: A form of DOPA-responsive dystonia. In the majority of cases, patients manifest progressive psychomotor retardation, dystonia and spasticity. Cognitive anomalies are also often present. The disease is due to severe dopamine and serotonin deficiencies in the central nervous system caused by a defect in BH4 synthesis. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. {ECO:0000269|PubMed:11443547, ECO:0000269|PubMed:16650784, ECO:0000269|PubMed:17159114). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Distoni, DOPA-Duyarlı, ± Hiperfenilalaninemi, 128230, Otozomal Resesif, Otozomal Dominant; DRD (Otozomal Dominant Dopa-Duyarlı Distoni) (GCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230, Autosomal recessive, Autosomal dominant; DRD (Autosomal dominant dopa-responsive dystonia) (GCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCH1	GCH1, DYT5, HPABH4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7674165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Distoni, DOPA-Duyarlı, ± Hiperfenilalaninemi, 128230, Otozomal Resesif, Otozomal Dominant; DRD (Otozomal Dominant Dopa-Duyarlı Distoni) (MLPA)	Dystonia, DOPA-responsive, with or without hyperphenylalaninemia, 128230, Autosomal recessive, Autosomal dominant; DRD (Autosomal dominant dopa-responsive dystonia) (MLPA)	GCH1	GCH1, DYT5, HPABH4B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7874165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Distoni, Jüvenil-Onset, 607371, Otozomal Dominant; DJO (Gelişimsel Malformasyonlar-Sağırlık-Distoni Sendromu) (ACTB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia, juvenile-onset, 607371, Autosomal dominant; DJO (Developmental malformations-deafness-dystonia syndrome) (ACTB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTB	ACTB, BRWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Distoni, Primer Servikal (DRD5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Dystonia, primary cervical (DRD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD5	DRD5, DRD1B, DRD1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika Inversa, 226600, Otozomal Resesif (Resesif Distrofik Epidermolizis Büllosa Inversa) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica inversa, 226600, Autosomal recessive (Recessive dystrophic epidermolysis bullosa inversa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika Inversa, 226600, Otozomal Resesif (Resesif Distrofik Epidermolizis Büllosa Inversa) (MLPA)	Epidermolysis bullosa dystrophica inversa, 226600, Autosomal recessive (Recessive dystrophic epidermolysis bullosa inversa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika Inversa, 226600, Otozomal Resesif (Resesif Distrofik Epidermolizis Büllosa Inversa) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica inversa, 226600, Autosomal recessive (Recessive dystrophic epidermolysis bullosa inversa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Distrofika Inversa, 226600, Otozomal Resesif (Resesif Distrofik Epidermolizis Büllosa Inversa) (Prenatal) (MLPA)	Epidermolysis bullosa dystrophica inversa, 226600, Autosomal recessive (Recessive dystrophic epidermolysis bullosa inversa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Bart Tipi, 132000, Otozomal Dominant (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica, Bart type, 132000, Autosomal dominant (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Bart Tipi, 132000, Otozomal Dominant (MLPA)	Epidermolysis bullosa dystrophica, Bart type, 132000, Autosomal dominant (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Bart Tipi, 132000, Otozomal Dominant (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica, Bart type, 132000, Autosomal dominant (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Bart Tipi, 132000, Otozomal Dominant (Prenatal) (MLPA)	Epidermolysis bullosa dystrophica, Bart type, 132000, Autosomal dominant (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Localisata Varyanti (Jeneralize Dominant Distrofik Epidermolizis Büllosa) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica, localisata variant (Generalized dominant dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Localisata Varyanti (Jeneralize Dominant Distrofik Epidermolizis Büllosa) (MLPA)	Epidermolysis bullosa dystrophica, localisata variant (Generalized dominant dystrophic epidermolysis bullosa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Localisata Varyanti (Jeneralize Dominant Distrofik Epidermolizis Büllosa) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica, localisata variant (Generalized dominant dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Distrofika, Localisata Varyanti (Jeneralize Dominant Distrofik Epidermolizis Büllosa) (Prenatal) (MLPA)	Epidermolysis bullosa dystrophica, localisata variant (Generalized dominant dystrophic epidermolysis bullosa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 1, Hipohidrotik, X'e Bağlı 305100, X'e Bağlı; XHED (X'e Bağlı Hipohidrotik Ektodermal Displazi) (EDA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100, X-linked recessive; XHED (X-linked hypohidrotic ectodermal dysplasia) (EDA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDA	EDA, ED1, ECTD1, EDA, HED1, STHAGX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 1, hypohidrotic, X-linked (XHED) [MIM:305100]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. It is the most common form of over 150 clinically distinct ectodermal dysplasias. {ECO:0000269|PubMed:10469321, ECO:0000269|PubMed:10951256, ECO:0000269|PubMed:11279189, ECO:0000269|PubMed:11295832, ECO:0000269|PubMed:11343303, ECO:0000269|PubMed:11378824, ECO:0000269|PubMed:12225002, ECO:0000269|PubMed:12932274, ECO:0000269|PubMed:17256800, ECO:0000269|PubMed:18231121, ECO:0000269|PubMed:19127222, ECO:0000269|PubMed:19438931, ECO:0000269|PubMed:20486090, ECO:0000269|PubMed:20979233, ECO:0000269|PubMed:22008666, ECO:0000269|PubMed:22350046, ECO:0000269|PubMed:8696334, ECO:0000269|PubMed:9507389, ECO:0000269|PubMed:9630076, ECO:0000269|PubMed:9683615, ECO:0000269|PubMed:9736768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi 1, Hipohidrotik, X'e Bağlı 305100, X'e Bağlı; XHED (X'e Bağlı Hipohidrotik Ektodermal Displazi) (Prenatal) (EDA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100, X-linked recessive; XHED (X-linked hypohidrotic ectodermal dysplasia) (EDA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDA	EDA, ED1, ECTD1, EDA, HED1, STHAGX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 1, hypohidrotic, X-linked (XHED) [MIM:305100]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. It is the most common form of over 150 clinically distinct ectodermal dysplasias. {ECO:0000269|PubMed:10469321, ECO:0000269|PubMed:10951256, ECO:0000269|PubMed:11279189, ECO:0000269|PubMed:11295832, ECO:0000269|PubMed:11343303, ECO:0000269|PubMed:11378824, ECO:0000269|PubMed:12225002, ECO:0000269|PubMed:12932274, ECO:0000269|PubMed:17256800, ECO:0000269|PubMed:18231121, ECO:0000269|PubMed:19127222, ECO:0000269|PubMed:19438931, ECO:0000269|PubMed:20486090, ECO:0000269|PubMed:20979233, ECO:0000269|PubMed:22008666, ECO:0000269|PubMed:22350046, ECO:0000269|PubMed:8696334, ECO:0000269|PubMed:9507389, ECO:0000269|PubMed:9630076, ECO:0000269|PubMed:9683615, ECO:0000269|PubMed:9736768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 10A, Hipohidrotik / Saç / Tırnak Tipi, Otozomal Dominant, 129490, Otozomal Dominant; ECTD10A (Hipohidrotik Ektodermal Displazi) (EDAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490, Autosomal dominant; ECTD10A (Hypohidrotic ectodermal dysplasia) (EDAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDAR	EDAR, DL, ED3, EDA3, HRM1, ECTD10A, ECTD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (ECTD10A) [MIM:129490]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal dominant condition characterized by hypotrichosis, abnormal or missing teeth, and hypohidrosis due to the absence of sweat glands. {ECO:0000269|PubMed:10431241, ECO:0000269|PubMed:18231121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi 10A, Hipohidrotik / Saç / Tırnak Tipi, Otozomal Dominant, 129490, Otozomal Dominant; ECTD10A (Hipohidrotik Ektodermal Displazi) (Prenatal) (EDAR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490, Autosomal dominant; ECTD10A (Hypohidrotic ectodermal dysplasia) (EDAR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDAR	EDAR, DL, ED3, EDA3, HRM1, ECTD10A, ECTD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (ECTD10A) [MIM:129490]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal dominant condition characterized by hypotrichosis, abnormal or missing teeth, and hypohidrosis due to the absence of sweat glands. {ECO:0000269|PubMed:10431241, ECO:0000269|PubMed:18231121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 10B, Hipohidrotik / Saç / Diş Tipi, Otozomal Resesif, 224900, Otozomal Resesif; ECTD10B (Hipohidrotik Ektodermal Displazi) (EDAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900, Autosomal recessive; ECTD10B (Hypohidrotic ectodermal dysplasia) (EDAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDAR	EDAR, DL, ED3, EDA3, HRM1, ECTD10A, ECTD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (ECTD10A) [MIM:129490]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal dominant condition characterized by hypotrichosis, abnormal or missing teeth, and hypohidrosis due to the absence of sweat glands. {ECO:0000269|PubMed:10431241, ECO:0000269|PubMed:18231121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ektodermal Displazi 10B, Hipohidrotik / Saç / Diş Tipi, Otozomal Resesif, 224900, Otozomal Resesif; ECTD10B (Hipohidrotik Ektodermal Displazi) (Prenatal) (EDAR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 10B, hypohidrotic/hairtooth type, autosomal recessive, 224900, Autosomal recessive; ECTD10B (Hypohidrotic ectodermal dysplasia) (EDAR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDAR	EDAR, DL, ED3, EDA3, HRM1, ECTD10A, ECTD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant (ECTD10A) [MIM:129490]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal dominant condition characterized by hypotrichosis, abnormal or missing teeth, and hypohidrosis due to the absence of sweat glands. (ECO:0000269|PubMed:10431241, ECO:0000269|PubMed:18231121). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 11A, Hipohidrotik / Saç / Diş Tipi, Otozomal Dominant, 614940, Otozomal Dominant; ECTD11A (Hipohidrotik Ektodermal Displazi) (EDARADD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 11A, hypohidrotic/hairtooth type, autosomal dominant, 614940, Autosomal dominant; ECTD11A (Hypohidrotic ectodermal dysplasia) (EDARADD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDARADD	EDARADD, ED3, EDA3, ECTD11B, ECTD11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive (ECTD11B) [MIM:614941]: A disorder due to abnormal development of two or more ectodermal structures, and characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. (ECO:0000269|PubMed:11780064, ECO:0000269|PubMed:20222921). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ektodermal Displazi 11A, Hipohidrotik / Saç / Diş Tipi, Otozomal Dominant, 614940, Otozomal Dominant; ECTD11A (Hipohidrotik Ektodermal Displazi) (Prenatal) (EDARADD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 11A, hypohidrotic/hairtooth type, autosomal dominant, 614940, Autosomal dominant; ECTD11A (Hypohidrotic ectodermal dysplasia) (EDARADD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDARADD	EDARADD, ED3, EDA3, ECTD11B, ECTD11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive (ECTD11B) [MIM:614941]: A disorder due to abnormal development of two or more ectodermal structures, and characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. (ECO:0000269|PubMed:11780064, ECO:0000269|PubMed:20222921). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 11B, Hipohidrotik / Saç / Diş Tipi, Otozomal Resesif, 614941, Otozomal Resesif; ECTD11B (Hipohidrotik Ektodermal Displazi) (EDARADD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 11B, hypohidrotic/hairtooth type, autosomal recessive, 614941, Autosomal recessive; ECTD11B (Hypohidrotic ectodermal dysplasia) (EDARADD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDARADD	EDARADD, ED3, EDA3, ECTD11B, ECTD11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive (ECTD11B) [MIM:614941]: A disorder due to abnormal development of two or more ectodermal structures, and characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. (ECO:0000269|PubMed:11780064, ECO:0000269|PubMed:20222921). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ektodermal Displazi 11B, Hipohidrotik / Saç / Diş Tipi, Otozomal Resesif, 614941, Otozomal Resesif; ECTD11B (Hipohidrotik Ektodermal Displazi) (Prenatal) (EDARADD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 11B, hypohidrotic/hairtooth type, autosomal recessive, 614941, Autosomal recessive; ECTD11B (Hypohidrotic ectodermal dysplasia) (EDARADD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDARADD	EDARADD, ED3, EDA3, ECTD11B, ECTD11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive (ECTD11B) [MIM:614941]: A disorder due to abnormal development of two or more ectodermal structures, and characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. (ECO:0000269|PubMed:11780064, ECO:0000269|PubMed:20222921). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 12, Hipohidrotik / Saç / Diş / Tırnak Tipi, 617337, Otozomal Dominant; ECTD12 (KDF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 12, hypohidrotic/hairtoothinail type, 617337, Autosomal dominant; ECTD12 (KDF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDF1	KDF1, C1orf172, ECTD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektodermal Displazi 12, Hipohidrotik / Saç / Diş / Tırnak Tipi, 617337, Otozomal Dominant; ECTD12 (Prenatal) (KDF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 12, hypohidrotic/hairtoothinail type, 617337, Autosomal dominant; ECTD12 (KDF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KDF1	KDF1, C1orf172, ECTD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 13, Saç / Diş Tipi, 617392, Otozomal Resesif; ECTD13 (KREMEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 13, hair/tooth type, 617392, Autosomal recessive; ECTD13 (KREMEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KREMEN1	kREMEM1, KRM1, ECTD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Ektodermal Displazi 13, Saç / Diş Tipi, 617392, Otozomal Resesif; ECTD13 (Prenatal) (KREMEN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 13, hair/tooth type, 617392, Autosomal recessive; ECTD13 (KREMEN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KREMEN1	kREMEM1, KRM1, ECTD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 2, Clouston Tipi, 129500, Otozomal Dominant (Hidrotik Ektodermal Displazi) (GJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 2, Clouston type, 129500, Autosomal dominant (Hidrotic ectodermal dysplasia) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 2, Clouston type (ECTD2) [MIM:129500]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD2 is an autosomal dominant condition characterized by atrichosis, nail hypoplasia and deformities, hyperpigmentation of the skin, normal teeth, normal sweat and sebaceous gland function. Palmoplantar hyperkeratosis is a frequent feature. Hearing impairment has been detected in few cases. (ECO:0000269|PubMed:11017065, ECO:0000269|PubMed:11874494). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi 2, Clouston Tipi, 129500, Otozomal Dominant (Hidrotik Ektodermal Displazi) (GJB6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 2, Clouston type, 129500, Autosomal dominant (Hidrotic ectodermal dysplasia) (GJB6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJB6	GJB6, CX30, DFNA3B, DFNB1B, ECTD2, HED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 2, Clouston type (ECTD2) [MIM:129500]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD2 is an autosomal dominant condition characterized by atrichosis, nail hypoplasia and deformities, hyperpigmentation of the skin, normal teeth, normal sweat and sebaceous gland function. Palmoplantar hyperkeratosis is a frequent feature. Hearing impairment has been detected in few cases. (ECO:0000269|PubMed:11017065, ECO:0000269|PubMed:11874494). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 3, Witkop Tipi, 189500, Otozomal Dominant (Hipodonti - Tırnak Displazisi) (MSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 3, Witkop type, 189500, Autosomal dominant (Hypodontia-dysplasia of nails syndrome) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 1 (STHAG1) [MIM:106600]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). Tooth agenesis selective type 1 can be associated with orofacial cleft in some patients. (ECO:0000269|PubMed:12097313, ECO:0000269|PubMed:8696335). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi 3, Witkop Tipi, 189500, Otozomal Dominant (Hipodonti - Tırnak Displazisi) (Prenatal) (MSX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 3, Witkop type, 189500, Autosomal dominant (Hypodontia-dysplasia of nails syndrome) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tooth agenesis selective 1 (STHAG1) [MIM:106600]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). Tooth agenesis selective type 1 can be associated with orofacial cleft in some patients. (ECO:0000269|PubMed:12097313, ECO:0000269|PubMed:8696335). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 4, Saç / Tırnak Tipi, 602032, Otozomal Resesif; ECTD4 (Saf Saç ve Tırnak Ektodermal Displazisi) (KRT85 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 4, hair/nail type, 602032, Autosomal recessive; ECTD4 (Pure hair and nail ectodermal dysplasia) (KRT85 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT85	KRT85, KRTHB5, HBS, ECTD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektodermal Displazi 4, Saç / Tırnak Tipi, 602032, Otozomal Resesif; ECTD4 (Saf Saç ve Tırnak Ektodermal Displazisi) (Prenatal) (KRT85 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 4, hair/nail type, 602032, Autosomal recessive; ECTD4 (Pure hair and nail ectodermal dysplasia) (KRT85 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT85	KRT85, KRTHB5, HBS, ECTD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 7, Saç / Tırnak Tipi, 614929, Otozomal Resesif; ECTD7 (Saf Saç ve Tırnak Ektodermal Displazisi) (KRT74 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 7, hair/nail type, 614929, Autosomal recessive; ECTD7 (Pure hair and nail ectodermal dysplasia) (KRT74 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT74	KRT74, K6IRS4, KRT6IRS4, HTS52, HYPT3, ADWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 3 (HYPT3) [MIM:613981]: A condition characterized by the presence of less than the normal amount of hair. Affected individuals have normal hair in early childhood but experience progressive hair loss limited to the scalp beginning in the middle of the first decade and almost complete baldness by the third decade. Body hair, beard, eyebrows, axillary hair, teeth, and nails develop normally. (ECO:0000269	PubMed:21188418). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi 7, Saç / Tırnak Tipi, 614929, Otozomal Resesif; ECTD7 (Saf Saç ve Tırnak Ektodermal Displazisi) (Prenatal) (KRT74 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 7, hair/nail type, 614929, Autosomal recessive; ECTD7 (Pure hair and nail ectodermal dysplasia) (KRT74 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT74	KRT74, K6IRS4, KRT6IRS4, HTS52, HYPT3, ADWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypotrichosis 3 (HYPT3) [MIM:613981]: A condition characterized by the presence of less than the normal amount of hair. Affected individuals have normal hair in early childhood but experience progressive hair loss limited to the scalp beginning in the middle of the first decade and almost complete baldness by the third decade. Body hair, beard, eyebrows, axillary hair, teeth, and nails develop normally. (ECO:0000269	PubMed:21188418). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi 9, Saç / Tırnak Tipi, 614931, Otozomal Resesif; ECTD9 (Saf Saç ve Tırnak Ektodermal Displazisi) (HOXC13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia 9, hair/nail type, 614931, Autosomal recessive; ECTD9 (Pure hair and nail ectodermal dysplasia) (HOXC13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXC13	HOXC13, HOX3G, ECTD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 9, hair/nail type (ECTD9) [MIM:614931]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD9 is characterized by hypotrichosis and nail dystrophy without non-ectodermal or other ectodermal manifestations. Hypotrichosis usually occurs after birth with varying degrees of severity, ranging from mild hair loss to complete atrichia, including the loss of scalp hair, beard, eyebrows, eyelashes, axillary hair, and pubic hair. Nail dystrophy affects all 20 digits by causing short fragile nails or spoon nails (koilonychia). (ECO:0000269	PubMed:23063621). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Ektodermal Displazi 9, Saç / Tırnak Tipi, 614931, Otozomal Resesif; ECTD9 (Saf Saç ve Tırnak Ektodermal Displazisi) (Prenatal) (HOXC13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia 9, hair/nail type, 614931, Autosomal recessive; ECTD9 (Pure hair and nail ectodermal dysplasia) (HOXC13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXC13	HOXC13, HOX3G, ECTD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia 9, hair/nail type (ECTD9) [MIM:614931]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures such as hair, teeth, nails and sweat glands, with or without any additional clinical sign. Each combination of clinical features represents a different type of ectodermal dysplasia. ECTD9 is characterized by hypotrichosis and nail dystrophy without non-ectodermal or other ectodermal manifestations. Hypotrichosis usually occurs after birth with varying degrees of severity, ranging from mild hair loss to complete atrichia, including the loss of scalp hair, beard, eyebrows, eyelashes, axillary hair, and pubic hair. Nail dystrophy affects all 20 digits by causing short fragile nails or spoon nails (koilonychia). (ECO:0000269	PubMed:23063621). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi-Sindaktili Sendromu 1, 613573; EDSS1 (Ektodermal Displazi-Sindaktili Sendromu) (NECTIN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia-syndactyly syndrome 1, 613573; EDSS1 (Ectodermal dysplasia-syndactyly syndrome) (NECTIN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NECTIN4	PVRL4, PRR4, EDSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia-syndactyly syndrome 1 (EDSS1) [MIM:613573]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. EDSS1 is characterized by the association of hair and teeth abnormalities with cutaneous syndactyly of the hands and/or feet. Hair morphologic abnormalities include twists at irregular intervals (pili torti) and swelling along the shafts, particularly associated with areas of breakage. Dental findings consist of abnormally widely spaced teeth, with peg- shaped and conical crowns. Patients have normal sweating. (ECO:0000269	PubMed:20691405). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ektodermal Displazi-Sindaktili Sendromu 1, 613573; EDSS1 (Ektodermal Displazi-Sindaktili Sendromu) (Prenatal) (NECTIN4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia-syndactyly syndrome 1, 613573; EDSS1 (Ectodermal dysplasia-syndactyly syndrome) (NECTIN4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NECTIN4	PVRL4, PRR4, EDSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia-syndactyly syndrome 1 (EDSS1) [MIM:613573]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. EDSS1 is characterized by the association of hair and teeth abnormalities with cutaneous syndactyly of the hands and/or feet. Hair morphologic abnormalities include twists at irregular intervals (pili torti) and swelling along the shafts, particularly associated with areas of breakage. Dental findings consist of abnormally widely spaced teeth, with peg- shaped and conical crowns. Patients have normal sweating. (ECO:0000269	PubMed:20691405). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Anhidrotik-İmmun Yetmezlik, Osteopetrozis ve Lenfödem; Oledaid (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (IKBKG geni) (Tüm Gen Dizi Analizi) (Postnatal)	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH IMMUNODEFICIENCY, OSTEOPETROSIS, AND LYMPHEDEMA, OLEDAID (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
Ektodermal Displazi, Anhidrotik-İmmun Yetmezlik, Osteopetrozis ve Lenfödem; Oledaid (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (MLPA)	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH IMMUNODEFICIENCY, OSTEOPETROSIS, AND LYMPHEDEMA, OLEDAID (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektodermal Displazi, Anhidrotik-İmmun Yetmezlik, Osteopetrozis ve Lenfödem; Oledaid (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (Prenatal) (IKBKG geni) (Tüm Gen Dizi Analizi) (Prenatal)	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH IMMUNODEFICIENCY, OSTEOPETROSIS, AND LYMPHEDEMA, OLEDAID (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Anhidrotik-İmmun Yetmezlik, Osteopetrozis ve Lenfödem; Oledaid (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (Prenatal) (MLPA)	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH IMMUNODEFICIENCY, OSTEOPETROSIS, AND LYMPHEDEMA, OLEDAID (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (Prenatal) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Anhidrotik, T Hücre İmmün Yetmezliği, 612132, Otozomal Dominant (Hipohidrotik Ektodermal Displazi) (NFKBIA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132, Autosomal dominant (Hypohidrotic ectodermal dysplasia) (NFKBIA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFKBIA	NFKBIA, IKBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency autosomal dominant (ADEDAID) [MIM:612132]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. This form of ectodermal dysplasia is associated with decreased production of pro-inflammatory cytokines and certain interferons, rendering patients susceptible to infection. (ECO:0000269|PubMed:14523047, ECO:0000269|PubMed:18412279). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ektodermal Displazi, Anhidrotik, T Hücre İmmün Yetmezliği, 612132, Otozomal Dominant (Hipohidrotik Ektodermal Displazi) (Prenatal) (NFKBIA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132, Autosomal dominant (Hypohidrotic ectodermal dysplasia) (NFKBIA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFKBIA	NFKBIA, IKBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency autosomal dominant (ADEDAID) [MIM:612132]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. This form of ectodermal dysplasia is associated with decreased production of pro-inflammatory cytokines and certain interferons, rendering patients susceptible to infection. (ECO:0000269|PubMed:14523047, ECO:0000269|PubMed:18412279). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Ektrodaktili ve Makiler Distrofi, 225280, Otozomal Resesif; EEMS (EEM Sendromu) (CDH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280, Autosomal recessive; EEMS (EEM syndrome) (CDH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH3	CDH3, CDHP, PCAD, HJMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome (EEMS) [MIM:225280]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal recessive condition characterized by features of ectodermal dysplasia such as sparse eyebrows and scalp hair, and selective tooth agenesis associated with macular dystrophy and ectrodactyly. (ECO:0000269	PubMed:15805154). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ektodermal Displazi, Ektrodaktili ve Makiler Distrofi, 225280, Otozomal Resesif; EEMS (EEM Sendromu) (Prenatal) (CDH3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280, Autosomal recessive; EEMS (EEM syndrome) (CDH3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDH3	CDH3, CDHP, PCAD, HJMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome (EEMS) [MIM:225280]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal recessive condition characterized by features of ectodermal dysplasia such as sparse eyebrows and scalp hair, and selective tooth agenesis associated with macular dystrophy and ectrodactyly. (ECO:0000269	PubMed:15805154). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Hipohidrotik, İmmün Yetmezlik, 300291 (Hipohidrotik Ektodermal Displazi) (IKBKG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (Hypohidrotic ectodermal dysplasia) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
Ektodermal Displazi, Hipohidrotik, İmmün Yetmezlik, 300291 (Hipohidrotik Ektodermal Displazi) (MLPA)	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (Hypohidrotic ectodermal dysplasia) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ektodermal Displazi, Hipohidrotik, İmmün Yetmezlik, 300291 (Hipohidrotik Ektodermal Displazi) (Prenatal) (IKBKG gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (Hypohidrotic ectodermal dysplasia) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi, Hipohidrotik, İmmün Yetmezlik, 300291 (Hipohidrotik Ektodermal Displazi) (Prenatal) (MLPA)	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (Hypohidrotic ectodermal dysplasia) (Prenatal) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi / Kısa Boy Sendromu, 616029, Otozomal Resesif; ECTD5 (Tırnak ve Diş Anormallikleri-Marjinal Palmoplantar Keratoderma-Oral Hiperpigmentasyon Sendromu) (GRHL2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia/short stature syndrome, 616029, Autosomal recessive; ECTD5 (Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome) (GRHL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRHL2	GRHL2, TFCP2L3, DFNA28, ECTD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektodermal Displazi / Kısa Boy Sendromu, 616029, Otozomal Resesif; ECTD5 (Tırnak ve Diş Anormallikleri-Marjinal Palmoplantar Keratoderma-Oral Hiperpigmentasyon Sendromu) (Prenatal) (GRHL2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal dysplasia/short stature syndrome, 616029, Autosomal recessive; ECTD5 (Nail and teeth abnormalities-marginal palmoplantar keratoderma-oral hyperpigmentation syndrome) (GRHL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRHL2	GRHL2, TFCP2L3, DFNA28, ECTD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal Displazi / Deri Frajilitesi Sendromu, 604536 (Epidermolizis Bulloza Simpleks, Plakofilin Eksikliği) (PKP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal dysplasia/skin fragility syndrome, 604536 (Epidermolysis bullosa simplex due to plakophilin deficiency) (PKP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKP1	PKP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektodermal, Displazi, Anhidrotik, Lenfödem ve İmmün Yetmezlik, 300301 (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (IKBKG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ On Ice
Ektodermal, Displazi, Anhidrotik, Lenfödem ve İmmün Yetmezlik, 300301 (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (MLPA)	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ektodermal, Displazi, Anhidrotik, Lenfödem ve İmmün Yetmezlik, 300301 (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (Prenatal) (IKBKG gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektodermal, Displazi, Anhidrotik, Lenfödem ve İmmün Yetmezlik, 300301 (Anhidrotik Ektodermal Displazi-İmmün Yetmezlik-Osteopetroz-Lenfödem Sendromu) (Prenatal) (MLPA)	Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (Anhidrotic ectodermal dysplasia-immunodeficiency-osteopetrosis-lymphedema syndrome) (Prenatal) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ektopia Lentis 2, İzole, Otozomal Resesif; Ectol2 (İzole Ektopia Lentis) (ADAMTSL4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	ECTOPIA LENTIS 2, ISOLATED, AUTOSOMAL RECESSIVE; ECTOL2 (Isolated ectopia lentis) (ADAMTSL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTSL4	ADAMTSL4, TSRC1, ECTOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectopia lentis 2, isolated, autosomal recessive (ECTOL2) [MIM:225100]: An ocular abnormality characterized by partial or complete displacement of the lens from its space resulting from defective zonule formation. {ECO:0000269	PubMed:19200529}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektopia Lentis Et Pupilla, 225200, Otozomal Resesif (İzole Ektopia Lentis) (ADAMTSL4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectopia lentis et pupillae, 225200, Autosomal recessive (isolated ectopia lentis) (ADAMTSL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTSL4	ADAMTSL4, TSRC1, ECTOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectopia lentis 2, isolated, autosomal recessive (ECTOL2) [MIM:225100]: An ocular abnormality characterized by partial or complete displacement of the lens from its space resulting from defective zonule formation. {ECO:0000269	PubMed:19200529}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ektopia Lentis, Ailesel, 129600, Otozomal Dominant; ECTOL1 (İzole Ektopia Lentis) (FBN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectopia lentis, familial, 129600, Autosomal dominant; ECTOL1 (Isolated ectopia lentis) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSO2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektopia Lentis, Ailesel, 129600, Otozomal Dominant; ECTOL1 (İzole Ektopia Lentis) (MLPA)	Ectopia lentis, familial, 129600, Autosomal dominant; ECTOL1 (Isolated ectopia lentis) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSO2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ektrodaktili, Ektodermal Displazi ve Dudak / Damak Sendromu 3, 604292, Otozomal Dominant; EEC3 (EEC Sendromu) (TP63 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292, Autosomal dominant; EEC3 (EEC syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ektrodaktili, Ektodermal Displazi ve Dudak / Damak Sendromu 3, 604292, Otozomal Dominant; EEC3 (EEC Sendromu) (Prenatal) (TP63 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292, Autosomal dominant; EEC3 (EEC syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
EDICT Sendromu, 614303, Otozomal Dominant (EDICT Sendromu) (MIR184 gen) (Tüm Gen Dizi Analizi) (Postnatal)	EDICT syndrome, 614303, Autosomal dominant (EDICT syndrome) (MIR184 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIR184	MIR184, MIRN184, KTCNCT, EDICT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDICT Sendromu, 614303, Otozomal Dominant (EDICT Sendromu) (Prenatal) (MIR184 gen) (Tüm Gen Dizi Analizi) (Prenatal)	EDICT syndrome, 614303, Autosomal dominant (EDICT syndrome) (MIR184 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MIR184	MIR184, MIRN184, KTCNCT, EDICT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Etavirenz, Merkezi Sinir Sistemi Toksisitesi, Yatkınlık, 614546 (Efavirenz Toksisitesi) (CYP2B6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Efavirenz central nervous system toxicity, susceptibility to, 614546 (Efavirenz toxicity) (CYP2B6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2B6	CYP2B6, CYP2B, EFVM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Etavirenz, Merkezi Sinir Sistemi Toksisitesi, Yatkınlık, 614546 (Efavirenz Toksisitesi) (MLPA)	Efavirenz central nervous system toxicity, susceptibility to, 614546 (Efavirenz toxicity) (MLPA)	CYP2B6	CYP2B6, CYP2B, EFVM	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Etavirenz, Zayıf Metabolizma, 614546 (Efavirenz Toksisitesi) (CYP2B6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Efavirenz, poor metabolism of, 614546 (Efavirenz toxicity) (CYP2B6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2B6	CYP2B6, CYP2B, EFVM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Etavirenz, Zayıf Metabolizma, 614546 (Efavirenz Toksisitesi) (MLPA)	Efavirenz, poor metabolism of, 614546 (Efavirenz toxicity) (MLPA)	CYP2B6	CYP2B6, CYP2B, EFVM	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tenaskin 10 Eksikliğine Bağlı, 606408, Otozomal Resesif (Ehlers-Danlos Sendromu, Tenaskin-X Eksikliğine Bağlı) (TNXB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408, Autosomal recessive (Ehlers-Danlos syndrome due to tenascin-X deficiency) (TNXB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNXB	TNXB, TNX, TNXB1, TNXBS, TNXB2, EDS3, VLR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome due to tenascin X deficiency (EDSTNXD) [MIM:606408]: An Ehlers-Danlos-like syndrome characterized by hyperextensible skin, hypermobile joints, and tissue fragility. Tenascin-X-deficient patients, however, lack atrophic scars, a major diagnostic criteria for classic Ehlers-Danlos. Delayed wound healing, which is also common in classic EDS, is only present in a subset of patients. {ECO:0000269	PubMed:23768946}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tenaskin 10 Eksikliğine Bağlı, 606408, Otozomal Resesif (Ehlers-Danlos Sendromu, Tenaskin-X Eksikliğine Bağlı) (MLPA)	Ehlers-Danlos syndrome due to tenascin X deficiency, 606408, Autosomal recessive (Ehlers-Danlos syndrome due to tenascin-X deficiency) (MLPA)	TNXB	TNXB, TNX, TNXB1, TNXBS, TNXB2, EDS3, VLR8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome due to tenascin X deficiency (EDSTNXD) [MIM:606408]: An Ehlers-Danlos-like syndrome characterized by hyperextensible skin, hypermobile joints, and tissue fragility. Tenascin-X-deficient patients, however, lack atrophic scars, a major diagnostic criteria for classic Ehlers-Danlos. Delayed wound healing, which is also common in classic EDS, is only present in a subset of patients. {ECO:0000269	PubMed:23768946}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Progresif Kifoskolyoz, Miyopati ve İşitme Kaybı, 614557, Otozomal Resesif; EDSKMH (Ehlers-Danlos Sendromu, Kifoskolyotik ve Sağırlık Tipi) (FKBP14 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss, 614557, Autosomal recessive; EDSKMH (Ehlers-Danlos syndrome, kyphoscoliotic and deafness type) (FKBP14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKBP14	FKBP14, EDSKMH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome, with progressive kyphoscoliosis, myopathy, and hearing loss (EDSKMH) [MIM:614557]: A syndrome with features of Ehlers-Danlos syndrome types VIA and VIB on the one hand, and the collagen VI-related congenital myopathies Ullrich congenital muscular dystrophy and Bethlem myopathy on the other hand. Clinically, this disorder is characterized by the following features: severe generalized hypotonia at birth with marked muscle weakness that improve in infancy; early-onset progressive kyphoscoliosis; joint hypermobility without contractures; hyperelastic skin with follicular hyperkeratosis, easy bruising, and occasional abnormal scarring; myopathy; hearing impairment, which is predominantly sensorineural; normal ratio of lysyl pyridinoline to hydroxylysyl pyridinoline (LP/HP) in urine. {ECO:0000269	PubMed:22265013}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ehlers-Danlos Sendromu, Kısa Boy ve Ekstremite Anomalileri, 130070, Otozomal Resesif; EDSSLA (Ehlers-Danlos Sendromu, Progeroid Tipi) (B4GALT7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome with short stature and limb anomalies, 130070, Autosomal recessive; EDSSLA (Ehlers-Danlos syndrome, progeroid type) (B4GALT7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B4GALT7	B4GALT7, XGALT1, XGPT1, EDSSLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Kardiyak Valvüler Form, 225320, Otozomal Resesif (Ehlers-Danlos Sendromu, Kalp Kapağı Tipi) (COL1A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, cardiac valvular form, 225320, Autosomal recessive (Ehlers-Danlos syndrome, cardiac valvular type) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Kardiyak Valvüler Form, 225320, Otozomal Resesif (Ehlers-Danlos Sendromu, Kalp Kapağı Tipi) (MLPA)	Ehlers-Danlos syndrome, cardiac valvular form, 225320, Autosomal recessive (Ehlers-Danlos syndrome, cardiac valvular type) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Klasik Tip, 130000, Otozomal Dominant (Ehlers-Danlos Sendromu, Klasik Tipi (COL5A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, classic type, 130000, Autosomal dominant (Ehlers-Danlos syndrome, classic type) (COL5A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL5A2	COL5A2, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome, classic type (EDS) [MIM:130000]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. {ECO:0000269|PubMed:9425233, ECO:0000269|PubMed:9783710}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Ehlers-Danlos Sendromu, Klasik Tip, 130000, Otozomal Dominant (Ehlers-Danlos Sendromu, Klasik Tip) (COL5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, classic type, 130000, Autosomal dominant (Ehlers-Danlos syndrome, classic type) (COL5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL5A1	COL5A1, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome, classic type (EDS) [MIM:130000]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. (ECO:0000269|PubMed:10602121, ECO:0000269|PubMed:11992482, ECO:0000269|PubMed:15580559, ECO:0000269|PubMed:18972565, ECO:0000269|PubMed:9042913). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ehlers-Danlos Sendromu, Klasik Tip, 130000, Otozomal Dominant (Ehlers-Danlos Sendromu, Klasik Tip) (MLPA)	Ehlers-Danlos syndrome, classic type, 130000, Autosomal dominant (Ehlers-Danlos syndrome, classic type) (MLPA)	COL5A1	COL5A1, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome, classic type (EDS) [MIM:130000]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. (ECO:0000269|PubMed:10602121, ECO:0000269|PubMed:11992482, ECO:0000269|PubMed:15580559, ECO:0000269|PubMed:18972565, ECO:0000269|PubMed:9042913). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ehlers-Danlos Sendromu, Klasik, 130000, Otozomal Dominant (Ehlers-Danlos Sendromu, Klasik Tip) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, classic, 130000, Autosomal dominant (Ehlers-Danlos syndrome, classic type) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Klasik, 130000, Otozomal Dominant (Ehlers-Danlos Sendromu, Klasik Tip) (MLPA)	Ehlers-Danlos syndrome, classic, 130000, Autosomal dominant (Ehlers-Danlos syndrome, classic type) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Muskülokontraktürel Tip 1, 601776, Otozomal Resesif; EDSMC1 (Ehlers-Danlos Sendromu, Muskülokontraktürel Tip) (CHST14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, musculocontractural type 1, 601776, Autosomal recessive; EDSMC1 (Ehlers-Danlos syndrome, musculocontractural type) (CHST14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHST14	CHST14, D4ST1, ATCS, EDSMC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Kas Kontraktürü Tip 2, 615539, Otozomal Resesif; EDSMC2 (Ehlers-Danlos Sendromu, Muskülokontraktürel Tip) (DSE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, musculocontractural type 2, 615539, Autosomal recessive; EDSMC2 (Ehlers-Danlos syndrome, musculocontractural type) (DSE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSE	DSE, SART2, EDSMC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Ehlers-Danlos Sendromu, Periodontal Tipi, 1, 130080, Otozomal Dominant (Ehlers-Danlos Sendromu, Periodontit Tip) (C1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, periodontal type, 1, 130080, Autosomal dominant (Ehlers-Danlos syndrome, periodontitis type) (C1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1R	C1R, EDSPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Periodontal Tip, 2, 617174, Otozomal Dominant; EDSPD2 (Ehlers-Danlos Sendromu, Periodontit Tip) (C1S geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, periodontal type, 2, 617174, Autosomal dominant; EDSPD2 (Ehlers-Danlos syndrome, periodontitis type) (C1S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1S	C1S, EDSPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Progeroid Tip, 2, 615349, Otozomal Resesif; EDSP2 (Ehlers-Danlos Sendromu, Progeroid Tip) (B3GALT6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, progeroid type, 2, 615349, Autosomal recessive; EDSP2 (Ehlers-Danlos syndrome, progeroid type) (B3GALT6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GALT6	B3GALT6, SEMDJL1, EDSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 4, 130050, Otozomal Dominant (Ehlers-Danlos Sendromu, Vasküler Tip) (COL3A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, type IV, 130050, Autosomal dominant (Ehlers-Danlos syndrome, vascular type) (COL3A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL3A1	COL3A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 4, 130050, Otozomal Dominant (Ehlers-Danlos Sendromu, Vasküler Tip) (MLPA)	Ehlers-Danlos syndrome, type IV, 130050, Autosomal dominant (Ehlers-Danlos syndrome, vascular type) (MLPA)	COL3A1	COL3A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 6, 225400, Otozomal Resesif; EDS6 (Ehlers-Danlos Sendromu, Kifoskolyotik Tip) (PLOD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, type VI, 225400, Autosomal recessive; EDS6 (Ehlers-Danlos syndrome, kyphoscoliotic type) (PLOD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLOD1	PLOD1, LH1, LLH, EDS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 6 (EDS6) [MIM:225400]: A connective tissue disorder characterized by generalized joint hypermobility, hyperextensible skin, atrophic cutaneous scars due to tissue fragility, progressive kyphoscoliosis already present at birth, ocular manifestations, arterial rupture, easy bruising, severe neonatal muscle hypotonia and delayed motor development. (ECO:0000269|PubMed:10686424, ECO:0000269|PubMed:15666309, ECO:0000269|PubMed:15854030, ECO:0000269|PubMed:15979919, ECO:0000269|PubMed:8163671, ECO:0000269|PubMed:9617436). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 7A, 130060, Otozomal Dominant (Ehlers-Danlos Sendromu, Artrokalazis Tip) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, type VIIA, 130060, Autosomal dominant (Ehlers-Danlos syndrome, arthrochalasis type) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 7A, 130060, Otozomal Dominant (Ehlers-Danlos Sendromu, Artrokalazis Tip) (MLPA)	Ehlers-Danlos syndrome, type VIIA, 130060, Autosomal dominant (Ehlers-Danlos syndrome, arthrochalasis type) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 7B, 130060, Otozomal Dominant (Ehlers-Danlos Sendromu, Artrokalazis Tip) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, type VIIB, 130060, Autosomal dominant (Ehlers-Danlos syndrome, arthrochalasis type) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. (ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:3680255). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 7B, 130060, Otozomal Dominant (Ehlers-Danlos Sendromu, Artrokalazis Tip) (MLPA)	Ehlers-Danlos syndrome, type VIIB, 130060, Autosomal dominant (Ehlers-Danlos syndrome, arthrochalasis type) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. (ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:3680255). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ehlers-Danlos Sendromu, Tip 7C, 225410, Otozomal Resesif (Ehlers-Danlos Sendromu, Dermatoparaksis Tipi) (ADAMTS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ehlers-Danlos syndrome, type VIIC, 225410, Autosomal recessive (Ehlers-Danlos syndrome, dermatosparaxis type) (ADAMTS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTS2	ADAMTS2, NPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7C (EDS7C) [MIM:225410]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by extremely fragile tissues, hyperextensible skin and easy bruising. Facial skin contains numerous folds, as in the cutis laxa syndrome. (ECO:0000269	PubMed:10417273). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ehlers-Danlos sendromu, vasküler tip (COL3A1 2q31, TNXB 6p21.3.) (MLPA)	Ehlers-Danlos syndrome, vascular type (COL3A1 2q31, TNXB 6p21.3.) (MLPA)	COL3A1 2q31, TNXB 6p21.3.	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Eiken Sendromu, 600002, Otozomal Resesif (Eiken Sendromu) (PTH1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Eiken syndrome, 600002, Autosomal recessive (Eiken syndrome) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Eliptositoz-1, 611804, EL1 (Kalıtsal Eliptositoz) (EPB41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Elliptocytosis-1, 611804, EL1 (Hereditary elliptocytosis) (EPB41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPB41	EPB41, EL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 1 (EL1) [MIM:611804]: A Rhesus-linked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. (ECO:0000269	PubMed:3467321). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eliptositozis-2, 130600, Otozomal Dominant; EL2 (Kalıtsal Eliptositoz) (SPTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Elliptocytosis-2, 130600, Autosomal dominant; EL2 (Hereditary elliptocytosis) (SPTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTA1	SPTA1, EL2, SPH3, HS3, HPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 2 (EL2) [MIM:130600]: A Rhesus-unlinked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. (ECO:0000269|PubMed:1541680, ECO:0000269|PubMed:1638030, ECO:0000269|PubMed:1679439, ECO:0000269|PubMed:1878597, ECO:0000269|PubMed:2384601, ECO:0000269|PubMed:2568861, ECO:0000269|PubMed:2568862, ECO:0000269|PubMed:2794061, ECO:0000269|PubMed:7772539, ECO:0000269|PubMed:8018926, ECO:0000269|PubMed:8183371, ECO:0000269|PubMed:8364215). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Eliptositozis-3 (Kalıtsal Sferositoz) (SPTB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Elliptocytosis-3 (Hereditary spherocytosis) (SPTB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTB	SPTB, SPH2, EL3, HS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 3 (EL3) [MIM:182870]: A Rhesus-unlinked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. (ECO:0000269|PubMed:1975598, ECO:0000269|PubMed:7883966, ECO:0000269|PubMed:8018926, ECO:0000269|PubMed:8226774). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ellis van Creveld sendromu (4p16.2 (EVC-EVC2)) (MLPA)	Ellis van Creveld syndrome (4p16.2 (EVC-EVC2)) (MLPA)	4p16.2 (EVC-EVC2)	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ellis van Creveld sendromu (4p16.2 (EVC-EVC2)) (MLPA) (Prenatal)	Ellis van Creveld syndrome (4p16.2 (EVC-EVC2)) (MLPA) (Prenatal)	4p16.2 (EVC-EVC2)	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif (Ellis Van Creveld Sendromu) (EVC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ellis-van Creveld syndrome, 225500, Autosomal recessive (Ellis Van Creveld syndrome) (EVC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EVC	EVC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif (Ellis Van Creveld Sendromu) (MLPA)	Ellis-van Creveld syndrome, 225500, Autosomal recessive (Ellis Van Creveld syndrome) (MLPA)	EVC	EVC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif (Ellis Van Creveld Sendromu) (Prenatal) (EVC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ellis-van Creveld syndrome, 225500, Autosomal recessive (Ellis Van Creveld syndrome) (EVC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EVC	EVC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif (Ellis Van Creveld Sendromu) (Prenatal) (MLPA)	Ellis-van Creveld syndrome, 225500, Autosomal recessive (Ellis Van Creveld syndrome) (Prenatal) (MLPA)	EVC	EVC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif; EVC (Ellis Van Creveld Sendromu) (EVC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ellis-van Creveld syndrome, 225500, Autosomal recessive; EVC (Ellis Van Creveld syndrome) (EVC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EVC2	LBN, EVC2, WAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif; EVC (Ellis Van Creveld Sendromu) (MLPA)	Ellis-van Creveld syndrome, 225500, Autosomal recessive; EVC (Ellis Van Creveld syndrome) (MLPA)	EVC2	LBN, EVC2, WAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Ellis Van Creveld Sendromu, 225500, Otozomal Resesif; EVC (Ellis Van Creveld Sendromu) (Prenatal) (EVC2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ellis-van Creveld syndrome, 225500, Autosomal recessive; EVC (Ellis Van Creveld syndrome) (EVC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EVC2	LBN, EVC2, WAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ellis Van Creveld Sendromu, 225500, Otozomal Resesif; EVC (Ellis Van Creveld Sendromu) (Prenatal) (MLPA)	Ellis-van Creveld syndrome, 225500, Autosomal recessive; EVC (Ellis Van Creveld syndrome) (Prenatal) (MLPA)	EVC2	LBN, EVC2, WAD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Emanuel Sendromu, 609029, Kalıtsal Kromozomal Dengesizlik (Emanuel Sendromu) (Konjenital Anomali)	Emanuel syndrome, 609029, Inherited chromosomal imbalance (Emanuel syndrome) (Chromosome Analysis)	.	DER22t11-22	Kromozom analizi/ Karyotype analysis	Heparinli Kan (2-4 ml)	*	15	45	.	.	21	Postnatal	.	Heparinized blood sample
Emberger Sendromu, 614038, Otozomal Dominant (Sağırlık-Lenfödem-Lösemi Sendromu) (GATA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emberger syndrome, 614038, Autosomal dominant (Deafness-lymphedema-leukemia syndrome) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Emberger Sendromu, 614038, Otozomal Dominant (Sağırlık-Lenfödem-Lösemi Sendromu) (GATA2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Emberger syndrome, 614038, Autosomal dominant (Deafness-lymphedema-leukemia syndrome) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 1, X'e Bağlı, 310300, X'e Bağlı; EDMD1 (Emery-Dreifuss Müsküler Distrofi) (EMD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 1, X-linked, 310300, X-linked recessive; EDMD1 (Emery-Dreifuss muscular dystrophy) (EMD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EMD	EMD, EDMD, STA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Emery-Dreifuss muscular dystrophy 1, X-linked (EDMD1) [MIM:310300]: A form of Emery-Dreifuss muscular dystrophy, a degenerative myopathy characterized by weakness and atrophy of muscle without involvement of the nervous system, early contractures of the elbows, Achilles tendons and spine, and cardiomyopathy associated with cardiac conduction defects. {ECO:0000269|PubMed:10323252, ECO:0000269|PubMed:11587540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 2, AD, 181350, Otozomal Dominant; EDMD2 (Emery-Dreifuss Müsküler Distrofi) (LMNA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 2, AD, 181350, Autosomal dominant; EDMD2 (Emery-Dreifuss muscular dystrophy) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 2, AD, 181350, Otozomal Dominant; EDMD2 (Emery-Dreifuss Müsküler Distrofi) (MLPA)	Emery-Dreifuss muscular dystrophy 2, AD, 181350, Autosomal dominant; EDMD2 (Emery-Dreifuss muscular dystrophy) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 3 AR, 616516, Otozomal Resesif; EDMD3 (Emery-Dreifuss Müsküler Distrofi) (LMNA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 3, AR, 616516, Autosomal recessive; EDMD3 (Emery-Dreifuss muscular dystrophy) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 3 AR, 616516, Otozomal Resesif; EDMD3 (Emery-Dreifuss Müsküler Distrofi) (MLPA)	Emery-Dreifuss muscular dystrophy 3, AR, 616516, Autosomal recessive; EDMD3 (Emery-Dreifuss muscular dystrophy) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 4, Otozomal Dominant, 612998, Otozomal Dominant; EDMD4 (Emery-Dreifuss Müsküler Distrofi) (SYNE1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 4, autosomal dominant, 612998, Autosomal dominant; EDMD4 (Emery-Dreifuss muscular dystrophy) (SYNE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNE1	SYNE1, KIAA0796, KIAA1756, KIAA1262, SCAR8, EDMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 5, Otozomal Dominant, 612999, Otozomal Dominant; EDMD5 (Emery-Dreifuss Müsküler Distrofi) (SYNE2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 5, autosomal dominant, 612999, Autosomal dominant; EDMD5 (Emery-Dreifuss muscular dystrophy) (SYNE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNE2	SYNE2, NUANCE, KIAA1011, EDMD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Emery-Dreifuss muscular dystrophy 5, autosomal dominant (EDMD5) [MIM:612999]: A form of Emery-Dreifuss muscular dystrophy, a degenerative myopathy characterized by weakness and atrophy of muscle without involvement of the nervous system, early contractures of the elbows, Achilles tendons and spine, and cardiomyopathy associated with cardiac conduction defects. {ECO:0000269	PubMed:17761684}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 6, X'e Bağlı, 300696, X'e Bağlı (Emery-Dreifuss Müsküler Distrofi) (FHL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 6, X-linked, 300696, X-linked recessive (Emery-Dreifuss muscular dystrophy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Emery-Dreifuss Müsküler Distrofi 7, AD, 614302, Otozomal Dominant; EDMD7 (Emery-Dreifuss Müsküler Distrofi) (TMEM43 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emery-Dreifuss muscular dystrophy 7, AD, 614302, Autosomal dominant; EDMD7 (Emery-Dreifuss muscular dystrophy) (TMEM43 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM43	TMEM43, ARVD5, ARVC5, EDMD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Emery-Dreifuss muscular dystrophy 7, autosomal dominant (EDMD7) [MIM:614302]: A form of Emery-Dreifuss muscular dystrophy, a degenerative myopathy characterized by weakness and atrophy of muscle without involvement of the nervous system, early contractures of the elbows, Achilles tendons and spine, and cardiomyopathy associated with cardiac conduction defects. {ECO:0000269	PubMed:21391257}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
EML4/ALK (FISH)	EML4/ALK (FISH)	2p21-2p23.2-p23.1	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
AAT Eksikliğine Bağlı Amfizem, 613490, Otozomal Resesif; A1ATD (Alfa-1-Antitripsin Eksikliği) (SERPINA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emphysema due to AAT deficiency, 613490, Autosomal recessive; A1ATD (Alpha-1-antitrypsin deficiency) (SERPINA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA1	SERPINA1, PI, AAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
AAT Eksikliğine Bağlı Amfizem, 613490, Otozomal Resesif; A1ATD (Alfa-1-Antitripsin Eksikliği) (MLPA)	Emphysema due to AAT deficiency, 613490, Autosomal recessive; A1ATD (Alpha-1-antitrypsin deficiency) (MLPA)	SERPINA1	SERPINA1, PI, AAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amfizem-Siroz, AAT Eksikliğine Bağlı Olarak, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (SERPINA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Emphysema-cirrhosis, due to AAT deficiency, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (SERPINA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA1	SERPINA1, PI, AAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amfizem-Siroz, AAT Eksikliğine Bağlı Olarak, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (MLPA)	Emphysema-cirrhosis, due to AAT deficiency, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (MLPA)	SERPINA1	SERPINA1, PI, AAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amfizem-Siroz, AAT Eksikliğine Bağlı Olarak, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (Prenatal) (SERPINA1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Emphysema-cirrhosis, due to AAT deficiency, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (SERPINA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPINA1	SERPINA1, PI, AAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Amfizem-Siroz, AAT Eksikliğine Bağlı Olarak, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (Prenatal) (MLPA)	Emphysema-cirrhosis, due to AAT deficiency, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (Prenatal) (MLPA)	SERPINA1	SERPINA1, PI, AAT	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalokraniokutanöz Lipomatozis, 613001, Somatik Mozaik; ECCL (Ensefalokraniokütanöz Lipomatoz) (FGFR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalocraniocutaneous lipomatosis, 613001, Somatic mosaicism; ECCL (Encephalocraniocutaneous lipomatosis) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Ensefalokraniokutanöz Lipomatozis, 613001, Somatik Mozaik; ECCL (Ensefalokraniokütanöz Lipomatoz) (Prenatal) (FGFR1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalocraniocutaneous lipomatosis, 613001, Somatic mosaicism; ECCL (Encephalocraniocutaneous lipomatosis) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Ensefalopati, Letal, Defektif Mitokondriyal ve Peroksisomal Fisyona Bağlı 1, 614388, Otozomal Dominant; EMPF1 (Letal Ensefalopati, Mitokondriyal ve Peroksisomal Fisyon Defekti) (DNM1L gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission 1, 614388, Autosomal dominant; EMPF1 (Lethal encephalopathy due to mitochondrial and peroxisomal fission defect) (DNM1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM1L	DNM1L, DRP1, DVLP, DYMPLE, EMPF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission (EMPF) [MIM:614388]: A rare autosomal dominant systemic disorder resulting in lack of neurologic development and death in infancy. After birth, infants present in the first week of life with poor feeding and neurologic impairment, including hypotonia, little spontaneous movement, no tendon reflexes, no response to light stimulation, and poor visual fixation. Other features include mildly elevated plasma concentration of very-long-chain fatty acids, lactic acidosis, microcephaly, deep-set eyes, optic atrophy and hypoplasia, and an abnormal gyral pattern in both frontal lobes associated with dysmyelination. {ECO:0000269|PubMed:17460227}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Ensefalopati, Letal, Defektif Mitokondriyal ve Peroksisomal Fisyona Bağlı 1, 614388, Otozomal Dominant; EMPF1 (Letal Ensefalopati, Mitokondriyal ve Peroksisomal Fisyon Defekti) (Prenatal) (DNM1L gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission 1, 614388, Autosomal dominant; EMPF1 (Lethal encephalopathy due to mitochondrial and peroxisomal fission defect) (DNM1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNM1L	DNM1L, DRP1, DVLP, DYMPLE, EMPF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Encephalopathy, lethal, due to defective mitochondrial and peroxisomal fission (EMPF) [MIM:614388]: A rare autosomal dominant systemic disorder resulting in lack of neurologic development and death in infancy. After birth, infants present in the first week of life with poor feeding and neurologic impairment, including hypotonia, little spontaneous movement, no tendon reflexes, no response to light stimulation, and poor visual fixation. Other features include mildly elevated plasma concentration of very-long-chain fatty acids, lactic acidosis, microcephaly, deep-set eyes, optic atrophy and hypoplasia, and an abnormal gyral pattern in both frontal lobes associated with dysmyelination. {ECO:0000269|PubMed:17460227}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Defektif Mitokondriyal ve Peroksisomal Fisyona Bağlı 2, 617086 (için Otozomal Resesif; EMPF2 (MFF gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086, Autosomal recessive; EMPF2 (MFF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFF	MFF, C2orf33, EMPF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Defektif Mitokondriyal ve Peroksisomal Fisyona Bağlı 2, 617086 İçin Otozomal Resesif; EMPF2 (Prenatal) (MFF gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy due to defective mitochondrial and peroxisomal fission 2, 617086, Autosomal recessive; EMPF2 (MFF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MFF	MFF, C2orf33, EMPF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Akut, Enfeksiyon Kaynaklı, 3, Yatkınlık, 608033, Otozomal Dominant; IIAE3 (Aktesil Akut Nekrotizan Ensefalopati) (RANBP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, acute, infection-induced, 3, susceptibility to, 608033, Autosomal dominant; IIAE3 (Familial acute necrotizing encephalopathy) (RANBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RANBP2	RANBP2, NUP358, ANE1, IIAE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Encephalopathy, acute, infection-induced, 3 (IIAE3) [MIM:608033]: A rapidly progressive encephalopathy manifesting in susceptible individuals with seizures and coma. It can occur within days in otherwise healthy children after common viral infections such as influenza and parainfluenza, without evidence of viral infection of the brain or inflammatory cell infiltration. Brain T2-weighted magnetic resonance imaging reveals characteristic symmetric lesions present in the thalami, pons and brainstem. {ECO:0000269|PubMed:19118815}. Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations in the RANBP2 gene predispose to IIAE3, but by themselves are insufficient to make the phenotype fully penetrant; additional genetic and environmental factors are required (PubMed:19118815). {ECO:0000269|PubMed:19118815}.,	EDTA Blood Sample
Ensefalopati, Akut, Enfeksiyon Kaynaklı, 3, Yatkınlık, 608033, Otozomal Dominant; IIAE3 (Aktesil Akut Nekrotizan Ensefalopati) (Prenatal) (RANBP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, acute, infection-induced, 3, susceptibility to, 608033, Autosomal dominant; IIAE3 (Familial acute necrotizing encephalopathy) (RANBP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RANBP2	RANBP2, NUP358, ANE1, IIAE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Encephalopathy, acute, infection-induced, 3 (IIAE3) [MIM:608033]: A rapidly progressive encephalopathy manifesting in susceptible individuals with seizures and coma. It can occur within days in otherwise healthy children after common viral infections such as influenza and parainfluenza, without evidence of viral infection of the brain or inflammatory cell infiltration. Brain T2-weighted magnetic resonance imaging reveals characteristic symmetric lesions present in the thalami, pons and brainstem. {ECO:0000269|PubMed:19118815}. Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations in the RANBP2 gene predispose to IIAE3, but by themselves are insufficient to make the phenotype fully penetrant; additional genetic and environmental factors are required (PubMed:19118815). {ECO:0000269|PubMed:19118815}.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Akut, Enfeksiyon Kaynaklı, 4, Yatkınlık, 614212, Otozomal Dominant, Otozomal Resesif (Çocukluk Akut Nekrotizan Ensefalopati) (CPT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, acute, infection-induced, 4, susceptibility to, 614212, Autosomal recessive, Autosomal dominant (Acute necrotizing encephalopathy of childhood) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Akut, Enfeksiyon Kaynaklı, 4, Yatkınlık, 614212, Otozomal Dominant, Otozomal Resesif (Çocukluk Akut Nekrotizan Ensefalopati) (Prenatal) (CPT2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, acute, infection-induced, 4, susceptibility to, 614212, Autosomal recessive, Autosomal dominant (Acute necrotizing encephalopathy of childhood) (CPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPT2	CPT2, IIAE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, GLUT1 eksikliğine bağlı (SLC2A1, ST XBP1) (MLPA)	Encephalopathy, due to GLUT1 deficiency (SLC2A1, ST XBP1) (MLPA)	SLC2A1, ST XBP1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ensefalopati, GLUT1 eksikliğine bağlı (SLC2A1, ST XBP1) (MLPA) (Prenatal)	Encephalopathy, due to GLUT1 deficiency (SLC2A1, ST XBP1) (MLPA) (Prenatal)	SLC2A1, ST XBP1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Etilmalonik Ee (Etilmalonik Ensefalopati) (ETHE1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	ENCEPHALOPATHY, ETHYLMALONIC, EE (Ethylmalonic encephalopathy) (ETHE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETHE1	ETHE1, HSCO, D83198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Etilmalonik Ee (Etilmalonik Ensefalopati) (Prenatal) (ETHE1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	ENCEPHALOPATHY, ETHYLMALONIC, EE (Ethylmalonic encephalopathy) (ETHE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ETHE1	ETHE1, HSCO, D83198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Ailesel, Nöroserpin İnklüzyon Cisimcikleri, 604218, Otozomal Dominant; FENIB (Ailesel Ensefalopati, Nöroserpin İnklüzyon Cisimcikleri Ib) (SERPIN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218, Autosomal dominant; FENIB (Familial encephalopathy with neuroserpin inclusion bodies) (SERPIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPIN1	SERPIN1, PI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Ailesel, Nöroserpin İnklüzyon Cisimcikleri, 604218, Otozomal Dominant; FENIB (Ailesel Ensefalopati, Nöroserpin İnklüzyon Cisimcikleri Ib) (Prenatal) (SERPIN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218, Autosomal dominant; FENIB (Familial encephalopathy with neuroserpin inclusion bodies) (SERPIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPIN1	SERPIN1, PI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Neonatal Ağır, 300673, X'e Bağlı (Ağır Neonatal Başlangıçlı Ensefalopati-Mikrosefali) (MECP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, neonatal severe, 300673, X-linked recessive (Severe neonatal-onset encephalopathy with microcephaly) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, ALTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Neonatal Ağır, 300673, X'e Bağlı (Ağır Neonatal Başlangıçlı Ensefalopati-Mikrosefali) (MLPA)	Encephalopathy, neonatal severe, 300673, X-linked recessive (Severe neonatal-onset encephalopathy with microcephaly) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, ALTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ensefalopati, Neonatal Ağır, 300673, X'e Bağlı (Ağır Neonatal Başlangıçlı Ensefalopati-Mikrosefali) (Prenatal) (MECP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, neonatal severe, 300673, X-linked recessive (Severe neonatal-onset encephalopathy with microcephaly) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, ALTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Neonatal Ağır, 300673, X'e Bağlı (Ağır Neonatal Başlangıçlı Ensefalopati-Mikrosefali) (Prenatal) (MLPA)	Encephalopathy, neonatal severe, 300673, X-linked recessive (Severe neonatal-onset encephalopathy with microcephaly) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, ALTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Progresif, Erken Başlangıçlı, Beyin Atrofisi ve İnce Korpus Kallosum, 617193, Otozomal Resesif; PEBAT (TBCD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193, Autosomal recessive; PEBAT (TBCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBCD	TBCD, PEBAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Progresif, Erken Başlangıçlı, Beyin Atrofisi ve İnce Korpus Kallosum, 617193, Otozomal Resesif; PEBAT (Prenatal) (TBCD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, progressive, early-onset, with brain atrophy and thin corpus callosum, 617193, Autosomal recessive; PEBAT (TBCD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBCD	TBCD, PEBAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Beyin Ödemi ve / Veya Lökoensefalopati Progresif, Erken Başlangıçlı, 617186, Otozomal Resesif; PEBE (NAXE gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186, Autosomal recessive; PEBE (NAXE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAXE	NAXE, APOA1BP, AIBP, PEBEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Beyin Ödemi ve / Veya Lökoensefalopati Progresif, Erken Başlangıçlı, 617186, Otozomal Resesif; PEBE (Prenatal) (NAXE gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, progressive, early-onset, with brain edema and/or leukoencephalopathy, 617186, Autosomal recessive; PEBE (NAXE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAXE	NAXE, APOA1BP, AIBP, PEBEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Progresif, Amyotrofi ve Optik Atrofi, 617207, Otozomal Resesif; PEAMO (Sanjad-Sakati Sendromu) (TBCE gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207, Autosomal recessive; PEAMO (Sanjad-Sakati syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Progresif, Amyotrofi ve Optik Atrofi, 617207, Otozomal Resesif; PEAMO (Sanjad-Sakati Sendromu) (Prenatal) (TBCE gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, progressive, with amyotrophy and optic atrophy, 617207, Autosomal recessive; PEAMO (Sanjad-Sakati syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ensefalopati, Progresif, Lipodistrofi Olan Veya Olmayan, 615924, Otozomal Resesif; PELD (Ağır Nörodejeneratif Sendrom-Lipodistrofi) (BSCL2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Encephalopathy, progressive, with or without lipodystrophy, 615924, Autosomal recessive; PELD (Severe neurodegenerative syndrome with lipodystrophy) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ensefalopati, Progresif, Lipodistrofi Olan Veya Olmayan, 615924, Otozomal Resesif; PELD (Ağır Nörodejeneratif Sendrom-Lipodistrofi) (Prenatal) (BSCL2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Encephalopathy, progressive, with or without lipodystrophy, 615924, Autosomal recessive; PELD (Severe neurodegenerative syndrome with lipodystrophy) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Son Evre Böbrek Hastalığı, Nondiyabetik, Yatkınlık, 612551; FSGS4 (Sporadik İdyopatik Steroide Dirençli Nefrotik Sendrom) (APOL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	End-stage renal disease, nondiabetic, susceptibility to, 612551; FSGS4 (Sporadic idiopathic steroid-resistant nephrotic syndrome) (APOL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOL1	APOL1, FSGS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 4 (FSGS4) [MIM:612551]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly mortal state requiring either dialysis therapy or kidney transplantation. {ECO:0000269|PubMed:20635188, ECO:0000269|PubMed:20647424}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Endokrin-Serebroosteodisplazi, 612651, Otozomal Resesif; ECO (Endokrin-Serebro-Osteodisplazi Sendromu) (ICK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Endocrine-cerebrosteodysplasia, 612651, Autosomal recessive; ECO (Endocrine-cerebro-osteodysplasia syndrome) (ICK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ICK	ICK, MRK, KIAA0936, ECO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endocrine-cerebrosteodysplasia (ECO) [MIM:612651]: Previously unidentified neonatal lethal recessive disorder with multiple anomalies involving the endocrine, cerebral, and skeletal systems. {ECO:0000269|PubMed:19185282}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Endokrin-Serebroosteodisplazi, 612651, Otozomal Resesif; ECO (Endokrin-Serebro-Osteodisplazi Sendromu) (Prenatal) (ICK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Endocrine-cerebroosteodysplasia, 612651, Autosomal recessive; ECO (Endocrine-cerebro-osteodysplasia syndrome) (ICK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ICK	ICK, MRK, KIAA0936, ECO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Endocrine-cerebroosteodysplasia (ECO) [MIM:612651]: Previously unidentified neonatal lethal recessive disorder with multiple anomalies involving the endocrine, cerebral, and skeletal systems. {ECO:0000269	PubMed:19185282}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Endometrial Kanser, Ailesel, 608089 (MSH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Endometrial cancer, familial, 608089 (MSH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH6	MSH6, GTBP, HNPCC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Endometrial Kanser, Ailesel, 608089 (MLPA)	Endometrial cancer, familial, 608089 (MLPA)	MSH6	MSH6, GTBP, HNPCC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Endometrial Kanser, Yatkınlık, 608089 (MLH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Endometrial cancer, susceptibility to, 608089 (MLH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH3	MLH3, HNPCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Endometriyal Karsinom, Somatik, 608089 (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Endometrial carcinoma, somatic, 608089 (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Endometriyal Karsinom, Somatik, 608089 (MSH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Endometrial carcinoma, somatic, 608089 (MSH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH3	MSH3, FAP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		20	60	.	.	30	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Endometriyal Karsinom, Somatik, 608089 (CDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Endometrial carcinoma, somatic, 608089 (CDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH1	CDH1, UVO, LCAM, ECAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Endosteal Hiperostoz, Otozomal Dominant (Otozomal Dominant Osteoskleroz, Worth Tipi) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ENDOSTEAL HYPEROSTOSIS, AUTOSOMAL DOMINANT (Autosomal dominant osteosclerosis, Worth type) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Güçlenmiş S-Kon Sendromu, 268100, Otozomal Resesif; ESCS (Goldmann-Favre Sendromu) (NR2E3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Enhanced S-cone syndrome, 268100, Autosomal recessive; ESCS (Goldmann-Favre syndrome) (NR2E3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR2E3	NR2E3, PNR, ESCS, RP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Enhanced S cone syndrome (ESCS) [MIM:268100]: Autosomal recessive retinopathy in which patients have increased sensitivity to blue light perception of blue light is mediated by what is normally the least populous cone photoreceptor subtype, the S (short wavelength, blue) cones. ESCS is also associated with visual loss, with night blindness occurring from early in life, varying degrees of L- (long, red)- and M (middle, green)-cone vision, and retinal degeneration. {ECO:0000269|PubMed:10655056, ECO:0000269|PubMed:11071390, ECO:0000269|PubMed:12963616, ECO:0000269|PubMed:15459973, ECO:0000269|PubMed:16225923, ECO:0000269|PubMed:18294254, ECO:0000269|PubMed:19006237}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Genişlemiş Vestibüler Kanal, 600791, Otozomal Resesif; DFNB4 (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (FOXI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Enlarged vestibular aqueduct, 600791, Autosomal recessive; DFNB4 (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (FOXI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXI1	FOXI1, FKHL10, FREAC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Genişlemiş Vestibüler Kanal, Dijenik, 600791, Otozomal Resesif (Otozomal Resesif Sendromik Olmayan Sensorinöral İşitme Kaybı Tipi DFNB) (KCNJ10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Enlarged vestibular aqueduct, digenic, 600791, Autosomal recessive (Autosomal recessive non-syndromic sensorineural deafness type DFNB) (KCNJ10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ10	KCNJ10, SESAME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Enolaz Eksikliği (ENO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Enolase deficiency (ENO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENO1	ENO1, PPH, MPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Enolaz Eksikliği (Prenatal) (ENO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Enolase deficiency (ENO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ENO1	ENO1, PPH, MPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Enterokinaz Eksikliği, 226200, Otozomal Resesif (Konjenital Enteropati, Enteropeptidaz Eksikliğine Bağlı) (TMPRSS15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Enterokinase deficiency, 226200, Autosomal recessive (Congenital enteropathy due to enteropeptidase deficiency) (TMPRSS15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMPRSS15	PRSS7, ENTK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Enterokinase deficiency (ENTKD) [MIM:226200]: Life-threatening intestinal malabsorption disorder characterized by diarrhea and failure to thrive. {ECO:0000269	PubMed:11719902}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Enterokinaz Eksikliği, 226200, Otozomal Resesif (Konjenital Enteropati, Enteropeptidaz Eksikliğine Bağlı) (Prenatal) (TMPRSS15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Enterokinase deficiency, 226200, Autosomal recessive (Congenital enteropathy due to enteropeptidase deficiency) (TMPRSS15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMPRSS15	PRSS7, ENTK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Enterokinase deficiency (ENTKD) [MIM:226200]: Life-threatening intestinal malabsorption disorder characterized by diarrhea and failure to thrive. {ECO:0000269	PubMed:11719902}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
EPCR 4600A>G Mutasyonu (. gen) (Dizi Analizi) (Postnatal)	EPCR 4600A>G mutation (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
EPCR 4678G>C Mutasyonu (. gen) (Dizi Analizi) (Postnatal)	EPCR 4678G>C mutation (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Epidermal Nevus, Somatik, 162900 (Yünsü Saç Nevus) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermal nevus, somatic, 162900 (Woolly hair nevus) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Epidermodisplazia Verruciformis, 226400, Otozomal Resesif (Epidermodisplazi Verruciformis) (TMC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermodysplasia verruciformis, 226400, Autosomal recessive (Epidermodysplasia verruciformis) (TMC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMC8	TMC8, EVER2, EV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermodisplazia Verruciformis, 226400, Otozomal Resesif (Epidermodisplazi Verruciformis) (Prenatal) (TMC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermodysplasia verruciformis, 226400, Autosomal recessive (Epidermodysplasia verruciformis) (TMC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMC8	TMC8, EVER2, EV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermodisplazia Verruciformis, 226400, Otozomal Resesif; EV (Epidermodisplazi Verruciformis) (TMC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermodysplasia verruciformis, 226400, Autosomal recessive; EV (Epidermodysplasia verruciformis) (TMC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMC6	TMC6, EVER1, EV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermodisplazia Verruciformis, 226400, Otozomal Resesif; EV (Epidermodisplazi Verruciformis) (Prenatal) (TMC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermodysplasia verruciformis, 226400, Autosomal recessive; EV (Epidermodysplasia verruciformis) (TMC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMC6	TMC6, EVER1, EV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Distrofika AD, 131750, Otozomal Dominant (Generalize Dominant Distrofik Epidermolizis Bülloza) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica, AD, 131750, Autosomal dominant (Generalized dominant dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Distrofika AD, 131750, Otozomal Dominant (Generalize Dominant Distrofik Epidermolizis Bülloza) (MLPA)	Epidermolysis bullosa dystrophica, AD, 131750, Autosomal dominant (Generalized dominant dystrophic epidermolysis bullosa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Distrofika AD, 131750, Otozomal Dominant (Generalize Dominant Distrofik Epidermolizis Bülloza) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica, AD, 131750, Autosomal dominant (Generalized dominant dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Distrofika AD, 131750, Otozomal Dominant (Generalize Dominant Distrofik Epidermolizis Bülloza) (Prenatal) (MLPA)	Epidermolysis bullosa dystrophica, AD, 131750, Autosomal dominant (Generalized dominant dystrophic epidermolysis bullosa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Distrofika, OR, 226600, Otozomal Resesif (Siddelli Jeneralize Resesif Distrofik Epidermolizis Bülloza) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica, AR, 226600, Autosomal recessive (Severe generalized recessive dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Distrofika, OR, 226600, Otozomal Resesif (Siddelli Jeneralize Resesif Distrofik Epidermolizis Bülloza) (MLPA)	Epidermolysis bullosa dystrophica, AR, 226600, Autosomal recessive (Severe generalized recessive dystrophic epidermolysis bullosa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Distrofika, OR, 226600, Otozomal Resesif (Siddelli Jeneralize Resesif Distrofik Epidermolizis Bülloza) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica, AR, 226600, Autosomal recessive (Severe generalized recessive dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Distrofika, OR, 226600, Otozomal Resesif (Siddelli Jeneralize Resesif Distrofik Epidermolizis Bülloza) (Prenatal) (MLPA)	Epidermolysis bullosa dystrophica, AR, 226600, Autosomal recessive (Severe generalized recessive dystrophic epidermolysis bullosa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Distrofik, Otozomal Resesif, Modifiye Edici, 226600, Otozomal Resesif; RDEB (Resesif Distrofik Epidermolizis Bülloza Inversa) (MMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa dystrophica, autosomal recessive, modifier of, 226600, Autosomal recessive; RDEB (Recessive dystrophic epidermolysis bullosa inversa) (MMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP1	MMP1, CLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Distrofik, Otozomal Resesif, Modifiye Edici, 226600, Otozomal Resesif; RDEB (Resesif Distrofik Epidermolizis Bülloza Inversa) (Prenatal) (MMP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa dystrophica, autosomal recessive, modifier of, 226600, Autosomal recessive; RDEB (Recessive dystrophic epidermolysis bullosa inversa) (MMP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP1	MMP1, CLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, El ve Ayaklar, 131800, Otozomal Dominant (Lokalize Epidermolizis Bülloza Simpleks) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa of hands and feet, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, El ve Ayaklar, 131800, Otozomal Dominant (Lokalize Epidermolizis Bülloza Simpleks) (Prenatal) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa of hands and feet, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolisis Bullosa paneli (Miks 1) (3p21.31 COL7A1, 12q13.13 KRT5) (MLPA)	Epidermolysis Bullosa panel (Mix 1) (3p21.31 COL7A1, 12q13.13 KRT5) (MLPA)	3p21.31 COL7A1, 12q13.13 KRT5	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolisis Bullosa paneli (Miks 1) (3p21.31 COL7A1, 12q13.13 KRT5) (MLPA) (Prenatal)	Epidermolysis Bullosa panel (Mix 1) (3p21.31 COL7A1, 12q13.13 KRT5) (MLPA) (Prenatal)	3p21.31 COL7A1, 12q13.13 KRT5	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolisis Bullosa paneli (Miks 2) (17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2) (MLPA)	Epidermolysis Bullosa panel (Mix 2) (17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2) (MLPA)	17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolisis Bullosa paneli (Miks 2) (17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2) (MLPA) (Prenatal)	Epidermolysis Bullosa panel (Mix 2) (17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2) (MLPA) (Prenatal)	17q21.2 KRT14; 18q11.2 LAMA3; 1q32.2 LAMB3; 1q25.3 LAMC2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epidermolizis Bülloza Pruriginosa, 604129, Otozomal Resesif, Otozomal Dominant (Distrofik Epidermolizis Bülloza Pruriginosa) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa pruriginosa, 604129, Autosomal recessive, Autosomal dominant (Dystrophic epidermolysis bullosa pruriginosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Pruriginosa, 604129, Otozomal Resesif, Otozomal Dominant (Distrofik Epidermolizis Bülloza Pruriginosa) (MLPA)	Epidermolysis bullosa pruriginosa, 604129, Autosomal recessive, Autosomal dominant (Dystrophic epidermolysis bullosa pruriginosa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Pruriginosa, 604129, Otozomal Resesif, Otozomal Dominant (Distrofik Epidermolizis Bülloza Pruriginosa) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa pruriginosa, 604129, Autosomal recessive, Autosomal dominant (Dystrophic epidermolysis bullosa pruriginosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	.	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Pruriginosa, 604129, Otozomal Resesif, Otozomal Dominant (Distrofik Epidermolizis Bülloza Pruriginosa) (Prenatal) (MLPA)	Epidermolysis bullosa pruriginosa, 604129, Autosomal recessive, Autosomal dominant (Dystrophic epidermolysis bullosa pruriginosa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks-Müsküler Distrofi, 226670, Otozomal Resesif, EBSMD (Müsküler Distrofi-Epidermolizis Bulosa Simpleks) (PLEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex with muscular dystrophy, 226670, Autosomal recessive; EBSMD (Epidermolysis bullosa simplex with muscular dystrophy) (PLEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks-Müsküler Distrofi, 226670, Otozomal Resesif, EBSMD (Müsküler Distrofi-Epidermolizis Bulosa Simpleks) (Prenatal) (PLEC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex with muscular dystrophy, 226670, Autosomal recessive; EBSMD (Epidermolysis bullosa simplex with muscular dystrophy) (PLEC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Tırnak Distrofisi, 616487, Otozomal Resesif; EBSND (Müsküler Distrofi-Epidermolizis Bulosa Simpleks) (PLEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex with nail dystrophy, 616487, Autosomal recessive; EBSND (Epidermolysis bullosa simplex with muscular dystrophy) (PLEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Tırnak Distrofisi, 616487, Otozomal Resesif; EBSND (Müsküler Distrofi-Epidermolizis Bulosa Simpleks) (Prenatal) (PLEC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex with nail dystrophy, 616487, Autosomal recessive; EBSND (Epidermolysis bullosa simplex with muscular dystrophy) (PLEC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks-Pilor Atrezisi, 612138, Otozomal Resesif; EBSPA (Epidermolizis Bulosa Simpleks-Pilorik Atrezi) (PLEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex with pyloric atresia, 612138, Autosomal recessive; EBSPA (Epidermolysis bullosa simplex with pyloric atresia) (PLEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks-Pilor Atrezisi, 612138, Otozomal Resesif; EBSPA (Epidermolizis Bulosa Simpleks-Pilorik Atrezi) (PLEC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex with pyloric atresia, 612138, Autosomal recessive; EBSPA (Epidermolysis bullosa simplex with pyloric atresia) (PLEC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks-MP, 131960, Otozomal Dominant; EBSMP (Epidermolizis Bülloza Simpleks-MP) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex-MP, 131960, Autosomal dominant; EBSMP (Epidermolysis bullosa simplex with mottled pigmentation) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks-MP, 131960, Otozomal Dominant; EBSMP (Epidermolizis Bülloza Simpleks-MP) (MLPA)	Epidermolysis bullosa simplex-MP, 131960, Autosomal dominant; EBSMP (Epidermolysis bullosa simplex with mottled pigmentation) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks-MP, 131960, Otozomal Dominant; EBSMP (Epidermolizis Bülloza Simpleks-MP) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex-MP, 131960, Autosomal dominant; EBSMP (Epidermolysis bullosa simplex with mottled pigmentation) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks-MP, 131960, Otozomal Dominant; EBSMP (Epidermolizis Bülloza Simpleks-MP) (Prenatal) (MLPA)	Epidermolysis bullosa simplex-MP, 131960, Autosomal dominant; EBSMP (Epidermolysis bullosa simplex with mottled pigmentation) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Resesif 1; Ebsb1 (KRT14 İlişkili Epidermolizis Bülloza Simpleks) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1; EBSB1 (KRT14-related epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Resesif 1; Ebsb1 (KRT14 İlişkili Epidermolizis Bülloza Simpleks) (MLPA)	EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1; EBSB1 (KRT14-related epidermolysis bullosa simplex) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Resesif 1; Ebsb1 (KRT14 İlişkili Epidermolizis Bülloza Simpleks) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1; EBSB1 (KRT14-related epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Resesif 1; Ebsb1 (KRT14 İlişkili Epidermolizis Bülloza Simpleks) (Prenatal) (MLPA)	EPIDERMOLYSIS BULLOSA SIMPLEX, AUTOSOMAL RECESSIVE 1; EBSB1 (KRT14-related epidermolysis bullosa simplex) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Resesif 2, 615425, Otozomal Resesif; EBSB2 (Epidermolizis Bülloza Simpleks, BP230 Eksikliğine Bağlı) (DST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, autosomal recessive 2, 615425, Autosomal recessive; EBSB2 (Epidermolysis bullosa simplex due to BP230 deficiency) (DST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DST	DST, BPAG1, DMH, D6S1101, HSAN6, EBSB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Otozomal Reseasif 2, 615425, Otozomal Resesif; EBSB2 (Epidermolizis Bülloza Simpleks, BP230 Eksikliğine Bağlı) (Prenatal) (DST geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, autosomal recessive 2, 615425, Autosomal recessive; EBSB2 (Epidermolysis bullosa simplex due to BP230 deficiency) (DST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DST	DST, BPAG1, DMH, D6S1101, HSAN6, EBSB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant (Epidermolysis bullosa simplex, Dowling-Meara type) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (MLPA)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant (Epidermolysis bullosa simplex, Dowling-Meara type) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (Prenatal) (KRT14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant (Epidermolysis bullosa simplex, Dowling-Meara type) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT14	KRT14	.	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (Prenatal) (MLPA)	KRT14	KRT14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa simplex, Dowling-Meara type (DM- EBS) [MIM:131760]: A severe form of intraepidermal epidermolysis bullosa characterized by generalized herpetiform blistering, milia formation, dystrophic nails, and mucous membrane involvement. {ECO:0000269|PubMed:10730767, ECO:0000269|PubMed:12655565, ECO:0000269|PubMed:1372711, ECO:0000269|PubMed:16786515, ECO:0000269|PubMed:16882168, ECO:0000269|PubMed:21623745, ECO:0000269|PubMed:8757772, ECO:0000269|PubMed:9036937, ECO:0000269|PubMed:9406827, ECO:0000269|PubMed:9989794, ECO:0000269|Ref.16}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (MLPA)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epidermolysis bullosa simplex, Dowling-Meara type (DM- EBS) [MIM:131760]: A severe form of intraepidermal epidermolysis bullosa characterized by generalized herpetiform blistering, milia formation, dystrophic nails, and mucous membrane involvement. {ECO:0000269|PubMed:10730767, ECO:0000269|PubMed:12655565, ECO:0000269|PubMed:1372711, ECO:0000269|PubMed:16786515, ECO:0000269|PubMed:16882168, ECO:0000269|PubMed:21623745, ECO:0000269|PubMed:8757772, ECO:0000269|PubMed:9036937, ECO:0000269|PubMed:9406827, ECO:0000269|PubMed:9989794, ECO:0000269|Ref.16}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa simplex, Dowling-Meara type (DM- EBS) [MIM:131760]: A severe form of intraepidermal epidermolysis bullosa characterized by generalized herpetiform blistering, milia formation, dystrophic nails, and mucous membrane involvement. {ECO:0000269|PubMed:10730767, ECO:0000269|PubMed:12655565, ECO:0000269|PubMed:1372711, ECO:0000269|PubMed:16786515, ECO:0000269|PubMed:16882168, ECO:0000269|PubMed:21623745, ECO:0000269|PubMed:8757772, ECO:0000269|PubMed:9036937, ECO:0000269|PubMed:9406827, ECO:0000269|PubMed:9989794, ECO:0000269|Ref.16}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi, 131760, Otozomal Dominant; EBSDM (Epidermolizis Bülloza Simpleks, Dowling-Meara Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Dowling-Meara type, 131760, Autosomal dominant; EBSDM (Epidermolysis bullosa simplex, Dowling-Meara type) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa simplex, Dowling-Meara type (DM- EBS) [MIM:131760]: A severe form of intraepidermal epidermolysis bullosa characterized by generalized herpetiform blistering, milia formation, dystrophic nails, and mucous membrane involvement. {ECO:0000269|PubMed:10730767, ECO:0000269|PubMed:12655565, ECO:0000269|PubMed:1372711, ECO:0000269|PubMed:16786515, ECO:0000269|PubMed:16882168, ECO:0000269|PubMed:21623745, ECO:0000269|PubMed:8757772, ECO:0000269|PubMed:9036937, ECO:0000269|PubMed:9406827, ECO:0000269|PubMed:9989794, ECO:0000269|Ref.16}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epidermolizis Büllosa Simpleks, Jeneralize, Skar ve Saç Kaybı, 617294, Otozomal Dominant; EBSSH (KLHL24 geni) (Tüm Gen Dizi Analizi (Postnatal)	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294, Autosomal dominant; EBSSH (KLHL24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL24	KLHL24, KRIP6, EBSSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Jeneralize, Skar ve Saç Kaybı, 617294, Otozomal Dominant; EBSSH (Prenatal) (KLHL24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, 617294, Autosomal dominant; EBSSH (KLHL24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLHL24	KLHL24, KRIP6, EBSSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (MLPA)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (MLPA)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (Prenatal) (KRT14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Koebner Tipi, 131900, Otozomal Dominant (Jeneralize Epidermolizis Büllosa Simpleks, Non-Dowling-Meara Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Koebner type, 131900, Autosomal dominant (Generalized epidermolysis bullosa simplex, non-Dowling-Meara type) (Prenatal) (MLPA)	KRT14	KRT14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Lokalize (Lokalize Epidermolizis Büllosa Simpleks) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Lokalize (Lokalize Epidermolizis Büllosa Simpleks) (Prenatal) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	EPIDERMOLYSIS BULLOSA SIMPLEX, LOCALIZED (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Ogna Tipi, 131950, Otozomal Dominant; EBSOG (Epidermolizis Büllosa Simpleks, Ogna Tipi) (PLEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Ogna type, 131950, Autosomal dominant; EBSOG (Epidermolysis bullosa simplex, Ogna type) (PLEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa simplex, Ogna type (O-EBS) [MIM:131950]: A form of intraepidermal epidermolysis bullosa characterized by generalized skin bruising, skin fragility with non-scarring blistering and small hemorrhagic blisters on hands. At the ultrastructural level, it is differentiated from classical cases of K-EBS, WC-EBS and DM-EBS, by the occurence of blisters originating in basal cells above hemidesmosomes, and abnormal hemidesmosome intracellular attachment plates.. (ECO:0000269	PubMed:11851880). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Ogna Tipi, 131950, Otozomal Dominant; EBSOG (Epidermolizis Büllosa Simpleks, Ogna Tipi) (Prenatal) (PLEC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Ogna type, 131950, Autosomal dominant; EBSOG (Epidermolysis bullosa simplex, Ogna type) (PLEC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa simplex, Ogna type (O-EBS) [MIM:131950]: A form of intraepidermal epidermolysis bullosa characterized by generalized skin bruising, skin fragility with non-scarring blistering and small hemorrhagic blisters on hands. At the ultrastructural level, it is differentiated from classical cases of K-EBS, WC-EBS and DM-EBS, by the occurence of blisters originating in basal cells above hemidesmosomes, and abnormal hemidesmosome intracellular attachment plates. (ECO:0000269	PubMed:11851880). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (MLPA)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (MLPA)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (Prenatal) (KRT14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Resesif 1, 601001, Otozomal Resesif (KRT14 İlişkili Epidermolizis Büllosa Simpleks) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, recessive 1, 601001, Autosomal recessive (KRT14-related epidermolysis bullosa simplex) (MLPA)	KRT14	KRT14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (KRT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (MLPA)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (MLPA)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (Prenatal) (KRT14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa Simpleks, Weber-Cockayne Tipi, 131800, Otozomal Dominant (Lokalize Epidermolizis Büllosa Simpleks) (Prenatal) (MLPA)	Epidermolysis bullosa simplex, Weber-Cockayne type, 131800, Autosomal dominant (Localized epidermolysis bullosa simplex) (Prenatal) (MLPA)	KRT14	KRT14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa, Jeneralize Atrofik Benign, 226650, Otozomal Resesif (LAMA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, generalized atrophic benign, 226650, Autosomal recessive (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa, Jeneralize Atrofik Benign, 226650, Otozomal Resesif (MLPA)	Epidermolysis bullosa, generalized atrophic benign, 226650, Autosomal recessive (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Büllosa, Jeneralize Atrofik Benign, 226650, Otozomal Resesif (Prenatal) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, generalized atrophic benign, 226650, Autosomal recessive (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa, Jeneralize Atrofik Benign, 226650, Otozomal Resesif (Prenatal) (MLPA)	Epidermolysis bullosa, generalized atrophic benign, 226650, Autosomal recessive (Prenatal) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Büllosa, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolysis Bullosa, Herlitz Tipi) (LAMC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMC2	LAMC2, LAMNB2, LAMB2T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (LAMB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB3	LAMB3, AI1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (MLPA)	LAMC2	LAMC2, LAMNB2, LAMB2T	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (LAMC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMC2	LAMC2, LAMNB2, LAMB2T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (LAMB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMB3	LAMB3, AI1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMC2	LAMC2, LAMNB2, LAMB2T	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMC2	LAMC2, LAMNB2, LAMB2T	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:11810295, ECO:0000269|PubMed:8012393). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Herlitz Tipi, 226700, Otozomal Resesif (Junctional Epidermolisis Bullosa, Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, Herlitz type, 226700, Autosomal recessive (Junctional epidermolysis bullosa, Herlitz type) (Prenatal) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. (ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Localisata Varyant, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (COL17A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, localisata variant, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (COL17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL17A1	COL17A1, BPAG2, ERED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Localisata Varyant, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (Prenatal) (COL17A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, localisata variant, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (COL17A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL17A1	COL17A1, BPAG2, ERED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (LAMC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (LAMC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMC2	LAMC2, LAMNB2, LAMB2T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (LAMB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (LAMB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB3	LAMB3, AI1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (MLPA)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (MLPA)	LAMC2	LAMC2, LAMNB2, LAMB2T	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (MLPA)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (Prenatal) (LAMC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (LAMC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMC2	LAMC2, LAMNB2, LAMB2T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (Prenatal) (LAMB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (LAMB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMB3	LAMB3, AI1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (Prenatal) (MLPA)	LAMC2	LAMC2, LAMNB2, LAMB2T	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Junktional Epidermolizis Bülloza, Non-Herlitz Tipi) (Prenatal) (MLPA)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Junctional epidermolysis bullosa, non-Herlitz type) (Prenatal) (MLPA)	LAMB3	LAMB3, AI1A	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Lokalize Epidermolizis Bülloza Simpleks) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized atrophic benign epidermolysis bullosa (GABEB) [MIM:226650]: A non-lethal, adult form of junctional epidermolysis bullosa characterized by life-long blistering of the skin, associated with hair and tooth abnormalities. [ECO:0000269	PubMed:10792571]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Lokalize Epidermolizis Bülloza Simpleks) (Prenatal) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Localized epidermolysis bullosa simplex) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Generalized atrophic benign epidermolysis bullosa (GABEB) [MIM:226650]: A non-lethal, adult form of junctional epidermolysis bullosa characterized by life-long blistering of the skin, associated with hair and tooth abnormalities. {ECO:0000269	PubMed:10792571}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Epidermolizis Bülloza, Lokalize, Kavşak Tipi (Juntional), Non-Herlitz Tipi) (COL17A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Localized junctional epidermolysis bullosa, non-Herlitz type) (COL17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL17A1	COL17A1, BPAG2, ERED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Non-Herlitz Tipi, 226650, Otozomal Resesif (Epidermolizis Bülloza, Lokalize, Kavşak Tipi (Juntional), Non-Herlitz Tipi) (Prenatal) (COL17A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, non-Herlitz type, 226650, Autosomal recessive (Localized junctional epidermolysis bullosa, non-Herlitz type) (COL17A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL17A1	COL17A1, BPAG2, ERED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Pilor Atrezisi, 226730, Otozomal Resesif (Junctional Epidermolisis Bulloza-Pilorik Atrezi Sendromu) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, with pyloric atresia, 226730, Autosomal recessive (Junctional epidermolysis bullosa-pyloric atresia syndrome) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Pilor Atrezisi, 226730, Otozomal Resesif (Junctional Epidermolisis Bulloza-Pilorik Atrezi Sendromu) (Prenatal) (ITGB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, with pyloric atresia, 226730, Autosomal recessive (Junctional epidermolysis bullosa-pyloric atresia syndrome) (ITGB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGB4	ITGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Pilorik Stenoz, 226730, Otozomal Resesif (Junctional Epidermolisis Bulloza-Pilorik Atrezi Sendromu) (ITGA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730, Autosomal recessive (Junctional epidermolysis bullosa-pyloric atresia syndrome) (ITGA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA6	ITGA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa letalis, with pyloric atresia (EB-PA) [MIM:226730]: An autosomal recessive, frequently lethal, epidermolysis bullosa with variable involvement of skin, nails, mucosa, and with variable effects on the digestive system. It is characterized by mucocutaneous fragility, aplasia cutis congenita, and gastrointestinal atresia, which most commonly affects the pylorus. Pyloric atresia is a primary manifestation rather than a scarring process secondary to epidermolysis bullosa. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolizis Bülloza, Junctional, Pilorik Stenoz, 226730, Otozomal Resesif (Junctional Epidermolisis Bulloza-Pilorik Atrezi Sendromu) (Prenatal) (ITGA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730, Autosomal recessive (Junctional epidermolysis bullosa-pyloric atresia syndrome) (ITGA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGA6	ITGA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa letalis, with pyloric atresia (EB-PA) [MIM:226730]: An autosomal recessive, frequently lethal, epidermolysis bullosa with variable involvement of skin, nails, mucosa, and with variable effects on the digestive system. It is characterized by mucocutaneous fragility, aplasia cutis congenita, and gastrointestinal atresia, which most commonly affects the pylorus. Pyloric atresia is a primary manifestation rather than a scarring process secondary to epidermolysis bullosa. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Letal Akantolitik, 609638, Otozomal Resesif, EBLA (Letal Akantolitik Epidermolitis Bülloza) (DSP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, lethal acantholytic, 609638, Autosomal recessive, EBLA (Lethal acantholytic epidermolysis bullosa) (DSP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Letal Akantolitik, 609638, Otozomal Resesif, EBLA (Letal Akantolitik Epidermolitis Bülloza) (Prenatal) (DSP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, lethal acantholytic, 609638, Autosomal recessive, EBLA (Lethal acantholytic epidermolysis bullosa) (DSP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Nonspesifik, Otozomal Resesif, 615028, Otozomal Resesif (Epidermolizis Bülloza Simpleks, Eksofilin 5 Eksikliğine Bağlı) (EXPH5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028, Autosomal recessive (Epidermolysis bullosa simplex due to exophilin 5 deficiency) (EXPH5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXPH5	EXPH5, SLAC2B, KIAA0624	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Nonspesifik, Otozomal Resesif, 615028, Otozomal Resesif (Epidermolizis Bülloza Simpleks, Eksofilin 5 Eksikliğine Bağlı) (Prenatal) (EXPH5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, nonspecific, autosomal recessive, 615028, Autosomal recessive (Epidermolysis bullosa simplex due to exophilin 5 deficiency) (EXPH5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EXPH5	EXPH5, SLAC2B, KIAA0624	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Spesifik Olmayan, Otozomal Resesif; Ebns (Epidermolizis Bülloza Simpleks, Eksofilin 5 Eksikliğine Bağlı) (EXPH5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	EPIDERMOLYSIS BULLOSA, NONSPECIFIC, AUTOSOMAL RECESSIVE; EBNS (Epidermolysis bullosa simplex due to exophilin 5 deficiency) (EXPH5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXPH5	EXPH5, SLAC2B, KIAA0624	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Spesifik Olmayan, Otozomal Resesif; Ebns (Epidermolizis Bülloza Simpleks, Eksofilin 5 Eksikliğine Bağlı) (Prenatal) (EXPH5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	EPIDERMOLYSIS BULLOSA, NONSPECIFIC, AUTOSOMAL RECESSIVE; EBNS (Epidermolysis bullosa simplex due to exophilin 5 deficiency) (EXPH5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EXPH5	EXPH5, SLAC2B, KIAA0624	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Pretibial, 131850, Otozomal Resesif, Otozomal Dominant (Pretibial Distrofik Epidermolizis Bülloza) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolysis bullosa, pretibial, 131850, Autosomal recessive, Autosomal dominant (Pretibial dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Pretibial, 131850, Otozomal Resesif, Otozomal Dominant (Pretibial Distrofik Epidermolizis Bülloza) (MLPA)	Epidermolysis bullosa, pretibial, 131850, Autosomal recessive, Autosomal dominant (Pretibial dystrophic epidermolysis bullosa) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülloza, Pretibial, 131850, Otozomal Resesif, Otozomal Dominant (Pretibial Distrofik Epidermolizis Bülloza) (Prenatal) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa, pretibial, 131850, Autosomal recessive, Autosomal dominant (Pretibial dystrophic epidermolysis bullosa) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza, Pretibial, 131850, Otozomal Resesif, Otozomal Dominant (Pretibial Distrofik Epidermolizis Bülloza) (Prenatal) (MLPA)	Epidermolysis bullosa, pretibial, 131850, Autosomal recessive, Autosomal dominant (Pretibial dystrophic epidermolysis bullosa) (Prenatal) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolitik Hiperkeratoz, 113800, Otozomal Resesif, Otozomal Dominant (Epidermolitik İktiyoz) (KRT10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolytic hyperkeratosis, 113800, Autosomal recessive, Autosomal dominant (Epidermolytic ichthyosis) (KRT10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT10	KRT10, EHK, BCIE, BIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10201536, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381287, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507150, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7508181, ECO:0000269|PubMed:7512983, ECO:0000269|Ref.7}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolitik Hiperkeratoz, 113800, Otozomal Resesif, Otozomal Dominant (Epidermolitik İktiyoz) (Prenatal) (KRT10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolytic hyperkeratosis, 113800, Autosomal recessive, Autosomal dominant (Epidermolytic ichthyosis) (KRT10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT10	KRT10, EHK, BCIE, BIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10201536, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381287, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507150, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7508181, ECO:0000269|PubMed:7512983, ECO:0000269|Ref.7}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolitik Hiperkeratoz, 113800, Otozomal Resesif, Otozomal Dominant; EHK (Epidermolitik İktiyoz) (KRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epidermolytic hyperkeratosis, 113800, Autosomal recessive, Autosomal dominant; EHK (Epidermolytic ichthyosis) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epidermolitik Hiperkeratoz, 113800, Otozomal Resesif, Otozomal Dominant; EHK (Epidermolitik İktiyoz) (Prenatal) (KRT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolytic hyperkeratosis, 113800, Autosomal recessive, Autosomal dominant; EHK (Epidermolytic ichthyosis) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülloza Simpleks-MCR, 609352 (Epidermolizis Bülloza Simpleks-Cincinate Göçmen Eritemi (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermolysis bullosa simplex-MCR, 609352 (Epidermolysis bullosa simplex with circinate migratory erythema) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Epidermolizis Bülbosa Simpleks-MCR, 609352 (Epidermolizis Bülbosa Simpleks-Cincinale Gömen Eritemi (MLPA)	Epidermiylysis bullosa simplex-MCR, 609352 (Epidermolysis bullosa simplex with circinate migratory erythema) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epidermolizis Bülbosa Simpleks-MCR, 609352 (Epidermolizis Bülbosa Simpleks-Cincinale Gömen Eritemi) (Prenatal) (KRT5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epidermiylysis bullosa simplex-MCR, 609352 (Epidermolysis bullosa simplex with circinate migratory erythema) (KRT5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT5	KRT5, DDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epidermolizis Bülbosa Simpleks-MCR, 609352 (Epidermolizis Bülbosa Simpleks-Cincinale Gömen Eritemi) (Prenatal) (MLPA)	Epidermiylysis bullosa simplex-MCR, 609352 (Epidermolysis bullosa simplex with circinate migratory erythema) (Prenatal) (MLPA)	KRT5	KRT5, DDD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize 8, 612899 (Aliesel Hipokalsiürik Hiperkalsemi) (CASR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy idiopathic generalized, susceptibility to, 8, 612899 (Familial hypocalciuric hypercalcemia) (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize 8, 612899 (Aliesel Hipokalsiürik Hiperkalsemi) (MLPA)	Epilepsy idiopathic generalized, susceptibility to, 8, 612899 (Familial hypocalciuric hypercalcemia) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epilepsi, Çocukluk Çağı Absans Tipi, Yatkınlık, 2, 607681, Otozomal Dominant; ECA2 (Çocukluk Çağı Absans Tipi Epilepsi) (GABRG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, childhood absence, susceptibility to, 2, 607681, Autosomal dominant; ECA2 (Childhood absence epilepsy) (GABRG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRG2	GABRG2, GEFSP3, CAE2, ECA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, childhood absence 2 (ECA2) [MIM:607681]: A subtype of idiopathic generalized epilepsy characterized by an onset at age 6-7 years, frequent absence seizures (several per day) and bilateral, synchronous, symmetric 3-Hz spike waves on EEG. Tonic-clonic seizures often develop in adolescence. Some individuals manifest febrile seizures. Absence seizures may either remit or persist into adulthood. {ECO:0000269	PubMed:11326275}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Çocukluk Çağı Absans Tipi, Yatkınlık, 4, 611136; EIG13 (Çocukluk Çağı Absans Tipi Epilepsi) (GABRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, childhood absence, susceptibility to, 4, 611136; EIG13 (Childhood absence epilepsy) (GABRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRA1	GABRA1, EJM5, ECA4, EIEE19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, childhood absence 4 (ECA4) [MIM:611136]: A subtype of idiopathic generalized epilepsy characterized by an onset at age 6-7 years, frequent absence seizures (several per day) and bilateral, synchronous, symmetric 3-Hz spike waves on EEG. Tonic-clonic seizures often develop in adolescence. Absence seizures may either remit or persist into adulthood. {ECO:0000269	PubMed:16718694}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Çocukluk Çağı Absans Tipi, Yatkınlık, 5, 612269; ECA5 (Çocukluk Çağı Absans Tipi Epilepsi) (GABRB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, childhood absence, susceptibility to, 5, 612269; ECA5 (Childhood absence epilepsy) (GABRB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRB3	GABRB3, ECA5, EIEE43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, childhood absence 5 (ECA5) [MIM:612269]: A subtype of idiopathic generalized epilepsy characterized by an onset at age 6-7 years, frequent absence seizures (several per day) and bilateral, synchronous, symmetric 3-Hz spike waves on EEG. Tonic-clonic seizures often develop in adolescence. Absence seizures may either remit or persist into adulthood. {ECO:0000269	PubMed:18514161}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Çocukluk Çağı Absans Tipi, Yatkınlık, 6, 611942; ECA6 (Çocukluk Çağı Absans Tipi Epilepsi) (CACNA1H geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, childhood absence, susceptibility to, 6, 611942; ECA6 (Childhood absence epilepsy) (CACNA1H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1H	CACNA1H, EIG6, ECA6, HALD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, idiopathic generalized 6 (EIG6) [MIM:611942]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269	PubMed:15048902}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Erken Başlangıçlı Vitamin, B6 Vitamin Bağımlı, 617290, Otozomal Resesif; EPVB6D (Piridoksin Bağımlı Epilepsi) (PLPBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, early-onset, vitamin B6-dependent, 617290, Autosomal recessive; EPVB6D (Pyridoxine-dependent epilepsy) (PLPBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLPBP	PROSC, EPVB6D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Aliesel Fokal, Değişken Odaklı 1, 604364, Otozomal Dominant; FFEVF1 (Aliesel Değişken Odaklı Fokal Epilepsi) (DEPDC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial focal, with variable foci 1, 604364, Autosomal dominant; FFEVF1 (Familial focal epilepsy with variable foci) (DEPDC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DEPDC5	DEPDC5, KIAA0645, FFEVF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Aliesel Fokal, Değişken Odaklı 2, 617116, Otozomal Dominant; FFEVF2 (Aliesel Değişken Odaklı Fokal Epilepsi) (NPRL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial focal, with variable foci 2, 617116, Autosomal dominant; FFEVF2 (Familial focal epilepsy with variable foci) (NPRL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPRL2	NPR2L, NPRL2, FFEVF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Aliesel Fokal, Değişken Odaklı 3, 617118, Otozomal Dominant; FFEVF3 (Aliesel Değişken Odaklı Fokal Epilepsi) (NPRL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial focal, with variable foci 3, 617118, Autosomal dominant; FFEVF3 (Familial focal epilepsy with variable foci) (NPRL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPRL3	NPRL3, CGTHBA, FFEVF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Aliesel Temporal Lob, 1, 600512, Otozomal Dominant; ETL1 (Otozomal Dominant Epilepsi-İyitsel Bulgular) (LGI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial temporal lobe, 1, 600512, Autosomal dominant; ETL1 (Autosomal dominant epilepsy with auditory features) (LGI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LGI1	LGI1, EPT, ETL1, ADLTE, ADPEAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, familial temporal lobe, 1 (ETL1) [MIM:600512]: A focal form of epilepsy characterized by recurrent seizures that arise from foci within the temporal lobe. Seizures are usually accompanied by sensory symptoms, most often auditory in nature. {ECO:0000269|PubMed:11810107, ECO:0000269|PubMed:12205652, ECO:0000269|PubMed:12601709, ECO:0000269|PubMed:12771769, ECO:0000269|PubMed:15079010, ECO:0000269|PubMed:17296837, ECO:0000269|PubMed:17562837, ECO:0000269|PubMed:18625862, ECO:0000269|PubMed:19552651}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Aliesel Temporal Lob, 5, 614417, Otozomal Dominant; ETL5 (Benign Aliesel Mesial Temporal Lob Epilepsisi) (CPA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial temporal lobe, 5, 614417, Autosomal dominant; ETL5 (Benign familial mesial temporal lobe epilepsy) (CPA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPA6	CPA6, CPAH, ETL5, FEB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, familial temporal lobe, 5 (ETL5) [MIM:614417]: A focal form of epilepsy characterized by recurrent seizures that arise from foci within the temporal lobe. Seizures are usually accompanied by sensory symptoms, most often auditory in nature. {ECO:0000269	PubMed:21922598}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Aliesel Temporal Lob, 7, 616436, Otozomal Dominant; ETL7 (Otozomal Dominant Epilepsi-İyitsel Bulgular) (RELN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial temporal lobe, 7, 616436, Autosomal dominant; ETL7 (Autosomal dominant epilepsy with auditory features) (RELN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RELN	RELN, RL, LIS2, ETL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Aliesel Temporal Lob, 8, 616461, Otozomal Dominant; ETL8 (Otozomal Dominant Epilepsi-İyitsel Bulgular) (GAL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, familial temporal lobe, 8, 616461, Autosomal dominant; ETL8 (Autosomal dominant epilepsy with auditory features) (GAL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAL	GAL, GALN, GLNN, ETL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, familial temporal lobe, 8 (ETL8) [MIM:616461]: A focal form of epilepsy characterized by recurrent seizures that arise from foci within the temporal lobe. Seizures are usually accompanied by sensory symptoms, most often auditory in nature. {ECO:0000269	PubMed:25691535}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Fokal, Konuşma Bozukluğu ve ± Mental Retardasyon 245570, Otozomal Dominant; FESD (Rolandik Epilepsi-Konuşma Dispraksi Sendromu) (GRIN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570, Autosomal dominant; FESD (Rolandic epilepsy-speech dyspraxia syndrome) (GRIN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIN2A	GRIN2A, NMDAR2A, FESD, LKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, focal, with speech disorder and with or without mental retardation (FESD) [MIM:245570]: A highly variable neurologic disorder with features ranging from severe early-onset seizures associated with delayed psychomotor development, persistent speech difficulties, and mental retardation to a more benign entity characterized by childhood onset of mild or asymptomatic seizures associated with transient speech difficulties followed by remission of seizures in adolescence and normal psychomotor development. The disorder encompasses several clinical entities, including Landau-Kleffner syndrome, epileptic encephalopathy with continuous spike and wave during slow-wave sleep, autosomal dominant rolandic epilepsy, mental retardation and speech dyspraxia, and benign epilepsy with centrotemporal spikes. {ECO:0000269|PubMed:20890276, ECO:0000269|PubMed:23033978, ECO:0000269|PubMed:23933818, ECO:0000269|PubMed:23933819, ECO:0000269|PubMed:23933820, ECO:0000269|PubMed:24504326, ECO:0000269|PubMed:24903190}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Fokal, Konuşma Bozukluğu ve ± Mental Retardasyon 245570, Otozomal Dominant; FESD (Rolandik Epilepsi-Konuşma Dispraksi Sendromu) (Prenatal) (GRIN2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, focal, with speech disorder and with or without mental retardation, 245570, Autosomal dominant; FESD (Rolandic epilepsy-speech dyspraxia syndrome) (GRIN2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIN2A	GRIN2A, NMDAR2A, FESD, LKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, focal, with speech disorder and with or without mental retardation (FESD) [MIM:245570]: A highly variable neurologic disorder with features ranging from severe early-onset seizures associated with delayed psychomotor development, persistent speech difficulties, and mental retardation to a more benign entity characterized by childhood onset of mild or asymptomatic seizures associated with transient speech difficulties followed by remission of seizures in adolescence and normal psychomotor development. The disorder encompasses several clinical entities, including Landau-Kleffner syndrome, epileptic encephalopathy with continuous spike and wave during slow-wave sleep, autosomal dominant rolandic epilepsy, mental retardation and speech dyspraxia, and benign epilepsy with centrotemporal spikes. {ECO:0000269|PubMed:20890276, ECO:0000269|PubMed:23033978, ECO:0000269|PubMed:23933818, ECO:0000269|PubMed:23933819, ECO:0000269|PubMed:23933820, ECO:0000269|PubMed:24504326, ECO:0000269|PubMed:24903190}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Jeneralize, Ateşli (Febril) Nöbet, Tip 1, 604233, Otozomal Dominant (Jeneralize Epilepsi-Febril Nöbetler Artı) (SCN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, generalized, with febrile seizures plus, type 1, 604233, Autosomal dominant (Generalized epilepsy with febrile seizures-plus) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEE52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Epilepsi, Jeneralize, Ateşli (Febril) Nöbet, Tip 2, 604403, Otozomal Dominant (Jeneralize Epilepsi-Febril Nöbetler Arlı) (SCN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, generalized, with febrile seizures plus, type 2, 604403, Autosomal dominant (Generalized epilepsy with febrile seizures-plus) (SCN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18586737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117757	EDTA Blood Sample
Epilepsi, Jeneralize, Ateşli (Febril) Nöbet, Tip 2, 604403, Otozomal Dominant (Jeneralize Epilepsi-Febril Nöbetler Arlı) (MLPA)	Epilepsy, generalized, with febrile seizures plus, type 2, 604403, Autosomal dominant (Generalized epilepsy with febrile seizures-plus) (MLPA)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18586737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117757	EDTA Blood Sample
Epilepsi, Jeneralize, Ateşli (Febril) Nöbet, Tip 3, 611277, Otozomal Dominant (Jeneralize Epilepsi-Febril Nöbetler Arlı) (GABRG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, generalized, with febrile seizures plus, type 3, 611277, Autosomal dominant (Generalized epilepsy with febrile seizures-plus) (GABRG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GABRG2	GABRG2, GEFSP3, CAE2, ECA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Epilepsi, Jeneralize, Ateşli (Febril) Nöbet, Tip 5, Yatkınlık, 613060, Otozomal Dominant (Jüvenil Myoklonik Epilepsi) (GABRD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, generalized, with febrile seizures plus, type 5, susceptibility to, 613060, Autosomal dominant (Juvenile myoclonic epilepsy) (GABRD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRD	GABRD, GEFSP5, EIG10, EJM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 5 (GEFS+5) [MIM:613060]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:15115768}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Jeneralize, Ateşli (Febril) Nöbetlerle Birlikte, Tip 7, 613863, Otozomal Dominant (Jeneralize Epilepsi-Febril Nöbetler Arlı) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, generalized, with febrile seizures plus, type 7, 613863, Autosomal dominant (Generalized epilepsy with febrile seizures-plus) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, İşitme Kaybı ve Mental Retardasyon Sendromu, 616577, Otozomal Resesif; EHLMRS (Mikrosefali-Mental Retardasyon-İşitme Kaybı-Epilepsi-Anormal Kas Tonusu Sendromu) (SPATA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, hearing loss, and mental retardation syndrome, 616577, Autosomal recessive; EHLMRS (Microcephaly-Intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome) (SPATA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPATA5	SPATA5, SPAF, EHLMRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, İşitme Kaybı ve Mental Retardasyon Sendromu, 616577, Otozomal Resesif; EHLMRS (Mikrosefali-Mental Retardasyon-İşitme Kaybı-Epilepsi-Anormal Kas Tonusu Sendromu) (Prenatal) (SPATA5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, hearing loss, and mental retardation syndrome, 616577, Autosomal recessive; EHLMRS (Microcephaly-intellectual disability-sensorineural hearing loss-epilepsy-abnormal muscle tone syndrome) (SPATA5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPATA5	SPATA5, SPAF, EHLMRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, 10, 613060, Otozomal Dominant; EIG10 (Jeneralize Epilepsi-Febril Nöbetler Arlı) (GABRD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, idiopathic generalized, 10, 613060, Autosomal dominant; EIG10 (Generalized epilepsy with febrile seizures-plus) (GABRD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRD	GABRD, GEFSP5, EIG10, EJM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 5 (GEFS+5) [MIM:613060]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:15115768}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, Yatkınlık, 11, 607628, Otozomal Dominant; EIG11 (Jüvenil Myoklonik Epilepsi) (CLCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, idiopathic generalized, susceptibility to, 11, 607628, Autosomal dominant; EIG11 (Juvenile myoclonic epilepsy) (CLCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN2	CLCN2, EGMA, ECA2, EGI11, EJM8, LKPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, idiopathic generalized 11 (EIG11) [MIM:607628]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269|PubMed:19191339}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, Yatkınlık, 12, 614847, Otozomal Dominant (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, idiopathic generalized, susceptibility to, 12, 614847, Autosomal dominant (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, Yatkınlık, 12, 614847, Otozomal Dominant (MLPA)	Epilepsy, idiopathic generalized, susceptibility to, 12, 614847, Autosomal dominant (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, Yatkınlık, 14, 616685, Otozomal Dominant (SLC12A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, idiopathic generalized, susceptibility to, 14, 616685, Autosomal dominant (SLC12A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC12A5	SLC12A5, KCC2, KIAA1176, EIEE34, EIG14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, İdyopatik Jeneralize, Yatkınlık, 9, 607682, Otozomal Dominant; EIG9 (Jüvenil Miyoklonik Epilepsi) (CACNB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, idiopathic generalized, susceptibility to, 9, 607682, Autosomal dominant; EIG9 (Juvenile myoclonic epilepsy) (CACNB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNB4	CACNB4, EJM6, EA5, EIG9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Juvenile myoclonic epilepsy 6 (EJM6) [MIM:607682]: A subtype of idiopathic generalized epilepsy. Patients have afebrile seizures only, with onset in adolescence (rather than in childhood) and myoclonic jerks which usually occur after awakening and are triggered by sleep deprivation and fatigue. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Jüvenil Absans Tipi, Yatkınlık, 1, 607631, Otozomal (Jüvenil Absans Epilepsi) (EFHC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, juvenile absence, susceptibility to, 1, 607631, Autosomal dominant; EJA1 (Juvenile absence epilepsy) (EFHC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFHC1	EFHC1, FLJ10466, EJM1, JAE, EJA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Juvenile absence epilepsy 1 (JAE1) [MIM:607631]: A subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures. {ECO:0000305|PubMed:17159113}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Jüvenil Absans Tipi, Yatkınlık, 2, 607628, Otozomal Dominant (Jüvenil Miyoklonik Epilepsi) (CLCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, juvenile absence, susceptibility to, 2, 607628, Autosomal dominant (Juvenile myoclonic epilepsy) (CLCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN2	CLCN2, EGMA, ECA2, EGI11, EJM8, LKPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, idiopathic generalized 11 (EIG11) [MIM:607628]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269|PubMed:19191339}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Jüvenil Miyoklonik, Yatkınlık, 5, 611136 (Jüvenil Miyoklonik Epilepsi) (GABRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, juvenile myoclonic, susceptibility to, 5, 611136 (Juvenile myoclonic epilepsy) (GABRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRA1	GABRA1, EJM5, ECA4, EIEE19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, childhood absence 4 (ECA4) [MIM:611136]: A subtype of idiopathic generalized epilepsy characterized by an onset at age 6-7 years, frequent absence seizures (several per day) and bilateral, synchronous, symmetric 3-Hz spike waves on EEG. Tonic-clonic seizures often develop in adolescence. Absence seizures may either remit or persist into adulthood. {ECO:0000269|PubMed:16738694}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Jüvenil Miyoklonik, Yatkınlık, 613060, Otozomal Dominant (Jüvenil Miyoklonik Epilepsi) (GABRD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, juvenile myoclonic, susceptibility to, 613060, Autosomal dominant (Juvenile myoclonic epilepsy) (GABRD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRD	GABRD, GEFSP5, EIG10, EJM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 5 (GEFS+5) [MIM:613060]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:15115768}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

Epilepsi, Jüvenil Miyoklonik, Yatkınlık, 8, 607628, Otozomal Dominant (Jüvenil Miyoklonik Epilepsi) (CLCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, juvenile myoclonic, susceptibility to, 8, 607628, Autosomal dominant (Juvenile myoclonic epilepsy) (CLCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN2	CLCN2, EGMA, ECA2, EGI11, EJM8, LKPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, idiopathic generalized 11 (EIG11) [MIM:607628]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269	PubMed:19191339}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Miyoklonik, Allesel Erişkin, 2, 607876, Otozomal Dominant; FAME2 (Benign Yetişkin Allesel Miyoklonik Epilepsi) (ADRA2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, myoclonic, familial adult, 2, 607876, Autosomal dominant; FAME2 (Benign adult familial myoclonic epilepsy) (ADRA2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRA2B	ADRA2B, ADRA2L1, FAME2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, familial adult myoclonic, 2 (FAME2) [MIM:607876]: A form of cortical myoclonic tremor with epilepsy, a syndrome characterized by cortical myoclonus and variable occurrence of epileptic seizures. Usually, myoclonic tremor is the presenting symptom, characterized by tremulous finger movements and myoclonic jerks of the limbs increased by action and posture. In a minority of patients, seizures are the presenting symptom; both complex partial as well as generalized tonic clonic seizures are described. Some patients exhibit mild cognitive impairment. {ECO:0000269	PubMed:24114805}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Miyoklonik, Allesel Yetişkin, 5, 615400, Otozomal Resesif; FAME5 (Benign Yetişkin Allesel Miyoklonik Epilepsi) (CNTN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, myoclonic, familial adult, 5, 615400, Autosomal recessive; FAME5 (Benign adult familial myoclonic epilepsy) (CNTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTN2	CNTN2, TAX, TAX1, FAME5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, familial adult myoclonic, 5 (FAME5) [MIM:615400]: A form of cortical myoclonic tremor with epilepsy, a syndrome characterized by cortical myoclonus and variable occurrence of epileptic seizures. Usually, myoclonic tremor is the presenting symptom, characterized by tremulous finger movements and myoclonic jerks of the limbs increased by action and posture. In a minority of patients, seizures are the presenting symptom; both complex partial as well as generalized tonic clonic seizures are described. Some patients exhibit mild cognitive impairment. {ECO:0000269	PubMed:23518707}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Noktumal Frontal Lob, 1, 600513, Otozomal Dominant; ENFL1 (Otozomal Dominant Noktürnal Frontal Lob Epilepsi) (CHRNA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, nocturnal frontal lobe, 1, 600513, Autosomal dominant; ENFL1 (Autosomal dominant nocturnal frontal lobe epilepsy) (CHRNA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA4	CHRNA4, ENFL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, nocturnal frontal lobe, 1 (ENFL1) [MIM:600513]: An autosomal dominant focal epilepsy characterized by nocturnal seizures with hyperkinetic automatisms and poorly organized stereotyped movements. {ECO:0000269|PubMed:10583623, ECO:0000269|PubMed:14623738, ECO:0000269|PubMed:7550350}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Noktumal Frontal Lob, 3, 605375, ENFL3 (Otozomal Dominant Noktürnal Frontal Lob Epilepsi) (CHRNB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, nocturnal frontal lobe, 3, 605375; ENFL3 (Autosomal dominant nocturnal frontal lobe epilepsy) (CHRNB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNB2	CHRNB2, EFNL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, nocturnal frontal lobe, 3 (ENFL3) [MIM:605375]: An autosomal dominant focal epilepsy characterized by nocturnal seizures with hyperkinetic automatisms and poorly organized stereotyped movements. {ECO:0000269|PubMed:11062464, ECO:0000269|PubMed:11044662}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Noktumal Frontal Lob, 5, 615005, Otozomal Dominant; ENFL5 (Otozomal Dominant Noktürnal Frontal Lob Epilepsi) (KCNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, nocturnal frontal lobe, 5, 615005, Autosomal dominant; ENFL5 (Autosomal dominant nocturnal frontal lobe epilepsy) (KCNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNT1	KCNT1, KIAA1422, EIEE14, ENFL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Noktumal Frontal Lob, Tip 4, 610353, Otozomal Dominant; ENFL4 (Otozomal Dominant Noktürnal Frontal Lob Epilepsi) (CHRNA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, nocturnal frontal lobe, type 4, 610353, Autosomal dominant; ENFL4 (Autosomal dominant nocturnal frontal lobe epilepsy) (CHRNA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA2	CHRNA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, nocturnal frontal lobe, 4 (ENFL4) [MIM:610353]: An autosomal dominant focal epilepsy characterized by nocturnal seizures associated with fear sensation, tongue movements, and nocturnal wandering, closely resembling nightmares and sleep walking. {ECO:0000269	PubMed:16826524}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 1B, 612437, Otozomal Resesif; EPM1B (Unverricht-Lundborg Hastalığı) (PRICKLE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 1B, 612437, Autosomal recessive; EPM1B (Unverricht-Lundborg disease) (PRICKLE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRICKLE1	PRICKLE1, RILP, EPM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 1B, 612437, Otozomal Resesif; EPM1B (Unverricht-Lundborg Hastalığı) (Prenatal) (PRICKLE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 1B, 612437, Autosomal recessive; EPM1B (Unverricht-Lundborg disease) (PRICKLE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRICKLE1	PRICKLE1, RILP, EPM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 2A (Lafora), 254780, Otozomal Resesif (Lafora Hastalığı) (EPM2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 2A (Lafora), 254780, Autosomal recessive (Lafora disease) (EPM2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPM2A	EPM2A, MELF, EPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 2A (Lafora), 254780, Otozomal Resesif (Lafora Hastalığı) (Prenatal) (EPM2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 2A (Lafora), 254780, Autosomal recessive (Lafora disease) (EPM2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPM2A	EPM2A, MELF, EPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 2B (Lafora), 254780, Otozomal Resesif (Lafora Hastalığı) (NHLRC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 2B (Lafora), 254780, Autosomal recessive (Lafora disease) (NHLRC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NHLRC1	NHLRC1, EPM2A, EPM2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 2 (EPM2) [MIM:254780]: An autosomal recessive and severe form of adolescent-onset progressive epilepsy. Typically, as seizures increase in frequency, cognitive function declines towards dementia, and affected individuals die usually within 10 years after onset. EPM2 occurs worldwide, but it is particularly common in the mediterranean countries of southern Europe and northern Africa, in southern India and in the Middle East. At the cellular level, it is characterized by accumulation of starch-like polyglucosans called Lafora bodies (LBs) that are most abundant in organs with the highest glucose metabolism: brain, heart, liver and skeletal muscle. Among other conditions involving polyglucosans, EPM2 is unique in that the inclusions are in neuronal dendrites but not axons and the forming polyglucosan fibrils are associated with the endoplasmic reticulum. {ECO:0000269|PubMed:12958597, ECO:0000269|PubMed:15781812, ECO:0000269|PubMed:16021330, ECO:0000269|PubMed:18311786, ECO:0000269|PubMed:21505799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 2B (Lafora), 254780, Otozomal Resesif (Lafora Hastalığı) (Prenatal) (NHLRC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 2B (Lafora), 254780, Autosomal recessive (Lafora disease) (NHLRC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NHLRC1	NHLRC1, EPM2A, EPM2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 2 (EPM2) [MIM:254780]: An autosomal recessive and severe form of adolescent-onset progressive epilepsy. Typically, as seizures increase in frequency, cognitive function declines towards dementia, and affected individuals die usually within 10 years after onset. EPM2 occurs worldwide, but it is particularly common in the mediterranean countries of southern Europe and northern Africa, in southern India and in the Middle East. At the cellular level, it is characterized by accumulation of starch-like polyglucosans called Lafora bodies (LBs) that are most abundant in organs with the highest glucose metabolism: brain, heart, liver and skeletal muscle. Among other conditions involving polyglucosans, EPM2 is unique in that the inclusions are in neuronal dendrites but not axons and the forming polyglucosan fibrils are associated with the endoplasmic reticulum. {ECO:0000269|PubMed:12958597, ECO:0000269|PubMed:15781812, ECO:0000269|PubMed:16021330, ECO:0000269|PubMed:18311786, ECO:0000269|PubMed:21505799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 3, ± Hücre İçi İnklüzyonlar, 611726, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 3) (KCTD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726, Autosomal recessive (Progressive myoclonic epilepsy type 3) (KCTD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCTD7	KCTD7, EPM3, CLN14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 3, with or without intracellular inclusions (EPM3) [MIM:611726]: An autosomal recessive, severe, progressive myoclonic epilepsy with early onset. Multifocal myoclonic seizures begin between 16 and 24 months of age after normal initial development. Neurodegeneration and regression occur with seizure onset. Other features include mental retardation, dysarthria, truncal ataxia, and loss of fine finger movements. EEG shows slow dysrhythmia, multifocal and occasionally generalized epileptiform discharges. In some patients, ultrastructural findings on skin biopsies identify intracellular accumulation of autofluorescent lipopigment storage material, consistent with neuronal ceroid lipofuscinosis. {ECO:0000269|PubMed:17455289, ECO:0000269|PubMed:22606975, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:22693283, ECO:0000269|PubMed:22748208}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 3, ± Hücre İçi İnklüzyonlar, 611726, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 3) (Prenatal) (KCTD7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 3, with or without intracellular inclusions, 611726, Autosomal recessive (Progressive myoclonic epilepsy type 3) (KCTD7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCTD7	KCTD7, EPM3, CLN14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 3, with or without intracellular inclusions (EPM3) [MIM:611726]: An autosomal recessive, severe, progressive myoclonic epilepsy with early onset. Multifocal myoclonic seizures begin between 16 and 24 months of age after normal initial development. Neurodegeneration and regression occur with seizure onset. Other features include mental retardation, dysarthria, truncal ataxia, and loss of fine finger movements. EEG shows slow dysrhythmia, multifocal and occasionally generalized epileptiform discharges. In some patients, ultrastructural findings on skin biopsies identify intracellular accumulation of autofluorescent lipopigment storage material, consistent with neuronal ceroid lipofuscinosis. {ECO:0000269|PubMed:17455289, ECO:0000269|PubMed:22606975, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:22693283, ECO:0000269|PubMed:22748208}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 4, ± Böbrek Yetmezliği, 254900, Otozomal Resesif (Aksiyon Miyoklonusu-Böbrek Yetmezliği Sendromu) (SCARB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900, Autosomal recessive (Action myoclonus-renal failure syndrome) (SCARB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCARB2	SCARB2, CD36L2, LIMPII, AMRF, EPM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 4, with or without renal failure (EPM4) [MIM:254900]: An autosomal recessive progressive myoclonic epilepsy associated with renal failure in some cases. Cognitive function is preserved. Myoclonus is a brief, involuntary twitching of a muscle or a group of muscles. {ECO:0000269|PubMed:18308289, ECO:0000269|PubMed:18424452, ECO:0000269|PubMed:19454373, ECO:0000269|PubMed:21670406}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Epilepsi, Progresif Miyoklonik 4, ± Böbrek Yetmezliği, 254900, Otozomal Resesif (Aksiyon Miyoklonusu-Böbrek Yetmezliği Sendromu) (Prenatal) (SCARB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 4, with or without renal failure, 254900, Autosomal recessive (Action myoclonus-renal failure syndrome) (SCARB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCARB2	SCARB2, CD36L2, LIMPII, AMRF, EPM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 4, with or without renal failure (EPM4) [MIM:254900]: An autosomal recessive progressive myoclonic epilepsy associated with renal failure in some cases. Cognitive function is preserved. Myoclonus is a brief, involuntary twitching of a muscle or a group of muscles. {ECO:0000269|PubMed:18309289, ECO:0000269|PubMed:18424452, ECO:0000269|PubMed:19454373, ECO:0000269|PubMed:21670406}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 6, 614018, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 6) (GOSR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 6, 614018, Autosomal recessive (Progressive myoclonic epilepsy type 6) (GOSR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GOSR2	GOSR2, GS27, EPM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 6 (EPM6) [MIM:614018]: A neurologic disorder characterized by onset of ataxia in the first years of life, followed by action myoclonus and seizures later in childhood, and loss of independent ambulation in the second decade. Cognition is not usually affected, although mild memory difficulties may occur in the third decade. {ECO:0000269	PubMed:21549339}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 6, 614018, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 6) (Prenatal) (GOSR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 6, 614018, Autosomal recessive (Progressive myoclonic epilepsy type 6) (GOSR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GOSR2	GOSR2, GS27, EPM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 6 (EPM6) [MIM:614018]: A neurologic disorder characterized by onset of ataxia in the first years of life, followed by action myoclonus and seizures later in childhood, and loss of independent ambulation in the second decade. Cognition is not usually affected, although mild memory difficulties may occur in the third decade. {ECO:0000269	PubMed:21549339}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 7, 616187, Otozomal Dominant; EPM7 (Progresif Miyoklonik Epilepsi Tip 7) (KCNC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic 7, 616187, Autosomal dominant; EPM7 (Progressive myoclonic epilepsy type 7) (KCNC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNC1	KCNC1, EPM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 7 (EPM7) [MIM:616187]: A neurologic disorder characterized by progressive myoclonic epilepsy, manifesting in the first or second decades of life. Cognitive function may decline in some patients. Myoclonus is a brief, involuntary twitching of a muscle or a group of muscles. {ECO:0000269	PubMed:25401298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik 7, 616187, Otozomal Dominant; EPM7 (Progresif Miyoklonik Epilepsi Tip 7) (Prenatal) (KCNC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic 7, 616187, Autosomal dominant; EPM7 (Progressive myoclonic epilepsy type 7) (KCNC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNC1	KCNC1, EPM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 7 (EPM7) [MIM:616187]: A neurologic disorder characterized by progressive myoclonic epilepsy, manifesting in the first or second decades of life. Cognitive function may decline in some patients. Myoclonus is a brief, involuntary twitching of a muscle or a group of muscles. {ECO:0000269	PubMed:25401298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 10, 616640, Otozomal Resesif (Erken Başlangıçlı Lafora Cisimciği Hastalığı) (PRDM8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic, 10, 616640, Autosomal recessive (Early-onset Lafora body disease) (PRDM8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM8	PRDM8, EPM10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 10, 616640, Otozomal Resesif (Erken Başlangıçlı Lafora Cisimciği Hastalığı) (Prenatal) (PRDM8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic, 10, 616640, Autosomal recessive (Early-onset Lafora body disease) (PRDM8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRDM8	PRDM8, EPM10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 8, 616230, Otozomal Resesif; EPM8 (Progresif Miyoklonik Epilepsi Tip 8) (CERS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic, 8, 616230, Autosomal recessive; EPM8 (Progressive myoclonic epilepsy type 8) (CERS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CERS1	CERS1, LASS1, UOG1, EPM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, progressive myoclonic 8 (EPM8) [MIM:616230]: A severe form of progressive myoclonic epilepsy characterized by myoclonus, generalized tonic-clonic seizures and moderate to severe cognitive impairment. {ECO:0000269	PubMed:24782409}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 8, 616230, Otozomal Resesif; EPM8 (Progresif Miyoklonik Epilepsi Tip 8) (Prenatal) (CERS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic, 8, 616230, Autosomal recessive; EPM8 (Progressive myoclonic epilepsy type 8) (CERS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CERS1	CERS1, LASS1, UOG1, EPM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, progressive myoclonic 8 (EPM8) [MIM:616230]: A severe form of progressive myoclonic epilepsy characterized by myoclonus, generalized tonic-clonic seizures and moderate to severe cognitive impairment. {ECO:0000269	PubMed:24782409}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 9, 616540, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 9) (LMNB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, progressive myoclonic, 9, 616540, Autosomal recessive (Progressive myoclonic epilepsy type 9) (LMNB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNB2	LMNB2, LMN2, EPM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, Progresif Miyoklonik, 9, 616540, Otozomal Resesif (Progresif Miyoklonik Epilepsi Tip 9) (Prenatal) (LMNB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, progressive myoclonic, 9, 616540, Autosomal recessive (Progressive myoclonic epilepsy type 9) (LMNB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMNB2	LMNB2, LMN2, EPM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epilepsi, Piridoksin Bağımlı, 266100, Otozomal Resesif; EPD (Piridoksin Bağımlı Epilepsi) (ALDH7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, pyridoxine-dependent, 266100, Autosomal recessive; EPD (Pyridoxine-dependent epilepsy) (ALDH7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH7A1	ALDH7A1, ATQ1, EPD, PDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epilepsi, X'e Bağlı, Değişken Öğrenme Zorluğu ve Davranış Bozukluğu Olan, 300491, X'e Bağlı, X'e Bağlı Dominant (X'e Bağlı Epilepsi-Mental Retardasyon-Davranış Bozuklukları Sendromu) (SYN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491, X-linked recessive, X-linked dominant (X-linked epilepsy-learning disabilities-behavior disorders syndrome) (SYN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYN1	SYN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy X-linked, with variable learning disabilities and behavior disorders (XELBD) [MIM:300491]: A neurologic disorder characterized by variable combinations of epilepsy, learning difficulties, macrocephaly, and aggressive behavior. {ECO:0000269	PubMed:14985377}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epilepsi, X'e Bağlı, Değişken Öğrenme Zorluğu ve Davranış Bozukluğu Olan, 300491, X'e Bağlı, X'e Bağlı Dominant (X'e Bağlı Epilepsi-Mental Retardasyon-Davranış Bozuklukları Sendromu) (Prenatal) (SYN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491, X-linked recessive, X-linked dominant (X-linked epilepsy-learning disabilities-behavior disorders syndrome) (SYN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYN1	SYN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy X-linked, with variable learning disabilities and behavior disorders (XELBD) [MIM:300491]: A neurologic disorder characterized by variable combinations of epilepsy, learning difficulties, macrocephaly, and aggressive behavior. {ECO:0000269	PubMed:14985377}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Çocuklukta Başlayan, 615369, Otozomal Dominant; EEOC (Lennox-Gastaut Sendromu) (CHD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, childhood-onset, 615369, Autosomal dominant; EEOC (Lennox-Gastaut syndrome) (CHD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHD2	CHD2, EEOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, childhood-onset (EEOC) [MIM:615369]: A severe form of epilepsy characterized by onset of multiple seizure types in the first few years of life and associated with poor prognosis. Affected individuals have cognitive regression and intellectual disability. {ECO:0000269	PubMed:23708187}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Çocuklukta Başlayan, 615369, Otozomal Dominant; EEOC (Lennox-Gastaut Sendromu) (Prenatal) (CHD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, childhood-onset, 615369, Autosomal dominant; EEOC (Lennox-Gastaut syndrome) (CHD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHD2	CHD2, EEOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, childhood-onset (EEOC) [MIM:615369]: A severe form of epilepsy characterized by onset of multiple seizure types in the first few years of life and associated with poor prognosis. Affected individuals have cognitive regression and intellectual disability. {ECO:0000269	PubMed:23708187}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 1, 308350, X'e Bağlı Resesif; EIEE1 (Erken İnfantil Epileptik Ensefalopati) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 1, 308350, X-linked recessive; EIEE1 (Early infantile epileptic encephalopathy) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 1, 308350, X'e Bağlı Resesif; EIEE1 (Erken İnfantil Epileptik Ensefalopati) (MLPA)	Epileptic encephalopathy, early infantile, 1, 308350, X-linked recessive; EIEE1 (Early infantile epileptic encephalopathy) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 1, 308350, X'e Bağlı Resesif; EIEE1 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 1, 308350, X-linked recessive; EIEE1 (Early infantile epileptic encephalopathy) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 1, 308350, X'e Bağlı Resesif; EIEE1 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 1, 308350, X-linked recessive; EIEE1 (Early infantile epileptic encephalopathy) (Prenatal) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 11, 613721, Otozomal Dominant; EIEE11 (Erken İnfantil Epileptik Ensefalopati) (SCN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 11, 613721, Autosomal dominant; EIEE11 (Early infantile epileptic encephalopathy) (SCN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN2A	SCN2A, SCN2A1, BFIC3, EIEE11, BFIS3, BFNIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 11, 613721, Otozomal Dominant; EIEE11 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (SCN2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 11, 613721, Autosomal dominant; EIEE11 (Early infantile epileptic encephalopathy) (SCN2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN2A	SCN2A, SCN2A1, BFIC3, EIEE11, BFIS3, BFNIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 12, 613722, Otozomal Resesif; EIEE12 (West Sendromu) (PLCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 12, 613722, Autosomal recessive; EIEE12 (West syndrome) (PLCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCB1	PLCB1, KIAA0581, PLCB1A, PLCB1B, EIEE12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 12 (EIEE12) [MIM:613722]: A form of epilepsy characterized by frequent tonic seizures or spasms beginning in infancy with a specific EEG finding of suppression-burst patterns, characterized by high-voltage bursts alternating with almost flat suppression phases. Patients may progress to West syndrome, which is characterized by tonic spasms with clustering, arrest of psychomotor development, and hypsarrhythmia on EEG. {ECO:0000269	PubMed:20833646}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 12, 613722, Otozomal Resesif; EIEE12 (West Sendromu) (Prenatal) (PLCB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 12, 613722, Autosomal recessive; EIEE12 (West syndrome) (PLCB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLCB1	PLCB1, KIAA0581, PLCB1A, PLCB1B, EIEE12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 12 (EIEE12) [MIM:613722]: A form of epilepsy characterized by frequent tonic seizures or spasms beginning in infancy with a specific EEG finding of suppression-burst patterns, characterized by high-voltage bursts alternating with almost flat suppression phases. Patients may progress to West syndrome, which is characterized by tonic spasms with clustering, arrest of psychomotor development, and hypsarrhythmia on EEG. {ECO:0000269	PubMed:20833646}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 13, 614558, Otozomal Dominant; EIEE13 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (SCN8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 13, 614558, Autosomal dominant; EIEE13 (Undetermined early-onset epileptic encephalopathy) (SCN8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN8A	SCN8A, CIAT, EIEE13, BFIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 13, 614558, Otozomal Dominant; EIEE13 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (SCN8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 13, 614558, Autosomal dominant; EIEE13 (Undetermined early-onset epileptic encephalopathy) (SCN8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN8A	SCN8A, CIAT, EIEE13, BFIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epileptik Ensefalopati, Erken İnfantil, 14, 614959, Otozomal Dominant; EIEE14 (İnfantil Malign Göç Eden Parsiyel Nöbetler) (KCNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 14, 614959, Autosomal dominant; EIEE14 (Malignant migrating partial seizures of infancy) (KCNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNT1	KCNT1, KIAA1422, EIEE14, ENFL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 14 (EIEE14) [MIM:614959]: A rare epileptic encephalopathy of infancy that combines pharmacoresistant seizures with developmental delay. This severe neurologic disorder is characterized by onset in the first 6 months of life of refractory focal seizures and arrest of psychomotor development. ictal EEG shows discharges that arise randomly from various areas of both hemispheres and migrate from one brain region to another. {ECO:0000269|PubMed:23086397}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 14, 614959, Otozomal Dominant; EIEE14 (İnfantil Malign Göç Eden Parsiyel Nöbetler) (Prenatal) (KCNT1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 14, 614959, Autosomal dominant; EIEE14 (Malignant migrating partial seizures of infancy) (KCNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNT1	KCNT1, KIAA1422, EIEE14, ENFL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 14 (EIEE14) [MIM:614959]: A rare epileptic encephalopathy of infancy that combines pharmacoresistant seizures with developmental delay. This severe neurologic disorder is characterized by onset in the first 6 months of life of refractory focal seizures and arrest of psychomotor development. ictal EEG shows discharges that arise randomly from various areas of both hemispheres and migrate from one brain region to another. {ECO:0000269|PubMed:23086397}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 15, 615006, Otozomal Resesif; EIEE15 (West Sendromu) (ST3GAL3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 15, 615006, Autosomal recessive; EIEE15 (West syndrome) (ST3GAL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ST3GAL3	ST3GAL3, SIAT6, ST3GALII, MRT12, EIEE15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 15, 615006, Otozomal Resesif; EIEE15 (West Sendromu) (Prenatal) (ST3GAL3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 15, 615006, Autosomal recessive; EIEE15 (West syndrome) (ST3GAL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ST3GAL3	ST3GAL3, SIAT6, ST3GALII, MRT12, EIEE15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 16, 615338, Otozomal Resesif; EIEE16 (Progresif Miyoklonik Epilepsi-Distoni) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 16, 615338, Autosomal recessive; EIEE16 (Progressive myoclonic epilepsy with dystonia) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 16, 615338, Otozomal Resesif; EIEE16 (Progresif Miyoklonik Epilepsi-Distoni) (Prenatal) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 16, 615338, Autosomal recessive; EIEE16 (Progressive myoclonic epilepsy with dystonia) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 17, 615473, Otozomal Dominant; EIEE17 (Erken İnfantil Epileptik Ensefalopati) (GNAO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 17, 615473, Autosomal dominant; EIEE17 (Early infantile epileptic encephalopathy) (GNAO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAO1	GNAO1, EIEE17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 17 (EIEE17) [MIM:615473]: A severe neurologic disorder characterized by onset of intractable seizures in the first weeks or months of life and usually associated with EEG abnormalities. Affected infants have very poor psychomotor development and may have brain abnormalities, such as cerebral atrophy or thin corpus callosum. Some patients may show involuntary movements. {ECO:0000269|PubMed:23993195}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 17, 615473, Otozomal Dominant; EIEE17 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (GNAO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 17, 615473, Autosomal dominant; EIEE17 (Early infantile epileptic encephalopathy) (GNAO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNAO1	GNAO1, EIEE17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 17 (EIEE17) [MIM:615473]: A severe neurologic disorder characterized by onset of intractable seizures in the first weeks or months of life and usually associated with EEG abnormalities. Affected infants have very poor psychomotor development and may have brain abnormalities, such as cerebral atrophy or thin corpus callosum. Some patients may show involuntary movements. {ECO:0000269|PubMed:23993195}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 18, 615476, Otozomal Resesif; EIEE18 (Erken İnfantil Epileptik Ensefalopati-Supresyon Burst Olmadan) (SZT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 18, 615476, Autosomal recessive; EIEE18 (Early infantile epileptic encephalopathy without suppression burst) (SZT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SZT2	SZT2, KIAA0467, EIEE18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 18, 615476, Otozomal Resesif; EIEE18 (Erken İnfantil Epileptik Ensefalopati-Supresyon Burst Olmadan) (Prenatal) (SZT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 18, 615476, Autosomal recessive; EIEE18 (Early infantile epileptic encephalopathy without suppression burst) (SZT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SZT2	SZT2, KIAA0467, EIEE18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 19, 615744, Otozomal Dominant; EIEE19 (Dravet Syndrome) (GABRA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 19, 615744, Autosomal dominant; EIEE19 (Dravet syndrome) (GABRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRA1	GABRA1, EJM5, ECA4, EIEE19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 19, 615744, Otozomal Dominant; EIEE19 (Dravet Syndrome) (Prenatal) (GABRA1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 19, 615744, Autosomal dominant; EIEE19 (Dravet syndrome) (GABRA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GABRA1	GABRA1, EJM5, ECA4, EIEE19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 2, 300672, X'e Bağlı Dominant; EIEE2 (Erken İnfantil Epileptik Ensefalopati) (CDKL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 2, 300672, X-linked dominant; EIEE2 (Early infantile epileptic encephalopathy) (CDKL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKL5	CDKL5, STK9, ISSX, EIEE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 2 (EIEE2) [MIM:300672]: A severe form of epilepsy characterized by seizures or spasms beginning in infancy. Patients with epileptic encephalopathy early infantile type 2 manifest features resembling Rett syndrome such as microcephaly, lack of speech development, stereotypic hand movements. However, EIEE2 and Rett syndrome are considered two distinct entities. {ECO:0000269|PubMed:12736870, ECO:0000269|PubMed:15492925, ECO:0000269|PubMed:15499549, ECO:0000269|PubMed:15689447, ECO:0000269|PubMed:15917271, ECO:0000269|PubMed:16015284, ECO:0000269|PubMed:16611748, ECO:0000269|PubMed:17993579, ECO:0000269|PubMed:18790821, ECO:0000269|PubMed:18809835, ECO:0000269|PubMed:19241098, ECO:0000269|PubMed:19253388, ECO:0000269|PubMed:24564546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 2, 300672, X'e Bağlı Dominant; EIEE2 (Erken İnfantil Epileptik Ensefalopati) (MLPA)	Epileptic encephalopathy, early infantile, 2, 300672, X-linked dominant; EIEE2 (Early infantile epileptic encephalopathy) (MLPA)	CDKL5	CDKL5, STK9, ISSX, EIEE2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 2 (EIEE2) [MIM:300672]: A severe form of epilepsy characterized by seizures or spasms beginning in infancy. Patients with epileptic encephalopathy early infantile type 2 manifest features resembling Rett syndrome such as microcephaly, lack of speech development, stereotypic hand movements. However, EIEE2 and Rett syndrome are considered two distinct entities. {ECO:0000269|PubMed:12736870, ECO:0000269|PubMed:15492925, ECO:0000269|PubMed:15499549, ECO:0000269|PubMed:15689447, ECO:0000269|PubMed:15917271, ECO:0000269|PubMed:16015284, ECO:0000269|PubMed:16611748, ECO:0000269|PubMed:17993579, ECO:0000269|PubMed:18790821, ECO:0000269|PubMed:18809835, ECO:0000269|PubMed:19241098, ECO:0000269|PubMed:19253388, ECO:0000269|PubMed:24564546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 2, 300672, X'e Bağlı Dominant; EIEE2 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (CDKL5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 2, 300672, X-linked dominant; EIEE2 (Early infantile epileptic encephalopathy) (CDKL5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDKL5	CDKL5, STK9, ISSX, EIEE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 2 (EIEE2) [MIM:300672]: A severe form of epilepsy characterized by seizures or spasms beginning in infancy. Patients with epileptic encephalopathy early infantile type 2 manifest features resembling Rett syndrome such as microcephaly, lack of speech development, stereotypic hand movements. However, EIEE2 and Rett syndrome are considered two distinct entities. {ECO:0000269|PubMed:12736870, ECO:0000269|PubMed:15492925, ECO:0000269|PubMed:15499549, ECO:0000269|PubMed:15689447, ECO:0000269|PubMed:15917271, ECO:0000269|PubMed:16015284, ECO:0000269|PubMed:16611748, ECO:0000269|PubMed:17993579, ECO:0000269|PubMed:18790821, ECO:0000269|PubMed:18809835, ECO:0000269|PubMed:19241098, ECO:0000269|PubMed:19253388, ECO:0000269|PubMed:24564546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 2, 300672, X'e Bağlı Dominant; EIEE2 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 2, 300672, X-linked dominant; EIEE2 (Early infantile epileptic encephalopathy) (Prenatal) (MLPA)	CDKL5	CDKL5, STK9, ISSX, EIEE2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 2 (EIEE2) [MIM:300672]: A severe form of epilepsy characterized by seizures or spasms beginning in infancy. Patients with epileptic encephalopathy early infantile type 2 manifest features resembling Rett syndrome such as microcephaly, lack of speech development, stereotypic hand movements. However, EIEE2 and Rett syndrome are considered two distinct entities. {ECO:0000269|PubMed:12736870, ECO:0000269|PubMed:15492925, ECO:0000269|PubMed:15499549, ECO:0000269|PubMed:15689447, ECO:0000269|PubMed:15917271, ECO:0000269|PubMed:16015284, ECO:0000269|PubMed:16611748, ECO:0000269|PubMed:17993579, ECO:0000269|PubMed:18790821, ECO:0000269|PubMed:18809835, ECO:0000269|PubMed:19241098, ECO:0000269|PubMed:19253388, ECO:0000269|PubMed:24564546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 21, 615833, Otozomal Resesif; EIEE21 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (NECAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 21, 615833, Autosomal recessive; EIEE21 (Undetermined early-onset epileptic encephalopathy) (NECAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NECAP1	NECAP1, EIEE21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 21 (EIEE21) [MIM:615833]: A severe disease characterized by intractable seizures, profound global developmental delay, and persistent severe axial hypotonia as well as appendicular hypertonia. {ECO:0000269|PubMed:24399846}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Epileptik Ensefalopati, Erken Infantil, 21, 615833, Otozomal Resesif; EIEE21 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (NECAP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 21, 615833, Autosomal recessive; EIEE21 (Undetermined early-onset epileptic encephalopathy) (NECAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NECAP1	NECAP1, EIEE21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 21 (EIEE21) [MIM:615833]: A severe disease characterized by intractable seizures, profound global developmental delay, and persistent severe axial hypotonia as well as appendicular hypertonia. [ECO:0000269]PubMed:24399846). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 23, 615859, Otozomal Resesif; EIEE23 (Erken Başlangıçlı Epileptik Ensefalopati-Kortikal Körlük-Enteletkiel Yetersizlik-Fasiyal Dismorfizm Sendromu) (DOCK7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 23, 615859, Autosomal recessive; EIEE23 (Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome) (DOCK7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOCK7	DOCK7, KIAA1771, EIEE23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 23 (EIEE23) [MIM:615859]: A severe disease characterized by early-onset intractable epilepsy, dysmorphic features, intellectual disability, and cortical blindness. Brain imaging shows an abnormally marked pontobullar sulcus with mild pontine hypoplasia, white matter abnormalities, and atrophy in the occipital lobe. [ECO:0000269]PubMed:24814191). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 23, 615859, Otozomal Resesif; EIEE23 (Erken Başlangıçlı Epileptik Ensefalopati-Kortikal Körlük-Enteletkiel Yetersizlik-Fasiyal Dismorfizm Sendromu) (Prenatal) (DOCK7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 23, 615859, Autosomal recessive; EIEE23 (Early-onset epileptic encephalopathy-cortical blindness-intellectual disability-facial dysmorphism syndrome) (DOCK7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOCK7	DOCK7, KIAA1771, EIEE23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 23 (EIEE23) [MIM:615859]: A severe disease characterized by early-onset intractable epilepsy, dysmorphic features, intellectual disability, and cortical blindness. Brain imaging shows an abnormally marked pontobullar sulcus with mild pontine hypoplasia, white matter abnormalities, and atrophy in the occipital lobe. [ECO:0000269]PubMed:24814191). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 24, 615871, Otozomal Dominant; EIEE24 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (HCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 24, 615871, Autosomal dominant; EIEE24 (Undetermined early-onset epileptic encephalopathy) (HCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCN1	HCN1, BCNG1, EIEE24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 24 (EIEE24) [MIM:615871]: A disease characterized by early-onset seizures, intellectual disability of varying degrees, and behavioral disturbances or autistic features in most individuals. [ECO:0000269]PubMed:24747641). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 24, 615871, Otozomal Dominant; EIEE24 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (HCN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 24, 615871, Autosomal dominant; EIEE24 (Undetermined early-onset epileptic encephalopathy) (HCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HCN1	HCN1, BCNG1, EIEE24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 24 (EIEE24) [MIM:615871]: A disease characterized by early-onset seizures, intellectual disability of varying degrees, and behavioral disturbances or autistic features in most individuals. [ECO:0000269]PubMed:24747641). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 25, 615905, Otozomal Resesif; EIEE25 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (SLC13A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 25, 615905, Autosomal recessive; EIEE25 (Undetermined early-onset epileptic encephalopathy) (SLC13A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC13A5	SLC13A5, NACT, EIEE25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 25, 615905, Otozomal Resesif; EIEE25 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (SLC13A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 25, 615905, Autosomal recessive; EIEE25 (Undetermined early-onset epileptic encephalopathy) (SLC13A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC13A5	SLC13A5, NACT, EIEE25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 26, 616056, Otozomal Dominant; EIEE26 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (KCNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 26, 616056, Autosomal dominant; EIEE26 (Undetermined early-onset epileptic encephalopathy) (KCNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNB1	KCNB1, EIEE26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 26 (EIEE26) [MIM:616056]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. EIEE26 patients manifest multiple types of seizures, delayed psychomotor development, poor or absent speech, hypotonia, hypsarrhythmia. [ECO:0000269]PubMed:25164438). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 26, 616056, Otozomal Dominant; EIEE26 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (KCNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 26, 616056, Autosomal dominant; EIEE26 (Undetermined early-onset epileptic encephalopathy) (KCNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNB1	KCNB1, EIEE26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 26 (EIEE26) [MIM:616056]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. EIEE26 patients manifest multiple types of seizures, delayed psychomotor development, poor or absent speech, hypotonia, hypsarrhythmia. [ECO:0000269]PubMed:25164438). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 27, 616139, Otozomal Dominant; EIEE27 (West Sendromu) (GRIN2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 27, 616139, Autosomal dominant; EIEE27 (West syndrome) (GRIN2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIN2B	GRIN2B, NMDAR2B, MRD6, EIEE27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 27, 616139, Otozomal Dominant; EIEE27 (West Sendromu) (Prenatal) (GRIN2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 27, 616139, Autosomal dominant; EIEE27 (West syndrome) (GRIN2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIN2B	GRIN2B, NMDAR2B, MRD6, EIEE27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 28, 616211, Otozomal Resesif; EIEE28 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (WWOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 28, 616211, Autosomal recessive; EIEE28 (Undetermined early-onset epileptic encephalopathy) (WWOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WWOX	WWOX, FOR, SCAR12, EIEE28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 28, 616211, Otozomal Resesif; EIEE28 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (WWOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 28, 616211, Autosomal recessive; EIEE28 (Undetermined early-onset epileptic encephalopathy) (WWOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WWOX	WWOX, FOR, SCAR12, EIEE28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 29, 616339, Otozomal Resesif; EIEE29 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (AARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 29, 616339, Autosomal recessive; EIEE29 (Undetermined early-onset epileptic encephalopathy) (AARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AARS	AARS, CMT2N, EIEE29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 29, 616339, Otozomal Resesif; EIEE29 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (AARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 29, 616339, Autosomal recessive; EIEE29 (Undetermined early-onset epileptic encephalopathy) (AARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AARS	AARS, CMT2N, EIEE29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 3, 609304, Otozomal Resesif; EIEE3 (Erken İnfantil Epileptik Ensefalopati) (SLC25A22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 3, 609304, Autosomal recessive; EIEE3 (Early infantile epileptic encephalopathy) (SLC25A22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A22	SLC25A22, GC1, EIEE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 3, 609304, Otozomal Resesif; EIEE3 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (SLC25A22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 3, 609304, Autosomal recessive; EIEE3 (Early infantile epileptic encephalopathy) (SLC25A22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A22	SLC25A22, GC1, EIEE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 30, 616341, Otozomal Dominant; EIEE30 (Erken İnfantil Epileptik Ensefalopati) (SIK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 30, 616341, Autosomal dominant; EIEE30 (Early infantile epileptic encephalopathy) (SIK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIK1	SIK1, SNF1LK, MSK, EIEE30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 30 (EIEE30) [MIM:616341]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25839329). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 30, 616341, Otozomal Dominant; EIEE30 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (SIK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 30, 616341, Autosomal dominant; EIEE30 (Early infantile epileptic encephalopathy) (SIK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIK1	SIK1, SNF1LK, MSK, EIEE30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 30 (EIEE30) [MIM:616341]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25839329). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 31, 616346, Otozomal Dominant; EIEE31 (Lennox-Gastaut Sendromu) (DNM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 31, 616346, Autosomal dominant; EIEE31 (Lennox-Gastaut syndrome) (DNM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM1	DNM1, EIEE31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 31 (EIEE31) [MIM:616346]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25262651, ECO:0000269]PubMed:25533962). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 31, 616346, Otozomal Dominant; EIEE31 (Lennox-Gastaut Sendromu) (Prenatal) (DNM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 31, 616346, Autosomal dominant; EIEE31 (Lennox-Gastaut syndrome) (DNM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNM1	DNM1, EIEE31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 31 (EIEE31) [MIM:616346]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25262651, ECO:0000269]PubMed:25533962). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 32, 616366, Otozomal Dominant; EIEE32 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (KCNA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 32, 616366, Autosomal dominant; EIEE32 (Undetermined early-onset epileptic encephalopathy) (KCNA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNA2	KCNA2, EIEE32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 32 (EIEE32) [MIM:616366]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25471752, ECO:0000269]PubMed:25751627). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 32, 616366, Otozomal Dominant; EIEE32 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (KCNA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 32, 616366, Autosomal dominant; EIEE32 (Undetermined early-onset epileptic encephalopathy) (KCNA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNA2	KCNA2, EIEE32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 32 (EIEE32) [MIM:616366]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. [ECO:0000269]PubMed:25471752, ECO:0000269]PubMed:25751627). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 33, 616409, Otozomal Dominant; EIEE33 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (EEF1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 33, 616409, Autosomal dominant; EIEE33 (Undetermined early-onset epileptic encephalopathy) (EEF1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EEF1A2	EEF1A2, EIEE33, MRD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 33, 616409, Otozomal Dominant; EIEE33 (Bilinlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (EEF1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 33, 616409, Autosomal dominant; EIEE33 (Undetermined early-onset epileptic encephalopathy) (EEF1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EEF1A2	EEF1A2, EIEE33, MRD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epileptik Ensefalopati, Erken İnfantil, 34, 616645, Otozomal Resesif; EIEE34 (İnfantil Malign Göç Eden Parsiyel Nöbetler) (SLC12A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 34, 616645, Autosomal recessive; EIEE34 (Malignant migrating partial seizures of infancy) (SLC12A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC12A5	SLC12A5, KCC2, KIAA1176, EIEE34, EIG14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 34, 616645, Otozomal Resesif; EIEE34 (İnfantil Malign Göç Eden Parsiyel Nöbetler) (Prenatal) (SLC12A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 34, 616645, Autosomal recessive; EIEE34 (Malignant migrating partial seizures of infancy) (SLC12A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC12A5	SLC12A5, KCC2, KIAA1176, EIEE34, EIG14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 35, 616647, Otozomal Resesif; EIEE35 (İTPA İlişkili Ensefalopati) (ITPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 35, 616647, Autosomal recessive; EIEE35 (ITPA-related encephalopathy) (ITPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPA	ITPA, EIEE35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 35 (EIEE35) [MIM:616647]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. EIEE35 is characterized by onset of seizures in the first months of life associated with essentially no normal development. Many patients die in early childhood. {ECO:0000269	PubMed:26224535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 35, 616647, Otozomal Resesif; EIEE35 (ITPA İlişkili Ensefalopati) (Prenatal) (ITPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 35, 616647, Autosomal recessive; EIEE35 (ITPA-related encephalopathy) (ITPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITPA	ITPA, EIEE35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 35 (EIEE35) [MIM:616647]: A form of epileptic encephalopathy, a heterogeneous group of severe childhood onset epilepsies characterized by refractory seizures, neurodevelopmental impairment, and poor prognosis. Development is normal prior to seizure onset, after which cognitive and motor delays become apparent. EIEE35 is characterized by onset of seizures in the first months of life associated with essentially no normal development. Many patients die in early childhood. {ECO:0000269	PubMed:26224535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 36, 300884, X'e Bağlı Dominant; EIEE36 (ALG13-CDG) (ALG13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 36, 300884, X-linked dominant; EIEE36 (ALG13-CDG) (ALG13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG13	ALG13, GLT28D1, CDG1S, EIEE36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1S (CDG1S) [MIM:300884]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. {ECO:0000269	PubMed:22492991}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 36, 300884, X'e Bağlı Dominant; EIEE36 (ALG13-CDG) (Prenatal) (ALG13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 36, 300884, X-linked dominant; EIEE36 (ALG13-CDG) (ALG13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG13	ALG13, GLT28D1, CDG1S, EIEE36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 37, 616981, Otozomal Resesif; EIEE37 (FRRS1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 37, 616981, Autosomal recessive; EIEE37 (FRRS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRRS1L	FRRS1L, C9orf4, CG6, EIEE37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 37, 616981, Otozomal Resesif; EIEE37 (Prenatal) (FRRS1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 37, 616981, Autosomal recessive; EIEE37 (FRRS1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FRRS1L	FRRS1L, C9orf4, CG6, EIEE37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 38, 617020, Otozomal Resesif; EIEE38 (ARV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 38, 617020, Autosomal recessive; EIEE38 (ARV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARV1	ARV1, EIEE38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 38, 617020, Otozomal Resesif; EIEE38 (Prenatal) (ARV1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 38, 617020, Autosomal recessive; EIEE38 (ARV1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARV1	ARV1, EIEE38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 39, 612949, Otozomal Resesif; EIEE39 (Epileptik Ensefalopati-Global Serebral Demyelinasyon) (SLC25A12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 39, 612949, Autosomal recessive; EIEE39 (Epileptic encephalopathy with global cerebral demyelination) (SLC25A12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A12	SLC25A12, ARALAR, EIEE39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 39, 612949, Otozomal Resesif; EIEE39 (Epileptik Ensefalopati-Global Serebral Demyelinasyon) (Prenatal) (SLC25A12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 39, 612949, Autosomal recessive; EIEE39 (Epileptic encephalopathy with global cerebral demyelination) (SLC25A12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A12	SLC25A12, ARALAR, EIEE39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 4, 612164, Otozomal Dominant; EIEE4 (Erken İnfantil Epileptik Ensefalopati) (STXBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 4, 612164, Autosomal dominant; EIEE4 (Early infantile epileptic encephalopathy) (STXBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STXBP1	STXBP1, UNC18, EIEE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 4, 612164, Otozomal Dominant; EIEE4 (Erken İnfantil Epileptik Ensefalopati) (MLPA)	Epileptic encephalopathy, early infantile, 4, 612164, Autosomal dominant; EIEE4 (Early infantile epileptic encephalopathy) (MLPA)	STXBP1	STXBP1, UNC18, EIEE4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 4, 612164, Otozomal Dominant; EIEE4 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (STXBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 4, 612164, Autosomal dominant; EIEE4 (Early infantile epileptic encephalopathy) (STXBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STXBP1	STXBP1, UNC18, EIEE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 4, 612164, Otozomal Dominant; EIEE4 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 4, 612164, Autosomal dominant; EIEE4 (Early infantile epileptic encephalopathy) (Prenatal) (MLPA)	STXBP1	STXBP1, UNC18, EIEE4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 40, 617065, Otozomal Resesif; EIEE40 (West Sendromu) (GUF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 40, 617065, Autosomal recessive; EIEE40 (West syndrome) (GUF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUF1	GUF1, EF4, EIEE40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 40, 617065, Otozomal Resesif; EIEE40 (West Sendromu) (Prenatal) (GUF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 40, 617065, Autosomal recessive; EIEE40 (West syndrome) (GUF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GUF1	GUF1, EF4, EIEE40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 41, 617105, Otozomal Dominant; EIEE41 (Erken Miyoklonik Ensefalopati) (SLC1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 41, 617105, Autosomal dominant; EIEE41 (Early myoclonic encephalopathy) (SLC1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC1A2	SLC1A2, EAAT2, EIEE41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 41, 617105, Otozomal Dominant; EIEE41 (Erken Miyoklonik Ensefalopati) (Prenatal) (SLC1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 41, 617105, Autosomal dominant; EIEE41 (Early myoclonic encephalopathy) (SLC1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC1A2	SLC1A2, EAAT2, EIEE41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 42, 617106, Otozomal Dominant; EIEE42 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (CACNA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 42, 617106, Autosomal dominant; EIEE42 (Undetermined early-onset epileptic encephalopathy) (CACNA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 42, 617106, Otozomal Dominant; EIEE42 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (MLPA)	Epileptic encephalopathy, early infantile, 42, 617106, Autosomal dominant; EIEE42 (Undetermined early-onset epileptic encephalopathy) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 42, 617106, Otozomal Dominant; EIEE42 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (CACNA1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 42, 617106, Autosomal dominant; EIEE42 (Undetermined early-onset epileptic encephalopathy) (CACNA1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 42, 617106, Otozomal Dominant; EIEE42 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopatisi (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 42, 617106, Autosomal dominant; EIEE42 (Undetermined early-onset epileptic encephalopathy) (Prenatal) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 43, 617113, Otozomal Dominant; EIEE43 (Lennox-Gastaut Sendromu) (GABRB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 43, 617113, Autosomal dominant; EIEE43 (Lennox-Gastaut syndrome) (GABRB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRB3	GABRB3, ECA5, EIEE43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 43, 617113, Otozomal Dominant; EIEE43 (Lennox-Gastaut Sendromu) (Prenatal) (GABRB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 43, 617113, Autosomal dominant; EIEE43 (Lennox-Gastaut syndrome) (GABRB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GABRB3	GABRB3, ECA5, EIEE43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 44, 617132, Otozomal Resesif; EIEE44 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (UBA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 44, 617132, Autosomal recessive; EIEE44 (Undetermined early-onset epileptic encephalopathy) (UBA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBA5	UBA5, UBE1DC1, EIEE44, SCAR24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 44, 617132, Otozomal Resesif; EIEE44 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (UBA5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 44, 617132, Autosomal recessive; EIEE44 (Undetermined early-onset epileptic encephalopathy) (UBA5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBA5	UBA5, UBE1DC1, EIEE44, SCAR24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 45, 617153, Otozomal Dominant; EIEE45 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (GABRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 45, 617153, Autosomal dominant; EIEE45 (Undetermined early-onset epileptic encephalopathy) (GABRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRB1	GABRB1, EIEE45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 45, 617153, Otozomal Dominant; EIEE45 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (GABRB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 45, 617153, Autosomal dominant; EIEE45 (Undetermined early-onset epileptic encephalopathy) (GABRB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GABRB1	GABRB1, EIEE45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 46, 617162, Otozomal Dominant; EIEE46 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (GRIN2D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 46, 617162, Autosomal dominant; EIEE46 (Undetermined early-onset epileptic encephalopathy) (GRIN2D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIN2D	GRIN2D, NMDAR2D, EIEE46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 46, 617162, Otozomal Dominant; EIEE46 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (GRIN2D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 46, 617162, Autosomal dominant; EIEE46 (Undetermined early-onset epileptic encephalopathy) (GRIN2D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIN2D	GRIN2D, NMDAR2D, EIEE46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 47, 617166, Otozomal Dominant; EIEE47 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (FGF12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 47, 617166, Autosomal dominant; EIEE47 (Undetermined early-onset epileptic encephalopathy) (FGF12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF12	FGF12, FHF1, EIEE47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 47, 617166, Otozomal Dominant; EIEE47 (Belirlenemeyen Erken Başlangıçlı Epileptik Ensefalopati) (Prenatal) (FGF12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 47, 617166, Autosomal dominant; EIEE47 (Undetermined early-onset epileptic encephalopathy) (FGF12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGF12	FGF12, FHF1, EIEE47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epileptik Ensefalopati, Erken Infantil, 48, 617276, Otozomal Resesif; EIEE48 (Erken İnfantil Epileptik Ensefalopati) (AP3B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 48, 617276, Autosomal recessive; EIEE48 (Early infantile epileptic encephalopathy) (AP3B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP3B2	AP3B2, NAPTB, EIEE48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 48, 617276, Otozomal Resesif; EIEE48 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (AP3B2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 48, 617276, Autosomal recessive; EIEE48 (Early infantile epileptic encephalopathy) (AP3B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP3B2	AP3B2, NAPTB, EIEE48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 49, 617281, EIEE49 (Erken İnfantil Epileptik Ensefalopati) (DENND5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 49, 617281, EIEE49 (Early infantile epileptic encephalopathy) (DENND5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DENND5A	DENND5A, RAB6IP1, KIAA1091, EIEE49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 49, 617281, EIEE49 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (DENND5A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 49, 617281, EIEE49 (Early infantile epileptic encephalopathy) (DENND5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DENND5A	DENND5A, RAB6IP1, KIAA1091, EIEE49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 5, 613477, Otozomal Dominant; EIEE5 (West Sendromu) (SPTAN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 5, 613477, Autosomal dominant; EIEE5 (West syndrome) (SPTAN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTAN1	SPTAN1, NEAS, EIEE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 5 (EIEE5) [MIM:613477]: A disorder characterized by seizures associated with hypsarrhythmia, profound mental retardation with lack of visual attention and speech development, as well as spastic quadriplegia. {ECO:0000269|PubMed:20493457}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 5, 613477, Otozomal Dominant; EIEE5 (West Sendromu) (SPTAN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 5, 613477, Autosomal dominant; EIEE5 (West syndrome) (SPTAN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPTAN1	SPTAN1, NEAS, EIEE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 5 (EIEE5) [MIM:613477]: A disorder characterized by seizures associated with hypsarrhythmia, profound mental retardation with lack of visual attention and speech development, as well as spastic quadriplegia. {ECO:0000269|PubMed:20493457}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 50, 616457, Otozomal Resesif; EIEE50 (CAD-CDG) (CAD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 50, 616457, Autosomal recessive; EIEE50 (CAD-CDG) (CAD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAD	CAD, CDG1Z, EIEE50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1Z (CDG1Z) [MIM:616457]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. CDG1Z patients show abnormal glycosylation of red blood cell proteins, but glycosylation of serum transferrin is normal. {ECO:0000269|PubMed:25678555}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 50, 616457, Otozomal Resesif; EIEE50 (CAD-CDG) (Prenatal) (CAD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 50, 616457, Autosomal recessive; EIEE50 (CAD-CDG) (CAD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAD	CAD, CDG1Z, EIEE50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1Z (CDG1Z) [MIM:616457]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N-glycoproteins during embryonic development, differentiation, and maintenance of cell functions. CDG1Z patients show abnormal glycosylation of red blood cell proteins, but glycosylation of serum transferrin is normal. {ECO:0000269|PubMed:25678555}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 51, 617339, Otozomal Resesif; EIEE51 (Erken İnfantil Epileptik Ensefalopati) (MDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 51, 617339, Autosomal recessive; EIEE51 (Early infantile epileptic encephalopathy) (MDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MDH2	MDH2, EIEE51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 51, 617339, Otozomal Resesif; EIEE51 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (MDH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 51, 617339, Autosomal recessive; EIEE51 (Early infantile epileptic encephalopathy) (MDH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MDH2	MDH2, EIEE51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 52, 617350, Otozomal Resesif; EIEE52 (Erken İnfantil Epileptik Ensefalopati) (SCN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 52, 617350, Autosomal recessive; EIEE52 (Early infantile epileptic encephalopathy) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEE52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 52, 617350, Otozomal Resesif; EIEE52 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (SCN1B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 52, 617350, Autosomal recessive; EIEE52 (Early infantile epileptic encephalopathy) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, EIEE52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 53, 617389, EIEE53 (Erken İnfantil Epileptik Ensefalopati) (SYNJ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 53, 617389, EIEE53 (Early infantile epileptic encephalopathy) (SYNJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNJ1	SYNJ1, PARK20, EIEE53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 53, 617389, EIEE53 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (SYNJ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 53, 617389, EIEE53 (Early infantile epileptic encephalopathy) (SYNJ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYNJ1	SYNJ1, PARK20, EIEE53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 54, 617391, EIEE54 (Erken İnfantil Epileptik Ensefalopati) (HNRNPU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 54, 617391, EIEE54 (Early infantile epileptic encephalopathy) (HNRNPU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPU	HNRNPU, HNRPU, SAFA, EIEE54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 54, 617391, EIEE54 (Erken İnfantil Epileptik Ensefalopati) (Prenatal) (HNRNPU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 54, 617391, EIEE54 (Early infantile epileptic encephalopathy) (HNRNPU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HNRNPU	HNRNPU, HNRPU, SAFA, EIEE54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 6, 607208, Otozomal Dominant; EIEE6 (Dravet Sendromu) (SCN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 6, 607208, Autosomal dominant; EIEE6 (Dravet syndrome) (SCN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 6, 607208, Otozomal Dominant; EIEE6 (Dravet Sendromu) (MLPA)	Epileptic encephalopathy, early infantile, 6, 607208, Autosomal dominant; EIEE6 (Dravet syndrome) (MLPA)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 6, 607208, Otozomal Dominant; EIEE6 (Dravet Sendromu) (Prenatal) (SCN1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 6, 607208, Autosomal dominant; EIEE6 (Dravet syndrome) (SCN1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 6, 607208, Otozomal Dominant; EIEE6 (Dravet Sendromu) (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 6, 607208, Autosomal dominant; EIEE6 (Dravet syndrome) (Prenatal) (MLPA)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 7, 613720, Otozomal Dominant; EIEE7 (KCNQ2 İle İlişkili Epileptik Ensefalopati) (KCNQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 7, 613720, Autosomal dominant; EIEE7 (KCNQ2-related epileptic encephalopathy) (KCNQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ2	KCNQ2, EBN1, EIEE7, BFNS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 7, 613720, Otozomal Dominant; EIEE7 (KCNQ2 İle İlişkili Epileptik Ensefalopati) (Prenatal) (KCNQ2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 7, 613720, Autosomal dominant; EIEE7 (KCNQ2-related epileptic encephalopathy) (KCNQ2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNQ2	KCNQ2, EBN1, EIEE7, BFNS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 8, 300607, X'e Bağlı resesif; EIEE8 (Hiperekplektik Epilepsi Sendromu) (ARHGEF9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 8, 300607, X-linked recessive; EIEE8 (Hyperekplexia-epilepsy syndrome) (ARHGEF9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGEF9	ARHGEF9, PEM2, KIAA0424, EIEE8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 8, 300607, X'e Bağlı resesif; EIEE8 (Hiperekplektik Epilepsi Sendromu) (Prenatal) (ARHGEF9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 8, 300607, X-linked recessive; EIEE8 (Hyperekplexia-epilepsy syndrome) (ARHGEF9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARHGEF9	ARHGEF9, PEM2, KIAA0424, EIEE8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 9, 300088, X'e Bağlı; EIEE9 (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (PCDH19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epileptic encephalopathy, early infantile, 9, 300088, X-linked; EIEE9 (X-linked intellectual disability-epilepsy syndrome) (PCDH19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCDH19	PCDH19, KIAA1313, EFMR, EIEE9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 9 (EIEE9) [MIM:300088]: A condition characterized by seizure with onset in infancy or early childhood, cognitive impairment, and delayed development of variable severity in some patients. Additional features include autistic signs and psychosis. The disorder is sex-limited, with the phenotype being restricted to females. {ECO:0000269|PubMed:18469813, ECO:0000269|PubMed:19214208, ECO:0000269|PubMed:19752159, ECO:0000269|PubMed:20713952, ECO:0000269|PubMed:20830798, ECO:0000269|PubMed:21053371, ECO:0000269|PubMed:21480887, ECO:0000269|PubMed:21519002, ECO:0000269|PubMed:22050978, ECO:0000269|PubMed:22267240}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epileptik Ensefalopati, Erken Infantil, 9, 300088, X'e Bağlı; EIEE9 (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (MLPA)	Epileptic encephalopathy, early infantile, 9, 300088, X-linked; EIEE9 (X-linked intellectual disability-epilepsy syndrome) (MLPA)	PCDH19	PCDH19, KIAA1313, EFMR, EIEE9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epileptic encephalopathy, early infantile, 9 (EIEE9) [MIM:300088]: A condition characterized by seizure with onset in infancy or early childhood, cognitive impairment, and delayed development of variable severity in some patients. Additional features include autistic signs and psychosis. The disorder is sex-limited, with the phenotype being restricted to females. {ECO:0000269|PubMed:18469813, ECO:0000269|PubMed:19214208, ECO:0000269|PubMed:19752159, ECO:0000269|PubMed:20713952, ECO:0000269|PubMed:20830798, ECO:0000269|PubMed:21053371, ECO:0000269|PubMed:21480887, ECO:0000269|PubMed:21519002, ECO:0000269|PubMed:22050978, ECO:0000269|PubMed:22267240}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Epileptik Ensefalopati, Erken İnfantil, 9, 300088, X'e Bağlı; EIEE9 (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (Prenatal) (PCDH19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epileptic encephalopathy, early infantile, 9, 300088, X-linked; EIEE9 (X-linked intellectual disability-epilepsy syndrome) (PCDH19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCDH19	PCDH19, KIAA1313, EFMR, EIEE9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 9 (EIEE9) [MIM:300088]: A condition characterized by seizure with onset in infancy or early childhood, cognitive impairment, and delayed development of variable severity in some patients. Additional features include autistic signs and psychosis. The disorder is sex-limited, with the phenotype being restricted to females. {ECO:0000269|PubMed:18469813, ECO:0000269|PubMed:19214208, ECO:0000269|PubMed:19752159, ECO:0000269|PubMed:20713952, ECO:0000269|PubMed:20830798, ECO:0000269|PubMed:21053371, ECO:0000269|PubMed:21480887, ECO:0000269|PubMed:21519002, ECO:0000269|PubMed:22050978, ECO:0000269|PubMed:22627240}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epileptik Ensefalopati, Erken İnfantil, 9, 300088, X'e Bağlı; EIEE9 (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (Prenatal) (MLPA)	Epileptic encephalopathy, early infantile, 9, 300088, X-linked; EIEE9 (X-linked intellectual disability-epilepsy syndrome) (Prenatal) (MLPA)	PCDH19	PCDH19, KIAA1313, EFMR, EIEE9	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epileptic encephalopathy, early infantile, 9 (EIEE9) [MIM:300088]: A condition characterized by seizure with onset in infancy or early childhood, cognitive impairment, and delayed development of variable severity in some patients. Additional features include autistic signs and psychosis. The disorder is sex-limited, with the phenotype being restricted to females. {ECO:0000269|PubMed:18469813, ECO:0000269|PubMed:19214208, ECO:0000269|PubMed:19752159, ECO:0000269|PubMed:20713952, ECO:0000269|PubMed:20830798, ECO:0000269|PubMed:21053371, ECO:0000269|PubMed:21480887, ECO:0000269|PubMed:21519002, ECO:0000269|PubMed:22050978, ECO:0000269|PubMed:22627240}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifizyal Kondrodisplazi, Miura Tipi, 615923, Otozomal Dominant; ECDM (Uzun Boy-Skolyoz-Ayak Başparmağının Makrodaktilisi Sendromu) (NPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal chondrodysplasia, Miura type, 615923, Autosomal dominant; ECDM (Tall stature-scoliosis-macrodactyly of the great toes syndrome) (NPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acromesomelic dysplasia, Maroteaux type (AMDM) [MIM:602875]: An autosomal recessive acromesomelic chondrodysplasia. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs and handfoot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knobs-like fingers). AMDM is characterized by axial skeletal involvement with wedging of vertebral bodies. In AMDM all skeletal elements are present but show abnormal rates of linear growth. {ECO:0000269|PubMed:15146390}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epifizyal Kondrodisplazi, Miura Tipi, 615923, Otozomal Dominant; ECDM (Uzun Boy-Skolyoz-Ayak Başparmağının Makrodaktilisi Sendromu) (Prenatal) (NPR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal chondrodysplasia, Miura type, 615923, Autosomal dominant; ECDM (Tall stature-scoliosis-macrodactyly of the great toes syndrome) (NPR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifizyal Displazi, Multipl, 1, 132400, Otozomal Dominant; EDM1 (Multipl Epifizyal Displazi Tip 1) (COMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 1, 132400, Autosomal dominant; EDM1 (Multiple epiphyseal dysplasia type 1) (COMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COMP	COMP, EDM1, MED, PSACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 1 (EDM1) [MIM:132400]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:11565064, ECO:0000269|PubMed:21922596, ECO:0000269|PubMed:7670472, ECO:0000269|PubMed:9021009, ECO:0000269|PubMed:9184241, ECO:0000269|PubMed:9452026, ECO:0000269|PubMed:9463320, ECO:0000269|PubMed:9921895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epifizyal Displazi, Multipl, 1, 132400, Otozomal Dominant; EDM1 (Multipl Epifizyal Displazi Tip 1) (Prenatal) (COMP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 1, 132400, Autosomal dominant; EDM1 (Multiple epiphyseal dysplasia type 1) (COMP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COMP	COMP, EDM1, MED, PSACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 1 (EDM1) [MIM:132400]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:11565064, ECO:0000269|PubMed:21922596, ECO:0000269|PubMed:7670472, ECO:0000269|PubMed:9021009, ECO:0000269|PubMed:9184241, ECO:0000269|PubMed:9452026, ECO:0000269|PubMed:9463320, ECO:0000269|PubMed:9921895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifizyal Displazi, Multipl, 2, 600204, Otozomal Dominant; EDM2 (Multipl Epifizyal Displazi, Kollajen 9 Anomalisine Bağlı) (COL9A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 2, 600204, Autosomal dominant; EDM2 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A2	COL9A2, EDM2, STL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 2 (EDM2) [MIM:600204]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10364514}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epifizyal Displazi, Multipl, 2, 600204, Otozomal Dominant; EDM2 (Multipl Epifizyal Displazi, Kollajen 9 Anomalisine Bağlı) (Prenatal) (COL9A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 2, 600204, Autosomal dominant; EDM2 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL9A2	COL9A2, EDM2, STL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 2 (EDM2) [MIM:600204]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10364514}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifizyal Displazi, Multipl, 3, ± Miyopati, 600969, Otozomal Dominant; EDM3 (Multipl Epifizyal Displazi, Kollajen 9 Anomalisine Bağlı) (COL9A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969, Autosomal dominant; EDM3 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A3	COL9A3, EDM3, IDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 3 (EDM3) [MIM:600969]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10090888}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Epifizyal Displazi, Multipl, 3, ± Miyopati, 600969, Otozomal Dominant; EDM3 (Multipl Epifizyal Displazi, Kollajen 9 Anomalisine Bağlı) (Prenatal) (COL9A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969, Autosomal dominant; EDM3 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL9A3	COL9A3, EDM3, IDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 3 (EDM3) [MIM:600969]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10090888}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Epifiyal Displazi, Multipl, 4, 226900, Otozomal Resesif; EDM4 (Multipl Epifiyal Displazi Tip 4) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 4, 226900, Autosomal recessive; EDM4 (Multiple epiphyseal dysplasia type 4) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 4 (EDM4) [MIM:226900]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. Multiple epiphyseal dysplasia type 4 is a recessively inherited form, characterized by early childhood-onset hip dysplasia and recurrent patella dislocation. Short stature is not frequent. {ECO:0000269|PubMed:12966518, ECO:0000269|PubMed:21922596}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epifiyal Displazi, Multipl, 4, 226900, Otozomal Resesif; EDM4 (Multipl Epifiyal Displazi Tip 4) (Prenatal) (SLC26A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 4, 226900, Autosomal recessive; EDM4 (Multiple epiphyseal dysplasia type 4) (SLC26A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC26A2	SLC26A2, DTD, DTDST, DSS1708, EDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 4 (EDM4) [MIM:226900]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. Multiple epiphyseal dysplasia type 4 is a recessively inherited form, characterized by early childhood-onset hip dysplasia and recurrent patella dislocation. Short stature is not frequent. {ECO:0000269|PubMed:12966518, ECO:0000269|PubMed:21922596}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifiyal Displazi, Multipl, 5, 607078, Otozomal Dominant; EDM5 (Multipl Epifiyal Displazi Tip 5) (MATN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 5, 607078, Autosomal dominant; EDM5 (Multiple epiphyseal dysplasia type 5) (MATN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MATN3	MATN3, EDM5, HOA, OS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Epifiyal Displazi, Multipl, 5, 607078, Otozomal Dominant; EDM5 (Multipl Epifiyal Displazi Tip 5) (Prenatal) (MATN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 5, 607078, Autosomal dominant; EDM5 (Multiple epiphyseal dysplasia type 5) (MATN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MATN3	MATN3, EDM5, HOA, OS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifiyal Displazi, Multipl, 6, 614135, Otozomal Dominant; EDM6 (Multipl Epifiyal Displazi, Kollajen 9 Anomalisine Bağlı) (COL9A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, 6, 614135, Autosomal dominant; EDM6 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A1	COL9A1, EDM6, STL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epifiyal Displazi, Multipl, 6, 614135, Otozomal Dominant; EDM6 (Multipl Epifiyal Displazi, Kollajen 9 Anomalisine Bağlı) (Prenatal) (COL9A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, 6, 614135, Autosomal dominant; EDM6 (Multiple epiphyseal dysplasia due to collagen 9 anomaly) (COL9A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL9A1	COL9A1, EDM6, STL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifiyal Displazi, Multipl-Erken Başlangıçlı Diyabetes Mellitus, Wolcott-Rallison Sendromu, 226980, Otozomal Resesif (Wolcott-Rallison Sendromu) (EIF2AK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS, Wolcott-Rallison syndrome, 226980, Autosomal recessive (Wolcott-Rallison syndrome) (EIF2AK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2AK3	EIF2AK3, PEK, PERK, WRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epifiyal Displazi, Multipl-Erken Başlangıçlı Diyabetes Mellitus, Wolcott-Rallison Sendromu, 226980, Otozomal Resesif (Wolcott-Rallison Sendromu) (Prenatal) (EIF2AK3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS, Wolcott-Rallison syndrome, 226980, Autosomal recessive (Wolcott-Rallison syndrome) (EIF2AK3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2AK3	EIF2AK3, PEK, PERK, WRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifiyal Displazi, Multipl, Miyopi ve Sağırlık, 132450, Otozomal Dominant; EDMMD (Multipl Epifiyal Displazi, Beighton Tip) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Epiphyseal dysplasia, multiple, with myopia and deafness, 132450, Autosomal dominant; EDMMD (Multiple epiphyseal dysplasia, Beighton type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Epifiyal Displazi, Multipl, Miyopi ve Sağırlık, 132450, Otozomal Dominant; EDMMD (Multipl Epifiyal Displazi, Beighton Tip) (MLPA)	Epiphyseal dysplasia, multiple, with myopia and deafness, 132450, Autosomal dominant; EDMMD (Multiple epiphyseal dysplasia, Beighton type) (MLPA)	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Epifiyal Displazi, Multipl, Miyopi ve Sağırlık, 132450, Otozomal Dominant; EDMMD (Multipl Epifiyal Displazi, Beighton Tip) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Epiphyseal dysplasia, multiple, with myopia and deafness, 132450, Autosomal dominant; EDMMD (Multiple epiphyseal dysplasia, Beighton type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Epifiyal Displazi, Multipl, Miyopi ve Sağırlık, 132450, Otozomal Dominant; EDMMD (Multipl Epifiyal Displazi, Beighton Tip) (Prenatal) (MLPA)	Epiphyseal dysplasia, multiple, with myopia and deafness, 132450, Autosomal dominant; EDMMD (Multiple epiphyseal dysplasia, Beighton type) (Prenatal) (MLPA)	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Episodik Ataksi, Otozomal Dominant Tip 2, 108500; EA2 (Ailesel Paroksismal Ataksi) (CACNA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic ataxia, type 2, 108500, Autosomal dominant; EA2 (Familial paroxysmal ataxia) (CACNA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Episodik Ataksi, Otozomal Dominant Tip 2, 108500; EA2 (Ailesel Paroksismal Ataksi) (MLPA)	Episodic ataxia, type 2, 108500, Autosomal dominant; EA2 (Familial paroxysmal ataxia) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Episodik Ataksi, Tip 5, 613855, Otozomal Dominant; EA5 (Episodik Ataksi Tip 5) (CACNB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic ataxia, type 5, 613855, Autosomal dominant; EA5 (Episodic ataxia type 5) (CACNB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNB4	CACNB4, EJM6, EA5, EIG9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Episodik Ataksi, Tip 6, 612656, Otozomal Dominant; EA6 (Episodik Ataksi Tip 6) (SLC1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic ataxia, type 6, 612656, Autosomal dominant; EA6 (Episodic ataxia type 6) (SLC1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC1A3	SLC1A3, EAAT1, EA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic ataxia 6 (EA6) [MIM:612656]: A disorder characterized by episodic ataxia, seizures, migraine and alternating hemiplegia. {ECO:0000269|PubMed:16116111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Episodik Ataksi / Miyokimi Sendromu, 160120, Otozomal Dominant; EA1 (Ailesel Süreklİ Kas Lİf Aktivitesi) (KCNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic ataxia/myokymia syndrome, 160120, Autosomal dominant; EA1 (Hereditary continuous muscle fiber activity) (KCNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNA1	KCNA1, AEMK, EA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic ataxia 1 (EA1) [MIM:160120]: An autosomal dominant disorder characterized by brief episodes of ataxia and dysarthria. Neurological examination during and between the attacks demonstrates spontaneous, repetitive discharges in the distal musculature (myokymia) that arise from peripheral nerve. Nystagmus is absent. {ECO:0000269|PubMed:10355668, ECO:0000269|PubMed:11013453, ECO:0000269|PubMed:11026449, ECO:0000269|PubMed:15320332, ECO:0000269|PubMed:7842011, ECO:0000269|PubMed:8541859, ECO:0000269|PubMed:8871592, ECO:0000269|PubMed:9600245}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Episodik Kinesijenik Diskinezi 1, 128200, Otozomal Dominant; EKD1 (Paroksismal Kinesijenik Diskinezi) (PRRT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic kinesigenic dyskinesia 1, 128200, Autosomal dominant; EKD1 (Paroxysmal kinesigenic dyskinesia) (PRRT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRRT2	PRRT2, PKC, DYT10, EKD1, BFIS2, BFIC2, ICCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic kinesigenic dyskinesia 1 (EKD1) [MIM:128200]: An autosomal dominant neurologic condition characterized by recurrent and brief attacks of abnormal involuntary movements, triggered by sudden voluntary movement. These attacks usually have onset during childhood or early adulthood and can involve dystonic postures, chorea, or athetosis. {ECO:0000269|PubMed:22101681, ECO:0000269|PubMed:22120146, ECO:0000269|PubMed:22313361, ECO:0000269|PubMed:22209761}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations that produce truncation of the C-terminus of the protein alter subcellular location, from plasma membrane to cytoplasm {PubMed:22101681}. {ECO:0000269|PubMed:22101681}.	EDTA Blood Sample
Episodik Ağrı Sendromu, Ailesel, 2, 615551, Otozomal Dominant; FEPS2 (Sodyum Kanalopatisi İlişkili Small Fiber Nöropati) (SCN10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic pain syndrome, familial, 2, 615551, Autosomal dominant; FEPS2 (Sodium channelopathy-related small fiber neuropathy) (SCN10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN10A	SCN10A, FEPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic pain syndrome, familial, 2 (FEPS2) [MIM:615551]: An autosomal dominant neurologic disorder characterized by adult- onset of paroxysmal pain mainly affecting the distal lower extremities. {ECO:0000269|PubMed:23115331}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Episodik Ağrı Sendromu, Ailesel, 3, 615552, Otozomal Dominant; FEPS3 (Ailesel Episodik Ağrı Sendromu) (SCN11A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic pain syndrome, familial, 3, 615552, Autosomal dominant; FEPS3 (Familial episodic pain syndrome) (SCN11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN11A	SCN11A, HSAN7, FEPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Episodik Ağrı Sendromu, Ailesel, 615040, Otozomal Dominant (Ailesel Episodik Ağrı Sendromu) (TRPA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Episodic pain syndrome, familial, 615040, Autosomal dominant (Familial episodic pain syndrome) (TRPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPA1	TRPA1, ANKTM1, FEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic pain syndrome, familial, 1 (FEPS1) [MIM:615040]: An autosomal dominant neurologic disorder characterized by onset in infancy of episodic debilitating upper body pain triggered by fasting, cold, and physical stress. The period of intense pain is accompanied by breathing difficulties, tachycardia, sweating, generalized pallor, peribuccal cyanosis, and stiffness of the abdominal wall. Affected individuals do not manifest altered pain sensitivity outside the episodes. {ECO:0000269	PubMed:20547126}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epitelyal Tekrarlayan Erozyon Distrofi, 122400, Otozomal Dominant; ERED (Epitelyal Tekrarlayan Erozyon Distrofisi) (COL17A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Epithelial recurrent erosion dystrophy, 122400, Autosomal dominant; ERED (Epithelial recurrent erosion dystrophy) (COL17A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL17A1	COL17A1, BPAG2, ERED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epithelial recurrent erosion dystrophy (ERED) [MIM:122400]: A corneal dystrophy characterized by recurrent episodes of epithelial erosions from childhood, with occasional impairment of vision. Most patients have attacks of redness, photophobia, epiphora, and ocular pain. Exposure to sunlight or draught, dust and smoke and lack of sleep can precipitate attacks. {ECO:0000269	PubMed:25676728}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epstein Sendromu, 153650, Otozomal Dominant; EPSTNS (Epstein Sendromu) (MYH9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Epstein syndrome, 153650, Autosomal dominant; EPSTNS (Epstein syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Epstein Sendromu, 153650, Otozomal Dominant; EPSTNS (Epstein Sendromu) (Prenatal) (MYH9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Epstein syndrome, 153650, Autosomal dominant; EPSTNS (Epstein syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Eritermalji, Primer, 133020, Otozomal Dominant (Sodyum Kanalopatisi İlişkili Small Fiber Nöropati) (SCN9A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythermalgia, primary, 133020, Autosomal dominant (Sodium channelopathy-related small fiber neuropathy) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FE83B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Eritremia, Alfa- (MLPA)	Erythremias, alpha- (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Eritremia, Alfa- (Prenatal) (MLPA)	Erythremias, alpha- (Prenatal) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Eritremia, Beta- (HBB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythremias, beta- (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritremia, Beta- (MLPA)	Erythremias, beta- (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Eritremia, Beta- (Prenatal) (HBB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Erythremias, beta- (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN	*	.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Eritremia, Beta- (Prenatal) (MLPA)	Erythremias, beta- (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Eritrosit Amp Deaminase Eksikliği (Adenosin Monofosfat Deaminaz Eksikliği) (AMPD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	ERYTHROCYTE AMP DEAMINASE DEFICIENCY (Adenosine monophosphate deaminase deficiency) (AMPD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMPD3	AMPD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrosit Laktat Taşıyıcı Defekti, 245340, Otozomal Dominant (Metabolik Miyopati, Laktat Transporter Eksikliğine Bağlı) (SLC16A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocyte lactate transporter defect, 245340, Autosomal dominant (Metabolic myopathy due to lactate transporter defect) (SLC16A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC16A1	SLC16A1, MCT1, HHF7, MCT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrositoz (MLPA)	Erythrocytosis (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Eritrositoz (Prenatal) (MLPA)	Erythrocytosis (Prenatal) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Eritrositoz, Bisfosfogliserat Mutaz Eksikliğine Bağlı, 222800, Otozomal Resesif (Hemolitik Anemi, Difosfogliserat Mutaz Eksikliğine Bağlı) (BPGM gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocytosis due to bisphosphoglycerate mutase deficiency, 222800, Autosomal recessive (Hemolytic anemia due to diphosphoglycerate mutase deficiency) (BPGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BPGM	BPGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bisphosphoglycerate mutase deficiency (BPGMD) [MIM:222800]: A disease characterized by hemolytic anemia, splenomegaly, cholelithiasis and cholecystitis. {ECO:0000269	PubMed:1421379, ECO:0000269	PubMed:15054810}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eritrositoz, Ailesel, 2, 263400, Otozomal Resesif; ECYT2 (Chuvash Eritrositoz) (VHL gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocytosis, familial, 2, 263400, Autosomal recessive; ECYT2 (Chuvash erythrocytosis) (VHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VHL	VHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrositoz, Ailesel, 2, 263400, Otozomal Resesif; ECYT2 (Chuvash Eritrositoz) (MLPA)	Erythrocytosis, familial, 2, 263400, Autosomal recessive; ECYT2 (Chuvash erythrocytosis) (MLPA)	VHL	VHL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Eritrositoz, Ailesel, 3, 609820, Otozomal Dominant; ECYT3 (Otozomal Dominant Sekonder Polisitemi) (EGLN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocytosis, familial, 3, 609820, Autosomal dominant; ECYT3 (Autosomal dominant secondary polycythemia) (EGLN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGLN1	EGLN1, PHD2, HIFPH2, C1orf12 , ZMYND6, SM20, ECYT3, HALAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythrocytosis, familial, 3 (ECYT3) [MIM:609820]: An autosomal dominant disorder characterized by increased serum red blood cell mass, elevated serum hemoglobin and hematocrit, and normal serum erythropoietin levels. {ECO:0000269	PubMed:16407130, ECO:0000269	PubMed:17579185}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eritrositoz, Ailesel, 4, 611783; ECYT4 (Otozomal Dominant Sekonder Polisitemi) (EPAS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocytosis, familial, 4, 611783; ECYT4 (Autosomal dominant secondary polycythemia) (EPAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPAS1	EPAS1, MOP2, HIF2A, ECYT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythrocytosis, familial, 4 (ECYT4) [MIM:611783]: An autosomal dominant disorder characterized by increased serum red blood cell mass, elevated serum hemoglobin and hematocrit, and normal platelet and leukocyte counts. {ECO:0000269|PubMed:18184961, ECO:0000269|PubMed:18378852, ECO:0000269|PubMed:19208626, ECO:0000269|PubMed:22367913}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eritrositoz, Somatik, 133100 (SH2B3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrocytosis, somatic, 133100 (SH2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH2B3	SH2B3, LNK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrositoz, Somatik, 133100; ECYT1 (kodon 617 mutasyonları ve ekzon 12-14) (JAK2 gen) (Dizi Analizi) (Postnatal)	Erythrocytosis, somatic, 133100; ECYT1 (codon 617 mutations ve ekzon 12-14) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	JAK2, THCYT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrodermi, Konjenital, Palmoplantar Keratoderma, Hipotrikozis ve Hiper Ige İle 615508, Otozomal Resesif; EPKHE (Ağır Dermatit-Multipl Alerjiler-Metabolik Kilo Kaybı Sendromu) (DSG1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508, Autosomal recessive; EPKHE (Severe dermatitis-multiple allergies-metabolic wasting syndrome) (DSG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG1	DSG1, PPKS1, SPPK1, EPKHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrokeratodermia Variabilis ve Progressiva, 133200, Otozomal Resesif, Otozomal Dominant (Eritrokeratodermia variabilis) (GJA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrokeratodermia variabilis et progressiva, 133200, Autosomal recessive, Autosomal dominant (Erythrokeratodermia variabilis) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Eritrokeratodermia Variabilis ve Progressiva, 133200, Otozomal Resesif, Otozomal Dominant; EKVP (Eritrokeratodermia Variabilis) (GJB3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrokeratodermia variabilis et progressiva, 133200, Autosomal recessive, Autosomal dominant; EKVP (Erythrokeratodermia variabilis) (GJB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB3	GJB3, CX31, DFNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythrokeratodermia variabilis (EKV) [MIM:133200]: A genodermatosis characterized by the appearance of two independent skin lesions: transient figurate erythematous patches and hyperkeratosis that is usually localized but occasionally occurs in its generalized form. Clinical presentation varies significantly within a family and from one family to another. Palmoplantar keratoderma is present in around 50% of cases. {ECO:0000269	PubMed:10594760, ECO:0000269	PubMed:10798362, ECO:0000269	PubMed:9843209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Eritrokeratodermia Variabilis ve Progressiva, 133200, Otozomal Resesif, Otozomal Dominant; EKVP (Eritrokeratodermia Variabilis) (MLPA)	Erythrokeratodermia variabilis et progressiva, 133200, Autosomal recessive, Autosomal dominant; EKVP (Erythrokeratodermia variabilis) (MLPA)	GJB3	GJB3, CX31, DFNA2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Erythrokeratodermia variabilis (EKV) [MIM:133200]: A genodermatosis characterized by the appearance of two independent skin lesions: transient figurate erythematous patches and hyperkeratosis that is usually localized but occasionally occurs in its generalized form. Clinical presentation varies significantly within a family and from one family to another. Palmoplantar keratoderma is present in around 50% of cases. {ECO:0000269	PubMed:10594760, ECO:0000269	PubMed:10798362, ECO:0000269	PubMed:9843209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Eritrokeratodermia Variabilis, Eritema Giratum Repens, 133200, Otozomal Resesif, Otozomal Dominant (Eritrokeratodermia Variabilis) (GJB4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Erythrokeratodermia variabilis with erythema gyratum repens, 133200, Autosomal recessive, Autosomal dominant (Erythrokeratodermia variabilis) (GJB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB4	GJB4, CX30.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythrokeratodermia variabilis (EKV) [MIM:133200]: A genodermatosis characterized by the appearance of two independent skin lesions: transient figurate erythematous patches and hyperkeratosis that is usually localized but occasionally occurs in its generalized form. Clinical presentation varies significantly within a family and from one family to another. Palmoplantar keratoderma is present in around 50% of cases. {ECO:0000269	PubMed:11017804}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Escobar Sendromu, 265000, Otozomal Resesif; EVMPS (Otozomal Resesif Multipl Pterijum Sendromu (Escobar Sendromu)) (CHRNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Escobar syndrome, 265000, Autosomal recessive; EVMPS (Autosomal recessive multiple pterygium syndrome) (CHRNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNG	CHRNG, ACHRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Escobar Sendromu, 265000, Otozomal Resesif; EVMPS (Otozomal Resesif Multipl Pterijum Sendromu (Escobar Sendromu)) (Prenatal) (CHRNG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Escobar syndrome, 265000, Autosomal recessive; EVMPS (Autosomal recessive multiple pterygium syndrome) (CHRNG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNG	CHRNG, ACHRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Özofagus Kanseri, 133239, Otozomal Dominant (Özofageal Skuamoz Hücre Karsinoması) (DLEC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal cancer, 133239, Autosomal dominant (Squamous cell carcinoma of esophagus) (DLEC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLEC1	DLEC1, DLC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10215088}. Note=The gene represented in this entry may be involved in disease pathogenesis. DLEC1 silencing due to promoter methylation and aberrant transcription may be implicated in the development of esophageal cancer.;	EDTA Blood Sample
Özofagus Kanseri, Alkole Bağlı Yatkınlık (ALDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal cancer, alcohol-related, susceptibility to (ALDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH2	ALDH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Özofagus Kanseri, Somatik, 133239 (Özofageal Skuamoz Hücre Karsinması) (TGFBR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal cancer, somatic, 133239 (Squamous cell carcinoma of esophagus) (TGFBR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBR2	TGFBR2, HNPCC6, AAT3, MFS2, LDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10789724}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Özofagus Karsinomu, Somatik, 133239 (Özofageal Skuamoz Hücre Karsinması) (RNF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal carcinoma, somatic, 133239 (Squamous cell carcinoma of esophagus) (RNF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF6	RNF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:12154016}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Özofagus Karsinomu, Somatik, 133239 (Özofageal Skuamoz Hücre Karsinması) (DCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal carcinoma, somatic, 133239 (Squamous cell carcinoma of esophagus) (DCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCC	DCC, MRMV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10097140}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Özofagus Skuamöz Hücreli Karsinom, 133239, Otozomal Dominant (Özofageal Skuamoz Hücre Karsinoması) (LZTS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal squamous cell carcinoma, 133239, Autosomal dominant (Squamous cell carcinoma of esophagus) (LZTS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LZTS1	LZTS1, F37, FEZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10097140}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Özofagus Yassı Hücre Karsinomu, Somatik, 133239 (Özofageal Skuamoz Hücre Karsinması) (WWOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Esophageal squamous cell carcinoma, somatic, 133239 (Squamous cell carcinoma of esophagus) (WWOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WWOX	WWOX, FOR, SCAR12, EIEE28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:11956080}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Esansiyel Tremor, Yatkınlık, 190300, Otozomal Dominant (DRD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Essential tremor, susceptibility to, 190300, Autosomal dominant (DRD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD3	DRD3, ETM1, FET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tremor, hereditary essential 1 (ETM1) [MIM:190300]: A common movement disorder mainly characterized by postural tremor of the arms. Head, legs, trunk, voice, jaw, and facial muscles also may be involved. The condition can be aggravated by emotions, hunger, fatigue and temperature extremes, and may cause a functional disability or even incapacitation. Inheritance is autosomal dominant. {ECO:0000269|PubMed:16650084, ECO:0000269|PubMed:16809426}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Östrojen Direnci, 615363, Otozomal Resesif; ESTRR (Östrojen Direnci Sendromu) (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Estrogen resistance, 615363, Autosomal recessive; ESTRR (Estrogen resistance syndrome) (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Estrogen resistance (ESTRR) [MIM:615363]: A disorder characterized by partial or complete resistance to estrogens, in the presence of elevated estrogen serum levels. Clinical features include absence of the pubertal growth spurt, delayed bone maturation, unfused epiphyses, reduced bone mineral density, osteoporosis, continued growth into adulthood and very tall adult stature. Glucose intolerance, hyperinsulinemia and lipid abnormalities may also be present. {ECO:0000269|PubMed:23841731, ECO:0000269|PubMed:8961262}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Etilmalonik Ensefalopati, 602473, Otozomal Resesif (Etilmalonik Ensefalopati) (ETHE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ethylmalonic encephalopathy, 602473, Autosomal recessive (Ethylmalonic encephalopathy) (ETHE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETHE1	ETHE1, HSCO, D83198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Etilmalonik Ensefalopati, 602473, Otozomal Resesif (Etilmalonik Ensefalopati) (Prenatal) (ETHE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ethylmalonic encephalopathy, 602473, Autosomal recessive (Ethylmalonic encephalopathy) (ETHE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ETHE1	ETHE1, HSCO, D83198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Even-Plus Sendromu, 616854, Otozomal Resesif; EVPLS (HSPA9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Even-plus syndrome, 616854, Autosomal recessive; EVPLS (HSPA9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPA9	HSPA9, HSPA9B, MOT2, GRP75, EVPLS, SIDBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Even-Plus Sendromu, 616854, Otozomal Resesif; EVPLS (Prenatal) (HSPA9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Even-plus syndrome, 616854, Autosomal recessive; EVPLS (HSPA9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPA9	HSPA9, HSPA9B, MOT2, GRP75, EVPLS, SIDBA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ewing Sarkomu t(11;22)(q24;q12) (FLI1/EWSR1) (FISH)	Ewing sarcoma t(11;22)(q24;q12) (FLI1/EWSR1) (FISH)	11q24-22q12	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Ewing Sarkomu, 612219; ES (Ewing Sarcoma) (EWSR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ewing sarcoma, 612219; ES (Ewing sarcoma) (EWSR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EWSR1	EWSR1, EWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Egzersiz İntoleransı, Riboflavine Yanıtlı, 616839, Otozomal Resesif; RREI (SLC25A32 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exercise intolerance, riboflavin-responsive, 616839, Autosomal recessive; RREI (SLC25A32 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A32	SLC25A32, MFT, RREI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Eksfoliasyon Sendromu, Yatkınlık, 177650, Otozomal Dominant; XFS (LOXL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exfoliation syndrome, susceptibility to, 177650, Autosomal dominant; XFS (LOXL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LOXL1	LOXL1, LOXL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Exfoliation syndrome (XFS) [MIM:177650]: A disorder characterized by accumulation of abnormal fibrillar deposits in the anterior segment of the eye. In addition to being a cause of glaucoma and glaucomatous optic neuropathy, exfoliation syndrome has also been associated with lens zonule weakness, cataract formation, and systemic vascular complications due to deposition of exfoliation material in extraocular tissues. {ECO:0000269|PubMed:18037624, ECO:0000269|PubMed:19343041}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to exfoliation syndrome is conferred by a risk haplotype that includes two LOXL1 coding non-synonymous SNPs (Arg141Leu and Gly153Asp) and one intronic SNP. Arg141Leu and Gly153Asp are sufficient to confer disease susceptibility in some populations.;	EDTA Blood Sample
Ekzokrin Pankreas Yetersizliği, Diseritropoietik Anemi ve Kalvarial Hiperostoz, 612714, Otozomal Resesif (Pankreas Yetersizliği-Anemi-Hiperostoz Sendromu) (COX4I2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714, Autosomal recessive (Pancreatic insufficiency-anemia-hyperostosis syndrome) (COX4I2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX4I2	COX4I2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzokrin Pankreas Yetersizliği, Diseritropoietik Anemi ve Kalvarial Hiperostoz, 612714, Otozomal Resesif (Pankreas Yetersizliği-Anemi-Hiperostoz Sendromu) (Prenatal) (COX4I2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714, Autosomal recessive (Pancreatic insufficiency-anemia-hyperostosis syndrome) (COX4I2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX4I2	COX4I2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ekzostoz, Multipl, Tip 1, 133700, Otozomal Dominant (Multipl Osteokondroma) (EXT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exostoses, multiple, type 1, 133700, Autosomal dominant (Multiple osteochondromas) (EXT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXT1	EXT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzostoz, Multipl, Tip 1, 133700, Otozomal Dominant (Multipl Osteokondroma) (MLPA)	Exostoses, multiple, type 1, 133700, Autosomal dominant (Multiple osteochondromas) (MLPA)	EXT1	EXT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ekzostoz, Multipl, Tip 2, 133701, Otozomal Dominant (Multipl Osteokondroma) (EXT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exostoses, multiple, type 2, 133701, Autosomal dominant (Multiple osteochondromas) (EXT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXT2	EXT2, SSMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzostoz, Multipl, Tip 2, 133701, Otozomal Dominant (Multipl Osteokondroma) (MLPA)	Exostoses, multiple, type 2, 133701, Autosomal dominant (Multiple osteochondromas) (MLPA)	EXT2	EXT2, SSMS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Eksüdatif Vitreoretinopati 1, 133780, Otozomal Dominant; EVR1 (Allesel Eksudatif Vitreoretinopati) (FZD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exudative vitreoretinopathy 1, 133780, Autosomal dominant; EVR1 (Familial exudative vitreoretinopathy) (FZD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FZD4	FZD4, EVR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitreoretinopathy, exudative 1 (EVR1) [MIM:133780]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. In many ways the disease resembles retinopathy of prematurity but there is no evidence of prematurity or small birth weight in the patient history. (ECO:0000269|PubMed:12172548, ECO:0000269|PubMed:14507768, ECO:0000269|PubMed:15035989, ECO:0000269|PubMed:15223780, ECO:0000269|PubMed:15370539, ECO:0000269|PubMed:15488808, ECO:0000269|PubMed:15733276, ECO:0000269|PubMed:15981244, ECO:0000269|PubMed:17093393, ECO:0000269|PubMed:19172507, ECO:0000269|PubMed:19324941, ECO:0000269|PubMed:20340138). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eksüdatif Vitreoretinopati 2, X'e Bağlı, 305390; EVR2 (Allesel Eksudatif Vitreoretinopati) (NDP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exudative vitreoretinopathy 2, X-linked, 305390; EVR2 (Familial exudative vitreoretinopathy) (NDP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDP	NDP, ND, EVR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitreoretinopathy, exudative 2 (EVR2) [MIM:305390]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. (ECO:0000269|PubMed:16163268, ECO:0000269|PubMed:16970763, ECO:0000269|PubMed:17296899, ECO:0000269|PubMed:17325173, ECO:0000269|PubMed:8252044, ECO:0000269|PubMed:8946107, ECO:0000269|PubMed:9143917). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Eksüdatif Vitreoretinopati 4, 601813, Otozomal Resesif, Otozomal Dominant; EVR4 (Allesel Eksudatif Vitreoretinopati) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exudative vitreoretinopathy 4, 601813, Autosomal recessive, Autosomal dominant; EVR4 (Familial exudative vitreoretinopathy) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Eksüdatif Vitreoretinopati 5, 613310, Otozomal Dominant; EVR5 (Allesel Eksudatif Vitreoretinopati) (TSPAN12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exudative vitreoretinopathy 5, 613310, Autosomal dominant; EVR5 (Familial exudative vitreoretinopathy) (TSPAN12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSPAN12	TSPAN12, NET2, EVR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitreoretinopathy, exudative 5 (EVR5) [MIM:613310]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. (ECO:0000269|PubMed:20159111, ECO:0000269|PubMed:20159112, ECO:0000269|PubMed:22427576). Note=The disease is caused by mutations affecting the gene represented in this entry. TSPAN12 dominant and recessive mutations have been identified in patients with exudative vitreoretinopathy. Patients with mutations in both alleles of TSPAN12 have severe exudative vitreoretinopathy or retinal dysplasia. These mutations appear to result in a milder phenotype in heterozygous mutation carriers (PubMed:22427576). (ECO:0000269|PubMed:22427576).;	EDTA Blood Sample
Eksüdatif Vitreoretinopati 6, 616468, Otozomal Dominant; EVR6 (Allesel Eksudatif Vitreoretinopati) (ZNF408 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Exudative vitreoretinopathy 6, 616468, Autosomal dominant; EVR6 (Familial exudative vitreoretinopathy) (ZNF408 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF408	ZNF408, EVR6, RP72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fabry Hastalığı, 301500, X'e Bağlı (Fabry Hastalığı) (GLA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fabry disease, 301500, X-linked (Fabry disease) (GLA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLA	GLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fabry disease (FD) [MIM:301500]: Rare X-linked sphingolipidosis disease where glycolipid accumulates in many tissues. The disease consists of an inborn error of glycosphingolipid catabolism. FD patients show systemic accumulation of globotriaosylceramide (Gb3) and related glycosphingolipids in the plasma and cellular lysosomes throughout the body. Clinical recognition in males results from characteristic skin lesions (angiokeratomas) over the lower trunk. Patients may show ocular deposits, fotofle episodes, and burning pain in the extremities. Death results from renal failure, cardiac or cerebral complications of hypertension or other vascular disease. Heterozygous females may exhibit the disorder in an attenuated form, they are more likely to show corneal opacities. (ECO:0000269|PubMed:10090526, ECO:0000269|PubMed:10208848, ECO:0000269|PubMed:10666480, ECO:0000269|PubMed:10916280, ECO:0000269|PubMed:11076046, ECO:0000269|PubMed:11295840, ECO:0000269|PubMed:11668641, ECO:0000269|PubMed:11889412, ECO:0000269|PubMed:12694230, ECO:0000269|PubMed:1315715, ECO:0000269|PubMed:15162124, ECO:0000269|PubMed:15712228, ECO:0000269|PubMed:16533976, ECO:0000269|PubMed:1846223, ECO:0000269|PubMed:19621417, ECO:0000269|PubMed:2152885, ECO:0000269|PubMed:2171331, ECO:0000269|PubMed:2539398, ECO:0000269|PubMed:7504405.	EDTA Blood Sample
Fabry Hastalığı, 301500, X'e Bağlı (Fabry Hastalığı) (MLPA)	Fabry disease, 301500, X-linked (Fabry disease) (MLPA)	GLA	GLA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fabry disease (FD) [MIM:301500]: Rare X-linked sphingolipidosis disease where glycolipid accumulates in many tissues. The disease consists of an inborn error of glycosphingolipid catabolism. FD patients show systemic accumulation of globotriaosylceramide (Gb3) and related glycosphingolipids in the plasma and cellular lysosomes throughout the body. Clinical recognition in males results from characteristic skin lesions (angiokeratomas) over the lower trunk. Patients may show ocular deposits, fotofle episodes, and burning pain in the extremities. Death results from renal failure, cardiac or cerebral complications of hypertension or other vascular disease. Heterozygous females may exhibit the disorder in an attenuated form, they are more likely to show corneal opacities. (ECO:0000269|PubMed:10090526, ECO:0000269|PubMed:10208848, ECO:0000269|PubMed:10666480, ECO:0000269|PubMed:10916280, ECO:0000269|PubMed:11076046, ECO:0000269|PubMed:11295840, ECO:0000269|PubMed:11668641, ECO:0000269|PubMed:11889412, ECO:0000269|PubMed:12694230, ECO:0000269|PubMed:1315715, ECO:0000269|PubMed:15162124, ECO:0000269|PubMed:15712228, ECO:0000269|PubMed:16533976, ECO:0000269|PubMed:1846223, ECO:0000269|PubMed:19621417, ECO:0000269|PubMed:2152885, ECO:0000269|PubMed:2171331, ECO:0000269|PubMed:2539398, ECO:0000269|PubMed:7504405.	EDTA Blood Sample
Fabry Hastalığı, Kardiyak Varyant, 301500, X'e Bağlı (Fabry Hastalığı) (GLA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fabry disease, cardiac variant, 301500, X-linked (Fabry disease) (GLA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLA	GLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fabry disease (FD) [MIM:301500]: Rare X-linked sphingolipidosis disease where glycolipid accumulates in many tissues. The disease consists of an inborn error of glycosphingolipid catabolism. FD patients show systemic accumulation of globotriaosylceramide (Gb3) and related glycosphingolipids in the plasma and cellular lysosomes throughout the body. Clinical recognition in males results from characteristic skin lesions (angiokeratomas) over the lower trunk. Patients may show ocular deposits, fotofle episodes, and burning pain in the extremities. Death results from renal failure, cardiac or cerebral complications of hypertension or other vascular disease. Heterozygous females may exhibit the disorder in an attenuated form, they are more likely to show corneal opacities. (ECO:0000269|PubMed:10090526, ECO:0000269|PubMed:10208848, ECO:0000269|PubMed:10666480, ECO:0000269|PubMed:10916280, ECO:0000269|PubMed:11076046, ECO:0000269|PubMed:11295840, ECO:0000269|PubMed:11668641, ECO:0000269|PubMed:11889412, ECO:0000269|PubMed:12694230, ECO:0000269|PubMed:1315715, ECO:0000269|PubMed:15162124, ECO:0000269|PubMed:15712228, ECO:0000269|PubMed:16533976, ECO:0000269|PubMed:1846223, ECO:0000269|PubMed:19621417, ECO:0000269|PubMed:2152885, ECO:0000269|PubMed:2171331, ECO:0000269|PubMed:2539398, ECO:0000269|PubMed:7504405.	EDTA Blood Sample

Fabry Hastalığı, Kardiyak Varyant, 301500, X'e Bağlı (Fabry Hastalığı) (MLPA)	Fabry disease, cardiac variant, 301500, X-linked (Fabry disease) (MLPA)	GLA	GLA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fabry disease (FD) [MIM:301500]: Rare X-linked sphingolipidosis disease where glycolipid accumulates in many tissues. The disease consists of an inborn error of glycosphingolipid catabolism. FD patients show systemic accumulation of globotriaosylceramide (Gb3) and related glycosphingolipids in the plasma and cellular lysosomes throughout the body. Clinical recognition in males results from characteristic skin lesions (angiokeratomas) over the lower trunk. Patients may show ocular deposits, febrile episodes, and burning pain in the extremities. Death results from renal failure, cardiac or cerebral complications of hypertension or other vascular disease. Heterozygous females may exhibit the disorder in an attenuated form, they are more likely to show corneal opacities. {ECO:0000269|PubMed:10090526, ECO:0000269|PubMed:10208948, ECO:0000269|PubMed:10666480, ECO:0000269|PubMed:10916280, ECO:0000269|PubMed:11076046, ECO:0000269|PubMed:11295840, ECO:0000269|PubMed:11668641, ECO:0000269|PubMed:11889412, ECO:0000269|PubMed:12694230, ECO:0000269|PubMed:1315715, ECO:0000269|PubMed:15162124, ECO:0000269|PubMed:15712228, ECO:0000269|PubMed:16533976, ECO:0000269|PubMed:1846223, ECO:0000269|PubMed:19621417, ECO:0000269|PubMed:2152885, ECO:0000269|PubMed:2171331, ECO:0000269|PubMed:2539398, ECO:0000269|PubMed:7504405,	EDTA Blood Sample
Fasiyal Yarık, Otik, 1, 600251, Sporadik Vakalar; OBLFC1 (Tessier No 4 Fasiyal Yarık) (SPECC1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Facial clefting, oblique, 1, 600251, Isolated cases; OBLFC1 (Tessier number 4 facial cleft) (SPECC1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPECC1L	SPECC1L, KIAA0376, OBLFC1, GBBB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fasiyal Yarık, Otik, 1, 600251, Sporadik Vakalar; OBLFC1 (Tessier No 4 Fasiyal Yarık) (Prenatal) (SPECC1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Facial clefting, oblique, 1, 600251, Isolated cases; OBLFC1 (Tessier number 4 facial cleft) (SPECC1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPECC1L	SPECC1L, KIAA0376, OBLFC1, GBBB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fasiyal Parezi, Kalıtsal Konjenital, 3, 614744, Otozomal Resesif; HCFP3 (Konjenital Kalıtsal Fasiyal Paralizi-Değişken İşitme Kaybı Sendromu) (HOXB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Facial paresis, hereditary congenital, 3, 614744, Autosomal recessive; HCFP3 (Congenital hereditary facial paralysis with variable hearing loss) (HOXB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXB1	HOXB1, HOX2I, HCFP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Facial paresis, hereditary congenital, 3 (HCFP3) [MIM:614744]: A form of facial paresis, a disease characterized by isolated dysfunction of the facial nerve (CN VII). HCFP3 patients are affected by bilateral facial palsy, facial muscle weakness of muscles innervated by CN VII, hearing loss, and strabismus. {ECO:0000269|PubMed:22770681}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fasiyoskapulohumeral Müsküler Distrofi 1, 158900, Otozomal Dominant; FSHD1 (Fasioskapulohumeral Distrofi) (Tekrar Sayısı)	Facioscapulohumeral muscular dystrophy 1, 158900, Autosomal dominant; FSHD1 (Facioscapulohumeral dystrophy)(Repeat Analysis)	FSHD1	FSHD1, FSHD1A	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Faktör 5 ve Faktör 8, Kombine Eksikliği, 613625; FSF8D2 (Kombine Faktör 5 ve Faktör 8 Eksikliği) (MCFD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor V and factor VIII, combined deficiency of, 613625; FSF8D2 (Combined deficiency of factor V and factor VII) (MCFD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCFD2	MCFD2, FSF8D2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor V and factor VII combined deficiency 2 (FSF8D2) [MIM:613625]: A blood coagulation disorder characterized by bleeding symptoms similar to those in hemophilia or parahemophilia, that are caused by single deficiency of FV or FVIII, respectively. The most common symptoms are epistaxis, menorrhagia, and excessive bleeding during or after trauma. Plasma levels of coagulation factors V and VIII are in the range of 5 to 30% of normal. {ECO:0000269|PubMed:12717434, ECO:0000269|PubMed:18685427, ECO:0000269|PubMed:20491958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Faktör V Cambridge Mutasyonu (1001G>C) (ARG306THR) (R306T) (. geni) (Dizi Analizi) (Postnatal)	Factor V Cambridge mutation (1001G>C) (ARG306THR) (R306T) (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Faktör 5 Eksikliği, 227400, Otozomal Resesif (Konjenital Faktör 5 Eksikliği) (F5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor V deficiency, 227400, Autosomal recessive (Congenital factor V deficiency) (F5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Faktör V HongKong Mutasyonu (1090A>G) (ARG306GLY) (R306G) (. geni) (Dizi Analizi) (Postnatal)	Factor V HongKong mutation (1090A>G) (ARG306GLY) (R306G) (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Faktör V Leiden Mutasyonu (1691G>A) (ARG506GLN) (R506Q) (. geni) (Dizi Analizi) (Postnatal)	Factor V Leiden mutation (1691G>A) (ARG506GLN) (R506Q) (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Faktör V R2 Mutasyonu (4070A>G) (. geni) (Dizi Analizi) (Postnatal)	Factor V R2 mutation (4070A>G) (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Faktör 7 Eksikliği, 227500, Otozomal Resesif (Konjenital Faktör 7 Eksikliği) (F7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor VII deficiency, 227500, Autosomal recessive (Congenital factor VII deficiency) (F7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F7	F7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor VII deficiency (FA7D) [MIM:227500]: A hemorrhagic disease with variable presentation. The clinical picture can be very severe, with the early occurrence of intracerebral hemorrhages or repeated hemarthroses, or, in contrast, moderate with cutaneous-mucosal hemorrhages (epistaxis, menorrhagia) or hemorrhages provoked by a surgical intervention. Finally, numerous subjects are completely asymptomatic despite very low factor VII levels. {ECO:0000269|PubMed:10862079, ECO:0000269|PubMed:11091194, ECO:0000269|PubMed:11129332, ECO:0000269|PubMed:12472587, ECO:0000269|PubMed:14717781, ECO:0000269|PubMed:1634227, ECO:0000269|PubMed:18976247, ECO:0000269|PubMed:19432927, ECO:0000269|PubMed:19751712, ECO:0000269|PubMed:2070047, ECO:0000269|PubMed:21206266, ECO:0000269|PubMed:21372693, ECO:0000269|PubMed:7974346, ECO:0000269|PubMed:7981691, ECO:0000269|PubMed:8043443, ECO:0000269|PubMed:8204879, ECO:0000269|PubMed:8364544, ECO:0000269|PubMed:8652821, ECO:0000269|PubMed:8844208, ECO:0000269|PubMed:8883260, ECO:0000269|PubMed:8940045, ECO:0000269|PubMed:9414278, ECO:0000269|PubMed:9452082, ECO:0000269|PubMed:9574180, Note=The disease is caused	EDTA Blood Sample
Faktör 8 Eksikliği (F8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	FACTOR VIII DEFICIENCY (F8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F8	F8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Faktör 10 Eksikliği, 227600, Otozomal Resesif (Konjenital Faktör 10 Eksikliği) (F10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor X deficiency, 227600, Autosomal recessive (Congenital factor X deficiency) (F10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F10	F10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor X deficiency (FA10D) [MIM:227600]: A hemorrhagic disease with variable presentation. Affected individuals can manifest prolonged nasal and mucosal hemorrhage, menorrhagia, hematuria, and occasionally hemarthrosis. Some patients do not have clinical bleeding diathesis. {ECO:0000269|PubMed:10468877, ECO:0000269|PubMed:10746568, ECO:0000269|PubMed:11248282, ECO:0000269|PubMed:11728527, ECO:0000269|PubMed:12574802, ECO:0000269|PubMed:12945883, ECO:0000269|PubMed:15075089, ECO:0000269|PubMed:15650540, ECO:0000269|PubMed:17393015, ECO:0000269|PubMed:19135706, ECO:0000269|PubMed:1973167, ECO:0000269|PubMed:1985698, ECO:0000269|PubMed:25313940, ECO:0000269|PubMed:2790181, ECO:0000269|PubMed:7689871, ECO:0000269|PubMed:7860069, ECO:0000269|PubMed:8529633, ECO:0000269|PubMed:8845463, ECO:0000269|PubMed:8910490}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Faktör 11 Eksikliği, Otozomal Dominant 612416 (Konjenital Faktör 11 Eksikliği) (F11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor XI deficiency, autosomal dominant, 612416 (Congenital factor XI deficiency) (F11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F11	F11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor XI deficiency (FA11D) [MIM:612416]: A hemorrhagic disease characterized by reduced levels and activity of factor XI resulting in moderate bleeding symptoms, usually occurring after trauma or surgery. Patients usually do not present spontaneous bleeding but women can present with menorrhagia. Hemorrhages are usually moderate. {ECO:0000269|PubMed:10027710, ECO:0000269|PubMed:10606881, ECO:0000269|PubMed:11895778, ECO:0000269|PubMed:15026311, ECO:0000269|PubMed:15180874, ECO:0000269|PubMed:1547342, ECO:0000269|PubMed:15953011, ECO:0000269|PubMed:16607084, ECO:0000269|PubMed:18056151, ECO:0000269|PubMed:21457405, ECO:0000269|PubMed:21668437, ECO:0000269|PubMed:21998818, ECO:0000269|PubMed:22016685, ECO:0000269|PubMed:22159456, ECO:0000269|PubMed:22322133, ECO:0000269|PubMed:2813350, ECO:0000269|PubMed:7689972, ECO:0000269|PubMed:7888672, ECO:0000269|PubMed:9401068, ECO:0000269|PubMed:9787168}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Faktör 12 Eksikliği, 234000, Otozomal Resesif (Konjenital Faktör 12 Eksikliği) (F12 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Factor XII deficiency, 234000, Autosomal recessive (Congenital factor XII deficiency) (F12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F12	F12, HAF, HAE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor XII deficiency (FA12D) [MIM:234000]: An asymptomatic anomaly of in vitro blood coagulation. Its diagnosis is based on finding a low plasma activity of the factor in coagulating assays. It is usually only accidentally discovered through pre-operative blood tests. Factor XII deficiency is divided into two categories, a cross-reacting material (CRM)-negative group (negative F12 antigen detection) and a CRM-positive group (positive F12 antigen detection). (ECO:0000269|PubMed:10361128, ECO:0000269|PubMed:11776307, ECO:0000269|PubMed:15205584, ECO:0000269|PubMed:15617741, ECO:0000269|PubMed:2510163, ECO:0000269|PubMed:2882793, ECO:0000269|PubMed:8049433, ECO:0000269|PubMed:8528215, ECO:0000269|PubMed:9354665). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Faktör 12 Eksikliği, 234000, Otozomal Resesif (Konjenital Faktör 12 Eksikliği) (MLPA)	Factor XII deficiency, 234000, Autosomal recessive (Congenital factor XII deficiency) (MLPA)	F12	F12, HAF, HAE3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Factor XII deficiency (FA12D) [MIM:234000]: An asymptomatic anomaly of in vitro blood coagulation. Its diagnosis is based on finding a low plasma activity of the factor in coagulating assays. It is usually only accidentally discovered through pre-operative blood tests. Factor XII deficiency is divided into two categories, a cross-reacting material (CRM)-negative group (negative F12 antigen detection) and a CRM-positive group (positive F12 antigen detection). (ECO:0000269|PubMed:10361128, ECO:0000269|PubMed:11776307, ECO:0000269|PubMed:15205584, ECO:0000269|PubMed:15617741, ECO:0000269|PubMed:2510163, ECO:0000269|PubMed:2882793, ECO:0000269|PubMed:8049433, ECO:0000269|PubMed:8528215, ECO:0000269|PubMed:9354665). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Faktör XIII V34L Mutasyonu (177G>T) (VAL34LEU) (V34L) (.gen) (Dizi Analizi) (Postnatal)	Factor XIII V34L mutation (177G>T) (VAL34LEU) (V34L) (.gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Faktör 13A Eksikliği, 613225, Otozomal Resesif (Konjenital Faktör 13 Eksikliği) (F13A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor XIIIA deficiency, 613225, Autosomal recessive (Congenital factor XIII deficiency) (F13A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F13A1	F13A1, F13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor XIII subunit A deficiency (FA13AD) [MIM:613225]: An autosomal recessive hematologic disorder characterized by a life-long bleeding tendency, impaired wound healing and spontaneous abortion in affected women. (ECO:0000269|PubMed:1353995, ECO:0000269|PubMed:24286209, ECO:0000269|PubMed:24329762, ECO:0000269|PubMed:24899649). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Faktör 13B Eksikliği, 613235, Otozomal Resesif (Konjenital Faktör 13 Eksikliği) (F13B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Factor XIIIB deficiency, 613235, Autosomal recessive (Congenital factor XIII deficiency) (F13B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F13B	F13B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor XIII subunit B deficiency (FA13BD) [MIM:613235]: An autosomal recessive hematologic disorder characterized by a life-long bleeding tendency, impaired wound healing and spontaneous abortion in affected women. (ECO:0000269|PubMed:11313256, ECO:0000269|PubMed:20331752, ECO:0000269|PubMed:26247044, ECO:0000269|PubMed:8324218). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diş Sürmesinde Eksiklik, Primer, 125350, Otozomal Dominant; PFE (Primer Diş Sürme Yetersizliği) (PTH1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Failure of tooth eruption, primary, 125350, Autosomal dominant; PFE (Primary failure of tooth eruption) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary failure of tooth eruption (PFE) [MIM:125350]: Rare condition that has high penetrance and variable expressivity and in which tooth retention occurs without evidence of any obvious mechanical interference. Instead, malfunction of the eruptive mechanism itself appears to cause nonankylosed permanent teeth to fail to erupt, although the eruption pathway has been cleared by bone resorption. (ECO:0000269	PubMed:19061984). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Adenomatöz Polipozis 3, 616415, Otozomal Resesif; FAP3 (Atenüe Ailesel Adenomatöz Polipozis) (NTHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial adenomatous polyposis 3, 616415, Autosomal recessive; FAP3 (Attenuated familial adenomatous polyposis) (NTHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTHL1	NTHL1, OCTS3, FAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial adenomatous polyposis 3 (FAP3) [MIM:616415]: A form of familial adenomatous polyposis, a condition characterized by the development of multiple colorectal adenomatous polyps, benign neoplasms derived from glandular epithelium. Some affected individuals may develop colorectal carcinoma. (ECO:0000269	PubMed:25938944). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Adenomatöz Polipozis 4, 617100, Otozomal Resesif; FAP4 (MSH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial adenomatous polyposis 4, 617100, Autosomal recessive; FAP4 (MSH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH3	MSH3, FAP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel meme-over kanseri sendromu 1 (BRCA1 17q21.31) (MLPA)	Familial breast-over cancer syndrome 1 (BRCA1 17q21.31) (MLPA)	BRCA1 17q21.31	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel meme-over kanseri sendromu 1 (BRCA1) (MLPA)	Familial breast-over cancer syndrome 1 (BRCA1) (MLPA)	BRCA1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel meme-over kanseri sendromu 2 (BRCA2 13q13.1) (MLPA)	Familial breast-over cancer syndrome 2 (BRCA2 13q13.1) (MLPA)	BRCA2 13q13.1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel meme-over kanseri sendromu 2 (BRCA2) (MLPA)	Familial breast-over cancer syndrome 2 (BRCA2) (MLPA)	BRCA2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel kronik pankreatit paneli (PRSS1 7q34, SPINK1 5q32) (MLPA)	Familial chronic pancreatitis panel (PRSS1 7q34, SPINK1 5q32) (MLPA)	PRSS1 7q34, SPINK1 5q32	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel Soğuk Otoinflamatuvar Sendromu 2, 611762, Otozomal Dominant; FCAS2 (Kalıtsal Periyodik Ateş Sendromu, NLRP12 İlişkili) (NLRP12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial cold autoinflammatory syndrome 2, 611762, Autosomal dominant; FCAS2 (NLRP12-associated hereditary periodic fever syndrome) (NLRP12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP12	NLRP12, NALP12, PYPAF7, RNO, FCAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial cold autoinflammatory syndrome 2 (FCAS2) [MIM:611762]: A rare autosomal dominant systemic inflammatory disease characterized by recurrent episodes of maculopapular rash associated with arthralgias, myalgias, fever and chills, swelling of the extremities, and conjunctivitis after generalized exposure to cold. (ECO:0000269	PubMed:18230725). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Soğuk Otoinflamatuvar Sendromu 3, 614468, Otozomal Dominant; FCAS3 (PLCG2 İlişkili Antikor Eksikliği ve İmmün Disregülasyon) (PLCG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial cold autoinflammatory syndrome 3, 614468, Autosomal dominant; FCAS3 (PLCG2-associated antibody deficiency and immune dysregulation) (PLCG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCG2	PLCG2, FCAS3, APLAID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial cold autoinflammatory syndrome 3 (FCAS3) [MIM:614468]: An autosomal dominant immune disorder characterized by the development of cutaneous urticaria, erythema, and pruritis in response to cold exposure. Affected individuals have variable additional immunologic defects, including antibody deficiency, decreased numbers of B-cells, defective B-cells, increased susceptibility to infection, and increased risk of autoimmune disorders. (ECO:0000269	PubMed:22236196). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ailesel Soğuk Otoinflamatuvar Sendrom 4, 616115, Otozomal Dominant; FCAS4 (Ailesel Soğuk Ürtiker) (NLRC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial cold autoinflammatory syndrome 4, 616115, Autosomal dominant; FCAS4 (Familial cold urticaria) (NLRC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRC4	NLRC4, CARD12, CLAN, IPAF, AIFEC, FCAS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ailesel Soğuğa Bağlı İnflamatuvar Sendrom 1, 120100, Otozomal Dominant; FCAS1 (Ailesel Soğuk Ürtiker) (NLRP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Familial cold-induced inflammatory syndrome 1, 120100, Autosomal dominant; FCAS1 (Familial cold urticaria) (NLRP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP3	NLRP3, CIAS1, FCU, FCAS1, NALP3, PYPAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial cold autoinflammatory syndrome 1 (FCAS1) [MIM:120100]: A rare autosomal dominant systemic inflammatory disease characterized by recurrent episodes of maculopapular rash associated with arthralgias, myalgias, fever and chills, swelling of the extremities, and conjunctivitis after generalized exposure to cold. Rarely, some patients may also develop late-onset renal amyloidosis. (ECO:0000269|PubMed:11687797, ECO:0000269|PubMed:11992256, ECO:0000269|PubMed:12355493, ECO:0000269|PubMed:12522564, ECO:0000269|PubMed:15593220, ECO:0000269|PubMed:17284928, ECO:0000269|PubMed:24952504). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ailesel uzun QT sendromu paneli (KCNQ1 11p15.5, KCNH2 7q35) (MLPA)	Familial long QT syndrome panel (KCNQ1 11p15.5, KCNH2 7q35) (MLPA)	KCNQ1 11p15.5, KCNH2 7q35	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ailesel Akdeniz Ateşi, AD, 134610, Otozomal Dominant (Ailesel Akdeniz Ateşi) (MEFV gen) (Tüm Gen Dizi Analizi) (Postnatal)	Familial Mediterranean fever, AD, 134610, Autosomal dominant (Familial Mediterranean fever) (MEFV gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEFV	MEFV, MEF, FMF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Akdeniz Ateşi, AD, 134610, Otozomal Dominant (Ailesel Akdeniz Ateşi) (MLPA)	Familial Mediterranean fever, AD, 134610, Autosomal dominant (Familial Mediterranean fever) (MLPA)	MEFV	MEFV, MEF, FMF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Akdeniz Ateşi, AR, 249100, Otozomal Resesif; FMF (Ailesel Akdeniz Ateşi) (MEFV gen) (Tüm Gen Dizi Analizi) (Postnatal)	Familial Mediterranean fever, AR, 249100, Autosomal recessive; FMF (Familial Mediterranean fever) (MEFV gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEFV	MEFV, MEF, FMF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ailesel Akdeniz Ateşi, AR, 249100, Otozomal Resesif; FMF (Ailesel Akdeniz Ateşi) (MLPA)	Familial Mediterranean fever, AR, 249100, Autosomal recessive; FMF (Familial Mediterranean fever) (MLPA)	MEFV	MEFV, MEF, FMF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Fanconi Anemisi, Komplementasyon Grubu A, 227650, Otozomal Resesif; FANCA (Fanconi Anemisi) (FANCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group A, 227650, Autosomal recessive; FANCA (Fanconi anemia) (FANCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCA	FANCA, FACA, FA1, FA, FAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia, complementation group A (FANCA) [MIM:227650]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:10094191, ECO:0000269|PubMed:10210316, ECO:0000269|PubMed:10521298, ECO:0000269|PubMed:10807541, ECO:0000269|PubMed:11091222, ECO:0000269|PubMed:17924555, ECO:0000269|PubMed:9371798, ECO:0000269|PubMed:9399890, ECO:0000269|PubMed:9929978). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu A, 227650, Otozomal Resesif; FANCA (Fanconi Anemisi) (MLPA)	Fanconi anemia, complementation group A, 227650, Autosomal recessive; FANCA (Fanconi anemia) (MLPA)	FANCA	FANCA, FACA, FA1, FA, FAA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fanconi anemia, complementation group A (FANCA) [MIM:227650]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:10094191, ECO:0000269|PubMed:10210316, ECO:0000269|PubMed:10521298, ECO:0000269|PubMed:10807541, ECO:0000269|PubMed:11091222, ECO:0000269|PubMed:17924555, ECO:0000269|PubMed:9371798, ECO:0000269|PubMed:9399890, ECO:0000269|PubMed:9929978). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu A, 227650, Otozomal Resesif; FANCA (Fanconi Anemisi) (Prenatal) (FANCA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group A, 227650, Autosomal recessive; FANCA (Fanconi anemia) (FANCA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCA	FANCA, FACA, FA1, FA, FAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia, complementation group A (FANCA) [MIM:227650]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:10094191, ECO:0000269|PubMed:10210316, ECO:0000269|PubMed:10521298, ECO:0000269|PubMed:10807541, ECO:0000269|PubMed:11091222, ECO:0000269|PubMed:17924555, ECO:0000269|PubMed:9371798, ECO:0000269|PubMed:9399890, ECO:0000269|PubMed:9929978). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu A, 227650, Otozomal Resesif; FANCA (Fanconi Anemisi) (Prenatal) (MLPA)	Fanconi anemia, complementation group A, 227650, Autosomal recessive; FANCA (Fanconi anemia) (Prenatal) (MLPA)	FANCA	FANCA, FACA, FA1, FA, FAA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia, complementation group A (FANCA) [MIM:227650]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:10094191, ECO:0000269|PubMed:10210316, ECO:0000269|PubMed:10521298, ECO:0000269|PubMed:10807541, ECO:0000269|PubMed:11091222, ECO:0000269|PubMed:17924555, ECO:0000269|PubMed:9371798, ECO:0000269|PubMed:9399890, ECO:0000269|PubMed:9929978). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu B, 300514; FANCB (Fanconi Anemisi) (FANCB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group B, 300514; FANCB (Fanconi anemia) (FANCB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCB	FAAP95, FAAP90, FLJ34064, FANCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu B, 300514; FANCB (Fanconi Anemisi) (Prenatal) (FANCB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group B, 300514; FANCB (Fanconi anemia) (FANCB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCB	FAAP95, FAAP90, FLJ34064, FANCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group B (FANCB) [MIM:300514]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. Some severe FANCB cases manifest features of VACTERL syndrome with hydrocephalus. (ECO:0000269	PubMed:16679491). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu C, 227645, Otozomal Resesif; FANCC (Fanconi Anemisi) (FANCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group C, 227645, Autosomal recessive; FANCC (Fanconi anemia) (FANCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCC	FANCC, FACC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group C (FANCC) [MIM:227645]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:11520787, ECO:0000269|PubMed:15299030, ECO:0000269|PubMed:1574115, ECO:0000269|PubMed:8128956, ECO:0000269|PubMed:8499901, ECO:0000269|PubMed:8844212, ECO:0000269|PubMed:9242535). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu C, 227645, Otozomal Resesif; FANCC (Fanconi Anemisi) (Prenatal) (FANCC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group C, 227645, Autosomal recessive; FANCC (Fanconi anemia) (FANCC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCC	FANCC, FACC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group C (FANCC) [MIM:227645]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269|PubMed:11520787, ECO:0000269|PubMed:15299030, ECO:0000269|PubMed:1574115, ECO:0000269|PubMed:8128956, ECO:0000269|PubMed:8499901, ECO:0000269|PubMed:8844212, ECO:0000269|PubMed:9242535). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu D1, 605724, Otozomal Resesif; FANCD1, (Kalıtsal Kanser-Predispozisyon Sendromu, Bialelik BRCA2 Mutasyonlarına Bağlı) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group D1, 605724, Autosomal recessive; FANCD1 (Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu D1, 605724, Otozomal Resesif; FANCD1, (Kalıtsal Kanser-Predispozisyon Sendromu, Bialelik BRCA2 Mutasyonlarına Bağlı) (MLPA)	Fanconi anemia, complementation group D1, 605724, Autosomal recessive; FANCD1 (Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu D1, 605724, Otozomal Resesif; FANCD1, (Kalıtsal Kanser-Predispozisyon Sendromu, Bialelik BRCA2 Mutasyonlarına Bağlı) (Prenatal) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group D1, 605724, Autosomal recessive; FANCD1 (Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu D1, 605724, Otozomal Resesif; FANCD1, (Kalıtsal Kanser-Predispozisyon Sendromu, Bialelik BRCA2 Mutasyonlarına Bağlı) (Prenatal) (MLPA)	Fanconi anemia, complementation group D1, 605724, Autosomal recessive; FANCD1 (Inherited cancer-predisposing syndrome due to biallelic BRCA2 mutations) (Prenatal) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu D2, 227646, Otozomal Resesif; FANCD2 (Fanconi Anemisi) (FANCD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group D2, 227646, Autosomal recessive; FANCD2 (Fanconi anemia) (FANCD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCD2	FANCD2, FANCD, FACD, FAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group D2 (FANCD2) [MIM:227646]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269	PubMed:11239453). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu D2, 227646, Otozomal Resesif; FANCD2 (Fanconi Anemisi) (Prenatal) (FANCD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group D2, 227646, Autosomal recessive; FANCD2 (Fanconi anemia) (FANCD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCD2	FANCD2, FANCD, FACD, FAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group D2 (FANCD2) [MIM:227646]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. (ECO:0000269	PubMed:11239453). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Fanconi Anemisi, Komplementasyon E Grubu, 600901, Otozomal Resesif, FANCE (Fanconi Anemisi) (FANCE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group E, 600901, Autosomal recessive; FANCE (Fanconi anemia) (FANCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCE	FANCE, FACE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group E (FANCE) [MIM:600901]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:11001585, ECO:0000269|PubMed:17924555}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon E Grubu, 600901, Otozomal Resesif, FANCE (Fanconi Anemisi) (Prenatal) (FANCE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group E, 600901, Autosomal recessive; FANCE (Fanconi anemia) (FANCE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCE	FANCE, FACE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group E (FANCE) [MIM:600901]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:11001585, ECO:0000269|PubMed:17924555}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu F, 603467, FANCF (Fanconi Anemisi) (FANCF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group F, 603467, FANCF (Fanconi anemia) (FANCF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCF	FANCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group F (FANCF) [MIM:603467]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:10615118}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu F, 603467, FANCF (Fanconi Anemisi) (Prenatal) (FANCF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group F, 603467, FANCF (Fanconi anemia) (FANCF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCF	FANCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group F (FANCF) [MIM:603467]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:10615118}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu G, 614082, FANCG (Fanconi Anemisi) (FANCG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group G, 614082, FANCG (Fanconi anemia) (FANCG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCG	XRCC9, FANCG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu G, 614082, FANCG (Fanconi Anemisi) (Prenatal) (FANCG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group G, 614082, FANCG (Fanconi anemia) (FANCG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCG	XRCC9, FANCG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu I, 609053, FANCI (Fanconi Anemisi) (FANCI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group I, 609053, FANCI (Fanconi anemia) (FANCI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCI	FANCI, KIAA1794	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group I (FANCI) [MIM:609053]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:17452773}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu I, 609053, FANCI (Fanconi Anemisi) (Prenatal) (FANCI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group I, 609053, FANCI (Fanconi anemia) (FANCI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCI	FANCI, KIAA1794	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group I (FANCI) [MIM:609053]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:17452773}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu J, 609054, FANCJ (Fanconi Anemisi) (BRIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group J, 609054, FANCJ (Fanconi anemia) (BRIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRIP1	BRIP1, BACH1, FANCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu J 609054, FANCJ (Fanconi Anemisi) (Prenatal) (BRIP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group J, 609054, FANCJ (Fanconi anemia) (BRIP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRIP1	BRIP1, BACH1, FANCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu L, 614083, Otozomal Resesif, FANCL (Fanconi Anemisi) (FANCL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group L, 614083, Autosomal recessive; FANCL (Fanconi anemia) (FANCL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FANCL	PHF9, FANCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu L, 614083, Otozomal Resesif, FANCL (Fanconi Anemisi) (Prenatal) (FANCL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group L, 614083, Autosomal recessive; FANCL (Fanconi anemia) (FANCL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FANCL	PHF9, FANCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group L (FANCL) [MIM:614083]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:12973351}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu N, 610832, FANCN (Fanconi Anemisi) (PALB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group N, 610832, FANCN (Fanconi anemia) (PALB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PALB2	PALB2, FANCN, PNCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu N, 610832, FANCN (Fanconi Anemisi) (Prenatal) (PALB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group N, 610832, FANCN (Fanconi anemia) (PALB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PALB2	PALB2, FANCN, PNCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu O, 613390, Otozomal Resesif, FANCO (Fanconi Anemisi) (RAD51C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group O, 613390, Autosomal recessive; FANCO (Fanconi anemia) (RAD51C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51C	RAD51C, FANCO, BROVCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group O (FANCO) [MIM:613390]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:20400963}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu O, 613390, Otozomal Resesif, FANCO (Fanconi Anemisi) (Prenatal) (RAD51C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group O, 613390, Autosomal recessive; FANCO (Fanconi anemia) (RAD51C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAD51C	RAD51C, FANCO, BROVCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group O (FANCO) [MIM:613390]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269|PubMed:20400963}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu P, 613951, Otozomal Resesif, FANCP (Fanconi Anemisi) (SLX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group P, 613951, Autosomal recessive; FANCP (Fanconi anemia) (SLX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLX4	SLX4, BTBD12, MUS312, KIAA1784, KIAA1987, FANCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group P (FANCP) [MIM:613951]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. Some individuals affected by Fanconi anemia of complementation group P have skeletal anomalies. {ECO:0000269|PubMed:21240275, ECO:0000269|PubMed:21240277}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu P 613951, Otozomal Resesif, FANCP (Fanconi Anemisi) (Prenatal) (SLX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group P, 613951, Autosomal recessive; FANCP (Fanconi anemia) (SLX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLX4	SLX4, BTBD12, MUS312, KIAA1784, KIAA1987, FANCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group P (FANCP) [MIM:613951]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. Some individuals affected by Fanconi anemia of complementation group P have skeletal anomalies. {ECO:0000269|PubMed:21240275, ECO:0000269|PubMed:21240277}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu Q, 615272, Otozomal Resesif, FANCQ (Fanconi Anemisi) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group Q, 615272, Autosomal recessive; FANCQ (Fanconi anemia) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu Q, 615272, Otozomal Resesif, FANCQ (Fanconi Anemisi) (Prenatal) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group Q, 615272, Autosomal recessive; FANCQ (Fanconi anemia) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu R, 617244, Otozomal Dominant, FANCR (Fanconi Anemisi) (RAD51 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group R, 617244, Autosomal dominant; FANCR (Fanconi anemia) (RAD51 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51	RAD51A, RECA, MRMV2, FANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Fanconi Anemisi, Komplementasyon Grubu R, 617244, Otozomal Dominant; FANCR (Fanconi Anemisi) (Prenatal) (RAD51 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group R, 617244, Autosomal dominant; FANCR (Fanconi anemia) (RAD51 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAD51	RAD51A, RECA, MRMV2, FANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grup T, 616435, Otozomal Resesif; FANCT (Fanconi Anemisi) (UBE2T geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group T, 616435, Autosomal recessive; FANCT (Fanconi anemia) (UBE2T gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBE2T	UBE2T, HSPC150, FANCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi anemia complementation group T (FANCT) [MIM:616435]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269	PubMed:26046368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grup T, 616435, Otozomal Resesif; FANCT (Fanconi Anemisi) (Prenatal) (UBE2T geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group T, 616435, Autosomal recessive; FANCT (Fanconi anemia) (UBE2T gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBE2T	UBE2T, HSPC150, FANCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi anemia complementation group T (FANCT) [MIM:616435]: A disorder affecting all bone marrow elements and resulting in anemia, leukopenia and thrombopenia. It is associated with cardiac, renal and limb malformations, dermal pigmentary changes, and a predisposition to the development of malignancies. At the cellular level it is associated with hypersensitivity to DNA-damaging agents, chromosomal instability (increased chromosome breakage) and defective DNA repair. {ECO:0000269	PubMed:26046368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu U, 617247, Otozomal Resesif; FANCU (Fanconi Anemisi) (XRCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group U, 617247, Autosomal recessive; FANCU (Fanconi anemia) (XRCC2 gene) (Sequence Analysis-All Codno Exons) (Postnatal)	XRCC2	XRCC2, FANCU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemi, Komplementasyon Grubu U, 617247, Otozomal Resesif; FANCU (Fanconi Anemisi) (Prenatal) (XRCC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group U, 617247, Autosomal recessive; FANCU (Fanconi anemia) (XRCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XRCC2	XRCC2, FANCU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu 5, 617243, Otozomal Resesif; FANCV (Fanconi Anemisi) (MADZL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi anemia, complementation group V, 617243, Autosomal recessive; FANCV (Fanconi anemia) (MADZL2 gene) (Sequence Analysis-All Codno Exons) (Postnatal)	MADZL2	MADZL2, MAD2B, FANCV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Anemisi, Komplementasyon Grubu 5, 617243, Otozomal Resesif; FANCV (Fanconi Anemisi) (Prenatal) (MADZL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi anemia, complementation group V, 617243, Autosomal recessive; FANCV (Fanconi anemia) (MADZL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MADZL2	MADZL2, MAD2B, FANCV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Renotubular Sendrom 2, 613388, Otozomal Resesif; FRTS2 (Primer Fanconi Sendromu) (SLC34A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi renotubular syndrome 2, 613388, Autosomal recessive; FRTS2 (Primary Fanconi syndrome) (SLC34A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC34A1	SLC34A1, SLC17A2, NPT2, NPHLOP1, FRTS2, HCINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi renotubular syndrome 2 (FRTS2) [MIM:613388]: A disease due to a generalized dysfunction of the proximal kidney tubule resulting in decreased solute and water reabsorption. Patients have polydipsia and polyuria with phosphaturia, glycosuria and aminoaciduria. They may develop hypophosphatemic rickets or osteomalacia, acidosis and a tendency toward dehydration. Some eventually develop renal insufficiency. {ECO:0000269	PubMed:20335586}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Renotubular Sendrom 2, 613388, Otozomal Resesif; FRTS2 (Primer Fanconi Sendromu) (SLC34A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi renotubular syndrome 2, 613388, Autosomal recessive; FRTS2 (Primary Fanconi syndrome) (SLC34A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC34A1	SLC34A1, SLC17A2, NPT2, NPHLOP1, FRTS2, HCINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi renotubular syndrome 2 (FRTS2) [MIM:613388]: A disease due to a generalized dysfunction of the proximal kidney tubule resulting in decreased solute and water reabsorption. Patients have polydipsia and polyuria with phosphaturia, glycosuria and aminoaciduria. They may develop hypophosphatemic rickets or osteomalacia, acidosis and a tendency toward dehydration. Some eventually develop renal insufficiency. {ECO:0000269	PubMed:20335586}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Renotubular Sendrom 3, 615605, Otozomal Dominant (Primer Fanconi Sendromu) (EHHADH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi renotubular syndrome 3, 615605, Autosomal dominant; FRTS3 (Primary Fanconi syndrome) (EHHADH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EHHADH	EHHADH, PBFE, LBFP, FRTS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi renotubular syndrome 3 (FRTS3) [MIM:615605]: A disease due to a generalized dysfunction of the proximal kidney tubule resulting in decreased solute and water reabsorption. Patients have polydipsia and polyuria with phosphaturia, glycosuria and aminoaciduria. They may develop hypophosphatemic rickets or osteomalacia, acidosis and a tendency toward dehydration. Some eventually develop renal insufficiency. {ECO:0000269	PubMed:24401050}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi Renotubular Sendrom 3, 615605, Otozomal Dominant (Primer Fanconi Sendromu) (Prenatal) (EHHADH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi renotubular syndrome 3, 615605, Autosomal dominant; FRTS3 (Primary Fanconi syndrome) (EHHADH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EHHADH	EHHADH, PBFE, LBFP, FRTS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi renotubular syndrome 3 (FRTS3) [MIM:615605]: A disease due to a generalized dysfunction of the proximal kidney tubule resulting in decreased solute and water reabsorption. Patients have polydipsia and polyuria with phosphaturia, glycosuria and aminoaciduria. They may develop hypophosphatemic rickets or osteomalacia, acidosis and a tendency toward dehydration. Some eventually develop renal insufficiency. {ECO:0000269	PubMed:24401050}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Renotubular Sendrom 4, MODY, 616026, Otozomal Dominant; FRTS4 (Renal Kist - Diyabet Sendromu) (HNF4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026, Autosomal dominant; FRTS4 (Renal cysts and diabetes syndrome) (HNF4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fanconi Renotubular Sendrom 4, MODY, 616026, Otozomal Dominant; FRTS4 (Renal Kist - Diyabet Sendromu) (MLPA)	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026, Autosomal dominant; FRTS4 (Renal cysts and diabetes syndrome) (MLPA)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Fanconi Renotubular Sendrom 4, MODY, 616026, Otozomal Dominant; FRTS4 (Renal Kist - Diyabet Sendromu) (Prenatal) (HNF4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026, Autosomal dominant; FRTS4 (Renal cysts and diabetes syndrome) (HNF4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi Renotubular Sendrom 4, MODY, 616026, Otozomal Dominant; FRTS4 (Renal Kist - Diyabet Sendromu) (Prenatal) (MLPA)	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young, 616026, Autosomal dominant; FRTS4 (Renal cysts and diabetes syndrome) (Prenatal) (MLPA)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fanconi-Bickel Sendromu, 227810, Otozomal Resesif; FBS (Glikojen Depo Hastalığı, GLUT2 Eksikliğine Bağlı) (SLC2A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fanconi-Bickel syndrome, 227810, Autosomal recessive; FBS (Glycogen storage disease due to GLUT2 deficiency) (SLC2A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A2	SLC2A2, GLUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi-Bickel syndrome (FBS) [MIM:227810]: Rare, well- defined clinical entity, inherited in an autosomal recessive mode and characterized by hepatorenal glycogen accumulation, proximal renal tubular dysfunction, and impaired utilization of glucose and galactose. {ECO:0000269|PubMed:10987651, ECO:0000269|PubMed:11044475}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fanconi-Bickel Sendromu, 227810, Otozomal Resesif; FBS (Glikojen Depo Hastalığı, GLUT2 Eksikliğine Bağlı) (SLC2A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fanconi-Bickel syndrome, 227810, Autosomal recessive; FBS (Glycogen storage disease due to GLUT2 deficiency) (SLC2A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC2A2	SLC2A2, GLUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fanconi-Bickel syndrome (FBS) [MIM:227810]: Rare, well- defined clinical entity, inherited in an autosomal recessive mode and characterized by hepatorenal glycogen accumulation, proximal renal tubular dysfunction, and impaired utilization of glucose and galactose. {ECO:0000269|PubMed:10987651, ECO:0000269|PubMed:11044475}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Farber Lipogranulomatosis, 228000, Otozomal Resesif; FRBRL (Farber Hastalığı) (ASAH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Farber lipogranulomatosis, 228000, Autosomal recessive; FRBRL (Farber disease) (ASAH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASAH1	ASAH1, AC, SMAPME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Farber Lipogranulomatosis, 228000, Otozomal Resesif; FRBRL (Farber Hastalığı) (Prenatal) (ASAH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Farber lipogranulomatosis, 228000, Autosomal recessive; FRBRL (Farber disease) (ASAH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASAH1	ASAH1, AC, SMAPME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fasiyoskapulohumeral Müskiler Distrofi 2, Dijenik, 158901; FSHD2 (Fasioskapulohumeral Distrofi) (SMCHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fasioscapulohumeral muscular dystrophy 2, digenic, 158901; FSHD2 (Facioscapulohumeral dystrophy) (SMCHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMCHD1	SMCHD1, KIAA0650, BAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Facioscapulohumeral muscular dystrophy 2 (FSHD2) [MIM:158901]: A degenerative muscle disease characterized by slowly progressive weakness of the muscles of the face, upper-arm, and shoulder girdle. The onset of symptoms usually occurs in the first or second decade of life. Affected individuals usually present with impairment of upper extremity elevation. This tends to be followed by facial weakness, primarily involving the orbicularis oris and orbicularis oculi muscles. {ECO:0000269	PubMed:23143600}. Note=The disease is caused by mutations affecting the gene represented in this entry. SMCHD1 mutations lead to DUX4 expression in somatic tissues, including muscle cells, when an haplotype on chromosome 4 is permissive for DUX4 expression. Ectopic expression of DUX4 in skeletal muscle activates the expression of stem cell and germline genes, and, when overexpressed in somatic cells, DUX4 can ultimately lead to cell death.;	EDTA Blood Sample
Fatal Ailesel İnsomnia; FI (Fatal Ailesel İnsomnia) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	FATAL FAMILIAL INSOMNIA; FFI (Fatal familial insomnia) (PRNP gene) (Sequence Analysis-All Codno Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yağlı Karaciğer, Akut, Gebelik, 609016, Otozomal Resesif (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (HADHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fatty liver, acute, of pregnancy, 609016, Autosomal recessive (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Favism, 134700, Otozomal Dominant (G6PD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Favism, 134700, Autosomal dominant (G6PD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PD	G6PD, G6PD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fazio-Londe Hastalığı, 211500, Otozomal Resesif (Çocukluk Çağı Progresif Bulber Paralizi) (SLC52A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fazio-Londe disease, 211500, Autosomal recessive (Progressive bulbar paralysis of childhood) (SLC52A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC52A3	SLC52A3, C20orf54, RFT2, BVVLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fazio-Londe disease (FALOND) [MIM:211500]: A rare neurological disease characterized by progressive weakness of the muscles innervated by cranial nerves of the lower brain stem. It may present in childhood with severe neurological deterioration with hypotonia, respiratory insufficiency leading to premature death, or later in life with bulbar weakness which progresses to involve motor neurons throughout the neuroaxis. Clinical manifestations include dysarthria, dysphagia, facial weakness, tongue weakness, and fasciculations of the tongue and facial muscles. {ECO:0000269	PubMed:21110228}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Fazio-Londe Hastalığı, 211500, Otozomal Resesif (Çocukluk Çağı Progresif Bulber Paralizi) (Prenatal) (SLC52A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fazio-Londe disease, 211500, Autosomal recessive (Progressive bulbar paralysis of childhood) (SLC52A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC52A3	SLC52A3, C20orf54, RFT2, BVVLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fazio-Londe disease (FALOND) [MIM:211500]: A rare neurological disease characterized by progressive weakness of the muscles innervated by cranial nerves of the lower brain stem. It may present in childhood with severe neurological deterioration with hypotonia, respiratory insufficiency leading to premature death, or later in life with bulbar weakness which progresses to involve motor neurons throughout the neuroaxis. Clinical manifestations include dysarthria, dysphagia, facial weakness, tongue weakness, and fasciculations of the tongue and facial muscles. {ECO:0000269|PubMed:21110228}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FCGR bağlı immun yetmezlikler (Miks 1) (FCGR genleri 1q23.3) (MLPA)/ FCGR bağlı immun yetmezlikler (Miks 2) (FCGR genleri 1q23.3) (MLPA)	FCGR-linked immunodeficiencies (Mix 1) (FCGR genes 1q23.3) (MLPA) / FCGR-linked immunodeficiencies (Mix 2) (FCGR genes 1q23.3) (MLPA)	FCGR genleri 1q23.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
FCGR bağlı immun yetmezlikler (Miks 1) (FCGR genleri 1q23.3) (MLPA)/ FCGR bağlı immun yetmezlikler (Miks 2) (FCGR genleri 1q23.3) (MLPA) (Prenatal)	FCGR-linked immunodeficiencies (Mix 1) (FCGR genes 1q23.3) (MLPA) / FCGR-linked immunodeficiencies (Mix 2) (FCGR genes 1q23.3) (MLPA) (Prenatal)	FCGR genleri 1q23.3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 11, 614418, Otozomal Resesif; FEB11 (Ailesel Mezial Temporal Lob Epilepsisi-Febril Nöbetler) (CPA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Febrile seizures, familial, 11, 614418, Autosomal recessive; FEB11 (Familial mesial temporal lobe epilepsy with febrile seizures) (CPA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPA6	CPA6, CPAH, ETL5, FEB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 3A, 604403, Otozomal Dominant; GEFSP2 (Jeneralize Epilepsi-Febril Nöbetler Art) (SCN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Febrile seizures, familial, 3A, 604403, Autosomal dominant; GEFSP2 (Generalized epilepsy with febrile seizures-plus) (SCN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18566737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117753	EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 3A, 604403, Otozomal Dominant; GEFSP2 (Jeneralize Epilepsi-Febril Nöbetler Art) (MLPA)	Febrile seizures, familial, 3A, 604403, Autosomal dominant; GEFSP2 (Generalized epilepsy with febrile seizures-plus) (MLPA)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18566737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117753	EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 3B, 613863, Otozomal Dominant; GEFSP7 (Jeneralize Epilepsi-Febril Nöbetler Art) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Febrile seizures, familial, 3B, 613863, Autosomal dominant; GEFSP7 (Generalized epilepsy with febrile seizures-plus) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 4, 604352, Otozomal Dominant; FEB4 (Usher Sendromu) (ADGRV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Febrile seizures, familial, 4, 604352, Autosomal dominant; FEB4 (Usher syndrome) (ADGRV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRV1	ADGRV1, GPR98, MASS1, VLGR1, KIAA0586, FEB4, USH2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ateşli (Febril) Nöbetler, Ailesel, 8, 611277, Otozomal Dominant; GEFSP3 (Jeneralize Epilepsi-Febril Nöbetler Art) (GABRG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Febrile seizures, familial, 8, 611277, Autosomal dominant; GEFSP3 (Generalized epilepsy with febrile seizures-plus) (GABRG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABRG2	GABRG2, GEFSP3, CAE2, ECA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fechtner Sendromu, 153640, Otozomal Dominant; FTNS (Fechtner Sendromu) (MYH9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fechtner syndrome, 153640, Autosomal dominant; FTNS (Fechtner syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10972359, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fechtner Sendromu, 153640, Otozomal Dominant; FTNS (Fechtner Sendromu) (Prenatal) (MYH9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fechtner syndrome, 153640, Autosomal dominant; FTNS (Fechtner syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10972359, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Feingold Sendromu 1, 164280, Otozomal Dominant; FGLDS1 (Feingold Sendromu) (MYCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Feingold syndrome 1, 164280, Autosomal dominant; FGLDS1 (Feingold syndrome) (MYCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYCN	MYCN, NMYC, ODED, MODED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Feingold syndrome 1 (FGLDS1) [MIM:164280]: A syndrome characterized by variable combinations of esophageal and duodenal atresias, microcephaly, learning disability, mental retardation, and limb malformations. Hand and foot abnormalities may include hypoplastic thumbs, clinodactyly of second and fifth fingers, syndactyly (characteristically between second and third and fourth and fifth toes), and shortened or absent middle phalanges. Cardiac and renal malformations, vertebral anomalies, and deafness have also been described. {ECO:0000269|PubMed:15821734, ECO:0000269|PubMed:16906565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Feingold Sendromu 1, 164280, Otozomal Dominant; FGLDS1 (Feingold Sendromu) (Prenatal) (MYCN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Feingold syndrome 1, 164280, Autosomal dominant; FGLDS1 (Feingold syndrome) (MYCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYCN	MYCN, NMYC, ODED, MODED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Feingold syndrome 1 (FGLDS1) [MIM:164280]: A syndrome characterized by variable combinations of esophageal and duodenal atresias, microcephaly, learning disability, mental retardation, and limb malformations. Hand and foot abnormalities may include hypoplastic thumbs, clinodactyly of second and fifth fingers, syndactyly (characteristically between second and third and fourth and fifth toes), and shortened or absent middle phalanges. Cardiac and renal malformations, vertebral anomalies, and deafness have also been described. {ECO:0000269|PubMed:15821734, ECO:0000269|PubMed:16906565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Feingold Sendromu 2, 614326, Otozomal Dominant; FGLDS2 (Feingold Sendromu) (MIR17HG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Feingold syndrome 2, 614326, Autosomal dominant; FGLDS2 (Feingold syndrome) (MIR17HG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIR17HG	MIR17HG, MIRH1, MIHG1, MIRHG1, C13orf25, FGLDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Feingold Sendromu 2, 614326, Otozomal Dominant; FGLDS2 (Feingold Sendromu) (Prenatal) (MIR17HG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Feingold syndrome 2, 614326, Autosomal dominant; FGLDS2 (Feingold syndrome) (MIR17HG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MIR17HG	MIR17HG, MIRH1, MIHG1, MIRHG1, C13orf25, FGLDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif (Fetal Akinezi Deformasyon Sekansı) (RAPSN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive (Fetal akinesia deformation sequence) (RAPSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAPSN	RAPSN, CMS1D, CMS11, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:18179903, ECO:0000269|PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif (Fetal Akinezi Deformasyon Sekansı) (MUSK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive (Fetal akinesia deformation sequence) (MUSK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUSK	MUSK, CMS9, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:25537362, ECO:0000269|PubMed:25612909}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif (Fetal Akinezi Deformasyon Sekansı) (Prenatal) (RAPSN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive (Fetal akinesia deformation sequence) (RAPSN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAPSN	RAPSN, CMS1D, CMS11, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:18179903, ECO:0000269|PubMed:18252236}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif (Fetal Akinezi Deformasyon Sekansı) (Prenatal) (MUSK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive (Fetal akinesia deformation sequence) (MUSK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MUSK	MUSK, CMS9, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:25537362, ECO:0000269|PubMed:25612909}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif, FADS (Fetal Akinezi Deformasyon Sekansı) (DOK7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive, FADS (Fetal akinesia deformation sequence) (DOK7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOK7	DOK7, C4orf25, CMS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 10 (CMS10) [MIM:254300]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS10 is an autosomal recessive, post-synaptic form characterized by a typical 'limb girdle' pattern of muscle weakness with small, simplified neuromuscular junctions but normal acetylcholine receptor and acetylcholinesterase function. {ECO:0000269|PubMed:16917026, ECO:0000269|PubMed:17439981, ECO:0000269|PubMed:20012313, ECO:0000269|PubMed:22661499}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fetal Akinezi Deformasyon Sekansı 208150, Otozomal Resesif, FADS (Fetal Akinezi Deformasyon Sekansı) (Prenatal) (DOK7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal akinesia deformation sequence, 208150, Autosomal recessive, FADS (Fetal akinesia deformation sequence) (DOK7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOK7	DOK7, C4orf25, CMS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 10 (CMS10) [MIM:254300]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS10 is an autosomal recessive, post-synaptic form characterized by a typical 'limb girdle' pattern of muscle weakness with small, simplified neuromuscular junctions but normal acetylcholine receptor and acetylcholinesterase function. {ECO:0000269|PubMed:16917026, ECO:0000269|PubMed:17439981, ECO:0000269|PubMed:20012313, ECO:0000269|PubMed:22661499}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Kandan Kromozom Analizi	Fetal blood sample - Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Heparinli tüp, heparinli enjektör	*	.	.	15	30	20	Prenatal		Heparinized tube, heparinized injector
Fetal Hemoglobin Kantitatif Özellik Lokusu 1, 141749, Otozomal Dominant (Kalıtsal Fetal Hemoglobin Persistansı–Beta-Talasemi Sendromu) (HBG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal hemoglobin quantitative trait locus 1, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome) (HBG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBG2	HBG2, TNCY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusu 1, 141749, Otozomal Dominant (Kalıtsal Fetal Hemoglobin Persistansı–Beta-Talasemi Sendromu) (HBG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal hemoglobin quantitative trait locus 1, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome) (HBG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBG1	HBG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusu 1, 141749, Otozomal Dominant (Kalıtsal Fetal Hemoglobin Persistansı–Beta-Talasemi Sendromu) (Prenatal) (HBG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal hemoglobin quantitative trait locus 1, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome) (HBG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBG2	HBG2, TNCY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusu 1, 141749, Otozomal Dominant (Kalıtsal Fetal Hemoglobin Persistansı–Beta-Talasemi Sendromu) (Prenatal) (HBG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal hemoglobin quantitative trait locus 1, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-beta-thalassemia syndrome) (HBG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBG1	HBG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusu 1; Hbfqtl1 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusü 1; Hbfqtl1 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (MLPA)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusü 1; Hbfqtl1 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (Prenatal) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Hemoglobin Kantitatif Özellik Lokusü 1; Hbfqtl1 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (Prenatal) (MLPA)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 1; HBFQTL1 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Hemoglobin Kantitatif Karakter Loküsü 6; Hbfqtl6 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (KLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 6; HBFQTL6 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (KLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fetal Hemoglobin Kantitatif Karakter Loküsü 6; Hbfqtl6 (Fetal Hemoglobin Persistansı–Orak Hücre Hastalığı Sendromu) (Prenatal) (KLF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	FETAL HEMOGLOBIN QUANTITATIVE TRAIT LOCUS 6; HBFQTL6 (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (KLF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLF1	KLF1, EKLF, INLU, HBFQTL6, CDAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetal Hidantoin Sendromu (EPHX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fetal hydantoin syndrome (EPHX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHX1	EPHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fetal Hidantoin Sendromu (Prenatal) (EPHX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fetal hydantoin syndrome (EPHX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPHX1	EPHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FG Sendromu 2, 300321; FGS2 (Konjenital Kısa Bağırsak Sendromu) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	FG syndrome 2, 300321; FGS2 (Congenital short bowel syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
FG Sendromu 2, 300321; FGS2 (Konjenital Kısa Bağırsak Sendromu) (MLPA)	FG syndrome 2, 300321; FGS2 (Congenital short bowel syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
FG Sendromu 2, 300321; FGS2 (Konjenital Kısa Bağırsak Sendromu) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	FG syndrome 2, 300321; FGS2 (Congenital short bowel syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FG Sendromu 2, 300321; FGS2 (Konjenital Kısa Bağırsak Sendromu) (Prenatal) (MLPA)	FG syndrome 2, 300321; FGS2 (Congenital short bowel syndrome) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FG Sendromu 4, 300422; FGS4 (CASK geni) (Tüm Gen Dizi Analizi) (Postnatal)	FG syndrome 4, 300422; FGS4 (CASK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269|PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
FG Sendromu 4, 300422; FGS4 (Prenatal) (CASK geni) (Tüm Gen Dizi Analizi) (Prenatal)	FG syndrome 4, 300422; FGS4 (CASK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269|PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

FGFR1 Amplifikasyon (FISH)	FGFR1 amplification (FISH)	8p11.23	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinle gömü bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
FGFR1 Breakapart (FISH)	FGFR1 Breakapart (FISH)	8p11.23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
FGFR2 Breakapart (FISH)	FGFR2 Breakapart (FISH)	.	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinle gömü bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Deri Fibroblast kültürü	Fibroblast culture	.	.	Doku Kültürü/ Tissue Culture	Merkezimizden temin edilen transport besi yeri içinde/ Acil durumlarda steril serum fizyolojik içinde		15	30	.	.	21	Postnatal	.	Transport Medium suppliedfrom our center/ In urgency in Sterile Serum Physiologic
Fibroblast Kültürü	Fibroblast culture	.	.	Doku Kültürü/ Tissue Culture	Merkezimizden temin edilen transport besi yeri içinde/ Acil durumlarda steril serum fizyolojik içinde		15	30	.	.	21	Postnatal	.	Transport Medium suppliedfrom our center/ In urgency in Sterile Serum Physiologic
Fibrokalküloz Pankreatik Diyabet, Yatkınlık, 608189, Otozomal Resesif, Otozomal Dominant (Tropikal Pankreatit) (SPINK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrocalculous pancreatic diabetes, susceptibility to, 608189, Autosomal recessive, Autosomal dominant (Tropical pancreatitis) (SPINK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fibrokalküloz Pankreatik Diyabet, Yatkınlık, 608189, Otozomal Resesif, Otozomal Dominant (Tropikal Pankreatit) (MLPA)	Fibrocalculous pancreatic diabetes, susceptibility to, 608189, Autosomal recessive, Autosomal dominant (Tropical pancreatitis) (MLPA)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Fibrokondrogenezis 1, 228520, Otozomal Resesif, FBCG1 (Fibrokondrogenezis) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrochondrogenesis 1, 228520, Autosomal recessive; FBCG1 (Fibrochondrogenesis) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fibrokondrogenezis 1, 228520, Otozomal Resesif; FBCG1 (Fibrokondrogenezis) (Prenatal) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fibrochondrogenesis 1, 228520, Autosomal recessive; FBCG1 (Fibrochondrogenesis) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fibrokondrogenezis 2, 614524, Otozomal Resesif, Otozomal Dominant (Fibrokondrogenezis) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrochondrogenesis 2, 614524, Autosomal recessive, Autosomal dominant (Fibrochondrogenesis) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fibrokondrogenezis 2, 614524, Otozomal Resesif, Otozomal Dominant (Fibrokondrogenezis) (Prenatal) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fibrochondrogenesis 2, 614524, Autosomal recessive, Autosomal dominant (Fibrochondrogenesis) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fibrodisplasia Ossificans Progressiva, 135100, Otozomal Dominant; FOP (Fibrodisplazia Ossifikans Progressiva) (ACVR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrodysplasia ossificans progressiva, 135100, Autosomal dominant; FOP (Fibrodysplasia ossificans progressiva) (ACVR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACVR1	ACVR1, ACVRLK2, ALK2, FOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fibrodisplasia Ossificans Progressiva, 135100, Otozomal Dominant; FOP (Fibrodisplazia Ossifikans Progressiva) (Prenatal) (ACVR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fibrodysplasia ossificans progressiva, 135100, Autosomal dominant; FOP (Fibrodysplasia ossificans progressiva) (ACVR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACVR1	ACVR1, ACVRLK2, ALK2, FOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fibromatozis, Dijital, 1, 135300, Otozomal Dominant; GINGF1 (Ailesel Gingival Fibromatozi (SOS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibromatosis, gingival, 1, 135300, Autosomal dominant; GINGF1 (Hereditary gingival fibromatosis) (SOS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOS1	SOS1, GINGF, GF1, HGF, NS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekstraoküler Kas Fibrozis, Konjenital, 1, 135700, Otozomal Dominant; CFEOM1 (Ekstraoküler Kasların Konjenital Fibrozisi) (KIF21A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrosis of extraocular muscles, congenital, 1, 135700, Autosomal dominant; CFEOM1 (Congenital fibrosis of extraocular muscles) (KIF21A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF21A	KIF21A, KIAA1708, FEOM1, CFEOM1, CFEOM3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekstraoküler Kas Fibrozis, Konjenital, 2, 602078, Otozomal Resesif; CFEOM2 (Ekstraoküler Kasların Konjenital Fibrozisi) (PHOX2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrosis of extraocular muscles, congenital, 2, 602078, Autosomal recessive; CFEOM2 (Congenital fibrosis of extraocular muscles) (PHOX2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHOX2A	PHOX2A, ARIX, CFEOM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fibrosis of extraocular muscles, congenital, 2 (CFEOM2) [MIM:602078]: A congenital ocular motility disorder marked by restrictive ophthalmoplegia affecting extraocular muscles innervated by the oculomotor and/or trochlear nerves. It is clinically characterized by anchoring of the eyes in downward gaze, ptosis, and backward tilt of the head. Congenital fibrosis of extraocular muscles type 2 may result from the defective development of the oculomotor (nIII), trochlear (nIV) and abducens (nVI) cranial nerve nuclei. {ECO:0000269	PubMed:11600883}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Ekstraoküler Kas Fibrozis, Konjenital, 3A, 600638, Otozomal Dominant; CFEOM3A (Ekstraoküler Kasların Konjenital Fibrozisi) (TUBB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrosis of extraocular muscles, congenital, 3A, 600638, Autosomal dominant; CFEOM3A (Congenital fibrosis of extraocular muscles) (TUBB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB3	TUBB3, TUBB4, CFEOM3A, CDCBM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fibrosis of extraocular muscles, congenital, 3A (CFEOM3A) [MIM:600638]: A congenital ocular motility disorder marked by restrictive ophthalmoplegia affecting extraocular muscles innervated by the oculomotor and/or trochlear nerves. It is clinically characterized by anchoring of the eyes in downward gaze, ptosis, and backward tilt of the head. Congenital fibrosis of extraocular muscles type 3 presents as a non-progressive, autosomal dominant disorder with variable expression. Patients may be bilaterally or unilaterally affected, and their oculo-motility defects range from complete ophthalmoplegia (with the eyes fixed in a hypo- and exotropic position), to mild asymptomatic restrictions of ocular movement. Ptosis, refractive error, amblyopia, and compensatory head positions are associated with the more severe forms of the disorder. In some cases, this ocular phenotype is accompanied by additional features including developmental delay, corpus callosum agenesis, basal ganglia dysmorphism, facial weakness, polyneuropathy. {ECO:0000269	PubMed:20074521}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ekstraoküler Kas Fibrozu, Konjenital, 3B, 135700, Otozomal Dominant (Ekstraoküler Kasların Konjenital Fibrozisi) (KIF21A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrosis of extraocular muscles, congenital, 3B, 135700, Autosomal dominant (Congenital fibrosis of extraocular muscles) (KIF21A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF21A	KIF21A, KIAA1708, FEOM1, CFEOM1, CFEOM3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fibrosis of extraocular muscles, congenital, 1 (CFEOM1) [MIM:135700]: A congenital ocular motility disorder marked by restrictive ophthalmoplegia affecting extraocular muscles innervated by the oculomotor and/or trochlear nerves. It is clinically characterized by anchoring of the eyes in downward gaze, ptosis, and backward tilt of the head. Patients affected by congenital fibrosis of extraocular muscles type 1 show an absence of the superior division of the oculomotor nerve (cranial nerve III) and corresponding oculomotor subnuclei. {ECO:0000269|PubMed:14585441, ECO:0000269|PubMed:16157808, ECO:0000269|PubMed:17511870, ECO:0000269|PubMed:24715754}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Ekstraoküler Kas Fibrozis, Konjenital, 5, 616219, Otozomal Resesif, CFEOM5 (Duane Retraksiyon Sendromu) (COL25A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fibrosis of extraocular muscles, congenital, 5, 616219, Autosomal recessive; CFEOM5 (Duane retraction syndrome) (COL25A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL25A1	COL25A1, CLAC, CFEOM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fibrosis of extraocular muscles, congenital, 5 (CFEOM5) [MIM:616219]: An ocular motility disorder characterized by congenital dysinnervation of various cranial nerves to ocular muscles. Clinical features are ophthalmoplegia, anchoring of the eyes in downward gaze, ptosis, and backward tilt of the head. {ECO:0000269	PubMed:25500261}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Filippi Sendromu, 272440, Otozomal Resesif; FLPIS (Filippi Sendromu) (CKAP2L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Filippi syndrome, 272440, Autosomal recessive; FLPIS (Filippi syndrome) (CKAP2L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CKAP2L	CKAP2L, RADMIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Filippi syndrome (FLPIS) [MIM:272440]: A rare disorder characterized by microcephaly, pre- and postnatal growth failure, syndactyly, and distinctive facial features, including a broad nasal bridge and underdeveloped alae nasi. Some affected individuals have intellectual disability, seizures, undescended testicles in males, and teeth and hair abnormalities. {ECO:0000269	PubMed:25439729}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Filippi Sendromu, 272440, Otozomal Resesif; FLPIS (Filippi Sendromu) (Prenatal) (CKAP2L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Filippi syndrome, 272440, Autosomal recessive; FLPIS (Filippi syndrome) (CKAP2L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CKAP2L	CKAP2L, RADMIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Filippi syndrome (FLPIS) [MIM:272440]: A rare disorder characterized by microcephaly, pre- and postnatal growth failure, syndactyly, and distinctive facial features, including a broad nasal bridge and underdeveloped alae nasi. Some affected individuals have intellectual disability, seizures, undescended testicles in males, and teeth and hair abnormalities. {ECO:0000269	PubMed:25439729}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FILS Sendromu, 615139, Otozomal Resesif (Fasiyal Dismorfizm-İrmiksi Yetmezlik-Livedo-Kısa Boy Sendromu) (POLE geni) (Tüm Gen Dizi Analizi) (Postnatal)	FILS syndrome, 615139, Autosomal recessive (Facial dysmorphism-immunodeficiency-livedo-short stature syndrome) (POLE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLE	POLE1, CRCS12, FILS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FILS Sendromu, 615139, Otozomal Resesif (Fasiyal Dismorfizm-İrmiksi Yetmezlik-Livedo-Kısa Boy Sendromu) (Prenatal) (POLE geni) (Tüm Gen Dizi Analizi) (Prenatal)	FILS syndrome, 615139, Autosomal recessive (Facial dysmorphism-immunodeficiency-livedo-short stature syndrome) (POLE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLE	POLE1, CRCS12, FILS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FIP1L1/ CHIC2/ PDGFRA (CHIC2 Delesyonu/ FIP1L1 - PDGFRA Füzyonu) (FISH)	FIP1L1/CHIC2/PDGFRA Deletion/Fusion (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Balık Gözü Hastalığı, 136120, Otozomal Resesif; FED (LCAT Eksikliği) (LCAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fish-eye disease, 136120, Autosomal recessive; FED (LCAT deficiency) (LCAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LCAT	LCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fish-eye disease (FED) [MIM:136120]: A disorder of lipoprotein metabolism due to partial lecithin-cholesterol acyltransferase deficiency that affects only alpha-LCAT activity. FED is characterized by low plasma HDL and corneal opacities due to accumulation of cholesterol deposits in the cornea ('fish-eye'). {ECO:0000269|PubMed:1516702, ECO:0000269|PubMed:1571050, ECO:0000269|PubMed:15994445, ECO:0000269|PubMed:17327840, ECO:0000269|PubMed:21901787, ECO:0000269|PubMed:8620346, ECO:0000269|PubMed:9261271}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Balık Gözü Hastalığı, 136120, Otozomal Resesif; FED (LCAT Eksikliği) (Prenatal) (LCAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fish-eye disease, 136120, Autosomal recessive; FED (LCAT deficiency) (LCAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LCAT	LCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fish-eye disease (FED) [MIM:136120]: A disorder of lipoprotein metabolism due to partial lecithin-cholesterol acyltransferase deficiency that affects only alpha-LCAT activity. FED is characterized by low plasma HDL and corneal opacities due to accumulation of cholesterol deposits in the cornea ('fish-eye'). {ECO:0000269|PubMed:1516702, ECO:0000269|PubMed:1571050, ECO:0000269|PubMed:15994445, ECO:0000269|PubMed:17327840, ECO:0000269|PubMed:21901787, ECO:0000269|PubMed:8620346, ECO:0000269|PubMed:9261271}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fletcher Faktörü (Prekallikrein) Eksikliği, 612423, Otozomal Resesif (Konjenital Prekallikrein Eksikliği) (KLKB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fletcher factor (prekallikrein) deficiency, 612423, Autosomal recessive (Congenital prekallikrein deficiency) (KLKB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLKB1	KLKB1, KLK3, PKKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prekallikrein deficiency (PKK deficiency) [MIM:612423]: This disorder is a blood coagulation defect. {ECO:0000269|PubMed:14652634, ECO:0000269|PubMed:17598838}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Floating-Harbor Sendromu, 136140, Otozomal Dominant; FLHS (Floating-Harbor Sendromu) (SRCAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Floating-Harbor syndrome, 136140, Autosomal dominant; FLHS (Floating-Harbor syndrome) (SRCAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRCAP	SRCAP, SWR1, KIAA0309, FLHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Floating-Harbor syndrome (FLHS) [MIM:136140]: A rare genetic disorder characterized by proportionate short stature, delayed bone age, delayed speech development, and typical facial features. The face is triangular with deep-set eyes, long eyelashes, bulbous nose, wide columella, short philtrum, and thin lips. (ECO:0000269	PubMed:22265015). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Floating-Harbor Sendromu, 136140, Otozomal Dominant; FLHS (Floating-Harbor Sendromu) (Prenatal) (SRCAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Floating-Harbor syndrome, 136140, Autosomal dominant; FLHS (Floating-Harbor syndrome) (SRCAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRCAP	SRCAP, SWR1, KIAA0309, FLHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Floating-Harbor syndrome (FLHS) [MIM:136140]: A rare genetic disorder characterized by proportionate short stature, delayed bone age, delayed speech development, and typical facial features. The face is triangular with deep-set eyes, long eyelashes, bulbous nose, wide columella, short philtrum, and thin lips. (ECO:0000269	PubMed:22265015). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
FLT3 Aktive Edici Mutasyon (Ekzon14 - 20) (Dizi Analizi) (FLT3 geni) (Postnatal)	FLT3 activation (Exon14- 20) (FLT3 gene) (Sequence Analysis) (Postnatal)	FLT3	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269|PubMed:11090077, ECO:0000269|PubMed:11290608, ECO:0000269|PubMed:11442493, ECO:0000269|PubMed:14504097, ECO:0000269|PubMed:16266983, ECO:0000269|PubMed:18305215, ECO:0000269|PubMed:8946930, ECO:0000269|PubMed:9737679). Note=The gene represented in this entry may be involved in disease pathogenesis. Somatic mutations that lead to constitutive activation of FLT3 are frequent in AML patients. These mutations fall into two classes, the most common being in-frame internal tandem duplications of variable length in the juxtamembrane region that disrupt the normal regulation of the kinase activity. Likewise, point mutations in the activation loop of the kinase domain can result in a constitutively activated kinase.;	EDTA Blood Sample
FMF (Ailevi Akdeniz Ateşi) (Sık Görülen 12 Mutasyon) (MEFV geni) (Dizi Analizi) (Postnatal)	FMF (Familial Mediterranean Fever) (12 frequent mutations) (MEFV gene) (Dizi Analizi) (Postnatal)	MEFV	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
FMF (Ailevi Akdeniz Ateşi) (MEFV geni) (Tüm Gen Dizi Analizi) (Postnatal)	FMF (Familial Mediterranean Fever) (MEFV Gene) (All coding exons - Sequence analysis) (MEFV gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEFV	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial Mediterranean fever, autosomal dominant (ADFMF) [MIM:134610]: A hereditary periodic fever syndrome characterized by periodic fever, serosal inflammation and pain in the abdomen, chest or joints as seen also in the autosomal recessive form of the disease. It is associated with reactive renal amyloidosis and characterized by colchicine unresponsiveness. (ECO:0000269|PubMed:10787449, ECO:0000269|PubMed:14679589). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fokal Dermal Hipoplazi, 305600, X'e Bağlı Dominant; FDH (Fokal Dermal Hipoplazi) (PORCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Focal dermal hypoplasia, 305600, X-linked dominant; FDH (Focal dermal hypoplasia) (PORCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PORCN	PORCN, PORC, DHOF, FODH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fokal Dermal Hipoplazi, 305600, X'e Bağlı Dominant; FDH (Fokal Dermal Hipoplazi) (Prenatal) (PORCN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Focal dermal hypoplasia, 305600, X-linked dominant; FDH (Focal dermal hypoplasia) (PORCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PORCN	PORCN, PORC, DHOF, FODH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fokal Fasiyal Dermal Displazi 3, Setleis Tipi, 227260, Otozomal Resesif; FFDD3 (Fokal Fasiyal Dermal Displazi Tip 3) (TWIST2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Focal facial dermal dysplasia 3, Setleis type, 227260, Autosomal recessive; FFDD3 (Focal facial dermal dysplasia type III) (TWIST2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST2	TWIST2, DERMO1, SETLSS, FFDD3, BBRSAY, AMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fokal Fasiyal Dermal Displazi 3, Setleis Tipi, 227260, Otozomal Resesif; FFDD3 (Fokal Fasiyal Dermal Displazi Tip 3) (Prenatal) (TWIST2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Focal facial dermal dysplasia 3, Setleis type, 227260, Autosomal recessive; FFDD3 (Focal facial dermal dysplasia type III) (TWIST2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST2	TWIST2, DERMO1, SETLSS, FFDD3, BBRSAY, AMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fokal Fasiyal Dermal Displazi 4, 614974, Otozomal Resesif; FFDD4 (Fokal Fasiyal Dermal Displazi) (CYP26C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Focal facial dermal dysplasia 4, 614974, Autosomal recessive; FFDD4 (Focal facial dermal dysplasia) (CYP26C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP26C1	CYP26C1, FFDD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal facial dermal dysplasia 4 (FFDD4) [MIM:614974]: A form of focal facial dermal dysplasia, a group of developmental defects characterized by bitemporal or preauricular skin lesions resembling aplasia cutis congenita. Skin defects occur at the sites of facial fusion during embryogenesis, with temporal lesions situated at the junction between the frontonasal and maxillary facial prominences, and preauricular lesions at the meeting point of the maxillary and mandibular prominences. The ectodermal lesions show consistent histologic abnormalities: atrophy and flattening of the epidermis, replacement of the dermis by loose connective tissue, reduced levels of fragmented elastic tissue and absence of the subcutaneous tissues and adnexal structures. FFDD4 is characterized by isolated, preauricular skin lesions. (ECO:0000269	PubMed:23161670). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fokal Fasiyal Dermal Displazi 4, 614974, Otozomal Resesif; FFDD4 (Fokal Fasiyal Dermal Displazi) (Prenatal) (CYP26C1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Focal facial dermal dysplasia 4, 614974, Autosomal recessive; FFDD4 (Focal facial dermal dysplasia) (CYP26C1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP26C1	CYP26C1, FFDD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Focal facial dermal dysplasia 4 (FFDD4) [MIM:614974]: A form of focal facial dermal dysplasia, a group of developmental defects characterized by bitemporal or preauricular skin lesions resembling aplasia cutis congenita. Skin defects occur at the sites of facial fusion during embryogenesis, with temporal lesions situated at the junction between the frontonasal and maxillary facial prominences, and preauricular lesions at the meeting point of the maxillary and mandibular prominences. The ectodermal lesions show consistent histologic abnormalities: atrophy and flattening of the epidermis, replacement of the dermis by loose connective tissue, reduced levels of fragmented elastic tissue and absence of the subcutaneous tissues and adnexal structures. FFDD4 is characterized by isolated, preauricular skin lesions. (ECO:0000269	PubMed:23161670). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental 8, 616032, Otozomal Dominant; FSGS8 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (ANLN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental 8, 616032, Autosomal dominant; FSGS8 (Familial idiopathic steroid-resistant nephrotic syndrome) (ANLN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANLN	ANLN, FSFS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 8 (FSGS8) [MIM:616032]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. (ECO:0000269	PubMed:24676636). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental 9, 616220, Otozomal Resesif; FSGS9 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendromi) (CRB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental 9, 616220, Autosomal recessive; FSGS9 (Familial idiopathic steroid-resistant nephrotic syndrome) (CRB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRB2	CRB2, FSGS9, VMCKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Folat Malabsorpsiyonu, Kalıtsal, 229050, Otozomal Resesif (Kalıtsal Folat Malabsorpsiyonu) (SLC46A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Folate malabsorption, hereditary, 229050, Autosomal recessive (Hereditary folate malabsorption) (SLC46A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC46A1	SLC46A1, HCP1, PCFT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Beyin Defektleri (TDGF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Forebrain defects (TDGF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TDGF1	TDGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ön Beyin Defektleri (Prenatal) (TDGF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Forebrain defects (TDGF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TDGF1	TDGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Foveal Hipoplazi 1, 136520, Otozomal Dominant; FVH1 (Foveal Hipoplazi-Presenil Katarakt Sendromu) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Foveal hypoplasia 1, 136520, Autosomal dominant; FVH1 (Foveal hypoplasia-presenile cataract syndrome) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Foveal Hipoplazi 1, 136520, Otozomal Dominant; FVH1 (Foveal Hipoplazi-Presenil Katarakt Sendromu) (MLPA)	Foveal hypoplasia 1, 136520, Autosomal dominant; FVH1 (Foveal hypoplasia-presenile cataract syndrome) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGDS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Foveal Hipoplazi 2, + Optik Sinirin Yanlış Rotası ve / Veya Ön Segment Disgenezisi, 609218, Otozomal Resesif; FVH2 (Foveal Hipoplazi-Optik Sinir Çapraz/arma Defekt,- Ön Segment Disgenezisi Sendromu) (SLC38A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Foveal hypoplasia 2, with or without optic nerve misrouting and/or anterior segment dysgenesis, 609218, Autosomal recessive; FVH2 (Foveal hypoplasia-optic nerve decussation defect-anterior segment dysgenesis syndrome) (SLC38A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC38A8	SLC38A8, FVH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frajil X Sendromu, 300624, X'e Bağlı Dominant (Frajil X Sendromu) (Tekrar Sayısı)	Fragile X syndrome, 300624, X-linked dominant (Fragile X syndrome)(Repeat Analysis)	FMR1	FMR1, FRAXA, POF1	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Frajil X Sendromu, 300624, X'e Bağlı Dominant (Frajil X Sendromu) (MLPA)	Fragile X syndrome, 300624, X-linked dominant (Fragile X syndrome) (MLPA)	FMR1	FMR1, FRAXA, POF1	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Frajil X Sendromu, 300624, X'e Bağlı Dominant (Frajil X Sendromu) (Prenatal) (Tekrar Sayısı)	Fragile X syndrome, 300624, X-linked dominant (Fragile X syndrome) (Prenatal)(Repeat Analysis)	FMR1	FMR1, FRAXA, POF1	Tekrar sayısı/ Repeat Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frajil X Sendromu, 300624, X'e Bağlı Dominant (Frajil X Sendromu) (Prenatal) (MLPA)	Fragile X syndrome, 300624, X-linked dominant (Fragile X syndrome) (Prenatal) (MLPA)	FMR1	FMR1, FRAXA, POF1	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frajil-X Tremor / Ataksi Sendromu, 300623, X'e Bağlı Dominant; FXTAS (Frajil-X İle İlişkili Tremor / Ataksi Sendromu) (Tekrar Sayısı)	Fragile X tremor/ataxia syndrome, 300623, X-linked dominant; FXTAS (Fragile X-associated tremor/ataxia syndrome)(Repeat Analysis)	FMR1	FMR1, FRAXA, POF1	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Fragile X tremor/ataxia syndrome (FXTAS) [MIM:300623]: In FXTAS, the expanded repeats range in size from 55 to 200 repeats and are referred to as 'premutations'. Full repeat expansions with greater than 200 repeats results in fragile X mental retardation syndrome [MIM:300624]. Carriers of the premutation typically do not show the full fragile X syndrome phenotype, but comprise a subgroup that may have some physical features of fragile X syndrome or mild cognitive and emotional problems. (ECO:0000269	PubMed:11445641). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Fraji-X Tremor / Ataksi Sendromu, 300623, X'e Bağlı Dominant; FXTAS (Fraji-X ile İlişkili Tremor / Ataksi Sendromu) (MLPA)	Fragile X tremor/ataxia syndrome, 300623, X-linked dominant; FXTAS (Fragile X-associated tremor/ataxia syndrome) (MLPA)	FMR1	FMR1, FRAXA, POF1	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fragile X tremor/ataxia syndrome (FXTAS) [MIM:300623]: In FXTAS, the expanded repeats range in size from 55 to 200 repeats and are referred to as 'premutations'. Full repeat expansions with greater than 200 repeats results in fragile X mental retardation syndrome [MIM:300624]. Carriers of the premutation typically do not show the full fragile X syndrome phenotype, but comprise a subgroup that may have some physical features of fragile X syndrome or mild cognitive and emotional problems. {ECO:0000269	PubMed:11445641}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Frank-Ter Haar Sendromu, 249420, Otozomal Resesif; FTHS (Frank-Ter Haar Sendromu) (SH3PXD2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frank-ter Haar syndrome, 249420, Autosomal recessive; FTHS (Frank-Ter Haar syndrome) (SH3PXD2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH3PXD2B	SH3PXD2B, TKS4, KIAA1295, FTHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frank-Ter Haar syndrome (FTHS) [MIM:249420]: A syndrome characterized by brachycephaly, wide fontanels, prominent forehead, hypertelorism, prominent eyes, macrocornea with or without glaucoma, full cheeks, small chin, bowing of the long bones and flexion deformity of the fingers. {ECO:0000269	PubMed:20137777}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Frank-Ter Haar Sendromu, 249420, Otozomal Resesif; FTHS (Frank-Ter Haar Sendromu) (Prenatal) (SH3PXD2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frank-ter Haar syndrome, 249420, Autosomal recessive; FTHS (Frank-Ter Haar syndrome) (SH3PXD2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SH3PXD2B	SH3PXD2B, TKS4, KIAA1295, FTHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frank-Ter Haar syndrome (FTHS) [MIM:249420]: A syndrome characterized by brachycephaly, wide fontanels, prominent forehead, hypertelorism, prominent eyes, macrocornea with or without glaucoma, full cheeks, small chin, bowing of the long bones and flexion deformity of the fingers. {ECO:0000269	PubMed:20137777}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (GRIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (GRIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIP1	GRIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:22510445}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (FREM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (FREM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FREM2	FREM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:15838507}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (FRAS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (FRAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRAS1	FRAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:23473829}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (Prenatal) (GRIP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (GRIP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIP1	GRIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:22510445}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (Prenatal) (FREM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (FREM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FREM2	FREM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:15838507}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fraser Sendromu, 219000, Otozomal Resesif (Fraser Sendromu) (Prenatal) (FRAS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fraser syndrome, 219000, Autosomal recessive (Fraser syndrome) (FRAS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FRAS1	FRAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fraser syndrome (FRASS) [MIM:219000]: Multisystem malformation usually comprising cryptophthalmos, cutaneous syndactyly, ear abnormalities, renal agenesis and congenital heart defects. {ECO:0000269	PubMed:23473829}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frasier Sendromu, 136680, Otozomal Dominant, Somatik Mutasyon (Frasier Sendromu) (WT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frasier syndrome, 136680, Autosomal dominant, Somatic mutation (Frasier syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ Biopsy of affected site-transport media
Frasier Sendromu, 136680, Otozomal Dominant, Somatik Mutasyon (Frasier Sendromu) (MLPA)	Frasier syndrome, 136680, Autosomal dominant, Somatic mutation (Frasier syndrome) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		20	60	.	.	30	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ Biopsy of affected site-transport media
Frasier Sendromu, 136680, Otozomal Dominant, Somatik Mutasyon (Frasier Sendromu) (Prenatal) (WT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frasier syndrome, 136680, Autosomal dominant, Somatic mutation (Frasier syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frasier Sendromu, 136680, Otozomal Dominant, Somatik Mutasyon (Frasier Sendromu) (Prenatal) (MLPA)	Frasier syndrome, 136680, Autosomal dominant, Somatic mutation (Frasier syndrome) (Prenatal) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frias Sendromu, 609640, Otozomal Dominant (Büyüme Yetersizliği-Brakidaktili-Dismorfizm Sendromu) (2200)	Frias syndrome, 609640, Autosomal dominant (Growth deficiency-brachydactyly-dysmorphism syndrome) (440)	.	FRIASS, DEL14q22, C14DEL q22	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Frias Sendromu, 609640, Otozomal Dominant (Büyüme Yetersizliği-Brakidaktili-Dismorfizm Sendromu) (Prenatal)	Frias syndrome, 609640, Autosomal dominant (Growth deficiency-brachydactyly-dysmorphism syndrome) (Prenatal)	.	FRIASS, DEL14q22, C14DEL q22	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Friedreich Ataksisi-Korunmuş Refleksler, 229300, Otozomal Resesif; FRDA (Friedreich ataksisi) (FXN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Friedreich ataxia with retained reflexes, 229300, Autosomal recessive; FRDA (Friedreich ataxia) (FXN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FXN	FXN, FRDA, FARR, X25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Friedreich ataxia (FRDA) [MIM:229300]: Autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region. {ECO:0000269|PubMed:10732799, ECO:0000269|PubMed:10874325, ECO:0000269|PubMed:9150176, ECO:0000269|PubMed:9779809, ECO:0000269|PubMed:9989622, ECO:0000269|Ref.35, ECO:0000269|Ref.7, ECO:0000269|Ref.8}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Friedreich Ataksisi-Korunmuş Refleksler, 229300, Otozomal Resesif; FRDA (Friedreich Ataksisi) (MLPA)	Friedreich ataxia with retained reflexes, 229300, Autosomal recessive; FRDA (Friedreich ataxia) (MLPA)	FXN	FXN, FRDA, FARR, X25	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Friedreich ataxia (FRDA) [MIM:229300]: Autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region. {ECO:0000269|PubMed:10732799, ECO:0000269|PubMed:10874325, ECO:0000269|PubMed:9150176, ECO:0000269|PubMed:9779809, ECO:0000269|PubMed:9989622, ECO:0000269|Ref.35, ECO:0000269|Ref.7, ECO:0000269|Ref.8}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Friedreich Ataksisi, 229300, Otozomal Resesif (Friedreich Ataksisi) (Tekrar Sayısı)	Friedreich ataxia, 229300, Autosomal recessive (Friedreich ataxia)(Repeat Analysis)	FXN	FXN, FRDA, FARR, X25	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Friedreich ataxia (FRDA) [MIM:229300]: Autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region. {ECO:0000269|PubMed:10732799, ECO:0000269|PubMed:10874325, ECO:0000269|PubMed:9150176, ECO:0000269|PubMed:9779809, ECO:0000269|PubMed:9989622, ECO:0000269|Ref.35, ECO:0000269|Ref.7, ECO:0000269|Ref.8}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Friedreich Ataksisi, 229300, Otozomal Resesif (Friedreich Ataksisi) (MLPA)	Friedreich ataxia, 229300, Autosomal recessive (Friedreich ataxia) (MLPA)	FXN	FXN, FRDA, FARR, X25	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Friedreich ataxia (FRDA) [MIM:229300]: Autosomal recessive, progressive degenerative disease characterized by neurodegeneration and cardiomyopathy it is the most common inherited ataxia. The disorder is usually manifest before adolescence and is generally characterized by incoordination of limb movements, dysarthria, nystagmus, diminished or absent tendon reflexes, Babinski sign, impairment of position and vibratory senses, scoliosis, pes cavus, and hammer toe. In most patients, FRDA is due to GAA triplet repeat expansions in the first intron of the frataxin gene. But in some cases the disease is due to mutations in the coding region. {ECO:0000269|PubMed:10732799, ECO:0000269|PubMed:10874325, ECO:0000269|PubMed:9150176, ECO:0000269|PubMed:9779809, ECO:0000269|PubMed:9989622, ECO:0000269|Ref.35, ECO:0000269|Ref.7, ECO:0000269|Ref.8}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Frontometafizyal Displazi 1, 305620, X'e Bağlı Resesif; FMD1 (Frontometafizyal Displazi) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontometaphyseal dysplasia 1, 305620, X-linked recessive; FMD1 (Frontometaphyseal dysplasia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontometafizyal Displazi 1, 305620, X'e Bağlı Resesif; FMD1 (Frontometafizyal Displazi) (MLPA)	Frontometaphyseal dysplasia 1, 305620, X-linked recessive; FMD1 (Frontometaphyseal dysplasia) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Frontometafizyal Displazi 1, 305620, X'e Bağlı Resesif; FMD1 (Frontometafizyal Displazi) (MLPA)	Frontometaphyseal dysplasia 1, 305620, X-linked recessive; FMD1 (Frontometaphyseal dysplasia) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontometafizyal Displazi 2, 617137, Otozomal Dominant; FMD2 (Frontometafizyal Displazi) (MAP3K7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontometaphyseal dysplasia 2, 617137, Autosomal dominant; FMD2 (Frontometaphyseal dysplasia) (MAP3K7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP3K7	MAP3K7, TAK1, CSCF, FMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontometafizyal Displazi 2, 617137, Otozomal Dominant; FMD2 (Frontometafizyal Displazi) (Prenatal) (MAP3K7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frontometaphyseal dysplasia 2, 617137, Autosomal dominant; FMD2 (Frontometaphyseal dysplasia) (MAP3K7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP3K7	MAP3K7, TAK1, CSCF, FMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontonazal Displazi 1, 136760, Otozomal Resesif; FND1 (Frontonin) (ALX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontonasal dysplasia 1, 136760, Autosomal recessive; FND1 (Frontothiny) (ALX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALX3	ALX3, FND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frontonasal dysplasia 1 (FND1) [MIM:136760]: The term frontonasal dysplasia describes an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism; broadening of the nasal roo;t median facial cleft affecting the nose and/or upper lip and palate; unilateral or bilateral clefting of the alae nasi; lack of formation of the nasal tip; anterior cranium bifidum occultum; a V-shaped or widow's peak frontal hairline. {ECO:0000269|PubMed:19409524}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Frontonazal Displazi 1, 136760, Otozomal Resesif; FND1 (Frontonin) (Prenatal) (ALX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frontonasal dysplasia 1, 136760, Autosomal recessive; FND1 (Frontothiny) (ALX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALX3	ALX3, FND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frontonasal dysplasia 1 (FND1) [MIM:136760]: The term frontonasal dysplasia describes an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism; broadening of the nasal roo;t median facial cleft affecting the nose and/or upper lip and palate; unilateral or bilateral clefting of the alae nasi; lack of formation of the nasal tip; anterior cranium bifidum occultum; a V-shaped or widow's peak frontal hairline. {ECO:0000269|PubMed:19409524}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontonazal Displazi 2, 613451, Otozomal Resesif; FND2 (Frontonazal Displazi-Alopesi ve Genital Anomali) (ALX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontonasal dysplasia 2, 613451, Autosomal recessive; FND2 (Frontonasal dysplasia with alopecia and genital anomaly) (ALX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontonazal Displazi 2, 613451, Otozomal Resesif; FND2 (Frontonazal Displazi-Alopesi ve Genital Anomali) (MLPA)	Frontonasal dysplasia 2, 613451, Autosomal recessive; FND2 (Frontonasal dysplasia with alopecia and genital anomaly) (MLPA)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Frontonazal Displazi 2, 613451, Otozomal Resesif; FND2 (Frontonazal Displazi-Alopesi ve Genital Anomali) (Prenatal) (ALX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frontonasal dysplasia 2, 613451, Autosomal recessive; FND2 (Frontonasal dysplasia with alopecia and genital anomaly) (ALX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontonazal Displazi 2, 613451, Otozomal Resesif; FND2 (Frontonazal Displazi-Alopesi ve Genital Anomali) (Prenatal) (MLPA)	Frontonasal dysplasia 2, 613451, Autosomal recessive; FND2 (Frontonasal dysplasia with alopecia and genital anomaly) (Prenatal) (MLPA)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontonazal Displazi 3, 613456; FND3 (Frontonazal Displazi-Ağır Mikroftalmi-Ağır Fasiyal Yarık Sendromu) (ALX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontonasal dysplasia 3, 613456; FND3 (Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome) (ALX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALX1	ALX1, CART1, FND3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frontonasal dysplasia 3 (FND3) [MIM:613456]: The term frontonasal dysplasia describes an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism; broadening of the nasal roo;t median facial cleft affecting the nose and/or upper lip and palate; unilateral or bilateral clefting of the alae nasi; lack of formation of the nasal tip; anterior cranium bifidum occultum; a V-shaped or widow's peak frontal hairline. {ECO:0000269|PubMed:20451171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Frontonazal Displazi 3, 613456; FND3 (Frontonazal Displazi-Ağır Mikroftalmi-Ağır Fasiyal Yarık Sendromu) (Prenatal) (ALX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Frontonasal dysplasia 3, 613456, FND3 (Frontonasal dysplasia-severe microphthalmia-severe facial clefting syndrome) (ALX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALX1	ALX1, CART1, FND3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frontonasal dysplasia 3 (FND3) [MIM:613456]: The term frontonasal dysplasia describes an array of abnormalities affecting the eyes, forehead and nose and linked to midfacial dysraphia. The clinical picture is highly variable. Major findings include true ocular hypertelorism; broadening of the nasal roo;t median facial cleft affecting the nose and/or upper lip and palate; unilateral or bilateral clefting of the alae nasi; lack of formation of the nasal tip; anterior cranium bifidum occultum; a V-shaped or widow's peak frontal hairline. {ECO:0000269|PubMed:20451171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Frontotemporal Demans ve / Veya Amyotrofik Lateral Skleroz 1, 105550, Otozomal Dominant; FTDAL S1 (Frontotemporal Demans-Motor Nöron Hastalığı) (C9orf72 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal dementia and/or amyotrophic lateral sclerosis 1, 105550, Autosomal dominant; FTDALS1 (Frontotemporal dementia with motor neuron disease) (C9orf72 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C9orf72	C9orf72, FTDALS1, FTDALS, ALSFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontotemporal Demans ve / Veya Amyotrofik Lateral Skleroz 2, 615911, Otozomal Dominant; FTDALS2 (Frontotemporal Demans-Motor Nöron Hastalığı) (CHCHD10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal dementia and/or amyotrophic lateral sclerosis 2, 615911, Autosomal dominant; FTDALS2 (Frontotemporal dementia with motor neuron disease) (CHCHD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHCHD10	CHCHD10, FTDALS2, SMAJ, IMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontotemporal Demans ve / Veya Amyotrofik Lateral Skleroz 3, 616437, Otozomal Dominant; FTDALS3 (Amyotrofik Lateral Skleroz) (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal dementia and/or amyotrophic lateral sclerosis 3, 616437, Autosomal dominant; FTDALS3 (Amyotrophic lateral sclerosis) (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontotemporal Demans ve / Veya Amyotrofik Lateral Sklerozu 4, 616439, Otozomal Dominant; FTDALS4 (Frontotemporal Demans-Motor Nöron Hastalığı) (TBK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal dementia and/or amyotrophic lateral sclerosis 4, 616439, Autosomal dominant; FTDALS4 (Frontotemporal dementia with motor neuron disease) (TBK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBK1	TBK1, NAK, FTDALS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Frontotemporal Lob Dejenerasyonu-Ubiquitin Pozitif İnklüzyonlar, 607485, Otozomal Dominant (Frontotemporal Demans) (GRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal lobar degeneration with ubiquitin-positive inclusions, 607485, Autosomal dominant (Frontotemporal dementia) (GRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRN	GRN, CLN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ubiquitin-positive frontotemporal dementia (UP-FTD) [MIM:607485]: Frontotemporal dementia (FTD) is the second most common cause of dementia in people under the age of 65 years. It is an autosomal dominant neurodegenerative disease. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Frontotemporal Lob Dejenerasyonu, TARDBP İle İlgili, 612069, Otozomal Dominant (Frontotemporal Demans-Motor Nöron Hastalığı) (TARDBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Frontotemporal lobar degeneration, TARDBP-related, 612069, Autosomal dominant (Frontotemporal dementia with motor neuron disease) (TARDBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TARDBP	TARDBP, TDP43, ALS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 10 (ALS10) [MIM:612069]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5- 10% of the cases. {ECO:0000269|PubMed:16288693, ECO:0000269|PubMed:18309045, ECO:0000269|PubMed:18372902, ECO:0000269|PubMed:18396105, ECO:0000269|PubMed:18438952, ECO:0000269|PubMed:19224587, ECO:0000269|PubMed:19695877, ECO:0000269|PubMed:21220647, ECO:0000269|PubMed:21418058, ECO:0000269|PubMed:22456481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fruktoz intoleransı paneli (ALDOB FBP1 9q21) (MLPA)	Fructose intolerance panel (ALDOB FBP1 9q21) (MLPA)	ALDOB FBP1 9q21	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Fruktoz intoleransı paneli (ALDOB FBP1 9q21) (MLPA) (Prenatal)	Fructose intolerance panel (ALDOB FBP1 9q21) (MLPA) (Prenatal)	ALDOB FBP1 9q21	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fruktoz İntoleransı, 229600, Otozomal Resesif (Kalıtsal Fruktoz İntoleransı) (ALDOB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fructose intolerance, 229600, Autosomal recessive (Hereditary fructose intolerance) (ALDOB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDOB	ALDOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary fructose intolerance (HFI) [MIM:229600]: Autosomal recessive disease that results in an inability to metabolize fructose and related sugars. Complete exclusion of fructose results in dramatic recovery; however, if not treated properly, HFI subjects suffer episodes of hypoglycemia, general ill condition, and risk of death the remainder of life. {ECO:0000269|PubMed:10024431, ECO:0000269|PubMed:10970798, ECO:0000269|PubMed:12205126, ECO:0000269|PubMed:15532022, ECO:0000269|PubMed:15880727, ECO:0000269|PubMed:1967768, ECO:0000269|PubMed:2338380, ECO:0000269|PubMed:3383242, ECO:0000269|PubMed:8162030, ECO:0000269|PubMed:8299883}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Fruktoz İntoleransı, 229600, Otozomal Resesif (Kalıtsal Fruktoz İntoleransı) (MLPA)	Fructose intolerance, 229600, Autosomal recessive (Hereditary fructose intolerance) (MLPA)	ALDOB	ALDOB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary fructose intolerance (HFI) [MIM:229600]: Autosomal recessive disease that results in an inability to metabolize fructose and related sugars. Complete exclusion of fructose results in dramatic recovery; however, if not treated properly, HFI subjects suffer episodes of hypoglycemia, general ill condition, and risk of death the remainder of life. {ECO:0000269|PubMed:10024431, ECO:0000269|PubMed:10970798, ECO:0000269|PubMed:12205126, ECO:0000269|PubMed:15532022, ECO:0000269|PubMed:15880727, ECO:0000269|PubMed:1967768, ECO:0000269|PubMed:2336380, ECO:0000269|PubMed:3383242, ECO:0000269|PubMed:8162030, ECO:0000269|PubMed:8299883}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fruktoz İntoleransı, 229600, Otozomal Resesif (Kalıtsal Fruktoz İntoleransı) (Prenatal) (ALDOB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fructose intolerance, 229600, Autosomal recessive (Hereditary fructose intolerance) (ALDOB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDOB	ALDOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary fructose intolerance (HFI) [MIM:229600]: Autosomal recessive disease that results in an inability to metabolize fructose and related sugars. Complete exclusion of fructose results in dramatic recovery; however, if not treated properly, HFI subjects suffer episodes of hypoglycemia, general ill condition, and risk of death the remainder of life. {ECO:0000269|PubMed:10024431, ECO:0000269|PubMed:10970798, ECO:0000269|PubMed:12205126, ECO:0000269|PubMed:15532022, ECO:0000269|PubMed:15880727, ECO:0000269|PubMed:1967768, ECO:0000269|PubMed:2336380, ECO:0000269|PubMed:3383242, ECO:0000269|PubMed:8162030, ECO:0000269|PubMed:8299883}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fruktoz İntoleransı, 229600, Otozomal Resesif (Kalıtsal Fruktoz İntoleransı) (Prenatal) (MLPA)	Fructose intolerance, 229600, Autosomal recessive (Hereditary fructose intolerance) (Prenatal) (MLPA)	ALDOB	ALDOB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary fructose intolerance (HFI) [MIM:229600]: Autosomal recessive disease that results in an inability to metabolize fructose and related sugars. Complete exclusion of fructose results in dramatic recovery; however, if not treated properly, HFI subjects suffer episodes of hypoglycemia, general ill condition, and risk of death the remainder of life. {ECO:0000269|PubMed:10024431, ECO:0000269|PubMed:10970798, ECO:0000269|PubMed:12205126, ECO:0000269|PubMed:15532022, ECO:0000269|PubMed:15880727, ECO:0000269|PubMed:1967768, ECO:0000269|PubMed:2336380, ECO:0000269|PubMed:3383242, ECO:0000269|PubMed:8162030, ECO:0000269|PubMed:8299883}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fruktoz-1,6-Bisfosfataz Eksikliği, 229700, Otozomal Resesif; FBP1D (Fruktoz-1,6-Bisfosfataz Eksikliği) (FBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fructose-1,6-bisphosphatase deficiency, 229700, Autosomal recessive; FBP1D (Fructose-1,6-bisphosphatase deficiency) (FBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBP1	FBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fructose-1,6-bisphosphatase deficiency (FBP1D) [MIM:229700]: An autosomal recessive metabolic disorder characterized by impaired gluconeogenesis, and episodes of hypoglycemia and metabolic acidosis that can be lethal in newborn infants or young children. {ECO:0000269|PubMed:12126934, ECO:0000269|PubMed:9382095}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fruktoz-1,6-Bisfosfataz Eksikliği, 229700, Otozomal Resesif; FBP1D (Fruktoz-1,6-Bisfosfataz Eksikliği) (MLPA)	Fructose-1,6-bisphosphatase deficiency, 229700, Autosomal recessive; FBP1D (Fructose-1,6-bisphosphatase deficiency) (MLPA)	FBP1	FBP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fructose-1,6-bisphosphatase deficiency (FBP1D) [MIM:229700]: An autosomal recessive metabolic disorder characterized by impaired gluconeogenesis, and episodes of hypoglycemia and metabolic acidosis that can be lethal in newborn infants or young children. {ECO:0000269|PubMed:12126934, ECO:0000269|PubMed:9382095}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fruktoz-1,6-Bisfosfataz Eksikliği, 229700, Otozomal Resesif; FBP1D (Fruktoz-1,6-Bisfosfataz Eksikliği) (Prenatal) (FBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fructose-1,6-bisphosphatase deficiency, 229700, Autosomal recessive; FBP1D (Fructose-1,6-bisphosphatase deficiency) (FBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBP1	FBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fructose-1,6-bisphosphatase deficiency (FBP1D) [MIM:229700]: An autosomal recessive metabolic disorder characterized by impaired gluconeogenesis, and episodes of hypoglycemia and metabolic acidosis that can be lethal in newborn infants or young children. {ECO:0000269|PubMed:12126934, ECO:0000269|PubMed:9382095}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fruktoz-1,6-Bisfosfataz Eksikliği, 229700, Otozomal Resesif; FBP1D (Fruktoz-1,6-Bisfosfataz Eksikliği) (Prenatal) (MLPA)	Fructose-1,6-bisphosphatase deficiency, 229700, Autosomal recessive; FBP1D (Fructose-1,6-bisphosphatase deficiency) (Prenatal) (MLPA)	FBP1	FBP1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fructose-1,6-bisphosphatase deficiency (FBP1D) [MIM:229700]: An autosomal recessive metabolic disorder characterized by impaired gluconeogenesis, and episodes of hypoglycemia and metabolic acidosis that can be lethal in newborn infants or young children. {ECO:0000269|PubMed:12126934, ECO:0000269|PubMed:9382095}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fukosidoz, 230000, Otozomal Resesif (Fukosidozis) (FUCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fucosidosis, 230000, Autosomal recessive (Fucosidosis) (FUCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUCA1	FUCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fucosidosis (FUCA1D) [MIM:230000]: An autosomal recessive lysosomal storage disease characterized by accumulation of fucose- containing glycolipids and glycoproteins in various tissues. Clinical signs include facial dysmorphism, dysostosis multiplex, moderate hepatomegaly, severe intellectual deficit, deafness, and according to age, angiokeratomas. {ECO:0000269|PubMed:7874128, ECO:0000269|PubMed:8504303, ECO:0000269|PubMed:9762612}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fukosidoz, 230000, Otozomal Resesif (Fukosidozis) (Prenatal) (FUCA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fucosidosis, 230000, Autosomal recessive (Fucosidosis) (FUCA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FUCA1	FUCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fucosidosis (FUCA1D) [MIM:230000]: An autosomal recessive lysosomal storage disease characterized by accumulation of fucose- containing glycolipids and glycoproteins in various tissues. Clinical signs include facial dysmorphism, dysostosis multiplex, moderate hepatomegaly, severe intellectual deficit, deafness, and according to age, angiokeratomas. {ECO:0000269|PubMed:7874128, ECO:0000269|PubMed:8504303, ECO:0000269|PubMed:9762612}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fukosiltransferaz 6 Eksikliği, 613852 (FUT6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fucosyltransferase 6 deficiency, 613852 (FUT6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT6	FUT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fukosiltransferaz 6 Eksikliği, 613852 (Prenatal) (FUT6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fucosyltransferase 6 deficiency, 613852 (FUT6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FUT6	FUT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fuhrmann Sendromu, 228930, Otozomal Resesif (Fuhrmann Sendromu) (WNT7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fuhrmann syndrome, 228930, Autosomal recessive (Fuhrmann syndrome) (WNT7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT7A	WNT7A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fuhrmann syndrome (FUHRS) [MIM:228930]: Distinct limb- malformation disorder characterized also by various degrees of limb aplasia/hypoplasia and joint dysplasia. {ECO:0000269|PubMed:16826533}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fuhrmann Sendromu, 228930, Otozomal Resesif (Fuhrmann Sendromu) (Prenatal) (WNT7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fuhrmann syndrome, 228930, Autosomal recessive (Fuhrmann syndrome) (WNT7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT7A	WNT7A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fuhrmann syndrome (FUHRS) [MIM:228930]: Distinct limb- malformation disorder characterized also by various degrees of limb aplasia/hypoplasia and joint dysplasia. {ECO:0000269|PubMed:16826533}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fumaraz Eksikliği, 606812, Otozomal Resesif; FMRD (Fumarik Asitüri) (FH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fumarase deficiency, 606812, Autosomal recessive (Fumaric aciduria) (FH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FH	FH, HLRCC, MCUL1, FMRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fumaraz Eksikliği, 606812, Otozomal Resesif; FMRD (Fumarik Asitüri) (Prenatal) (FH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Fumarase deficiency, 606812, Autosomal recessive (Fumaric aciduria) (FH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FH	FH, HLRCC, MCUL1, FMRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fundus Albipunktatus, 136880, Otozomal Resesif, Otozomal Dominant (Fundus Albipunctatus) (RLBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fundus albipunctatus, 136880, Autosomal recessive, Autosomal dominant (Fundus albipunctatus) (RLBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RLBP1	RLBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. {ECO:0000269|PubMed:10102299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fundus Albipunktatus, 136880, Otozomal Resesif, Otozomal Dominant (Fundus Albipunctatus) (RDH5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fundus albipunctatus, 136880, Autosomal recessive, Autosomal dominant (Fundus albipunctatus) (RDH5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RDH5	RDH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Fundus Flavimaculatus, 248200, Otozomal Resesif (Stargardt Hastalığı) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fundus flavimaculatus, 248200, Autosomal recessive (Stargardt disease) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABA-Transaminaz Eksikliği, 613163, Otozomal Resesif (Gama-Aminobütirik Asit Transaminaz Eksikliği) (ABAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	GABA-transaminase deficiency, 613163, Autosomal recessive (Gamma-aminobutyric acid transaminase deficiency) (ABAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABAT	ABAT, GABAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABA-Transaminaz Eksikliği, 613163, Otozomal Resesif (Gama-Aminobütirik Asit Transaminaz Eksikliği) (Prenatal) (ABAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	GABA-transaminase deficiency, 613163, Autosomal recessive (Gamma-aminobutyric acid transaminase deficiency) (ABAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABAT	ABAT, GABAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Galaktokinaz Eksikliği,Katarakt, 230200, Otozomal Resesif (Galaktozemi) (GALK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Galactokinase deficiency with cataracts, 230200, Autosomal recessive (Galactosemia) (GALK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALK1	GALK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Galactosemia II (GALCT2) [MIM:230200]: Autosomal recessive deficiency characterized by congenital cataracts during infancy and presenile cataracts in the adult population. The cataracts are secondary to accumulation of galactitol in the lenses. {ECO:0000269|PubMed:10521295, ECO:0000269|PubMed:10790206, ECO:0000269|PubMed:11139256, ECO:0000269|PubMed:11231902, ECO:0000269|PubMed:15024738}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Galaktokinaz Eksikliği,Katarakt, 230200, Otozomal Resesif (Galaktozemi) (Prenatal) (GALK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Galactokinase deficiency with cataracts, 230200, Autosomal recessive (Galactosemia) (GALK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GALK1	GALK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Galactosemia II (GALCT2) [MIM:230200]: Autosomal recessive deficiency characterized by congenital cataracts during infancy and presenile cataracts in the adult population. The cataracts are secondary to accumulation of galactitol in the lenses. {ECO:0000269|PubMed:10521295, ECO:0000269|PubMed:10790206, ECO:0000269|PubMed:11139256, ECO:0000269|PubMed:11231902, ECO:0000269|PubMed:15024738}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Galaktoz Epimeraz Eksikliği, 230350, Otozomal Resesif (Galaktozemi) (GALE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Galactose epimerase deficiency, 230350, Autosomal recessive (Galactosemia) (GALE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALE	GALE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epimerase-deficiency galactosemia (EDG) [MIM:230350]: Clinical features include early-onset cataracts, liver damage, deafness and mental retardation. There are two clinically distinct forms of EDG. (1) A benign, or 'peripheral' form with no detectable GALE activity in red blood cells and characterized by mild symptoms. Some patients may suffer no symptoms beyond raised levels of galactose-1-phosphate in the blood. (2) A much rarer 'generalized' form with undetectable levels of GALE activity in all tissues and resulting in severe features such as restricted growth and mental development. {ECO:0000269|PubMed:11903335, ECO:0000269|PubMed:16301867, ECO:0000269|PubMed:9326324, ECO:0000269|PubMed:9538513, ECO:0000269|PubMed:9973283}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Galaktoz Epimeraz Eksikliği, 230350, Otozomal Resesif (Galaktozemi) (Prenatal) (GALE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Galactose epimerase deficiency, 230350, Autosomal recessive (Galactosemia) (GALE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GALE	GALE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epimerase-deficiency galactosemia (EDG) [MIM:230350]: Clinical features include early-onset cataracts, liver damage, deafness and mental retardation. There are two clinically distinct forms of EDG. (1) A benign, or 'peripheral' form with no detectable GALE activity in red blood cells and characterized by mild symptoms. Some patients may suffer no symptoms beyond raised levels of galactose-1-phosphate in the blood. (2) A much rarer 'generalized' form with undetectable levels of GALE activity in all tissues and resulting in severe features such as restricted growth and mental development. {ECO:0000269|PubMed:11903335, ECO:0000269|PubMed:16301867, ECO:0000269|PubMed:9326324, ECO:0000269|PubMed:9538513, ECO:0000269|PubMed:9973283}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Galaktozemi, 230400, Otozomal Resesif (Galaktozemi) (GALT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Galactosemia, 230400, Autosomal recessive (Galactosemia) (GALT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALT	GALT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Galactosemia (GALCT) [MIM:230400]: Inherited disorder of galactose metabolism that causes jaundice, cataracts, and mental retardation. {ECO:0000269|PubMed:10220154, ECO:0000269|PubMed:11754113, ECO:0000269|PubMed:11919338, ECO:0000269|PubMed:1373122, ECO:0000269|PubMed:1427861, ECO:0000269|PubMed:15841485, ECO:0000269|PubMed:1610789, ECO:0000269|PubMed:17041746, ECO:0000269|PubMed:17876724, ECO:0000269|PubMed:18956253, ECO:0000269|PubMed:1897530, ECO:0000269|PubMed:2011574, ECO:0000269|PubMed:22461411, ECO:0000269|PubMed:23022339, ECO:0000269|PubMed:25592817, ECO:0000269|PubMed:25614870, ECO:0000269|PubMed:7550229, ECO:0000269|PubMed:7887416, ECO:0000269|PubMed:7887417, ECO:0000269|PubMed:8499924, ECO:0000269|PubMed:8598637, ECO:0000269|PubMed:8741038, ECO:0000269|PubMed:8869397, ECO:0000269|PubMed:8956044, ECO:0000269|PubMed:9222760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Galaktozemi, 230400, Otozomal Resesif (Galaktozemi) (MLPA)	Galactosemia, 230400, Autosomal recessive (Galactosemia) (MLPA)	GALT	GALT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Galactosemia (GALCT) [MIM:230400]: Inherited disorder of galactose metabolism that causes jaundice, cataracts, and mental retardation. {ECO:0000269|PubMed:10220154, ECO:0000269|PubMed:11754113, ECO:0000269|PubMed:11919338, ECO:0000269|PubMed:1373122, ECO:0000269|PubMed:1427861, ECO:0000269|PubMed:15841485, ECO:0000269|PubMed:1610789, ECO:0000269|PubMed:17041746, ECO:0000269|PubMed:17876724, ECO:0000269|PubMed:18956253, ECO:0000269|PubMed:1897530, ECO:0000269|PubMed:2011574, ECO:0000269|PubMed:22461411, ECO:0000269|PubMed:23022339, ECO:0000269|PubMed:25592817, ECO:0000269|PubMed:25614870, ECO:0000269|PubMed:7550229, ECO:0000269|PubMed:7887416, ECO:0000269|PubMed:7887417, ECO:0000269|PubMed:8499924, ECO:0000269|PubMed:8598637, ECO:0000269|PubMed:8741038, ECO:0000269|PubMed:8869397, ECO:0000269|PubMed:8956044, ECO:0000269|PubMed:9222760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Galaktozemi, 230400, Otozomal Resesif (Galaktozemi) (Prenatal) (GALT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Galactosemia, 230400, Autosomal recessive (Galactosemia) (GALT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GALT	GALT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Galactosemia (GALCT) [MIM:230400]: Inherited disorder of galactose metabolism that causes jaundice, cataracts, and mental retardation. {ECO:0000269|PubMed:10220154, ECO:0000269|PubMed:11754113, ECO:0000269|PubMed:11919338, ECO:0000269|PubMed:1373122, ECO:0000269|PubMed:1427861, ECO:0000269|PubMed:15841485, ECO:0000269|PubMed:1610789, ECO:0000269|PubMed:17041746, ECO:0000269|PubMed:17876724, ECO:0000269|PubMed:18956253, ECO:0000269|PubMed:1897530, ECO:0000269|PubMed:2011574, ECO:0000269|PubMed:22461411, ECO:0000269|PubMed:23022339, ECO:0000269|PubMed:25592817, ECO:0000269|PubMed:25614870, ECO:0000269|PubMed:7550229, ECO:0000269|PubMed:7887416, ECO:0000269|PubMed:7887417, ECO:0000269|PubMed:8499924, ECO:0000269|PubMed:8598637, ECO:0000269|PubMed:8741038, ECO:0000269|PubMed:8869397, ECO:0000269|PubMed:8956044, ECO:0000269|PubMed:9222760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Galaktozemi, 230400, Otozomal Resesif (Galaktozemi) (Prenatal) (MLPA)	Galactosemia, 230400, Autosomal recessive (Galactosemia) (Prenatal) (MLPA)	GALT	GALT	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Galactosemia (GALCT) [MIM:230400]: Inherited disorder of galactose metabolism that causes jaundice, cataracts, and mental retardation. {ECO:0000269|PubMed:10220154, ECO:0000269|PubMed:11754113, ECO:0000269|PubMed:11919338, ECO:0000269|PubMed:1373122, ECO:0000269|PubMed:1427861, ECO:0000269|PubMed:15841485, ECO:0000269|PubMed:1610789, ECO:0000269|PubMed:17041746, ECO:0000269|PubMed:17876724, ECO:0000269|PubMed:18956253, ECO:0000269|PubMed:1897530, ECO:0000269|PubMed:2011574, ECO:0000269|PubMed:22461411, ECO:0000269|PubMed:23022339, ECO:0000269|PubMed:25592817, ECO:0000269|PubMed:25614870, ECO:0000269|PubMed:7550229, ECO:0000269|PubMed:7887416, ECO:0000269|PubMed:7887417, ECO:0000269|PubMed:8499924, ECO:0000269|PubMed:8598637, ECO:0000269|PubMed:8741038, ECO:0000269|PubMed:8869397, ECO:0000269|PubMed:8956044, ECO:0000269|PubMed:9222760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Galaktosiyalidoz, 256540, Otozomal Resesif; GSL (Galaktosialidoz) (CTSA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Galactosialidosis, 256540, Autosomal recessive; GSL (Galactosialidosis) (CTSA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSA	CTSA, PPGB, GSL, NGBE, GLB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Galactosialidosis (GSL) [MIM:256540]: A lysosomal storage disease associated with a combined deficiency of beta-galactosidase and neuraminidase, secondary to a defect in cathepsin A. All patients have clinical manifestations typical of a lysosomal disorder, such as coarse facies, cherry red spots, vertebral changes, foam cells in the bone marrow, and vacuolated lymphocytes. Three phenotypic subtypes are recognized. The early infantile form is associated with fetal hydrops, edema, ascites, visceromegaly, skeletal dysplasia, and early death. The late infantile type is characterized by hepatosplenomegaly, growth retardation, cardiac involvement, and a normal or mildly affected mental state. The juvenile/adult form is characterized by myoclonus, ataxia, angiokeratoma, mental retardation, neurologic deterioration, absence of visceromegaly, and long survival. {ECO:0000269|PubMed:10944848, ECO:0000269|PubMed:1756715, ECO:0000269|PubMed:8514852, ECO:0000269|PubMed:8968752}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Galaktosiyalidoz, 256540, Otozomal Resesif; GSL (Galaktosialidoz) (Prenatal) (CTSA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Galactosialidosis, 256540, Autosomal recessive; GSL (Galactosialidosis) (CTSA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTSA	CTSA, PPGB, GSL, NGBE, GLB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Galactosialidosis (GSL) [MIM:256540]: A lysosomal storage disease associated with a combined deficiency of beta-galactosidase and neuraminidase, secondary to a defect in cathepsin A. All patients have clinical manifestations typical of a lysosomal disorder, such as coarse facies, cherry red spots, vertebral changes, foam cells in the bone marrow, and vacuolated lymphocytes. Three phenotypic subtypes are recognized. The early infantile form is associated with fetal hydrops, edema, ascites, visceromegaly, skeletal dysplasia, and early death. The late infantile type is characterized by hepatosplenomegaly, growth retardation, cardiac involvement, and a normal or mildly affected mental state. The juvenile/adult form is characterized by myoclonus, ataxia, angiokeratoma, mental retardation, neurologic deterioration, absence of visceromegaly, and long survival. {ECO:0000269|PubMed:10944848, ECO:0000269|PubMed:1756715, ECO:0000269|PubMed:8514852, ECO:0000269|PubMed:8968752}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Safra Kesesi Hastalığı 1, 600803, Otozomal Resesif, Otozomal Dominant; GBD1 (Düşük Fosfolipid İlişkili Kolelitiazis) (ABCB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gallbladder disease 1, 600803, Autosomal recessive, Autosomal dominant; GBD1 (Low phospholipid associated cholelithiasis) (ABCB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB4	ABCB4, PGY3, MDR3, ICP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gallbladder disease 1 (GBD1) [MIM:600803]: One of the major digestive diseases. Gallstones composed of cholesterol (cholelithiasis) are the common manifestations in western countries. Most people with gallstones, however, remain asymptomatic through their lifetimes. {ECO:0000269|PubMed:11313316, ECO:0000269|PubMed:12891548, ECO:0000269|PubMed:22331132, ECO:0000269|PubMed:23533021, ECO:0000269|PubMed:24723470, ECO:0000269|Ref.2}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Kesesi Hastalığı 1, 600803, Otozomal Resesif, Otozomal Dominant; GBD1 (Düşük Fosfolipid İlişkili Kolelitiazis) (MLPA)	Gallbladder disease 1, 600803, Autosomal recessive, Autosomal dominant; GBD1 (Low phospholipid associated cholelithiasis) (MLPA)	ABCB4	ABCB4, PGY3, MDR3, ICP3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Gallbladder disease 1 (GBD1) [MIM:600803]: One of the major digestive diseases. Gallstones composed of cholesterol (cholelithiasis) are the common manifestations in western countries. Most people with gallstones, however, remain asymptomatic through their lifetimes. {ECO:0000269|PubMed:11313316, ECO:0000269|PubMed:12891548, ECO:0000269|PubMed:22331132, ECO:0000269|PubMed:23533021, ECO:0000269|PubMed:24723470, ECO:0000269|Ref.2}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Safra Kesesi Hastalığı 4, 611465 (ABCG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gallbladder disease 4, 611465 (ABCG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCG8	ABCG8, GBD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Galloway-Mowat Sendromu, 251300, Otozomal Resesif; GAMOS (Galloway-Mowat Sendromu) (WDR73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Galloway-Mowat syndrome, 251300, Autosomal recessive; GAMOS (Galloway-Mowat syndrome) (WDR73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR73	WDR73, HSPC264, GAMOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Galloway-Mowat syndrome (GAMOS) [MIM:251300]: A rare, autosomal recessive disease with onset in infancy, and characterized by microcephaly, central nervous system abnormalities resulting in severe neurological impairment and intellectual disability, and nephrotic syndrome. Most patients develop seizures and nephrotic syndrome late in childhood. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:25466283}.;	EDTA Blood Sample
Galloway-Mowat Sendromu, 251300, Otozomal Resesif; GAMOS (Galloway-Mowat Sendromu) (Prenatal) (WDR73 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Galloway-Mowat syndrome, 251300, Autosomal recessive; GAMOS (Galloway-Mowat syndrome) (WDR73 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR73	WDR73, HSPC264, GAMOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Galloway-Mowat syndrome (GAMOS) [MIM:251300]: A rare, autosomal recessive disease with onset in infancy, and characterized by microcephaly, central nervous system abnormalities resulting in severe neurological impairment and intellectual disability, and nephrotic syndrome. Most patients develop seizures and nephrotic syndrome late in childhood. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:25466283}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GAPO Sendromu, 230740, Otozomal Resesif (GAPO Sendromu) (ANTXR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GAPO syndrome, 230740, Autosomal recessive (GAPO syndrome) (ANTXR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANTXR1	ANTXR1, TEM8, ATR, GAPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GAPO syndrome (GAPO) [MIM:230740]: A disease characterized by growth retardation, alopecia, failure of tooth eruption, and progressive optic atrophy in some patients. {ECO:0000269|PubMed:23602711}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
GAPO Sendromu, 230740, Otozomal Resesif (GAPO Sendromu) (Prenatal) (ANTXR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GAPO syndrome, 230740, Autosomal recessive (GAPO syndrome) (ANTXR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANTXR1	ANTXR1, TEM8, ATR, GAPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GAPO syndrome (GAPO) [MIM:230740]: A disease characterized by growth retardation, alopecia, failure of tooth eruption, and progressive optic atrophy in some patients. {ECO:0000269|PubMed:23602711}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gardner Sendromu, 175100, Otozomal Dominant (Gardner Sendromu) (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gardner syndrome, 175100, Autosomal dominant (Gardner syndrome) (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gardner Sendromu, 175100, Otozomal Dominant (Gardner Sendromu) (MLPA)	Gardner syndrome, 175100, Autosomal dominant (Gardner syndrome) (MLPA)	APC	APC, GS, FPC, BTPS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
H. Pylori Enfeksiyonu Sonrası Gastrik Kanser Riski, 137215, Otozomal Dominant (Kalıtsal Diffüz Mide Kanseri) (IL1RN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer risk after H. pylori infection, 137215, Autosomal dominant (Hereditary diffuse gastric cancer) (IL1RN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL1RN	IL1RN, MVCD4, DIRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H. Pylori Enfeksiyonu Sonrası Gastrik Kanser Riski, 137215, Otozomal Dominant (Kalıtsal Diffüz Mide Kanseri) (IL1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer risk after H. pylori infection, 137215, Autosomal dominant (Hereditary diffuse gastric cancer) (IL1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL1B	IL1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mide Kanseri,Ailesel Diffüz, ± Dudak ve / Veya Damak Yarıkları, 137215, Otozomal Dominant (Kalıtsal Diffüz Mide Kanseri) (CDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, familial diffuse, with or without cleft lip and/or palate, 137215, Autosomal dominant (Hereditary diffuse gastric cancer) (CDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH1	CDH1, UVO, LCAM, ECAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary diffuse gastric cancer (HDGC) [MIM:137215]: A cancer predisposition syndrome with increased susceptibility to diffuse gastric cancer. Diffuse gastric cancer is a malignant disease characterized by poorly differentiated infiltrating lesions resulting in thickening of the stomach. Malignant tumors start in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. {ECO:0000269|PubMed:10319582, ECO:0000269|PubMed:12216071}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Heterozygous CDH1 germline mutations are responsible for familial cases of diffuse gastric cancer. Somatic mutations has also been found in patients with sporadic diffuse gastric cancer and lobular breast cancer.;	EDTA Blood Sample
Mide Kanseri, Kalıtsal Diffüz; Hdgc (Kalıtsal Diffüz Mide Kanseri) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	GASTRIC CANCER, HEREDITARY DIFFUSE; HDGC (Hereditary diffuse gastric cancer) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mide Kanseri, Kalıtsal Diffüz; Hdgc (Kalıtsal Diffüz Mide Kanseri) (MLPA)	GASTRIC CANCER, HEREDITARY DIFFUSE; HDGC (Hereditary diffuse gastric cancer) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mide Kanseri, Somatik, 137215 (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 137215 (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (ERBB2-HER2) (FISH)	Gastric cancer, somatic, 613659 (ERBB2- HER2) (FISH)	ERBB2	ERBB2, NGL, NEU, HER2	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	DISEASE: Hereditary diffuse gastric cancer (HDGC) [MIM:137215]: A cancer predisposition syndrome with increased susceptibility to diffuse gastric cancer. Diffuse gastric cancer is a malignant disease characterized by poorly differentiated infiltrating lesions resulting in thickening of the stomach. Malignant tumors start in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. Note=The gene represented in this entry is involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (IRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (IRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF1	IRF1, MAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Gastric cancer (GASC) [MIM:613659]: A malignant disease which starts in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. The term gastric cancer or gastric carcinoma refers to adenocarcinoma of the stomach that accounts for most of all gastric malignant tumors. Two main histologic types are recognized, diffuse type and intestinal type carcinomas. Diffuse tumors are poorly differentiated infiltrating lesions, resulting in thickening of the stomach. In contrast, intestinal tumors are usually exophytic, often ulcerating, and associated with intestinal metaplasia of the stomach, most often observed in sporadic disease. {ECO:0000269|PubMed:10395827, ECO:0000269|PubMed:9679752}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (CASP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (CASP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP10	CASP10, MCH4, ALPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Mide Kanseri, Somatik, 613659 (PIK3CA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (KLF6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (KLF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF6	KLF6, COPEB, BCD1, ZF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mide Kanseri, Somatik, 613659 (MUTYH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gastric cancer, somatic, 613659 (MUTYH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUTYH	MUTYH, MYH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Gastrointestinal Defektler ve İmmün Yetmezlik Sendromu, 243150, Otozomal Resesif, GIDID (Multipl Intestinal Atrezi) (TTC7A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gastrointestinal defects and immunodeficiency syndrome, 243150, Autosomal recessive, GIDID (Multiple intestinal atresia) (TTC7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC7A	TTC7A, TTC7, KIAA1140, MINAT, GIDID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gastrointestinal Stromal Tümör, 606764, Otozomal Dominant, Sporadik Vakalar (Gastrointestinal Stromal Tümör) (SDHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastrointestinal stromal tumor, 606764, Autosomal dominant, Isolated cases (Gastrointestinal stromal tumor) (SDHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHC	SDHC, PGL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gastrointestinal Stromal Tümör, 606764, Otozomal Dominant, Sporadik Vakalar, GIST (Gastrointestinal Stromal Tümör) (SDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastrointestinal stromal tumor, 606764, Autosomal dominant, Isolated cases, GIST (Gastrointestinal stromal tumor) (SDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHB	SDHB, SDH2, SDHIP, PGL4, CWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gastrointestinal Stromal Tümör, Ailesel, 606764, Otozomal Dominant, Sporadik Vakalar (Gastrointestinal Stromal Tümör) (KIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastrointestinal stromal tumor, familial, 606764, Autosomal dominant, Isolated cases (Gastrointestinal stromal tumor) (KIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIT	KIT, PBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gastrointestinal Stromal Tümör, Somatik, 606764 (Gastrointestinal Stromal Tümör) (PDGFRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gastrointestinal stromal tumor, somatic, 606764 (Gastrointestinal stromal tumor) (PDGFRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRA	PDGFRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Gastrointestinal stromal tumor (GIST) [MIM:606764]: Common mesenchymal neoplasms arising in the gastrointestinal tract, most often in the stomach. They are histologically, immunohistochemically, and genetically different from typical leiomyomas, leiomyosarcomas, and schwannomas. Most GISTs are composed of a fairly uniform population of spindle-shaped cells. Some tumors are dominated by epithelioid cells or contain a mixture of spindle and epithelioid morphologies. Primary GISTs in the gastrointestinal tract commonly metastasize in the omentum and mesenteries, often as multiple nodules. However, primary tumors may also occur outside of the gastrointestinal tract, in other intra-abdominal locations, especially in the omentum and mesentery. {ECO:0000269|PubMed:12522257, ECO:0000269|PubMed:15928335}. Note=The gene represented in this entry may be involved in disease pathogenesis. Mutations causing PDGFRA-constitutive activation have been found in gastrointestinal stromal tumors lacking KIT mutations (PubMed:12522257). {ECO:0000269|PubMed:12522257},.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Gaucher Hastalığı, Atipik, Saposin C Eksikliğine Bağlı, 610539 (Gaucher Hastalığı (PSAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	GAUCHER DISEASE, ATYPICAL, DUE TO SAPOSIN C DEFICIENCY, 610539 (Gaucher disease) (PSAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Atipik, Saposin C Eksikliğine Bağlı, 610539 (Gaucher Hastalığı) (Prenatal) (PSAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	GAUCHER DISEASE, ATYPICAL, DUE TO SAPOSIN C DEFICIENCY, 610539 (Gaucher disease) (PSAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Perinatal Letal, 608013, Otozomal Resesif (Gaucher Hastalığı) (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaucher disease, perinatal lethal, 608013, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Perinatal Letal, 608013, Otozomal Resesif (Gaucher Hastalığı) (MLPA)	Gaucher disease, perinatal lethal, 608013, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Perinatal Letal, 608013, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gaucher disease, perinatal lethal, 608013, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Perinatal Letal, 608013, Otozomal Resesif (Gaucher Hastalığı) Prenatal) (MLPA)	Gaucher disease, perinatal lethal, 608013, Autosomal recessive (Gaucher disease) (Prenatal) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 1, 230800, Otozomal Resesif (Gaucher Hastalığı) (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaucher disease, type I, 230800, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 1, 230800, Otozomal Resesif (Gaucher Hastalığı) (MLPA)	Gaucher disease, type I, 230800, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 1, 230800, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gaucher disease, type I, 230800, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 1, 230800, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (MLPA)	Gaucher disease, type I, 230800, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 2, 230900, Otozomal Resesif (Gaucher Hastalığı) (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaucher disease, type II, 230900, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 2, 230900, Otozomal Resesif (Gaucher Hastalığı) (MLPA)	Gaucher disease, type II, 230900, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 2, 230900, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gaucher disease, type II, 230900, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 2, 230900, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (MLPA)	Gaucher disease, type II, 230900, Autosomal recessive (Gaucher disease) (Prenatal) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 3, 231000, Otozomal Resesif (Gaucher Hastalığı) (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaucher disease, type III, 231000, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 3, 231000, Otozomal Resesif (Gaucher Hastalığı) (MLPA)	Gaucher disease, type III, 231000, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 3, 231000, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gaucher disease, type III, 231000, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 3, 231000, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (MLPA)	Gaucher disease, type III, 231000, Autosomal recessive (Gaucher disease) (Prenatal) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 3C, 231005, Otozomal Resesif (Gaucher Hastalığı) (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaucher disease, type IIIC, 231005, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 3C, 231005, Otozomal Resesif (Gaucher Hastalığı) (MLPA)	Gaucher disease, type IIIC, 231005, Autosomal recessive (Gaucher disease) (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gaucher Hastalığı, Tip 3C, 231005, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gaucher disease, type IIIC, 231005, Autosomal recessive (Gaucher disease) (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gaucher Hastalığı, Tip 3C, 231005, Otozomal Resesif (Gaucher Hastalığı) (Prenatal) (MLPA)	Gaucher disease, type IIIC, 231005, Autosomal recessive (Gaucher disease) (Prenatal) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bakış Paralizisi, Horizontal ve Progresif Skolyoz, 607313, Otozomal Resesif, HGPPS (Horizontal Bakış Felci-Progresif Skolyoz) (ROBO3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gaze palsy, horizontal, with progressive scoliosis, 607313, Autosomal recessive, HGPPS (Horizontal gaze palsy with progressive scoliosis) (ROBO3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROBO3	ROBO3, RBIG1, RIG1, HGPPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial horizontal gaze palsy with progressive scoliosis (HGPPS) [MIM:607313]: Patients show a medulla where motor and sensory projections appear uncrossed. {ECO:0000269|PubMed:15105459}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Geleofizik Displazi 1, 231050, Otozomal Resesif, GPHYSD1 (Geleofizik Displazi) (ADAMTSL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Geleophysic dysplasia 1, 231050, Autosomal recessive, GPHYSD1 (Geleophysic dysplasia) (ADAMTSL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTSL2	ADAMTSL2, KIAA0605, GPHYSD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Geleophysic dysplasia 1 (GPHYSD1) [MIM:231050]: An autosomal recessive disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have characteristic facial features including a 'happy' face with full cheeks, shortened nose, hypertelorism, long and flat philtrum, and thin upper lip. Other distinctive features include progressive cardiac valvular thickening often leading to an early death, toe walking, tracheal stenosis, respiratory insufficiency, and lysosomal-like storage vacuoles in various tissues. {ECO:0000269|PubMed:18677313, ECO:0000269|PubMed:21415077}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Geleofizik Displazi 1, 231050, Otozomal Resesif, GPHYSD1 (Geleofizik Displazi) (Prenatal) (ADAMTSL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Geleophysic dysplasia 1, 231050, Autosomal recessive, GPHYSD1 (Geleophysic dysplasia) (ADAMTSL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADAMTSL2	ADAMTSL2, KIAA0605, GPHYSD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Geleophysic dysplasia 1 (GPHYSD1) [MIM:231050]: An autosomal recessive disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have characteristic facial features including a 'happy' face with full cheeks, shortened nose, hypertelorism, long and flat philtrum, and thin upper lip. Other distinctive features include progressive cardiac valvular thickening often leading to an early death, toe walking, tracheal stenosis, respiratory insufficiency, and lysosomal-like storage vacuoles in various tissues. {ECO:0000269|PubMed:18677313, ECO:0000269|PubMed:21415077}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Geleofizik Displazi 2, 614185, Otozomal Dominant, GPHYSD2 (Geleofizik Displazi) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Geleophysic dysplasia 2, 614185, Autosomal dominant, GPHYSD2 (Geleophysic dysplasia) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Geleofizik Displazi 2, 614185, Otozomal Dominant, GPHYSD2 (Geleofizik Displazi) (MLPA)	Geleophysic dysplasia 2, 614185, Autosomal dominant, GPHYSD2 (Geleophysic dysplasia) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Geleofizik Displazi 2, 614185, Otozomal Dominant, GPHYSD2 (Geleofizik Displazi) (Prenatal) (FBN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Geleophysic dysplasia 2, 614185, Autosomal dominant, GPHYSD2 (Geleophysic dysplasia) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Geleofizik Displazi 2, 614185, Otozomal Dominant, GPHYSD2 (Geleofizik Displazi) (Prenatal) (MLPA)	Geleophysic dysplasia 2, 614185, Autosomal dominant, GPHYSD2 (Geleophysic dysplasia) (Prenatal) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Jeneralize Epilepsi ve Paroksismal Diskinezi, 609446, Otozomal Dominant; GEPD (Jeneralize Epilepsi-Paroksismal Diskinezi Sendromu) (KCNMA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Generalized epilepsy and paroxysmal dyskinesia, 609446, Autosomal dominant; GEPD (Generalized epilepsy-paroxysmal dyskinesia syndrome) (KCNMA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNMA1	KCNMA1, SLO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy and paroxysmal dyskinesia (GEPD) [MIM:609446]: Epilepsy is one of the most common and debilitating neurological disorders. Paroxysmal dyskinesias are neurological disorders characterized by sudden, unpredictable disabling attacks of involuntary movement often requiring life-long treatment. The coexistence of epilepsy and paroxysmal dyskinesia in the same individual or family is an increasingly recognized phenomenon. Patients manifest absence seizures, generalized tonic- clonic seizures, paroxysmal nonkinesigenic dyskinesia, involuntary dystonic or choreiform movements. Onset is usually in childhood and patients may have seizures only, dyskinesia only, or both. {ECO:0000269|PubMed:15937479}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Jeneralize Epilepsi ve Paroksismal Diskinezi, 609446, Otozomal Dominant; GEPD (Jeneralize Epilepsi-Paroksismal Diskinezi Sendromu) (Prenatal) (KCNMA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Generalized epilepsy and paroxysmal dyskinesia, 609446, Autosomal dominant; GEPD (Generalized epilepsy-paroxysmal dyskinesia syndrome) (KCNMA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNMA1	KCNMA1, SLO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Generalized epilepsy and paroxysmal dyskinesia (GEPD) [MIM:609446]: Epilepsy is one of the most common and debilitating neurological disorders. Paroxysmal dyskinesias are neurological disorders characterized by sudden, unpredictable, disabling attacks of involuntary movement often requiring life-long treatment. The coexistence of epilepsy and paroxysmal dyskinesia in the same individual or family is an increasingly recognized phenomenon. Patients manifest absence seizures, generalized tonic- clonic seizures, paroxysmal nonkinesigenic dyskinesia, involuntary dystonic or choreiform movements. Onset is usually in childhood and patients may have seizures only, dyskinesia only, or both. {ECO:0000269|PubMed:15937479}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jeneralize Epilepsi-Ateşli (Febril) Nöbetler, Tip 1; Gefsp1 (Jeneralize Epilepsi-Febril Nöbetler Arı) (SCN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 1; GEFSP1 (Generalized epilepsy with febrile seizures-plus) (SCN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1B	SCN1B, GEFSP1, BRGDA5, ATFB13, ÉIEÉS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Jeneralize Epilepsi, Ateşli (Febril) Nöbet, Tip 9, 616172, Otozomal Dominant; GEFSP9 (Jeneralize Epilepsi-Febril Nöbetler Arı) (STX1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Generalized epilepsy with febrile seizures plus, type 9, 616172, Autosomal dominant; GEFSP9 (Generalized epilepsy with febrile seizures-plus) (STX1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STX1B	STX1B, GEFSP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 9 (GEFSP9) [MIM:616172]: An autosomal dominant neurologic disorder characterized by febrile and/or afebrile seizures manifesting in early childhood. Seizure are variable and include generalized tonic-clonic, atonic, myoclonic, complex partial, and absence types. Most patients have remission of seizures later in childhood with no residual neurologic deficits. Rarely, patients may show mild developmental delay or mild intellectual disabilities. {ECO:0000269|PubMed:25362483}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Genetik Danışmanlık	Genetic counseling ( )	.	.	Klinik değerlendirme/ Clinical Evaluation	Tüm Tıbbi Dokümanlar ve Aile Fotoğrafları		0	0	.	.	0	Postnatal		Medical Documents and Family Photos
Genitopatellar Sendrom, 606170, Otozomal Dominant; GTPTS (Genitopatellar Sendromu) (KAT6B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Genitopatellar syndrome, 606170, Autosomal dominant; GTPTS (Genitopatellar syndrome) (KAT6B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KAT6B	KAT6B, MYST4, MORF, GTPTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Genitopatellar Sendrom, 606170, Otozomal Dominant; GTPTS (Genitopatellar Sendromu) (Prenatal) (KAT6B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Genitopatellar syndrome, 606170, Autosomal dominant; GTPTS (Genitopatellar syndrome) (KAT6B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KAT6B	KAT6B, MYST4, MORF, GTPTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Germ Hücreli Tümörler, 273300, Somatik Mutasyon (Testiküler Seminomatöz Germ Hücreli Tümör) (KIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Germ cell tumors, 273300, Somatic mutation (Testicular seminomatous germ cell tumor) (KIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIT	KIT, PBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Gerioderma Osteodisplastikum, 231070, Otozomal Resesif; GO (Gerioderma Osteodisplastika) (GORAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gerioderma osteodysplasticum, 231070, Autosomal recessive; GO (Gerioderma osteodysplastica) (GORAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GORAB	GORAB, SCYL1BP1, NTKLBP1, GO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gerioderma osteodysplasticum (GO) [MIM:231070]: A rare autosomal recessive disorder characterized by lax, wrinkled skin, joint laxity and a typical face with a prematurely aged appearance. Skeletal signs include severe osteoporosis leading to frequent fractures, malar and mandibular hypoplasia and a variable degree of growth retardation. {ECO:0000269|PubMed:18997784}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gerioderma Osteodisplastikum, 231070, Otozomal Resesif; GO (Gerioderma Osteodisplastika) (Prenatal) (GORAB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gerioderma osteodysplasticum, 231070, Autosomal recessive; GO (Gerioderma osteodysplastica) (GORAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GORAB	GORAB, SCYL1BP1, NTKLBP1, GO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Gerioderma osteodysplasticum (GO) [MIM:231070]: A rare autosomal recessive disorder characterized by lax, wrinkled skin, joint laxity and a typical face with a prematurely aged appearance. Skeletal signs include severe osteoporosis leading to frequent fractures, malar and mandibular hypoplasia and a variable degree of growth retardation. {ECO:0000269|PubMed:18997784}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gerstmann-Straussler Hastalığı, 137440, Otozomal Dominant; GSD (Gerstmann-Straussler-Scheinker Sendromu) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gerstmann-Straussler disease, 137440, Autosomal dominant; GSD (Gerstmann-Straussler-Scheinker syndrome) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gerstmann-Straussler Hastalığı, 137440, Otozomal Dominant; GSD (Gerstmann-Straussler-Scheinker Sendromu) (Prenatal) (PRNP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gerstmann-Straussler disease, 137440, Autosomal dominant; GSD (Gerstmann-Straussler-Scheinker syndrome) (PRNP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ghosal Hematodiaftaysal Sendrom, 231095, Otozomal Resesif (Ghosal Hematodiaftaysal Displazi) (TBXAS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ghosal hematodiaphyseal syndrome, 231095, Autosomal recessive (Ghosal hematodiaphyseal dysplasia) (TBXAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBXAS1	TBXAS1, GHOSAL, CYP5, BDPLT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ghosal Hematodiaftaysal Sendrom, 231095, Otozomal Resesif (Ghosal Hematodiaftaysal Displazi) (Prenatal) (TBXAS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ghosal hematodiaphyseal syndrome, 231095, Autosomal recessive (Ghosal hematodiaphyseal dysplasia) (TBXAS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBXAS1	TBXAS1, GHOSAL, CYP5, BDPLT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dev Aksonal Nöropati 2, Otozomal Dominant, 610100, Otozomal Dominant; GAN2 (Otozomal Dominant Charcot-Marie-Tooth Hastalığı, Tip 2-Dev Aksonlar) (DCAF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Giant axonal neuropathy 2, autosomal dominant, 610100, Autosomal dominant; GAN2 (Autosomal dominant Charcot-Marie-Tooth disease type 2 with giant axons) (DCAF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCAF8	DCAF8, WDR42A, GAN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Giant axonal neuropathy 2, autosomal dominant (GAN2) [MIM:610100]: An autosomal dominant peripheral axonal neuropathy characterized by onset of distal sensory impairment with lower extremity muscle weakness and atrophy after the second decade. Clinical features include foot deformities apparent in childhood, and cardiomyopathy in severely affected individuals. Sural nerve biopsy shows giant axonal swelling with neurofilament accumulation. {ECO:0000269|PubMed:24506946}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dev Aksonal Nöropati-1, 256850, Otozomal Resesif; GAN1 (Dev Aksonal Nöropati) (GAN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Giant axonal neuropathy-1, 256850, Autosomal recessive; GAN1 (Giant axonal neuropathy) (GAN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAN	GAN, GAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Giant axonal neuropathy 1, autosomal recessive (GAN1) [MIM:256850]: A severe autosomal recessive sensorimotor neuropathy affecting both the peripheral nerves and the central nervous system. Axonal loss and the presence of giant axonal swellings filled with neurofilaments on nerve biopsies are the hallmarks of this neurodegenerative disorder. {ECO:0000269|PubMed:11062483, ECO:0000269|PubMed:11971098, ECO:0000269|PubMed:12655563, ECO:0000269|PubMed:17578852, ECO:0000269|PubMed:17587580}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Dev Trombosit Bozukluğu, İzole, 231200, Otozomal Resesif (Bernard-Soulier Sendromu) (GP1BB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Giant platelet disorder, isolated, 231200, Autosomal recessive (Bernard-Soulier syndrome) (GP1BB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BB	GP1BB, BS, BDPLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Jigantizm, GHRF Hiperselesyonuna Bağlı (GHRH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gigantism due to GHRF hypersecretion (GHRH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHRH	GHRH, GHRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gilles de la Tourette Sendromu Yatkinlik, 137580, Otozomal Dominant (HDC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gilles de la Tourette syndrome, susceptibility to, 137580, Autosomal dominant (HDC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HDC	HDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gilles de la Tourette Sendromu, Yatkinlik, 137580, Otozomal Dominant (Prenatal) (HDC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gilles de la Tourette syndrome, susceptibility to, 137580, Autosomal dominant (HDC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HDC	HDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gillespie Sendromu, 206700; GLSP (Aniridi-Serebellar Ataksi-Entelektüel Yetersizlik Sendromu) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gillespie syndrome, 206700; GLSP (Aniridia-cerebellar ataxia-intellectual disability syndrome) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR1	ITPR1, SCA15, SCA16, SCA29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gillespie Sendromu, 206700; GLSP (Aniridi-Serebellar Ataksi-Entelektüel Yetersizlik Sendromu) (Prenatal) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gillespie syndrome, 206700; GLSP (Aniridia-cerebellar ataxia-intellectual disability syndrome) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITPR1	ITPR1, SCA15, SCA16, SCA29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gillessen-Kaesbach-Nishimura Sendromu, 263210, Otozomal Resesif; GIKANS (ALG9-CDG) (ALG9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gillessen-Kaesbach-Nishimura syndrome, 263210, Autosomal recessive; GIKANS (ALG9-CDG) (ALG9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG9	ALG9, DIBD1, CDG1L, GIKANS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gillessen-Kaesbach-Nishimura Sendromu, 263210, Otozomal Resesif; GIKANS (ALG9-CDG) (Prenatal) (ALG9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gillessen-Kaesbach-Nishimura syndrome, 263210, Autosomal recessive; GIKANS (ALG9-CDG) (ALG9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG9	ALG9, DIBD1, CDG1L, GIKANS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gitelman Sendromu, 263800, Otozomal Resesif; GTLMNS (Gitelman Sendromu) (SLC12A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gitelman syndrome, 263800, Autosomal recessive; GTLMNS (Gitelman syndrome) (SLC12A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC12A3	SLC12A3, NCCT, TSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gitelman syndrome (GS) [MIM:263800]: An autosomal recessive disorder characterized by hypokalemic alkalosis in combination with hypomagnesemia, low urinary calcium, and increased renin activity associated with normal blood pressure. Patients are often asymptomatic or present transient periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. The phenotype is highly heterogeneous in terms of age at onset and severity. Cardinal features such as hypocalciuria and hypomagnesemia might also change during the life cycle of a given patient. It has overlapping features with Bartter syndrome. {ECO:0000269|PubMed:10616841, ECO:0000269|PubMed:10988270, ECO:0000269|PubMed:11168953, ECO:0000269|PubMed:11940055, ECO:0000269|PubMed:12008755, ECO:0000269|PubMed:12112667, ECO:0000269|PubMed:15099170, ECO:0000269|PubMed:15687331, ECO:0000269|PubMed:16429844, ECO:0000269|PubMed:17654016, ECO:0000269|PubMed:17873326, ECO:0000269|PubMed:8528245, ECO:0000269|PubMed:8900229, ECO:0000269|PubMed:8954067, ECO:0000269|PubMed:9734597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Gitelman Sendromu, 263800, Otozomal Resesif; GTLMNS (Gitelman Sendromu) (MLPA)	Gitelman syndrome, 263800, Autosomal recessive; GTLMNS (Gitelman syndrome) (MLPA)	SLC12A3	SLC12A3, NCCT, TSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Gitelman syndrome (GS) [MIM:263800]: An autosomal recessive disorder characterized by hypokalemic alkalosis in combination with hypomagnesemia, low urinary calcium, and increased renin activity associated with normal blood pressure. Patients are often asymptomatic or present transient periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. The phenotype is highly heterogeneous in terms of age at onset and severity. Cardinal features such as hypocalciuria and hypomagnesemia might also change during the life cycle of a given patient. It has overlapping features with Bartter syndrome. {ECO:0000269|PubMed:10616841, ECO:0000269|PubMed:10988270, ECO:0000269|PubMed:11168953, ECO:0000269|PubMed:11940055, ECO:0000269|PubMed:12008755, ECO:0000269|PubMed:12112667, ECO:0000269|PubMed:15069170, ECO:0000269|PubMed:15687331, ECO:0000269|PubMed:16429844, ECO:0000269|PubMed:17654016, ECO:0000269|PubMed:17873326, ECO:0000269|PubMed:8528245, ECO:0000269|PubMed:8900229, ECO:0000269|PubMed:8954067, ECO:0000269|PubMed:9734597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gitelman Sendromu, 263800, Otozomal Resesif; GTLMNS (Gitelman Sendromu) (Prenatal) (SLC12A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gitelman syndrome, 263800, Autosomal recessive; GTLMNS (Gitelman syndrome) (SLC12A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC12A3	SLC12A3, NCCT, TSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Gitelman syndrome (GS) [MIM:263800]: An autosomal recessive disorder characterized by hypokalemic alkalosis in combination with hypomagnesemia, low urinary calcium, and increased renin activity associated with normal blood pressure. Patients are often asymptomatic or present transient periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. The phenotype is highly heterogeneous in terms of age at onset and severity. Cardinal features such as hypocalciuria and hypomagnesemia might also change during the life cycle of a given patient. It has overlapping features with Bartter syndrome. {ECO:0000269|PubMed:10616841, ECO:0000269|PubMed:10988270, ECO:0000269|PubMed:11168953, ECO:0000269|PubMed:11940055, ECO:0000269|PubMed:12008755, ECO:0000269|PubMed:12112667, ECO:0000269|PubMed:15069170, ECO:0000269|PubMed:15687331, ECO:0000269|PubMed:16429844, ECO:0000269|PubMed:17654016, ECO:0000269|PubMed:17873326, ECO:0000269|PubMed:8528245, ECO:0000269|PubMed:8900229, ECO:0000269|PubMed:8954067, ECO:0000269|PubMed:9734597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gitelman Sendromu, 263800, Otozomal Resesif; GTLMNS (Gitelman Sendromu) (Prenatal) (MLPA)	Gitelman syndrome, 263800, Autosomal recessive; GTLMNS (Gitelman syndrome) (Prenatal) (MLPA)	SLC12A3	SLC12A3, NCCT, TSC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Gitelman syndrome (GS) [MIM:263800]: An autosomal recessive disorder characterized by hypokalemic alkalosis in combination with hypomagnesemia, low urinary calcium, and increased renin activity associated with normal blood pressure. Patients are often asymptomatic or present transient periods of muscular weakness and tetany, usually accompanied by abdominal pain, vomiting and fever. The phenotype is highly heterogeneous in terms of age at onset and severity. Cardinal features such as hypocalciuria and hypomagnesemia might also change during the life cycle of a given patient. It has overlapping features with Bartter syndrome. {ECO:0000269|PubMed:10616841, ECO:0000269|PubMed:10988270, ECO:0000269|PubMed:11168953, ECO:0000269|PubMed:11940055, ECO:0000269|PubMed:12008755, ECO:0000269|PubMed:12112667, ECO:0000269|PubMed:15069170, ECO:0000269|PubMed:15687331, ECO:0000269|PubMed:16429844, ECO:0000269|PubMed:17654016, ECO:0000269|PubMed:17873326, ECO:0000269|PubMed:8528245, ECO:0000269|PubMed:8900229, ECO:0000269|PubMed:8954067, ECO:0000269|PubMed:9734597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glanzmann Trombastenisi, 273800, Otozomal Resesif (Glanzmann Trombastenisi) (ITGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glanzmann thrombasthenia, 273800, Autosomal recessive (Glanzmann thrombasthenia) (ITGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB3	ITGB3, GP3A, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glanzmann Trombastenisi, 273800, Otozomal Resesif; GT (Glanzmann Trombastenisi) (ITGA2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glanzmann thrombasthenia, 273800, Autosomal recessive; GT (Glanzmann thrombasthenia) (ITGA2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA2B	ITGA2B, GP2B, CD41B, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glass Sendromu, 612313, Otozomal Dominant (2q32q33 Mikrodelesyon Sendromu) (SATB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glass syndrome, 612313, Autosomal dominant (2q32q33 microdeletion syndrome) (SATB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SATB2	SATB2, KIAA1034, GLSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Glass Sendromu, 612313, Otozomal Dominant (2q32q33 Mikrodelesyon Sendromu) (Prenatal) (SATB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glass syndrome, 612313, Autosomal dominant (2q32q33 microdeletion syndrome) (SATB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SATB2	SATB2, KIAA1034, GLSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glokom 1, Açık Açılı, K, 613100 (NTF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 1, open angle, 1O, 613100 (NTF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTF4	NTF4 , NTF5, NT5, NT4, GLC1O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glokom 1, Açık Açılı, E, 137760, Otozomal Dominant (OPTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 1, open angle, E, 137760, Autosomal dominant (OPTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPTN	OPTN, GLC1E, FIP2, HYPL, NRP, ALS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glaucoma 1, open angle, E (GLC1E) [MIM:137760]: A form of primary open angle glaucoma (POAG). POAG is characterized by a specific pattern of optic nerve and visual field defects. The angle of the anterior chamber of the eye is open, and usually the intraocular pressure is increased. However, glaucoma can occur at any intraocular pressure. The disease is generally asymptomatic until the late stages, by which time significant and irreversible optic nerve damage has already taken place. {ECO:0000269|PubMed:11834836, ECO:0000269|PubMed:12939304, ECO:0000269|PubMed:14507044, ECO:0000269|PubMed:15226658, ECO:0000269|PubMed:15326130, ECO:0000269|PubMed:15557444}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Glokom 1, Açık Açılı, K, 603383; GLC1F (ASB10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 1, open angle, F, 603383; GLC1F (ASB10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASB10	ASB10, GLC1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glaucoma 1, open angle, F (GLC1F) [MIM:603383]: A form of primary open angle glaucoma (POAG). POAG is characterized by a specific pattern of optic nerve and visual field defects. The angle of the anterior chamber of the eye is open, and usually the intraocular pressure is increased. However, glaucoma can occur at any intraocular pressure. The disease is generally asymptomatic until the late stages, by which time significant and irreversible optic nerve damage has already taken place. {ECO:0000269|PubMed:22156576}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glokom 1, Açık Açılı, G, 609887; GLC1G (WDR36 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 1, open angle, G, 609887; GLC1G (WDR36 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR36	WDR36, TAWDRP, GLC1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glaucoma 1, open angle, G (GLC1G) [MIM:609887]: A form of primary open angle glaucoma (POAG). POAG is characterized by a specific pattern of optic nerve and visual field defects. The angle of the anterior chamber of the eye is open, and usually the intraocular pressure is increased. However, glaucoma can occur at any intraocular pressure. The disease is generally asymptomatic until the late stages, by which time significant and irreversible optic nerve damage has already taken place. {ECO:0000269|PubMed:15677485}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glokom 1A, Primer Açık Açılı, 137750, Otozomal Dominant; GLC1A (Jüvenil Glokom) (MYOC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 1A, primary open angle, 137750, Autosomal dominant; GLC1A (Juvenile glaucoma) (MYOC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYOC	MYOC, TIGR, GLC1A, JOAG, GPOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glaucoma 1, open angle, A (GLC1A) [MIM:137750]: A form of primary open angle glaucoma (POAG). POAG is characterized by a specific pattern of optic nerve and visual field defects. The angle of the anterior chamber of the eye is open, and usually the intraocular pressure is increased. However, glaucoma can occur at any intraocular pressure. The disease is generally asymptomatic until the late stages, by which time significant and irreversible optic nerve damage has already taken place. {ECO:0000269|PubMed:10196380, ECO:0000269|PubMed:10330365, ECO:0000269|PubMed:10340788, ECO:0000269|PubMed:10644174, ECO:0000269|PubMed:10798654, ECO:0000269|PubMed:10819638, ECO:0000269|PubMed:10873982, ECO:0000269|PubMed:10916185, ECO:0000269|PubMed:10980537, ECO:0000269|PubMed:11004290, ECO:0000269|PubMed:11774072, ECO:0000269|PubMed:12189160, ECO:0000269|PubMed:12356829, ECO:0000269|PubMed:12362081, ECO:0000269|PubMed:12442283, ECO:0000269|PubMed:12860809, ECO:0000269|PubMed:12872267, ECO:0000269|PubMed:15025728, ECO:0000269|PubMed:15255110, ECO:0000269|PubMed:15534471, ECO:0000269|PubMed:16401791, ECO:0000269|PubMed:17210859, ECO:0000269|PubMed:17499207, ECO:0000269|PubMed:9095863,	EDTA Blood Sample
Glokom 3, Primer Konjenital, D, 613086; GLC3D (Konjenital Glokom) (LTBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 3, primary congenital, D, 613086; GLC3D (Congenital glaucoma) (LTBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTBP2	LTBP2, LTBP3, GLC3D, MSPKA, WMS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glokom 3, Primer Konjenital, E, 617272, Otozomal Dominant (Konjenital Glokom) (TEK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 3, primary congenital, E, 617272, Autosomal dominant (Congenital glaucoma) (TEK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TEK	TEK, TIE2, VMCM, GLC3E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Glokom 3A, Primer Açık Açılı, Konjenital, Çocuk veya Yetişkin Başlangıç, 231300, Otozomal Resesif; GLC3A(Konjenital Glokom) (CYP1B1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300, Autosomal recessive; GLC3A (Congenital glaucoma) (CYP1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP1B1	CYP1B1, GLC3A, ASGD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glokom 3A, Primer Açık Açılı, Konjenital, Çocuk veya Yetişkin Başlangıç, 231300, Otozomal Resesif; GLC3A(Konjenital Glokom) (MLPA)	Glaucoma 3A, primary open angle, congenital, juvenile, or adult onset, 231300, Autosomal recessive; GLC3A (Congenital glaucoma) (MLPA)	CYP1B1	CYP1B1, GLC3A, ASGD6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Glokom, Normal Basınçlı, Yatkınlık, 606657 (OPTN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma, normal tension, susceptibility to, 606657 (OPTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPTN	OPTN, GLC1E, FIP2, HYPL, NRP, ALS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glokom, Normal Tansiyon, Yatkınlık, 606657 (Otozomal Dominant Optik Atrofi Plus Sendromu) (OPA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glaucoma, normal tension, susceptibility to, 606657 (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glioblastoma 3, 613029, Otozomal Resesif; GLM3 (Glial Tümör) (BRCA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glioblastoma 3, 613029, Autosomal recessive; GLM3 (Glial tumor) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glioblastoma 3, 613029, Otozomal Resesif; GLM3 (Glial Tümör) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Glioma Yatkınlık 1, 137800, Otozomal Dominant, Somatik Mutasyon (Glial Tümör) (TP53 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glioma susceptibility 1, 137800, Autosomal dominant, Somatic mutation (Glial tumor) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Glioma Yatkınlık 1, 137800, Otozomal Dominant, Somatik Mutasyon (Glial Tümör) (MLPA)	Glioma susceptibility 1, 137800, Autosomal dominant, Somatic mutation (Glial tumor) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		20	60	.	.	30	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Glioma Yatkınlık 2, 613028, GLM2 (Glial Tümör) (PTEN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glioma susceptibility 2, 613028, GLM2 (Glial tumor) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glioma Yatkınlık 2, 613028, GLM2 (Glial Tümör) (MLPA)	Glioma susceptibility 2, 613028, GLM2 (Glial tumor) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Glioma Yatkınlık 9, 616568, Otozomal Dominant (Anaplastik Oligodendrogliom) (POT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glioma susceptibility 9, 616568, Autosomal dominant (Anaplastic oligodendroglioma) (POT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POT1	POT1, CMM10, GLM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glioma, maligen (EGFR, TP53, PTEN + bazı diğer) (MLPA)	Glioma, malignant (EGFR, TP53, PTEN + some other) (MLPA)	EGFR, TP53, PTEN + bazı diğer	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Glioma, Yatkınlık, Somatik, 137800; GLM1 (Glial Tümör) (Kodon 132 ve IDH2 Kodon 172 Mutasyonları) (IDH1 gen) (Dizi Analizi) (Postnatal)	Glioma, susceptibility to, somatic, 137800; GLM1 (Glial tumor) (Codon 132 and IDH2 172 mutations) (IDH1 gene) (Sequence Analysis) (Postnatal)	IDH1, IDH2	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Glioma (GLM) [MIM:137800]: Gliomas are benign or malignant central nervous system neoplasms derived from glial cells. They comprise astrocytomas and glioblastoma multiforme that are derived from astrocytes, oligodendrogliomas derived from oligodendrocytes and ependymomas derived from ependymocytes. {ECO:0000269|PubMed:19117336, ECO:0000269|PubMed:19935646}. Note=The gene represented in this entry is involved in disease pathogenesis. Mutations affecting Arg-132 are tissue-specific, and suggest that this residue plays a unique role in the development of high-grade gliomas. Mutations of Arg-132 to Cys, His, Leu or Ser abolish magnesium binding and abolish the conversion of isocitrate to alpha-ketoglutarate. Instead, alpha-ketoglutarate is converted to R(-)-2-hydroxyglutarate. Elevated levels of R(-)-2-hydroxyglutarate are correlated with an elevated risk of malignant brain tumors. {ECO:0000269|PubMed:19935646}.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Yaygın Gelişimsel Gecikme, Aplastik-Hipoplastik Korpus Kallozum ve Dismorfik Yüz, 617260, Otozomal Dominant; GDACCF (ZNF148 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260, Autosomal dominant; GDACCF (ZNF148 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF148	ZNF148, ZFP148, GDACCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yaygın Gelişimsel Gecikme, Aplastik-Hipoplastik Korpus Kallozum ve Dismorfik Yüz, 617260, Otozomal Dominant; GDACCF (Prenatal) (ZNF148 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Global developmental delay, absent or hypoplastic corpus callosum, and dysmorphic facies, 617260, Autosomal dominant; GDACCF (ZNF148 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNF148	ZNF148, ZFP148, GDACCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glomerülokistik Böbrek Hastalığı-Hiperürisemi ve İzostenüri, 609886 (Otozomal Dominant Medüller Kistik Böbrek Hastalık ± Hiperürisemi) (UMOD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (UMOD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UMOD	UMOD, HNFJ1, FJHN, MCKD2, ADMCKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glomerülokistik Böbrek Hastalığı-Hiperürisemi ve İzostenüri, 609886 (Otozomal Dominant Medüller Kistik Böbrek Hastalık ± Hiperürisemi) (Prenatal) (UMOD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Glomerulocystic kidney disease with hyperuricemia and isosthenuria, 609886 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (UMOD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UMOD	UMOD, HNFJ1, FJHN, MCKD2, ADMCKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glomerülopati-Fibronektin Depozitleri 2, 601894, Otozomal Dominant; GFND2 (Fibronektin Glomerülopati) (FN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulopathy with fibronectin deposits 2, 601894, Autosomal dominant; GFND2 (Fibronectin glomerulopathy) (FN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FN1	FN1, FN, LETS, FNZ, GFND2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glomerulopathy with fibronectin deposits 2 (GFND2) [MIM:601894]: Genetically heterogeneous autosomal dominant disorder characterized clinically by proteinuria, microscopic hematuria, and hypertension that leads to end-stage renal failure in the second to fifth decade of life. {ECO:0000269	PubMed:18268355}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 1, 603278, Otozomal Dominant (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (ACTN4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 1, 603278, Autosomal dominant (Familial idiopathic steroid-resistant nephrotic syndrome) (ACTN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTN4	ACTN4, FSGS1, FSGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 1 (FSGS1) [MIM:603278]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. {ECO:0000269|PubMed:10700177, ECO:0000269|PubMed:19436095, ECO:0000269|PubMed:23890478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 2, 603965; FSGS2 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (TRPC6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 2, 603965; FSGS2 (Familial idiopathic steroid-resistant nephrotic syndrome) (TRPC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPC6	TRPC6, TRP6, FSGS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 2 (FSGS2) [MIM:603965]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. {ECO:0000269|PubMed:15879175, ECO:0000269|PubMed:15924139, ECO:0000269|PubMed:21734084}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 3, 607832; FSGS3 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (CD2AP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 3, 607832; FSGS3 (Familial idiopathic steroid-resistant nephrotic syndrome) (CD2AP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD2AP	CD2AP, CMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 3 (FSGS3) [MIM:607832]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. {ECO:0000269	PubMed:12764196}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 4, Yatkınlık, 612551 (Sporadik İdyopatik Steroide Dirençli Nefrotik Sendrom) (APOL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 4, susceptibility to, 612551 (Sporadic idiopathic steroid-resistant nephrotic syndrome) (APOL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOL1	APOL1, FSGS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 4 (FSGS4) [MIM:612551]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. {ECO:0000269|PubMed:20635188, ECO:0000269|PubMed:20647424}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 5, 613237; FSGS5 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (INF2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 5, 613237; FSGS5 (Familial idiopathic steroid-resistant nephrotic syndrome) (INF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INF2	INF2, FSGS5, C14orf173, CMTDIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Focal segmental glomerulosclerosis 5 (FSGS5) [MIM:613237]: A renal pathology defined by the presence of segmental sclerosis in glomeruli and resulting in proteinuria, reduced glomerular filtration rate and progressive decline in renal function. Renal insufficiency often progresses to end-stage renal disease, a highly morbid state requiring either dialysis therapy or kidney transplantation. {ECO:0000269|PubMed:20023859, ECO:0000269|PubMed:21258034, ECO:0000269|PubMed:21866090, ECO:0000269|PubMed:22971997, ECO:0000269|PubMed:25165188}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 6, 614131, Otozomal Resesif; FSGS6 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (MYO1E gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 6, 614131, Autosomal recessive; FSGS6 (Familial idiopathic steroid-resistant nephrotic syndrome) (MYO1E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO1E	MYO1E, MYO1C, FSGS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Glomerüloskleroz, Fokal Segmental, 7, 616002, Otozomal Dominant; FSGS7 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (PAX2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomerulosclerosis, focal segmental, 7, 616002, Autosomal dominant; FSGS7 (Familial idiopathic steroid-resistant nephrotic syndrome) (PAX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX2	PAX2, PAPRS, FSGS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glomuvenöz Malformasyonlar, 138000, Otozomal Dominant; GVM (Glomuvenöz Malformasyon) (GLMN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glomuvenous malformations, 138000, Autosomal dominant; GVM (Glomuvenous malformation) (GLMN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLMN	GLML, GVM, VMGLOM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glomuvenous malformations (GVMs) [MIM:138000]: Characterized by the presence of smooth-muscle-like glomus cells in the media surrounding distended vascular lumens. {ECO:0000269	PubMed:11845407}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glukokortikoid Eksikliği 2, 607398, Otozomal Resesif; GCCD2 (Ailesel Glukokortikoid Eksikliği) (MRAP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glucocorticoid deficiency 2, 607398, Autosomal recessive; GCCD2 (Familial glucocorticoid deficiency) (MRAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRAP	MRAP_FALP, C21orf61, GCCD2, FGD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glukokortikoid Eksikliği 4, ± Veya Mineralokortikoid Eksikliği, 614736, Otozomal Resesif; GCCD4 (Ailesel Glukokortikoid Eksikliği) (NNT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736, Autosomal recessive; GCCD4 (Familial glucocorticoid deficiency) (NNT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NNT	NNT, GCCD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glukokortikoid Eksikliği, ACTH Cevapsızlığına Bağlı, 202200, Otozomal Resesif; GCCD1 (Ailesel Glukokortikoid Eksikliği) (MC2R gen) (Tüm Gen Dizi Analizi) (Postnatal)	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200, Autosomal recessive; GCCD1 (Familial glucocorticoid deficiency) (MC2R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC2R	MC2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Glukokortikoid Direnci, 615962, Otozomal Dominant; GCCR (Glukokortikoid Direnci) (NR3C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glucocorticoid resistance, 615962, Autosomal dominant; GCCR (Glucocorticoid resistance) (NR3C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR3C1	NR3C1, GCR, GRL, GCRST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glucocorticoid resistance, generalized (GCCR) [MIM:615962]: An autosomal dominant disease characterized by increased plasma cortisol concentration and high urinary free cortisol, resistance to adrenal suppression by dexamethasone, and the absence of Cushing syndrome typical signs. Clinical features include hypoglycemia, hypertension, metabolic alkalosis, chronic fatigue and profound anxiety. {ECO:0000269|PubMed:11589680, ECO:0000269|PubMed:11701741, ECO:0000269|PubMed:12050230, ECO:0000269|PubMed:15769988, ECO:0000269|PubMed:1704018, ECO:0000269|PubMed:17635946, ECO:0000269|PubMed:7683692}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glukokortikoid Tedavisi, Yanıt, 614400; GCTR (GLCCI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glucocorticoid therapy, response to, 614400; GCTR (GLCCI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLCCI1	GLCCI1, TSSN1, GCTR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glikoz / Galaktoz Malabsorpsiyon, 606824, Otozomal Resesif, GGM (Glukoz-Galaktoz Malabsorpsiyon) (SLC5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glucose/galactose malabsorption, 606824, Autosomal recessive; GGM (Glucose-galactose malabsorption) (SLC5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC5A1	SLC5A1, SGLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital glucose/galactose malabsorption (GGM) [MIM:606824]: Intestinal monosaccharide transporter deficiency. It is an autosomal recessive disorder manifesting itself within the first weeks of life. It is characterized by severe diarrhea and dehydration which are usually fatal unless glucose and galactose are eliminated from the diet. {ECO:0000269|PubMed:10036327, ECO:0000269|PubMed:11406349, ECO:0000269|PubMed:2008213, ECO:0000269|PubMed:8195156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikoz / Galaktoz Malabsorpsiyon, 606824, Otozomal Resesif, GGM (Glukoz-Galaktoz Malabsorpsiyon) (Prenatal) (SLC5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glucose/galactose malabsorption, 606824, Autosomal recessive; GGM (Glucose-galactose malabsorption) (SLC5A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC5A1	SLC5A1, SGLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital glucose/galactose malabsorption (GGM) [MIM:606824]: Intestinal monosaccharide transporter deficiency. It is an autosomal recessive disorder manifesting itself within the first weeks of life. It is characterized by severe diarrhea and dehydration which are usually fatal unless glucose and galactose are eliminated from the diet. {ECO:0000269|PubMed:10036327, ECO:0000269|PubMed:11406349, ECO:0000269|PubMed:2008213, ECO:0000269|PubMed:8195156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GLUT1 Eksikliği Sendromu, 1, İnfantil Başlangıçlı, Ağır, 606777, Otozomal Resesif, Otozomal Dominant; GLUT1DS1 (Ensefalopati, GLUT1 Eksikliğine Bağlı) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777, Autosomal recessive, Autosomal dominant; GLUT1DS1 (Encephalopathy due to GLUT1 deficiency) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLUT1 Eksikliği Sendromu, 1, İnfantil Başlangıçlı, Ağır, 606777, Otozomal Resesif, Otozomal Dominant; GLUT1DS1 (Ensefalopati, GLUT1 Eksikliğine Bağlı) (MLPA)	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777, Autosomal recessive, Autosomal dominant; GLUT1DS1 (Encephalopathy due to GLUT1 deficiency) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
GLUT1 Eksikliği Sendromu, 1, İnfantil Başlangıçlı, Ağır, 606777, Otozomal Resesif, Otozomal Dominant; GLUT1DS1 (Ensefalopati, GLUT1 Eksikliğine Bağlı) (Prenatal) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777, Autosomal recessive, Autosomal dominant; GLUT1DS1 (Encephalopathy due to GLUT1 deficiency) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GLUT1 Eksikliği Sendromu, 1, İnfantil Başlangıçlı, Ağır, 606777, Otozomal Resesif, Otozomal Dominant; GLUT1DS1 (Ensefalopati, GLUT1 Eksikliğine Bağlı) (Prenatal) (MLPA)	GLUT1 deficiency syndrome 1, infantile onset, severe, 606777, Autosomal recessive, Autosomal dominant; GLUT1DS1 (Encephalopathy due to GLUT1 deficiency) (Prenatal) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GLUT1 Eksikliği Sendromu 2, Çocuklukta Başlayan, 612126, Otozomal Dominant; GLUT1DS2 (Paroksismal Efor Kaynaklı Diskinezi) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GLUT1 deficiency syndrome 2, childhood onset, 612126, Autosomal dominant; GLUT1DS2 (Paroxysmal exertion-induced dyskinesia) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLUT1 Eksikliği Sendromu 2, Çocuklukta Başlayan, 612126, Otozomal Dominant; GLUT1DS2 (Paroksismal Efor Kaynaklı Diskinezi) (MLPA)	GLUT1 deficiency syndrome 2, childhood onset, 612126, Autosomal dominant; GLUT1DS2 (Paroxysmal exertion-induced dyskinesia) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
GLUT1 Eksikliği Sendromu 2, Çocuklukta Başlayan, 612126, Otozomal Dominant; GLUT1DS2 (Paroksismal Efor Kaynaklı Diskinezi) (Prenatal) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GLUT1 deficiency syndrome 2, childhood onset, 612126, Autosomal dominant; GLUT1DS2 (Paroxysmal exertion-induced dyskinesia) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GLUT1 Eksikliği Sendromu 2, Çocuklukta Başlayan, 612126, Otozomal Dominant; GLUT1DS2 (Paroksismal Efor Kaynaklı Diskinezi) (Prenatal) (MLPA)	GLUT1 deficiency syndrome 2, childhood onset, 612126, Autosomal dominant; GLUT1DS2 (Paroxysmal exertion-induced dyskinesia) (Prenatal) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutamat Formiminotransferaz Eksikliği, 229100, Otozomal Resesif (Formiminoglutamik Asiduri) (FTCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutamate formiminotransferase deficiency, 229100, Autosomal recessive (Formiminoglutamic aciduria) (FTCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTCD	FTCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutamate formiminotransferase deficiency (FIGLUURIA) [MIM:229100]: Autosomal recessive disorder. Features of a severe phenotype, include elevated levels of formiminoglutamate (FIGLU) in the urine in response to histidine administration, megaloblastic anemia, and mental retardation. Features of a mild phenotype include high urinary excretion of FIGLU in the absence of histidine administration, mild developmental delay, and no hematological abnormalities. {ECO:0000269|PubMed:12815589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutamat Formiminotransferaz Eksikliği, 229100, Otozomal Resesif (Formiminoglutamik Asiduri) (Prenatal) (FTCD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutamate formiminotransferase deficiency, 229100, Autosomal recessive (Formiminoglutamic aciduria) (FTCD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FTCD	FTCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutamate formiminotransferase deficiency (FIGLUURIA) [MIM:229100]: Autosomal recessive disorder. Features of a severe phenotype, include elevated levels of formiminoglutamate (FIGLU) in the urine in response to histidine administration, megaloblastic anemia, and mental retardation. Features of a mild phenotype include high urinary excretion of FIGLU in the absence of histidine administration, mild developmental delay, and no hematological abnormalities. {ECO:0000269|PubMed:12815589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutamin Eksikliği, Konjenital, 610015, Otozomal Resesif (Konjenital Beyin Disgenezisi, Glutamin Sentaz Eksikliğine Bağlı) (GLUL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutamine deficiency, congenital, 610015, Autosomal recessive (Congenital brain dysgenesis due to glutamine synthetase deficiency) (GLUL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLUL	GLUL, GLNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital systemic glutamine deficiency (CSGD) [MIM:610015]: Rare developmental disorder with severe brain malformation resulting in multi-organ failure and neonatal death. Glutamine is largely absent from affected patients serum, urine and cerebrospinal fluid. {ECO:0000269|PubMed:16267323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutamin Eksikliği, Konjenital, 610015, Otozomal Resesif (Konjenital Beyin Disgenezisi, Glutamin Sentaz Eksikliğine Bağlı) (Prenatal) (GLUL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutamine deficiency, congenital, 610015, Autosomal recessive (Congenital brain dysgenesis due to glutamine synthetase deficiency) (GLUL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLUL	GLUL, GLNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital systemic glutamine deficiency (CSGD) [MIM:610015]: Rare developmental disorder with severe brain malformation resulting in multi-organ failure and neonatal death. Glutamine is largely absent from affected patients serum, urine and cerebrospinal fluid. {ECO:0000269|PubMed:16267323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutarik Asidemi 2A, 231680, Otozomal Resesif, MADD (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (ETFA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutaric acidemia IIA, 231680, Autosomal recessive; MADD (Multiple acyl-CoA dehydrogenase deficiency) (ETFA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETFA	ETFA, GA2, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutaric aciduria 2A (GA2A) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:1430199}. ECO:0000269|PubMed:1882842}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutarik Asidemi 2A, 231680, Otozomal Resesif, MADD (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (Prenatal) (ETFA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutaric acidemia IIA, 231680, Autosomal recessive; MADD (Multiple acyl-CoA dehydrogenase deficiency) (ETFA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ETFA	ETFA, GA2, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutaric aciduria 2A (GA2A) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:1430199}. ECO:0000269|PubMed:1882842}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutarik Asidemi 2B, 231680, Otozomal Resesif (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (ETFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutaric acidemia IIB, 231680, Autosomal recessive (Multiple acyl-CoA dehydrogenase deficiency) (ETFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETFB	ETFB, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutaric aciduria 2B (GA2B) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:12815589, ECO:0000269|PubMed:7912128}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutarik Asidemi 2B, 231680, Otozomal Resesif (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (Prenatal) (ETFB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutaric acidemia IIB, 231680, Autosomal recessive (Multiple acyl-CoA dehydrogenase deficiency) (ETFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ETFB	ETFB, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutaric aciduria 2B (GA2B) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:12815589, ECO:0000269|PubMed:7912128}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutarik Asidemi 2C, 231680, Otozomal Resesif (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (ETFDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutaric acidemia IIC, 231680, Autosomal recessive (Multiple acyl-CoA dehydrogenase deficiency) (ETFDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETFDH	ETFDH, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:12815589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutarik Asidemi 2C, 231680, Otozomal Resesif (Multipl Açil-CoA Dehidrojenaz Yetmezliği) (Prenatal) (ETFDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutaric acidemia IIC, 231680, Autosomal recessive (Multiple acyl-CoA dehydrogenase deficiency) (ETFDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ETFDH	ETFDH, MADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutaric aciduria 2C (GA2C) [MIM:231680]: An autosomal recessively inherited disorder of fatty acid, amino acid, and choline metabolism. It is characterized by multiple acyl-CoA dehydrogenase deficiencies resulting in large excretion not only of glutaric acid, but also of lactic, ethylmalonic, butyric, isobutyric, 2-methyl-butyric, and isovaleric acids. {ECO:0000269|PubMed:12815589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Glutarikasidüri 3 231690, Otozomal Resesif (Glutarik Asidemi Tip 3) (SUGCT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutaric aciduria III, 231690, Autosomal recessive (Glutaric acidemia type 3) (SUGCT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUGCT	C7orf10, GA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glutarikasidüri 3 231690, Otozomal Resesif (Glutarik Asidemi Tip 3) (Prenatal) (SUGCT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutaric aciduria III, 231690, Autosomal recessive (Glutaric acidemia type 3) (SUGCT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUGCT	C7orf10, GA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutarikasidüri, Tip I, 231670, Otozomal Resesif (Glutaril-CoA Dehidrojenaz Eksikliği) (GCDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutaricaciduria, type I, 231670, Autosomal recessive (Glutaryl-CoA dehydrogenase deficiency) (GCDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCDH	GCDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glutarikasidüri, Tip I, 231670, Otozomal Resesif (Glutaril-CoA Dehidrojenaz Eksikliği) (Prenatal) (GCDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutaricaciduria, type I, 231670, Autosomal recessive (Glutaryl-CoA dehydrogenase deficiency) (GCDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GCDH	GCDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutatyon Sentetaz Eksikliği, 266130, Otozomal Resesif; GSSD (Glutatyon Sentetaz Eksikliği) (GSS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutathione synthetase deficiency, 266130, Autosomal recessive; GSSD (Glutathione synthetase deficiency) (GSS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSS	GSS, GSHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutathione synthetase deficiency of erythrocytes (GLUSYNDE) [MIM:231900]: Mild form causing hemolytic anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glutatyon Sentetaz Eksikliği, 266130, Otozomal Resesif; GSSD (Glutatyon Sentetaz Eksikliği) (GSS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutathione synthetase deficiency, 266130, Autosomal recessive; GSSD (Glutathione synthetase deficiency) (GSS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GSS	GSS, GSHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutathione synthetase deficiency of erythrocytes (GLUSYNDE) [MIM:231900]: Mild form causing hemolytic anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glutatyonüriürl (Gama-Glutamil Transpeptidaz Eksikliği) (GGT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glutathionirnuria (Gamma-glutamyl transpeptidase deficiency) (GGT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GGT1	GGT1, GTG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glutatyonüriürl (Gama-Glutamil Transpeptidaz Eksikliği) (Prenatal) (GGT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glutathionirnuria (Gamma-glutamyl transpeptidase deficiency) (GGT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GGT1	GGT1, GTG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gliserol Kinaz Eksikliği, 307030, X'e Bağlı; GKD (İzole Gliserol Kinaz Eksikliği) (GK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycerol kinase deficiency, 307030, X-linked recessive; GKD (Isolated glycerol kinase deficiency) (GK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GK	GK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gliserol Kinaz Eksikliği, 307030, X'e Bağlı; GKD (İzole Gliserol Kinaz Eksikliği) (Prenatal) (GK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycerol kinase deficiency, 307030, X-linked recessive; GKD (Isolated glycerol kinase deficiency) (GK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GK	GK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin Ensefalopatisi, Normal Serum Glisin İle, 617301, Otozomal Resesif (Atipik Glisin Ensefalopati) (SLC6A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycine encephalopathy with normal serum glycine, 617301, Autosomal recessive (Atypical glycine encephalopathy) (SLC6A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A9	SLC6A9, GLYT1, GCENSG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glisin Ensefalopatisi, Normal Serum Glisin İle, 617301, Otozomal Resesif (Atipik Glisin Ensefalopati) (Prenatal) (SLC6A9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycine encephalopathy with normal serum glycine, 617301, Autosomal recessive (Atypical glycine encephalopathy) (SLC6A9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A9	SLC6A9, GLYT1, GCENSG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (GLDC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (GLDC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLDC	GLDC, HYGN1, GCSP, GCE, NKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:11592811, ECO:0000269|PubMed:1634607, ECO:0000269|PubMed:1996985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (GCSH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (GCSH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCSH	GCSH, NKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (MLPA)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (MLPA)	GLDC	GLDC, HYGN1, GCSP, GCE, NKH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:11592811, ECO:0000269|PubMed:1634607, ECO:0000269|PubMed:1996985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (Prenatal) (GLDC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (GLDC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLDC	GLDC, HYGN1, GCSP, GCE, NKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:11592811, ECO:0000269|PubMed:1634607, ECO:0000269|PubMed:1996985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (Prenatal) (GCSH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (GCSH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GCSH	GCSH, NKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif (Glisin Ensefalopatisi) (Prenatal) (MLPA)	Glycine encephalopathy, 605899, Autosomal recessive (Glycine encephalopathy) (Prenatal) (MLPA)	GLDC	GLDC, HYGN1, GCSP, GCE, NKH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:11592811, ECO:0000269|PubMed:1634607, ECO:0000269|PubMed:1996985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif; GCE (Glisin Ensefalopatisi) (AMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycine encephalopathy, 605899, Autosomal recessive; GCE (Glycine encephalopathy) (AMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMT	AMT, NKH, GCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:10873393, ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:26371980, ECO:0000269|PubMed:8005589, ECO:0000269|PubMed:9600239, ECO:0000269|PubMed:9621520}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glisin Ensefalopatisi, 605899, Otozomal Resesif; GCE (Glisin Ensefalopatisi) (Prenatal) (AMT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycine encephalopathy, 605899, Autosomal recessive; GCE (Glycine encephalopathy) (AMT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMT	AMT, NKH, GCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-ketotic hyperglycinemia (NKH) [MIM:605899]: Autosomal recessive disease characterized by accumulation of a large amount of glycine in body fluid and by severe neurological symptoms. {ECO:0000269|PubMed:10873393, ECO:0000269|PubMed:11286506, ECO:0000269|PubMed:26371980, ECO:0000269|PubMed:8005589, ECO:0000269|PubMed:9600239, ECO:0000269|PubMed:9621520}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glisin N-Metil Transferaz Eksikliği, 606664, Otozomal Resesif (Hipermetioninemi, Glisin N-Metiltransferaz Eksikliğine Bağlı) (GNMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycine N-methyltransferase deficiency, 606664, Autosomal recessive (Hypermethioninemia due to glycine N-methyltransferase deficiency) (GNMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNMT	GNMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glisin N-Metil Transferaz Eksikliği, 606664, Otozomal Resesif (Hipermetioninemi, Glisin N-Metiltransferaz Eksikliğine Bağlı) (Prenatal) (GNMT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycine N-methyltransferase deficiency, 606664, Autosomal recessive (Hypermethioninemia due to glycine N-methyltransferase deficiency) (GNMT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNMT	GNMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı, 0, Karaciğer, 240600, Otozomal Resesif; GSD0A (Glikojen Depo Hastalığı, Hepatik Glikojen Sentaz Eksikliğine Bağlı) (GYS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease 0, liver, 240600, Autosomal recessive; GSD0A (Glycogen storage disease due to hepatic glycogen synthase deficiency) (GYS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYS2	GYS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 0 (GSD0) [MIM:240600]: A metabolic disorder characterized by fasting hypoglycemia presenting in infancy or early childhood, high blood ketones and low alanine and lactate concentrations. Although feeding relieves symptoms, it often results in postprandial hyperglycemia and hyperlactatemia. {ECO:0000269	PubMed:9691087}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı, 0, Karaciğer, 240600, Otozomal Resesif; GSD0A (Glikojen Depo Hastalığı, Hepatik Glikojen Sentaz Eksikliğine Bağlı) (Prenatal) (GYS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease 0, liver, 240600, Autosomal recessive; GSD0A (Glycogen storage disease due to hepatic glycogen synthase deficiency) (GYS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GYS2	GYS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 0 (GSD0) [MIM:240600]: A metabolic disorder characterized by fasting hypoglycemia presenting in infancy or early childhood, high blood ketones and low alanine and lactate concentrations. Although feeding relieves symptoms, it often results in postprandial hyperglycemia and hyperlactatemia. {ECO:0000269	PubMed:9691087}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı, 0, Kas, 611556, Otozomal Resesif; GSD0B (Glikojen Depo Hastalığı, Kas ve Kalp Glikojen Sentaz Eksikliği) (GYS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease 0, muscle, 611556, Autosomal recessive; GSD0B (Glycogen storage disease due to muscle and heart glycogen synthase deficiency) (GYS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYS1	GYS1, GYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscle glycogen storage disease 0 (GSD0b) [MIM:611556]: Metabolic disorder characterized by fasting hypoglycemia presenting in infancy or early childhood. The role of muscle glycogen is to provide critical energy during bursts of activity and sustained muscle work. {ECO:0000269	PubMed:17928598}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı, 0, Kas, 611556, Otozomal Resesif; GSD0B (Glikojen Depo Hastalığı, Kas ve Kalp Glikojen Sentaz Eksikliği) (Prenatal) (GYS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease 0, muscle, 611556, Autosomal recessive; GSD0B (Glycogen storage disease due to muscle and heart glycogen synthase deficiency) (GYS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GYS1	GYS1, GYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscle glycogen storage disease 0 (GSD0b) [MIM:611556]: Metabolic disorder characterized by fasting hypoglycemia presenting in infancy or early childhood. The role of muscle glycogen is to provide critical energy during bursts of activity and sustained muscle work. {ECO:0000269	PubMed:17928598}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Glikojen Depo Hastalığı 1a, 232200, Otozomal Resesif, GSD1A (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (G6PC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease Ia, 232200, Autosomal recessive; GSD1A (Glycogen storage disease due to glucose-6-phosphatase deficiency) (G6PC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PC	G6PC, G6PT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 1A (GSD1A) [MIM:232200]: A metabolic disorder characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia. (ECO:0000269|PubMed:10070617, ECO:0000269|PubMed:10094563, ECO:0000269|PubMed:10447271, ECO:0000269|PubMed:10612834, ECO:0000269|PubMed:10738005, ECO:0000269|PubMed:10748407, ECO:0000269|PubMed:10874313, ECO:0000269|PubMed:10960498, ECO:0000269|PubMed:11058903, ECO:0000269|PubMed:11058910, ECO:0000269|PubMed:12373566, ECO:0000269|PubMed:15151508, ECO:0000269|PubMed:15316959, ECO:0000269|PubMed:15542400, ECO:0000269|PubMed:7573034, ECO:0000269|PubMed:7623438, ECO:0000269|PubMed:7655466, ECO:0000269|PubMed:8182131, ECO:0000269|PubMed:8733042, ECO:0000269|PubMed:9001800, ECO:0000269|PubMed:9332655, ECO:0000269|PubMed:9506659, ECO:0000269|PubMed:9700612, ECO:0000269|PubMed:9706613). Note=The disease is caused	EDTA Blood Sample
Glikojen Depo Hastalığı 1a, 232200, Otozomal Resesif, GSD1A (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (Prenatal) (G6PC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease Ia, 232200, Autosomal recessive; GSD1A (Glycogen storage disease due to glucose-6-phosphatase deficiency) (G6PC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	G6PC	G6PC, G6PT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 1A (GSD1A) [MIM:232200]: A metabolic disorder characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia. (ECO:0000269|PubMed:10070617, ECO:0000269|PubMed:10094563, ECO:0000269|PubMed:10447271, ECO:0000269|PubMed:10612834, ECO:0000269|PubMed:10738005, ECO:0000269|PubMed:10748407, ECO:0000269|PubMed:10874313, ECO:0000269|PubMed:10960498, ECO:0000269|PubMed:11058903, ECO:0000269|PubMed:11058910, ECO:0000269|PubMed:12373566, ECO:0000269|PubMed:15151508, ECO:0000269|PubMed:15316959, ECO:0000269|PubMed:15542400, ECO:0000269|PubMed:7573034, ECO:0000269|PubMed:7623438, ECO:0000269|PubMed:7655466, ECO:0000269|PubMed:8182131, ECO:0000269|PubMed:8733042, ECO:0000269|PubMed:9001800, ECO:0000269|PubMed:9332655, ECO:0000269|PubMed:9506659, ECO:0000269|PubMed:9700612, ECO:0000269|PubMed:9706613). Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı Ib 232220, Otozomal Resesif, GSD1B (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (SLC37A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease Ib, 232220, Autosomal recessive; GSD1B (Glycogen storage disease due to glucose-6-phosphatase deficiency) (SLC37A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC37A4	SLC37A4, G6PT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Glikojen Depo Hastalığı Ib 232220, Otozomal Resesif, GSD1B (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (Prenatal) (SLC37A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease Ib, 232220, Autosomal recessive; GSD1B (Glycogen storage disease due to glucose-6-phosphatase deficiency) (SLC37A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC37A4	SLC37A4, G6PT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı Ic 232240, Otozomal Resesif, GSD1C (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (SLC37A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease Ic, 232240, Autosomal recessive; GSD1C (Glycogen storage disease due to glucose-6-phosphatase deficiency) (SLC37A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC37A4	SLC37A4, G6PT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 1C (GSD1C) [MIM:232240]: A metabolic disorder characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia. (ECO:0000269|PubMed:10482962, ECO:0000269|PubMed:9758626). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Glikojen Depo Hastalığı Ic 232240, Otozomal Resesif, GSD1C (Glikojen Depo Hastalığı, Glukoz-6-Fosfataz Eksikliğine Bağlı) (Prenatal) (SLC37A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease Ic, 232240, Autosomal recessive; GSD1C (Glycogen storage disease due to glucose-6-phosphatase deficiency) (SLC37A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC37A4	SLC37A4, G6PT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 1C (GSD1C) [MIM:232240]: A metabolic disorder characterized by impairment of terminal steps of glycogenolysis and gluconeogenesis. Patients manifest a wide range of clinical symptoms and biochemical abnormalities, including hypoglycemia, severe hepatomegaly due to excessive accumulation of glycogen, kidney enlargement, growth retardation, lactic acidemia, hyperlipidemia, and hyperuricemia. (ECO:0000269|PubMed:10482962, ECO:0000269|PubMed:9758626). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 2, 232300, Otozomal Resesif, GSD2 (Glikojen Depo Hastalığı, Asit Maltaz Eksikliğine Bağlı) (Pompe Hastalığı) (GAA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease II, 232300, Autosomal recessive; GSD2 (Glycogen storage disease due to acid maltase deficiency) (Pompe Disease) (GAA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GAA	GAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 2 (GSD2) [MIM:232300]: A metabolic disorder with a broad clinical spectrum. The severe infantile form, or Pompe disease, presents at birth with massive accumulation of glycogen in muscle, heart and liver. Cardiomyopathy and muscular hypotonia are the cardinal features of this form whose life expectancy is less than two years. The juvenile and adult forms present as limb-girdle muscular dystrophy beginning in the lower limbs. Final outcome depends on respiratory muscle failure. Patients with the adult form can be free of clinical symptoms for most of their life but finally develop a slowly progressive myopathy. (ECO:0000269|PubMed:10189220, ECO:0000269|PubMed:10206684, ECO:0000269|PubMed:10338092, ECO:0000269|PubMed:10737124, ECO:0000269|PubMed:11071489, ECO:0000269|PubMed:11738358, ECO:0000269|PubMed:12601120, ECO:0000269|PubMed:12923862, ECO:0000269|PubMed:14643388, ECO:0000269|PubMed:14695532, ECO:0000269|PubMed:14972326, ECO:0000269|PubMed:15145338, ECO:0000269|PubMed:15688445, ECO:0000269|PubMed:16433701, ECO:0000269|PubMed:1652892, ECO:0000269|PubMed:16782080, ECO:0000269|PubMed:1684505, ECO:0000269|PubMed:16917947, ECO:0000269|PubMed:17643989, ECO:0000269|PubMed:18425781, ECO:0000269|PubMed:18429042,	EDTA Blood Sample
Glikojen Depo Hastalığı 2, 232300, Otozomal Resesif, GSD2 (Glikojen Depo Hastalığı, Asit Maltaz Eksikliğine Bağlı) (Pompe Hastalığı) (MLPA)	Glycogen storage disease II, 232300, Autosomal recessive; GSD2 (Glycogen storage disease due to acid maltase deficiency) (Pompe Disease) (MLPA)	GAA	GAA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Glycogen storage disease 2 (GSD2) [MIM:232300]: A metabolic disorder with a broad clinical spectrum. The severe infantile form, or Pompe disease, presents at birth with massive accumulation of glycogen in muscle, heart and liver. Cardiomyopathy and muscular hypotonia are the cardinal features of this form whose life expectancy is less than two years. The juvenile and adult forms present as limb-girdle muscular dystrophy beginning in the lower limbs. Final outcome depends on respiratory muscle failure. Patients with the adult form can be free of clinical symptoms for most of their life but finally develop a slowly progressive myopathy. (ECO:0000269|PubMed:10189220, ECO:0000269|PubMed:10206684, ECO:0000269|PubMed:10338092, ECO:0000269|PubMed:10737124, ECO:0000269|PubMed:11071489, ECO:0000269|PubMed:11738358, ECO:0000269|PubMed:12601120, ECO:0000269|PubMed:12923862, ECO:0000269|PubMed:14643388, ECO:0000269|PubMed:14695532, ECO:0000269|PubMed:14972326, ECO:0000269|PubMed:15145338, ECO:0000269|PubMed:15688445, ECO:0000269|PubMed:16433701, ECO:0000269|PubMed:1652892, ECO:0000269|PubMed:16782080, ECO:0000269|PubMed:1684505, ECO:0000269|PubMed:16917947, ECO:0000269|PubMed:17643989, ECO:0000269|PubMed:18425781, ECO:0000269|PubMed:18429042,	EDTA Blood Sample

Glikojen Depo Hastalığı 2, 232300, Otozomal Resesif, GSD2 (Glikojen Depo Hastalığı, Asit Maltaz Eksikliğine Bağlı) (Pompe Hastalığı) (Prenatal) (GAA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease II, 232300, Autosomal recessive; GSD2 (Glycogen storage disease due to acid maltase deficiency) (Pompe Disease) (GAA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GAA	GAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 2 (GSD2) [MIM:232300]: A metabolic disorder with a broad clinical spectrum. The severe infantile form, or Pompe disease, presents at birth with massive accumulation of glycogen in muscle, heart and liver. Cardiomyopathy and muscular hypotonia are the cardinal features of this form whose life expectancy is less than two years. The juvenile and adult forms present as limb-girdle muscular dystrophy beginning in the lower limbs. Final outcome depends on respiratory muscle failure. Patients with the adult form can be free of clinical symptoms for most of their life but finally develop a slowly progressive myopathy. {ECO:0000269|PubMed:10189220, ECO:0000269|PubMed:10206684, ECO:0000269|PubMed:10338092, ECO:0000269|PubMed:10737124, ECO:0000269|PubMed:11071489, ECO:0000269|PubMed:11738358, ECO:0000269|PubMed:12601120, ECO:0000269|PubMed:12923862, ECO:0000269|PubMed:14643388, ECO:0000269|PubMed:14695532, ECO:0000269|PubMed:14972326, ECO:0000269|PubMed:15145338, ECO:0000269|PubMed:15668445, ECO:0000269|PubMed:16433701, ECO:0000269|PubMed:1652892, ECO:0000269|PubMed:16782080, ECO:0000269|PubMed:1684505, ECO:0000269|PubMed:16917947, ECO:0000269|PubMed:17643989, ECO:0000269|PubMed:18425781, ECO:0000269|PubMed:18429042	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 2, 232300, Otozomal Resesif, GSD2 (Glikojen Depo Hastalığı, Asit Maltaz Eksikliğine Bağlı) (Pompe Hastalığı) (Prenatal) (MLPA)	Glycogen storage disease II, 232300, Autosomal recessive; GSD2 (Glycogen storage disease due to acid maltase deficiency) (Pompe Disease) (Prenatal) (MLPA)	GAA	GAA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 2 (GSD2) [MIM:232300]: A metabolic disorder with a broad clinical spectrum. The severe infantile form, or Pompe disease, presents at birth with massive accumulation of glycogen in muscle, heart and liver. Cardiomyopathy and muscular hypotonia are the cardinal features of this form whose life expectancy is less than two years. The juvenile and adult forms present as limb-girdle muscular dystrophy beginning in the lower limbs. Final outcome depends on respiratory muscle failure. Patients with the adult form can be free of clinical symptoms for most of their life but finally develop a slowly progressive myopathy. {ECO:0000269|PubMed:10189220, ECO:0000269|PubMed:10206684, ECO:0000269|PubMed:10338092, ECO:0000269|PubMed:10737124, ECO:0000269|PubMed:11071489, ECO:0000269|PubMed:11738358, ECO:0000269|PubMed:12601120, ECO:0000269|PubMed:12923862, ECO:0000269|PubMed:14643388, ECO:0000269|PubMed:14695532, ECO:0000269|PubMed:14972326, ECO:0000269|PubMed:15145338, ECO:0000269|PubMed:15668445, ECO:0000269|PubMed:16433701, ECO:0000269|PubMed:1652892, ECO:0000269|PubMed:16782080, ECO:0000269|PubMed:1684505, ECO:0000269|PubMed:16917947, ECO:0000269|PubMed:17643989, ECO:0000269|PubMed:18425781, ECO:0000269|PubMed:18429042	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 3a, 232400, Otozomal Resesif, GSD3 (Glikojen Depo Hastalığı, Glikojen Debranching Enzim Eksikliğine Bağlı, 3b) (AGL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease IIIa, 232400, Autosomal recessive; GSD3 (Glycogen storage disease due to glycogen debranching enzyme deficiency) (AGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGL	AGL, GDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 3 (GSD3) [MIM:232400]: A metabolic disorder associated with an accumulation of abnormal glycogen with short outer chains. It is clinically characterized by hepatomegaly, hypoglycemia, short stature, and variable myopathy. Glycogen storage disease type 3 includes different forms: GSD type 3A patients lack glycogen debrancher enzyme activity in both liver and muscle, while GSD type 3B patients are enzyme-deficient in liver only. In rare cases, selective loss of only 1 of the 2 debranching activities, glucosidase or transferase, results in GSD type 3C or type 3D, respectively. {ECO:0000269|PubMed:10571954}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 3a, 232400, Otozomal Resesif, GSD3 (Glikojen Depo Hastalığı, Glikojen Debranching Enzim Eksikliğine Bağlı, 3b) (Prenatal) (AGL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease IIIa, 232400, Autosomal recessive; GSD3 (Glycogen storage disease due to glycogen debranching enzyme deficiency) (AGL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGL	AGL, GDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 3 (GSD3) [MIM:232400]: A metabolic disorder associated with an accumulation of abnormal glycogen with short outer chains. It is clinically characterized by hepatomegaly, hypoglycemia, short stature, and variable myopathy. Glycogen storage disease type 3 includes different forms: GSD type 3A patients lack glycogen debrancher enzyme activity in both liver and muscle, while GSD type 3B patients are enzyme-deficient in liver only. In rare cases, selective loss of only 1 of the 2 debranching activities, glucosidase or transferase, results in GSD type 3C or type 3D, respectively. {ECO:0000269|PubMed:10571954}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 3b, 232400, Otozomal Resesif (Glikojen Depo Hastalığı, Glikojen Debranching Enzim Eksikliğine Bağlı, 3b) (AGL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease IIIb, 232400, Autosomal recessive (Glycogen storage disease due to glycogen debranching enzyme deficiency) (AGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGL	AGL, GDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 3 (GSD3) [MIM:232400]: A metabolic disorder associated with an accumulation of abnormal glycogen with short outer chains. It is clinically characterized by hepatomegaly, hypoglycemia, short stature, and variable myopathy. Glycogen storage disease type 3 includes different forms: GSD type 3A patients lack glycogen debrancher enzyme activity in both liver and muscle, while GSD type 3B patients are enzyme-deficient in liver only. In rare cases, selective loss of only 1 of the 2 debranching activities, glucosidase or transferase, results in GSD type 3C or type 3D, respectively. {ECO:0000269|PubMed:10571954}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 3b, 232400, Otozomal Resesif (Glikojen Depo Hastalığı, Glikojen Debranching Enzim Eksikliğine Bağlı, 3b) (Prenatal) (AGL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease IIIb, 232400, Autosomal recessive (Glycogen storage disease due to glycogen debranching enzyme deficiency) (AGL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGL	AGL, GDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 3 (GSD3) [MIM:232400]: A metabolic disorder associated with an accumulation of abnormal glycogen with short outer chains. It is clinically characterized by hepatomegaly, hypoglycemia, short stature, and variable myopathy. Glycogen storage disease type 3 includes different forms: GSD type 3A patients lack glycogen debrancher enzyme activity in both liver and muscle, while GSD type 3B patients are enzyme-deficient in liver only. In rare cases, selective loss of only 1 of the 2 debranching activities, glucosidase or transferase, results in GSD type 3C or type 3D, respectively. {ECO:0000269|PubMed:10571954}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 4, 232500, Otozomal Resesif, GSD4 (Glikojen Depo Hastalığı-Glikojen Dallandırıcı Enzim Eksikliğine Bağlı) (GBE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease IV, 232500, Autosomal recessive; GSD4 (Glycogen storage disease due to glycogen branching enzyme deficiency) (GBE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBE1	GBE1, GSD4, APBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glikojen Depo Hastalığı 4, 232500, Otozomal Resesif, GSD4 (Glikojen Depo Hastalığı-Glikojen Dallandırıcı Enzim Eksikliğine Bağlı) (Prenatal) (GBE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease IV, 232500, Autosomal recessive; GSD4 (Glycogen storage disease due to glycogen branching enzyme deficiency) (GBE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBE1	GBE1, GSD4, APBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 9c, 613027, Otozomal Resesif, GSD9C (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (PHKG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease IXc, 613027, Autosomal recessive; GSD9C (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKG2	PHKG2, GSD9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9C (GSD9C) [MIM:613027]: A metabolic disorder manifesting in infancy with hepatomegaly, growth retardation, hypotonia, liver dysfunction, and elevated plasma aminotransferases and lipids. These symptoms improve with age in most cases; however, some patients may develop hepatic fibrosis or cirrhosis. {ECO:0000269|PubMed:12930917, ECO:0000269|PubMed:8896567, ECO:0000269|PubMed:9245685}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 9c, 613027, Otozomal Resesif, GSD9C (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (Prenatal) (PHKG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease IXc, 613027, Autosomal recessive; GSD9C (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKG2	PHKG2, GSD9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9C (GSD9C) [MIM:613027]: A metabolic disorder manifesting in infancy with hepatomegaly, growth retardation, hypotonia, liver dysfunction, and elevated plasma aminotransferases and lipids. These symptoms improve with age in most cases; however, some patients may develop hepatic fibrosis or cirrhosis. {ECO:0000269|PubMed:12930917, ECO:0000269|PubMed:8896567, ECO:0000269|PubMed:9245685}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kalbin Glikojen Depo Hastalığı, Letal Konjenital, 261740, Otozomal Dominant (Fatal Konjenital Hipertrofik Kardiyomyopati, Glikojen Depo Hastalığına Bağlı) (PRKAG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease of heart, lethal congenital, 261740, Autosomal dominant (Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease) (PRKAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAG2	PRKAG2, WPWS, CMH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalbin Glikojen Depo Hastalığı, Letal Konjenital, 261740, Otozomal Dominant (Fatal Konjenital Hipertrofik Kardiyomyopati, Glikojen Depo Hastalığına Bağlı) (Prenatal) (PRKAG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease of heart, lethal congenital, 261740, Autosomal dominant (Fatal congenital hypertrophic cardiomyopathy due to glycogen storage disease) (PRKAG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKAG2	PRKAG2, WPWS, CMH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 5, Gsd5 (Glikojen Depo Hastalığı, Kas Glikojen Fosforilaz Eksikliğine Bağlı, Mcardle Hastalığı) (PYGM geni) (Tüm Gen Dizi Analizi) (Postnatal)	GLYCOGEN STORAGE DISEASE V, GSD5 (Glycogen storage disease due to muscle glycogen phosphorylase deficiency) (PYGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYGM	PYGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 5 (GSD5) [MIM:232600]: A metabolic disorder resulting in myopathy characterized by exercise intolerance, cramps, muscle weakness and recurrent myoglobinuria. {ECO:0000269|PubMed:10382911, ECO:0000269|PubMed:10382912, ECO:0000269|PubMed:10417800, ECO:0000269|PubMed:10590419, ECO:0000269|PubMed:10681080, ECO:0000269|PubMed:10714589, ECO:0000269|PubMed:10899452, ECO:0000269|PubMed:11706962, ECO:0000269|PubMed:12031624, ECO:0000269|PubMed:7603523, ECO:0000269|PubMed:8316268, ECO:0000269|PubMed:8535454, ECO:0000269|PubMed:9505649}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Glikojen Depo Hastalığı 5, GsdS (Glikojen Depo Hastalığı, Kas Glikojen Fosforilaz Eksikliğine Bağlı, Mcardle Hastalığı) (Prenatal) (PYGM geni) (Tüm Gen Dizi Analizi) (Prenatal)	GLYCOGEN STORAGE DISEASE V, GSD5 (Glycogen storage disease due to muscle glycogen phosphorylase deficiency) (PYGM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYGM	PYGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 5 (GSD5) [MIM:232600]: A metabolic disorder resulting in myopathy characterized by exercise intolerance, cramps, muscle weakness and recurrent myoglobinuria. {ECO:0000269|PubMed:10382911, ECO:0000269|PubMed:10382912, ECO:0000269|PubMed:10417800, ECO:0000269|PubMed:10530419, ECO:0000269|PubMed:10681080, ECO:0000269|PubMed:10714589, ECO:0000269|PubMed:10899452, ECO:0000269|PubMed:11706962, ECO:0000269|PubMed:12031624, ECO:0000269|PubMed:7603523, ECO:0000269|PubMed:8316268, ECO:0000269|PubMed:8535454, ECO:0000269|PubMed:9506549}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 6 232700, Otozomal Resesif, GSD6 (Glikojen Depo Hastalığı, Karaciğer Glikojen Fosforilaz Eksikliğine Bağlı) (PYGL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease VI, 232700, Autosomal recessive; GSD6 (Glycogen storage disease due to liver glycogen phosphorylase deficiency) (PYGL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYGL	PYGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 6 (GSD6) [MIM:232700]: A metabolic disorder characterized by mild to moderate hypoglycemia, mild ketosis, growth retardation, and prominent hepatomegaly. Heart and skeletal muscle are not affected. {ECO:0000269	PubMed:9529348}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 6 232700, Otozomal Resesif, GSD6 (Glikojen Depo Hastalığı, Karaciğer Glikojen Fosforilaz Eksikliğine Bağlı) (Prenatal) (PYGL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease VI, 232700, Autosomal recessive; GSD6 (Glycogen storage disease due to liver glycogen phosphorylase deficiency) (PYGL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYGL	PYGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 6 (GSD6) [MIM:232700]: A metabolic disorder characterized by mild to moderate hypoglycemia, mild ketosis, growth retardation, and prominent hepatomegaly. Heart and skeletal muscle are not affected. {ECO:0000269	PubMed:9529348}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 7 232800, Otozomal Resesif, GSD7 (Glikojen Depo Hastalığı, Kas Fosfofruktokinaz Eksikliğine Bağlı) (PFKM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease VII, 232800, Autosomal recessive; GSD7 (Glycogen storage disease due to muscle phosphofructokinase deficiency) (PFKM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PFKM	PFKM, GSD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 7 (GSD7) [MIM:232800]: A metabolic disorder characterized by exercise intolerance with associated nausea and vomiting, muscle cramping, exertional myopathy and compensated hemolysis. Short bursts of intense activity are particularly difficult. Severe muscle cramps and myoglobinuria develop after vigorous exercise. {ECO:0000269|PubMed:22133655, ECO:0000269|PubMed:24427140, ECO:0000269|PubMed:7513946, ECO:0000269|PubMed:7825568, ECO:0000269|PubMed:8889589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 7 232800, Otozomal Resesif, GSD7 (Glikojen Depo Hastalığı, Kas Fosfofruktokinaz Eksikliğine Bağlı) (Prenatal) (PFKM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease VII, 232800, Autosomal recessive; GSD7 (Glycogen storage disease due to muscle phosphofructokinase deficiency) (PFKM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PFKM	PFKM, GSD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 7 (GSD7) [MIM:232800]: A metabolic disorder characterized by exercise intolerance with associated nausea and vomiting, muscle cramping, exertional myopathy and compensated hemolysis. Short bursts of intense activity are particularly difficult. Severe muscle cramps and myoglobinuria develop after vigorous exercise. {ECO:0000269|PubMed:22133655, ECO:0000269|PubMed:24427140, ECO:0000269|PubMed:7513946, ECO:0000269|PubMed:7825568, ECO:0000269|PubMed:8889589}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 10, 261670, Otozomal Resesif, GSD10 (Glikojen Depo Hastalığı, Fosfoglukoserat Mutaz Eksikliği) (PGAM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease X, 261670, Autosomal recessive; GSD10 (Glycogen storage disease due to phosphoglycerate mutase deficiency) (PGAM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGAM2	PGAM2, PGAMM, GSD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glikojen Depo Hastalığı 10, 261670, Otozomal Resesif, GSD10 (Glikojen Depo Hastalığı, Fosfoglukoserat Mutaz Eksikliği) (Prenatal) (PGAM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease X, 261670, Autosomal recessive; GSD10 (Glycogen storage disease due to phosphoglycerate mutase deficiency) (PGAM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGAM2	PGAM2, PGAMM, GSD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 11, 612933, Otozomal Resesif, GSD11 (Glikojen Depo Hastalığı, Laktat Dehidrojenaz Eksikliğine Bağlı) (LDHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease XI, 612933, Autosomal recessive; GSD11 (Glycogen storage disease due to lactate dehydrogenase deficiency) (LDHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDHA	LDHA, LDH1, GSD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 11 (GSD11) [MIM:612933]: A metabolic disorder that results in exertional myoglobinuria, pain, cramps and easy fatigue. {ECO:0000269	PubMed:2334430}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 11, 612933, Otozomal Resesif, GSD11 (Glikojen Depo Hastalığı, Laktat Dehidrojenaz Eksikliğine Bağlı) (Prenatal) (LDHA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease XI, 612933, Autosomal recessive; GSD11 (Glycogen storage disease due to lactate dehydrogenase deficiency) (LDHA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LDHA	LDHA, LDH1, GSD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 11 (GSD11) [MIM:612933]: A metabolic disorder that results in exertional myoglobinuria, pain, cramps and easy fatigue. {ECO:0000269	PubMed:2334430}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 12, 611881, Otozomal Resesif, GSD12 (Glikojen Depo Hastalığı, Aldolaz A Eksikliğine Bağlı) (ALDOA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease XII, 611881, Autosomal recessive; GSD12 (Glycogen storage disease due to aldolase A deficiency) (ALDOA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDOA	ALDOA, GSD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 12 (GSD12) [MIM:611881]: A metabolic disorder associated with increased hepatic glycogen and hemolytic anemia. It may lead to myopathy with exercise intolerance and rhabdomyolysis. {ECO:0000269|PubMed:14615364, ECO:0000269|PubMed:14766013, ECO:0000269|PubMed:2825199, ECO:0000269|PubMed:8598869}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 12, 611881, Otozomal Resesif, GSD12 (Glikojen Depo Hastalığı, Aldolaz A Eksikliğine Bağlı) (Prenatal) (ALDOA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease XII, 611881, Autosomal recessive; GSD12 (Glycogen storage disease due to aldolase A deficiency) (ALDOA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDOA	ALDOA, GSD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 12 (GSD12) [MIM:611881]: A metabolic disorder associated with increased hepatic glycogen and hemolytic anemia. It may lead to myopathy with exercise intolerance and rhabdomyolysis. {ECO:0000269|PubMed:14615364, ECO:0000269|PubMed:14766013, ECO:0000269|PubMed:2825199, ECO:0000269|PubMed:8598869}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 13 612932, Otozomal Resesif, GSD13 (Glikojen Depo Hastalığı, Kas Beta Enoloz Eksikliğine Bağlı) (ENO3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease XIII, 612932, Autosomal recessive; GSD13 (Glycogen storage disease due to muscle beta-enolase deficiency) (ENO3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENO3	ENO3, GSD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 13 (GSD13) [MIM:612932]: A metabolic disorder that results in exercise-induced myalgias, generalized muscle weakness and fatigability. It is characterized by increased serum creatine kinase and decreased enolase 3 activity. Dramatically reduced protein levels with focal sarcoplasmic accumulation of glycogen-beta particles are detected on ultrastructural analysis. {ECO:0000269	PubMed:11506403}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı 13 612932, Otozomal Resesif, GSD13 (Glikojen Depo Hastalığı, Kas Beta Enoloz Eksikliğine Bağlı) (Prenatal) (ENO3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease XIII, 612932, Autosomal recessive; GSD13 (Glycogen storage disease due to muscle beta-enolase deficiency) (ENO3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ENO3	ENO3, GSD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 13 (GSD13) [MIM:612932]: A metabolic disorder that results in exercise-induced myalgias, generalized muscle weakness and fatigability. It is characterized by increased serum creatine kinase and decreased enolase 3 activity. Dramatically reduced protein levels with focal sarcoplasmic accumulation of glycogen-beta particles are detected on ultrastructural analysis. {ECO:0000269	PubMed:11506403}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı 15 613507, Otozomal Resesif, GSD15 (Glikojen Depo Hastalığı-Ağır Kardiyomiyopati, Glikogenin Eksikliğine Bağlı) (GYG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease XV, 613507, Autosomal recessive; GSD15 (Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency) (GYG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYG1	GYG1, GSD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 15 (GSD15) [MIM:613507]: A metabolic disorder resulting in muscle weakness, associated with the glycogen depletion in skeletal muscle, and cardiac arrhythmia, associated with the accumulation of abnormal storage material in the heart. The skeletal muscle shows a marked predominance of slow-twitch, oxidative muscle fibers and mitochondrial proliferation. {ECO:0000269	PubMed:20357282}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Glikojen Depo Hastalığı 15 613507, Otozomal Resesif, GSD15 (Glikojen Depo Hastalığı-Ağır Kardiyomiyopati, Glikogenin Eksikliğine Bağlı) (Prenatal) (GYG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease XV, 613507, Autosomal recessive; GSD15 (Glycogen storage disease with severe cardiomyopathy due to glycogenin deficiency) (GYG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GYG1	GYG1, GSD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 15 (GSD15) [MIM:613507]: A metabolic disorder resulting in muscle weakness, associated with the glycogen depletion in skeletal muscle, and cardiac arrhythmia, associated with the accumulation of abnormal storage material in the heart. The skeletal muscle shows a marked predominance of slow-twitch, oxidative muscle fibers and mitochondrial proliferation. {ECO:0000269	PubMed:20357282}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı, Tip 9a1, 306000, X'e Bağlı (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (PHKA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease, type IXa1, 306000, X-linked recessive (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKA2	PHKA2, PHK, XLG, PHK, PYKL, GSD9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9A (GSD9A) [MIM:306000]: A metabolic disorder resulting in a mild liver glycogenosis with clinical symptoms that include hepatomegaly, growth retardation, muscle weakness, elevation of glutamate-pyruvate transaminase and glutamate-oxaloacetate transaminase, hypercholesterolemia, hypertriglyceridemia, and fasting hyperketosis. Two subtypes are known: type 1 or classic type with no phosphorylase kinase activity in liver or erythrocytes, and type 2 or variant type with no phosphorylase kinase activity in liver, but normal activity in erythrocytes. Unlike other glycogenosis diseases, glycogen storage disease type 9A is generally a benign condition. Patients improve with age and are often asymptomatic as adults. Accurate diagnosis is therefore also of prognostic interest. {ECO:0000269|PubMed:10330341, ECO:0000269|PubMed:17689125, ECO:0000269|PubMed:7549948, ECO:0000269|PubMed:7847371, ECO:0000269|PubMed:8733133, ECO:0000269|PubMed:8733134, ECO:0000269|PubMed:9600238}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Glikojen Depo Hastalığı, Tip 9a1, 306000, X'e Bağlı (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (Prenatal) (PHKA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease, type IXa1, 306000, X-linked recessive (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKA2	PHKA2, PHK, XLG, PHK, PYKL, GSD9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9A (GSD9A) [MIM:306000]: A metabolic disorder resulting in a mild liver glycogenosis with clinical symptoms that include hepatomegaly, growth retardation, muscle weakness, elevation of glutamate-pyruvate transaminase and glutamate-oxaloacetate transaminase, hypercholesterolemia, hypertriglyceridemia, and fasting hyperketosis. Two subtypes are known: type 1 or classic type with no phosphorylase kinase activity in liver or erythrocytes, and type 2 or variant type with no phosphorylase kinase activity in liver, but normal activity in erythrocytes. Unlike other glycogenosis diseases, glycogen storage disease type 9A is generally a benign condition. Patients improve with age and are often asymptomatic as adults. Accurate diagnosis is therefore also of prognostic interest. {ECO:0000269|PubMed:10330341, ECO:0000269|PubMed:17689125, ECO:0000269|PubMed:7549948, ECO:0000269|PubMed:7847371, ECO:0000269|PubMed:8733133, ECO:0000269|PubMed:8733134, ECO:0000269|PubMed:9600238}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikojen Depo Hastalığı, Tip 9a2, 306000, X'e Bağlı Resesif (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (PHKA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycogen storage disease, type IXa2, 306000, X-linked recessive (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKA2	PHKA2, PHK, XLG, PHK, PYKL, GSD9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9A (GSD9A) [MIM:306000]: A metabolic disorder resulting in a mild liver glycogenosis with clinical symptoms that include hepatomegaly, growth retardation, muscle weakness, elevation of glutamate-pyruvate transaminase and glutamate-oxaloacetate transaminase, hypercholesterolemia, hypertriglyceridemia, and fasting hyperketosis. Two subtypes are known: type 1 or classic type with no phosphorylase kinase activity in liver or erythrocytes, and type 2 or variant type with no phosphorylase kinase activity in liver, but normal activity in erythrocytes. Unlike other glycogenosis diseases, glycogen storage disease type 9A is generally a benign condition. Patients improve with age and are often asymptomatic as adults. Accurate diagnosis is therefore also of prognostic interest. {ECO:0000269|PubMed:10330341, ECO:0000269|PubMed:17689125, ECO:0000269|PubMed:7549948, ECO:0000269|PubMed:7847371, ECO:0000269|PubMed:8733133, ECO:0000269|PubMed:8733134, ECO:0000269|PubMed:9600238}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikojen Depo Hastalığı, Tip 9a2, 306000, X'e Bağlı Resesif (Glikojen Depo Hastalığı, Karaciğer Fosforilaz Kinaz Eksikliğine Bağlı) (Prenatal) (PHKA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycogen storage disease, type IXa2, 306000, X-linked recessive (Glycogen storage disease due to liver phosphorylase kinase deficiency) (PHKA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKA2	PHKA2, PHK, XLG, PHK, PYKL, GSD9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9A (GSD9A) [MIM:306000]: A metabolic disorder resulting in a mild liver glycogenosis with clinical symptoms that include hepatomegaly, growth retardation, muscle weakness, elevation of glutamate-pyruvate transaminase and glutamate-oxaloacetate transaminase, hypercholesterolemia, hypertriglyceridemia, and fasting hyperketosis. Two subtypes are known: type 1 or classic type with no phosphorylase kinase activity in liver or erythrocytes, and type 2 or variant type with no phosphorylase kinase activity in liver, but normal activity in erythrocytes. Unlike other glycogenosis diseases, glycogen storage disease type 9A is generally a benign condition. Patients improve with age and are often asymptomatic as adults. Accurate diagnosis is therefore also of prognostic interest. {ECO:0000269|PubMed:10330341, ECO:0000269|PubMed:17689125, ECO:0000269|PubMed:7549948, ECO:0000269|PubMed:7847371, ECO:0000269|PubMed:8733133, ECO:0000269|PubMed:8733134, ECO:0000269|PubMed:9600238}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikoprotein 1a Eksikliği, 614200, Otozomal Dominant; BDPLT9 (Kanama Diyatezi, Kolajen Reseptör Defektine Bağlı) (ITGA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycoprotein Ia deficiency, 614200, Autosomal dominant; BDPLT9 (Bleeding diathesis due to a collagen receptor defect) (ITGA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA2	ITGA2, CD49B, BR, BDPLT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Glikoprotein 1a Eksikliği, 614200, Otozomal Dominant; BDPLT9 (Kanama Diyatezi, Kolajen Reseptör Defektine Bağlı) (Prenatal) (ITGA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycoprotein Ia deficiency, 614200, Autosomal dominant; BDPLT9 (Bleeding diathesis due to a collagen receptor defect) (ITGA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGA2	ITGA2, CD49B, BR, BDPLT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Glikosilfosfatidilinositol Eksikliği, 610293, Otozomal Resesif; GPID (Hiperkoagülabilite Sendromu, Glikosilfosfatidilinositol Eksikliğine Bağlı) (PIGM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Glycosylphosphatidylinositol deficiency, 610293, Autosomal recessive; GPID (Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency) (PIGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGM	PIGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycosylphosphatidylinositol deficiency (GPID) [MIM:610293]: Autosomal recessive trait that results in a propensity to venous thrombosis and seizures. Deficiency is due to a point mutation in the regulatory sequences of PIGM that disrupts binding of the transcription factor SP1 to its cognate promoter motif, leading to a strong reduction of expression. {ECO:0000269|PubMed:16767100}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Glikosilfosfatidilinositol Eksikliği, 610293, Otozomal Resesif; GPID (Hiperkoagülabilite Sendromu, Glikosilfosfatidilinositol Eksikliğine Bağlı) (Prenatal) (PIGM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Glycosylphosphatidylinositol deficiency, 610293, Autosomal recessive; GPID (Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency) (PIGM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGM	PIGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycosylphosphatidylinositol deficiency (GPID) [MIM:610293]: Autosomal recessive trait that results in a propensity to venous thrombosis and seizures. Deficiency is due to a point mutation in the regulatory sequences of PIGM that disrupts binding of the transcription factor SP1 to its cognate promoter motif, leading to a strong reduction of expression. {ECO:0000269|PubMed:16767100}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM1 Gangliosidozis, Tip 1, 230500, Otozomal Resesif (GM1 Gangliosidoz) (GLB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GM1-ganglicosidosis, type I, 230500, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM1-gangliosidosis 1 (GM1G1) [MIM:230500]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM1 gangliosides, glycoproteins and keratan sulfate primarily in neurons of the central nervous system. GM1-gangliosidosis type 1 is characterized by onset within the first three months of life, central nervous system degeneration, coarse facial features, hepatosplenomegaly, skeletal dysmorphology reminiscent of Hurler syndrome, and rapidly progressive psychomotor deterioration. Urinary oligosaccharide levels are high. It leads to death usually between the first and second year of life. {ECO:0000269|PubMed:10338095, ECO:0000269|PubMed:10737981, ECO:0000269|PubMed:10839995, ECO:0000269|PubMed:1487238, ECO:0000269|PubMed:15365997, ECO:0000269|PubMed:15714521, ECO:0000269|PubMed:15791924, ECO:0000269|PubMed:16538002, ECO:0000269|PubMed:16941474, ECO:0000269|PubMed:17309651, ECO:0000269|PubMed:1907800, ECO:0000269|PubMed:1909089, ECO:0000269|PubMed:1928092, ECO:0000269|PubMed:19472408, ECO:0000269|PubMed:25936995, ECO:0000269|PubMed:8213816, ECO:0000269|Ref.24, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
GM1 Gangliosidozis, Tip 1, 230500, Otozomal Resesif (GM1 Gangliosidoz) (Prenatal) (GLB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GM1-ganglicosidosis, type I, 230500, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM1-gangliosidosis 1 (GM1G1) [MIM:230500]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM1 gangliosides, glycoproteins and keratan sulfate primarily in neurons of the central nervous system. GM1-gangliosidosis type 1 is characterized by onset within the first three months of life, central nervous system degeneration, coarse facial features, hepatosplenomegaly, skeletal dysmorphology reminiscent of Hurler syndrome, and rapidly progressive psychomotor deterioration. Urinary oligosaccharide levels are high. It leads to death usually between the first and second year of life. {ECO:0000269|PubMed:10338095, ECO:0000269|PubMed:10737981, ECO:0000269|PubMed:10839995, ECO:0000269|PubMed:1487238, ECO:0000269|PubMed:15365997, ECO:0000269|PubMed:15714521, ECO:0000269|PubMed:15791924, ECO:0000269|PubMed:16538002, ECO:0000269|PubMed:16941474, ECO:0000269|PubMed:17309651, ECO:0000269|PubMed:1907800, ECO:0000269|PubMed:1909089, ECO:0000269|PubMed:1928092, ECO:0000269|PubMed:19472408, ECO:0000269|PubMed:25936995, ECO:0000269|PubMed:8213816, ECO:0000269|Ref.24, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM1 Gangliosidozis, Tip 2, 230600, Otozomal Resesif (GM1 Ganglisidoz) (GLB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GM1-ganglicosidosis, type II, 230600, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GM1 Gangliosidozis, Tip 2, 230600, Otozomal Resesif (GM1 Ganglisidoz) (Prenatal) (GLB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GM1-ganglicosidosis, type II, 230600, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM1 Gangliosidozis, Tip 3, 230650, Otozomal Resesif (GM1 Ganglisidoz) (GLB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GM1-ganglicosidosis, type III, 230650, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GM1 Gangliosidozis, Tip 3, 230650, Otozomal Resesif (GM1 Ganglisidoz) (Prenatal) (GLB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GM1-ganglicosidosis, type III, 230650, Autosomal recessive (GM1 ganglicosidosis) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM2-Gangliosidozis, AB Varyant, 272750, Otozomal Resesif (GM2 Gangliosidozis, AB Varyant) (GM2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	GM2-gangliosidosis, AB variant, 272750, Autosomal recessive (GM2 gangliosidosis, AB variant) (GM2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GM2A	GM2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM2-gangliosidosis AB (GM2GAB) [MIM:272750]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the presence of both normal hexosaminidase A and B. {ECO:0000269|PubMed:1915858, ECO:0000269|PubMed:8244332, ECO:0000269|PubMed:8900233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

GM2-Gangliosidozis, AB Varyant, 272750, Otozomal Resesif (GM2 Gangliosidozis, AB Varyant) (Prenatal) (GM2A gen) (Tüm Gen Dizi Analizi) (Prenatal)	GM2-gangliosidosis, AB variant, 272750, Autosomal recessive (GM2 gangliosidosis, AB variant) (GM2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GM2A	GM2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis AB (GM2CAB) [MIM:272750]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the presence of both normal hexosaminidase A and B. {ECO:0000269|PubMed:1915858, ECO:0000269|PubMed:8244332, ECO:0000269|PubMed:8900233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM2-Gangliosidozis, Çeşitli Formlar, 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (HEXA geni) (Tüm Gen Dizi Analizi) (Postnatal)	GM2-gangliosidosis, several forms, 272800, Autosomal recessive (Tay-Sachs disease) (HEXA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEXA	HEXA, TSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532389, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
GM2-Gangliosidozis, Çeşitli Formlar, 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (MLPA)	GM2-gangliosidosis, several forms, 272800, Autosomal recessive (Tay-Sachs disease) (MLPA)	HEXA	HEXA, TSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532389, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
GM2-Gangliosidozis, Çeşitli Formlar, 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (Prenatal) (HEXA geni) (Tüm Gen Dizi Analizi) (Prenatal)	GM2-gangliosidosis, several forms, 272800, Autosomal recessive (Tay-Sachs disease) (HEXA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HEXA	HEXA, TSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532389, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GM2-Gangliosidozis, Çeşitli Formlar, 272800, Otozomal Resesif (Tay-Sachs Hastalığı) (Prenatal) (MLPA)	GM2-gangliosidosis, several forms, 272800, Autosomal recessive (Tay-Sachs disease) (Prenatal) (MLPA)	HEXA	HEXA, TSD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. It is characterized by GM2 gangliosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532389, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gnatodiafizyal Displazi, 166260, Otozomal Dominant; GDD (Gnatodiafizyal Displazi) (ANO5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gnathodiaphyseal dysplasia, 166260, Autosomal dominant; GDD (Gnathodiaphyseal dysplasia) (ANO5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO5	ANO5, TMEM16E, GDD1, LGMD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Gnatodiafizyal Displazi, 166260, Otozomal Dominant; GDD (Gnatodiafizyal Displazi) (Prenatal) (ANO5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gnathodiaphyseal dysplasia, 166260, Autosomal dominant; GDD (Gnathodiaphyseal dysplasia) (ANO5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANO5	ANO5, TMEM16E, GDD1, LGMD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Guatr, Multinodüler 1, ± Sertoli-Leydig Hücreli Tümör, 138800, Otozomal Dominant; MNG1 (Ailesel Multinodüler Guatr) (DICER1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors, 138800, Autosomal dominant; MNG1 (Familial multinodular goiter) (DICER1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DICER1	DICER1, HERNA, KIAA0028, MNG1, RMSE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Goiter multinodular 1, with or without Sertoli-Leydig cell tumors (MNG1) [MIM:138800]: A common disorder characterized by nodular overgrowth of the thyroid gland. Some individuals may also develop Sertoli-Leydig cell tumors, usually of the ovary. {ECO:0000269|PubMed:21205968}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Goldberg Shprintzen Megakolon Sendromu, 609460, Otozomal Resesif; GOSHS (Goldberg-Shprintzen megacolon syndrome) (KIF1BP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Goldberg-Shprintzen megacolon syndrome, 609460, Autosomal recessive; GOSHS (Goldberg-Shprintzen megacolon syndrome) (KIF1BP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1BP	KIAA1279	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Goldberg Shprintzen Megakolon Sendromu, 609460, Otozomal Resesif; GOSHS (Goldberg Shprintzen Megakolon Sendromu) (Prenatal) (KIF1BP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Goldberg-Shprintzen megacolon syndrome, 609460, Autosomal recessive; GOSHS (Goldberg-Shprintzen megacolon syndrome) (KIF1BP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF1BP	KIAA1279	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gonadal Gelişim Bozukluğu paneli (DMRT1, CYP17A1, SRD5A2, HSD17B3) (MLPA)	Gonadal Developmental Disorder panel (DMRT1, CYP17A1, SRD5A2, HSD17B3) (MLPA)	DMRT1, CYP17A1, SRD5A2, HSD17B3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Gonadotropin Salgılayan Hormon Reseptörü 2; Gnrhr2 (GNRHR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GONADOTROPIN-RELEASING HORMONE RECEPTOR 2; GNRHR2 (GNRHR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNRHR2	GNRHR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gut Yatkınlık 4, 612671, Otozomal Dominant (SLC17A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gout susceptibility 4, 612671, Autosomal dominant (SLC17A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A3	SLC17A3, NPT4, UAQTL4, GOUT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gut, PRPS İlişkili, 300661, X'e Bağlı (Fosforibosilpirofosfat Sentetaz Süperaktivitesi) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gout, PRPS-related, 300661, X-linked recessive (Phosphoribosylpyrophosphate synthetase superactivity) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phosphoribosylpyrophosphate synthetase superactivity (PRPS1 superactivity) [MIM:300661]: Familial disorder characterized by excessive purine production, gout and uric acid urolithiasis. {ECO:0000269|PubMed:7593598, ECO:0000269|Ref.12}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Gracile Kemik Displazisi, 602361, Otozomal Dominant; GCLEB (Osteokraniostenozis) (FAM111A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gracile bone dysplasia, 602361, Autosomal dominant; GCLEB (Osteocraniostenosis) (FAM111A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM111A	FAM111A, KIAA1895, KCS2, GCLEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gracile Kemik Displazisi, 602361, Otozomal Dominant; GCLEB (Osteokraniostenozis) (Prenatal) (FAM111A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Gracile bone dysplasia, 602361, Autosomal dominant; GCLEB (Osteocraniostenosis) (FAM111A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAM111A	FAM111A, KIAA1895, KCS2, GCLEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GRACILE Sendromu, 603358 (GRACILE Sendromu) (BCS1L gen) (Tüm Gen Dizi Analizi) (Postnatal)	GRACILE syndrome, 603358 (GRACILE syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRACILE Sendromu, 603358 (GRACILE Sendromu) (Prenatal) (BCS1L gen) (Tüm Gen Dizi Analizi) (Prenatal)	GRACILE syndrome, 603358 (GRACILE syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Graft-Versus-Host Hastalığı, Karşı Koruma, 614395, GVHDS (Greft Versus Host Hastalığı) (IL10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Graft-versus-host disease, protection against, 614395, GVHDS (Graft versus host disease) (IL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10	IL10, CSIF, GVHDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Grange Sendromu, 602531, Otozomal Resesif; GRNG (Grange Sendromu) (YY1AP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Grange syndrome, 602531, Autosomal recessive; GRNG (Grange syndrome) (YY1AP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YY1AP1	YY1AP1, YAP, HCCA2, GRNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Grange Sendromu, 602531, Otozomal Resesif; GRNG (Grange Sendromu) (Prenatal) (YY1AP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Grange syndrome, 602531, Autosomal recessive; GRNG (Grange syndrome) (YY1AP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	YY1AP1	YY1AP1, YAP, HCCA2, GRNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal Resesif, Sitokrom B-Negatif (Kronik Granülomatöz Hastalık) (CYBA gen) (Tüm Gen Dizi Analizi) (Postnatal)	GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-NEGATIVE (Chronic granulomatous disease) (CYBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYBA	CYBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-negative, autosomal recessive (ARCGD) [MIM:233690]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10759707, ECO:0000269|PubMed:10910929, ECO:0000269|PubMed:10914676, ECO:0000269|PubMed:1415254, ECO:0000269|PubMed:1763037, ECO:0000269|PubMed:18422995, ECO:0000269|PubMed:2243141, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8168815}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal Resesif, Sitokrom B-Negatif (Kronik Granülomatöz Hastalık) (Prenatal) (CYBA gen) (Tüm Gen Dizi Analizi) (Prenatal)	GRANULOMATOUS DISEASE, CHRONIC, AUTOSOMAL RECESSIVE, CYTOCHROME b-NEGATIVE (Chronic granulomatous disease) (CYBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYBA	CYBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-negative, autosomal recessive (ARCGD) [MIM:233690]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:10759707, ECO:0000269|PubMed:10910929, ECO:0000269|PubMed:10914676, ECO:0000269|PubMed:1415254, ECO:0000269|PubMed:1763037, ECO:0000269|PubMed:18422995, ECO:0000269|PubMed:2243141, ECO:0000269|PubMed:23910690, ECO:0000269|PubMed:8168815}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal Resesif, Sitokrom B Pozitif, Tip 3, 613960, Otozomal Resesif; CDG3 (Kronik Granülomatöz Hastalık) (NCF4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960, Autosomal recessive; CDG3 (Chronic granulomatous disease) (NCF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NCF4	NCF4, P40PHOX, CGD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-positive 3, autosomal recessive (CGD3) [MIM:613960]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:19692703}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kronik Granülomatöz Hastalık, Otozomal Resesif, Sitokrom B Pozitif, Tip 3, 613960, Otozomal Resesif; CDG3 (Kronik Granülomatöz Hastalık) (Prenatal) (NCF4 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Granulomatous disease, chronic, autosomal recessive, cytochrome b-positive, type III, 613960, Autosomal recessive; CDG3 (Chronic granulomatous disease) (NCF4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NCF4	NCF4, P40PHOX, CGD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Granulomatous disease, chronic, cytochrome-b-positive 3, autosomal recessive (CGD3) [MIM:613960]: A disorder characterized by the inability of neutrophils and phagocytes to kill microbes that they have ingested. Patients suffer from life-threatening bacterial/fungal infections. {ECO:0000269|PubMed:19692703}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kronik Granülomatöz Hastalık, X'e Bağlı; Cdgx (Kronik Granülomatöz Hastalık) (CYBB gen) (Tüm Gen Dizi Analizi) (Postnatal)	GRANULOMATOUS DISEASE, CHRONIC, X-LINKED, CDGX (Chronic granulomatous disease) (CYBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. {ECO:0000269|PubMed:21278736}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Kronik Granülomatöz Hastalık, X'e Bağlı; Cdgx (Kronik Granülomatöz Hastalık) (Prenatal) (CYBB gen) (Tüm Gen Dizi Analizi) (Prenatal)	GRANULOMATOUS DISEASE, CHRONIC, X-LINKED, CDGX (Chronic granulomatous disease) (CYBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. {ECO:0000269|PubMed:21278736}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gri Trombosit Sendromu, 139090, Otozomal Resesif; GPS (Gri Trombosit Sendromu) (NBEAL2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Gray platelet syndrome, 139090, Autosomal recessive; GPS (Gray platelet syndrome) (NBEAL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBEAL2	NBEAL2, KIAA0540, GPS, BDPLT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gri Trombosit Sendromu, 139090, Otozomal Resesif; GPS (Gri Trombosit Sendromu) (Prenatal) (NBEAL2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Gray platelet syndrome, 139090, Autosomal recessive; GPS (Gray platelet syndrome) (NBEAL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NBEAL2	NBEAL2, KIAA0540, GPS, BDPLT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Greenberg İskelet Displazisi, 215140, Otozomal Resesif; GRBGD (Greenberg Displazisi) (LBR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Greenberg skeletal dysplasia, 215140, Autosomal recessive; GRBGD (Greenberg dysplasia) (LBR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Greenberg İskelet Displazisi, 215140, Otozomal Resesif; GRBGD (Greenberg Displazisi) (Prenatal) (LBR gen) (Tüm Gen Dizi Analizi) (Prenatal)	Greenberg skeletal dysplasia, 215140, Autosomal recessive; GRBGD (Greenberg dysplasia) (LBR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Greig Sefalopolisindaktili Sendromu, 175700, Otozomal Dominant; GCPS (Greig Sefalopolisindaktili Sendromu) (GLI3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Greig cephalopolysyndactyly syndrome, 175700, Autosomal dominant; GCPS (Greig cephalopolysyndactyly syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Greig Sefalopolisindaktili Sendromu, 175700, Otozomal Dominant; GCPS (Greig Sefalopolisindaktili Sendromu) (MLPA)	Greig cephalopolysyndactyly syndrome, 175700, Autosomal dominant; GCPS (Greig cephalopolysyndactyly syndrome) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Greig Sefalopolisindaktili Sendromu, 175700, Otozomal Dominant; GCPS (Greig Sefalopolisindaktili Sendromu) (Prenatal) (GLI3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Greig cephalopolysyndactyly syndrome, 175700, Autosomal dominant; GCPS (Greig cephalopolysyndactyly syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Greig Sefalopolisindaktili Sendromu, 175700, Otozomal Dominant; GCPS (Greig Sefalopolisindaktili Sendromu) (Prenatal) (MLPA)	Greig cephalopolysyndactyly syndrome, 175700, Autosomal dominant; GCPS (Greig cephalopolysyndactyly syndrome) (Prenatal) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Griscelli Sendromu, Tip 1, 214450, Otozomal Resesif; GS1 (Griscelli Hastalığı) (MYO5A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Griscelli syndrome, type 1, 214450, Autosomal recessive; GS1 (Griscelli disease) (MYO5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO5A	MYO5A, MYH12, GS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Griscelli Sendromu, Tip 1, 214450, Otozomal Resesif; GS1 (Griscelli Hastalığı) (Prenatal) (MYO5A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Griscelli syndrome, type 1, 214450, Autosomal recessive; GS1 (Griscelli disease) (MYO5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYO5A	MYO5A, MYH12, GS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Griscelli Sendromu, Otozomal Resesif Tip 2, 607624; GS2 (Griscelli Hastalığı) (RAB27A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Griscelli syndrome, type 2, 607624, Autosomal recessive; GS2 (Griscelli disease) (RAB27A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB27A	RAB27A, RAM, GS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Griscelli Sendromu, Otozomal Resesif Tip 2, 607624; GS2 (Griscelli Hastalığı) (Prenatal) (RAB27A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Griscelli syndrome, type 2, 607624, Autosomal recessive; GS2 (Griscelli disease) (RAB27A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB27A	RAB27A, RAM, GS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Griscelli Sendromu, Tip 3, 609227, Otozomal Resesif; GS3 (Griscelli Hastalığı) (MLPH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Griscelli syndrome, type 3, 609227, Autosomal recessive; GS3 (Griscelli disease) (MLPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLPH	MLPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Griscelli syndrome 3 (GS3) [MIM:609227]: Rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes, without other clinical manifestations. {ECO:0000269|PubMed:12897212}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Griscelli Sendromu, Tip 3, 609227, Otozomal Resesif; GS3 (Griscelli Hastalığı) (Prenatal) (MLPH gen) (Tüm Gen Dizi Analizi) (Prenatal)	Griscelli syndrome, type 3, 609227, Autosomal recessive; GS3 (Griscelli disease) (MLPH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MLPH	MLPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Griscelli syndrome 3 (GS3) [MIM:609227]: Rare autosomal recessive disorder characterized by pigmentary dilution of the skin and hair, the presence of large clumps of pigment in hair shafts, and an accumulation of melanosomes in melanocytes, without other clinical manifestations. {ECO:0000269|PubMed:12897212}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyüme Hormonu Eksikliği (GHD) panel (GH1, LHX4, POU1F1, HESX1, PROP1, GHRHR, LHX3) (MLPA)	Growth Hormone Deficiency (GHD) panel (GH1, LHX4, POU1F1, HESX1, PROP1, GHRHR, LHX3 (MLPA)	GH1, LHX4, POU1F1, HESX1, PROP1, GHRHR, LHX3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği-Hipofiz Anomalisi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (HESX1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency with pituitary anomalies, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (HESX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HESX1	HESX1, RPX, CPHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. {ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620787}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Büyüme Hormonu Eksikliği-Hipofiz Anomalisi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (MLPA)	Growth hormone deficiency with pituitary anomalies, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (MLPA)	HESX1	HESX1, RPX, CPHD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. (ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole Parsiyel, 615925, Otozomal Resesif, Otozomal Dominant, GHDP (Kısa Boy, GHSR Eksikliğine Bağlı) (GHSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency, isolated partial, 615925, Autosomal recessive, Autosomal dominant; GHDP (Short stature due to GHSR deficiency) (GHSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHSR	GHSR, GHDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1A, 262400, Otozomal Resesif; IGHD1A (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (GH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency, isolated, type IA, 262400, Autosomal recessive; IGHD1A (Non-acquired isolated growth hormone deficiency) (GH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GH1	GH1, GHN, IGHD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1A, 262400, Otozomal Resesif; IGHD1A (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (MLPA)	Growth hormone deficiency, isolated, type IA, 262400, Autosomal recessive; IGHD1A (Non-acquired isolated growth hormone deficiency) (MLPA)	GH1	GH1, GHN, IGHD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1B, 612781 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (GH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency, isolated, type IB, 612781 (Non-acquired isolated growth hormone deficiency) (GH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GH1	GH1, GHN, IGHD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1B, 612781 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (MLPA)	Growth hormone deficiency, isolated, type IB, 612781 (Non-acquired isolated growth hormone deficiency) (MLPA)	GH1	GH1, GHN, IGHD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1B, 612781; IGHD1B (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (GHRHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency, isolated, type IB, 612781; IGHD1B (Non-acquired isolated growth hormone deficiency) (GHRHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHRHR	GHRHR, GHRFR, IGHD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 1B, 612781; IGHD1B (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (MLPA)	Growth hormone deficiency, isolated, type IB, 612781; IGHD1B (Non-acquired isolated growth hormone deficiency) (MLPA)	GHRHR	GHRHR, GHRFR, IGHD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 2, 173100, Otozomal Dominant; IGHD2 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (GH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone deficiency, isolated, type II, 173100, Autosomal dominant; IGHD2 (Non-acquired isolated growth hormone deficiency) (GH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GH1	GH1, GHN, IGHD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Growth hormone deficiency, isolated, 2 (IGHD2) [MIM:173100]: An autosomal dominant deficiency of growth hormone leading to short stature. Clinical severity is variable. Patients have a positive response and immunologic tolerance to growth hormone therapy. (ECO:0000269|PubMed:11502836, ECO:0000269|PubMed:9152628). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Büyüme Hormonu Eksikliği, İzole, Tip 2, 173100, Otozomal Dominant; IGHD2 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (MLPA)	Growth hormone deficiency, isolated, type II, 173100, Autosomal dominant; IGHD2 (Non-acquired isolated growth hormone deficiency) (MLPA)	GH1	GH1, GHN, IGHD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Growth hormone deficiency, isolated, 2 (IGHD2) [MIM:173100]: An autosomal dominant deficiency of growth hormone leading to short stature. Clinical severity is variable. Patients have a positive response and immunologic tolerance to growth hormone therapy. (ECO:0000269|PubMed:11502836, ECO:0000269|PubMed:9152628). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Büyüme Hormonu Duyarsızlığı, İmmün Yetmezlik, 245590 (Laron Sendromu-İmmun Yetmezlik) (STAT5B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone insensitivity with immunodeficiency, 245590 (Laron syndrome with immunodeficiency) (STAT5B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT5B	STAT5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Growth hormone insensitivity with immunodeficiency (GHI) [MIM:245590]: A disease characterized by short stature, growth hormone deficiency in the presence of normal to elevated circulating concentrations of growth hormone, resistance to hexogeneous growth hormone therapy, and recurrent infections. (ECO:0000269|PubMed:13679528, ECO:0000269|PubMed:15827093, ECO:0000269|PubMed:22419735). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Büyüme Hormonu Duyarsızlık, Parsiyel, 604271; GHIP (Kısa Boy, Parsiyel GHR Eksikliğine Bağlı) (GHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth hormone insensitivity, partial, 604271; GHIP (Short stature due to partial GHR deficiency) (GHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHR	GHR, GHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Kısıtlaması, Ağır, Belirgin Yüz Bulguları, 616489, X'e Bağlı; GRDF (Silver-Russell Sendromu) (IGF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth restriction, severe, with distinctive facies, 616489, X-linked recessive; GRDF (Silver-Russell syndrome) (IGF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGF2	IGF2, GRDF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Kısıtlaması, Ağır, Belirgin Yüz Bulguları, 616489, X'e Bağlı; GRDF (Silver-Russell Sendromu) (MLPA)	Growth restriction, severe, with distinctive facies, 616489, X-linked recessive; GRDF (Silver-Russell syndrome) (MLPA)	IGF2	IGF2, GRDF	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Büyüme Kısıtlaması, Ağır, Belirgin Yüz Bulguları, 616489, X'e Bağlı; GRDF (Silver-Russell Sendromu) (Prenatal) (IGF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Growth restriction, severe, with distinctive facies, 616489, X-linked recessive; GRDF (Silver-Russell syndrome) (IGF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGF2	IGF2, GRDF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyüme Kısıtlaması, Ağır, Belirgin Yüz Bulguları, 616489, X'e Bağlı; GRDF (Silver-Russell Sendromu) (Prenatal) (MLPA)	Growth restriction, severe, with distinctive facies, 616489, X-linked recessive; GRDF (Silver-Russell syndrome) (Prenatal) (MLPA)	IGF2	IGF2, GRDF	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyüme Geriliği, Sağırlık ve Mental Retardasyon, IGF1 Eksikliğine Bağlı, 608747, Otozomal Resesif (Büyüme Gecikmesi, İnsülin Benzeri Büyüme Faktörü Tip 1 (IGF1) Eksikliğine Bağlı) (IGF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747, Autosomal recessive (Growth delay due to insulin-like growth factor type 1 deficiency) (IGF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGF1	IGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Insulin-like growth factor I deficiency (IGF1 deficiency) [MIM:608747]: Autosomal recessive disorder characterized by growth retardation, sensorineural deafness and mental retardation. (ECO:0000269	PubMed:8857020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Büyüme Geriliği, Sağırlık ve Mental Retardasyon, IGF1 Eksikliğine Bağlı, 608747, Otozomal Resesif (Büyüme Gecikmesi, İnsülin Benzeri Büyüme Faktörü Tip 1 (IGF1) Eksikliğine Bağlı) (Prenatal) (IGF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747, Autosomal recessive (Growth delay due to insulin-like growth factor type 1 deficiency) (IGF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGF1	IGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Insulin-like growth factor I deficiency (IGF1 deficiency) [MIM:608747]: Autosomal recessive disorder characterized by growth retardation, sensorineural deafness and mental retardation. (ECO:0000269	PubMed:8857020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyüme Geriliği, Gelişimsel Gecikme, Yüzde Dismorfizm 612938, Otozomal Resesif (Letal Polimalformatif Sendrom, Boissel Tip) (FTO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth retardation, developmental delay, facial dysmorphism, 612938, Autosomal recessive (Lethal polymalformative syndrome, Boissel type) (FTO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTO	FTO, GDFD, BMIQ14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Geriliği, Gelişimsel Gecikme, Yüzde Dismorfizm 612938, Otozomal Resesif (Letal Polimalformatif Sendrom, Boissel Tip) (Prenatal) (FTO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Growth retardation, developmental delay, facial dysmorphism, 612938, Autosomal recessive (Lethal polymalformative syndrome, Boissel type) (FTO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FTO	FTO, GDFD, BMIQ14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyüme Geriliği, Entelektüel Gelişimsel Bozukluk, Hipotoni ve Hepatopati, 617093, Otozomal Resesif; GRIDHH (IARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093, Autosomal recessive; GRIDHH (IARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IARS	IARS, GRIDHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyüme Geriliği, Entelektüel Gelişimsel Bozukluk, Hipotoni ve Hepatopati, 617093, Otozomal Resesif; GRIDHH (Prenatal) (IARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Growth retardation, intellectual developmental disorder, hypotonia, and hepatopathy, 617093, Autosomal recessive; GRIDHH (IARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IARS	IARS, GRIDHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
GTPaz, Çok Büyük İnterferon-İndüklenebilir, Psödogen 1; GVINP1 (GVINP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GTPase, VERY LARGE INTERFERON-INDUCIBLE, PSEUDOGENE 1; GVINP1 (GVINP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GVINP1	GVINP1, VLIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alesel Guillain-Barre Sendromu; GBS (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	GUILLAIN-BARRE SYNDROME, FAMILIAL; GBS (Acute inflammatory demyelinating polyradiculoneuropathy) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Alesel Guillain-Barre Sendromu; GBS (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati (MLPA)	GUILLAIN-BARRE SYNDROME, FAMILIAL; GBS (Acute inflammatory demyelinating polyradiculoneuropathy) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Alesel Guillain-Barre Sendromu; GBS (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati) (Prenatal) (PMP22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	GUILLAIN-BARRE SYNDROME, FAMILIAL; GBS (Acute inflammatory demyelinating polyradiculoneuropathy) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Alesel Guillain-Barre Sendromu; GBS (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati) (Prenatal) (MLPA)	GUILLAIN-BARRE SYNDROME, FAMILIAL; GBS (Acute inflammatory demyelinating polyradiculoneuropathy) (Prenatal) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Guttmacher Sendromu, 176305, Otozomal Dominant (Guttmacher Sendromu) (HOXA13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Guttmacher syndrome, 176305, Autosomal dominant (Guttmacher syndrome) (HOXA13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA13	HOXA13, HOX1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Guttmacher Sendromu, 176305, Otozomal Dominant (Guttmacher Sendromu) (Prenatal) (HOXA13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Guttmacher syndrome, 176305, Autosomal dominant (Guttmacher syndrome) (HOXA13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXA13	HOXA13, HOX1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koroid ve Retinanın Girat Atrofisi, ± Ornitinemi, 258870, Otozomal Resesif; GACR (Koroid ve Retinanın Girat Atrofisi) (OAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870, Autosomal recessive; GACR (Gyrate atrophy of choroid and retina) (OAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OAT	OAT, GACR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperornithinemia with gyrate atrophy of choroid and retina (HOGA) [MIM:258870]: A disorder clinically characterized by a triad of progressive chorioretinal degeneration, early cataract formation, and type II muscle fiber atrophy. Characteristic chorioretinal atrophy with progressive constriction of the visual fields leads to blindness at the latest during the sixth decade of life. Patients generally have normal intelligence. (ECO:0000269|PubMed:1612597, ECO:0000269|PubMed:1737786, ECO:0000269|PubMed:23076989, ECO:0000269|PubMed:2793865, ECO:0000269|PubMed:3375340, ECO:0000269|PubMed:7668253, ECO:0000269|PubMed:7887415). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Koroid ve Retinanın Girat Atrofisi, ± Ornitinemi, 258870, Otozomal Resesif; GACR (Koroid ve Retinanın Girat Atrofisi) (Prenatal) (OAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Gyrate atrophy of choroid and retina with or without ornithinemia, 258870, Autosomal recessive; GACR (Gyrate atrophy of choroid and retina) (OAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OAT	OAT, GACR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperornithinemia with gyrate atrophy of choroid and retina (HOGA) [MIM:258870]: A disorder clinically characterized by a triad of progressive chorioretinal degeneration, early cataract formation, and type II muscle fiber atrophy. Characteristic chorioretinal atrophy with progressive constriction of the visual fields leads to blindness at the latest during the sixth decade of life. Patients generally have normal intelligence. (ECO:0000269|PubMed:1612597, ECO:0000269|PubMed:1737786, ECO:0000269|PubMed:23076989, ECO:0000269|PubMed:2793865, ECO:0000269|PubMed:3375340, ECO:0000269|PubMed:7668253, ECO:0000269|PubMed:7887415). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

H. Pylori Enfeksiyonu, Duyarlılık, 600263 (PTPRZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	H. pylori infection, susceptibility to, 600263 (PTPRZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRZ1	PTPRZ1, PTP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H. Pylori Enfeksiyonu, Duyarlılık, 600263 (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	H. pylori infection, susceptibility to, 600263 (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Haddad Sendromu, 209880, Otozomal Dominant (Haddad sendromu) (ASCL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Haddad syndrome, 209880, Autosomal dominant (Haddad syndrome) (ASCL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASCL1	ASCL1, ASH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Haddad Sendromu, 209880, Otozomal Dominant (Haddad sendromu) (Prenatal) (ASCL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Haddad syndrome, 209880, Autosomal dominant (Haddad syndrome) (ASCL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASCL1	ASCL1, ASH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hailey-Hailey Hastalığı, 169600, Otozomal Dominant; BCPM (Ailesel Benign Kronik Pemfigus (Hailey-Hailey Hastalığı)) (ATP2C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hailey-Hailey disease, 169600, Autosomal dominant; BCPM (Familial benign chronic pemphigus) (ATP2C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2C1	ATP2C1, BCPM, HHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Haim-Munk Sendromu, 245010, Otozomal Resesif; HMS (Haim-Munk Sendromu) (CTSC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Haim-Munk syndrome, 245010, Autosomal recessive; HMS (Haim-Munk syndrome) (CTSC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSC	CTSC, CPPI, PALS, PLS, HMS, PDON1, JPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Haim-Munk Sendromu, 245010, Otozomal Resesif; HMS (Haim-Munk Sendromu) (Prenatal) (CTSC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Haim-Munk syndrome, 245010, Autosomal recessive; HMS (Haim-Munk syndrome) (CTSC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTSC	CTSC, CPPI, PALS, PLS, HMS, PDON1, JPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hajdu-Cheney Sendromu, 102500, Otozomal Dominant; HJCYS (Akroosteolitis Dominant Tip) (NOTCH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hajdu-Cheney syndrome, 102500, Autosomal dominant; HJCYS (Acroosteolysis dominant type) (NOTCH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH2	NOTCH2, AGS2, HJCYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hajdu-Cheney syndrome (HJCYS) [MIM:102500]: A rare skeletal disorder characterized by the association of facial anomalies, acro-osteolysis, general osteoporosis, insufficient ossification of the skull, and periodontal disease (premature loss of permanent teeth). Other features include cleft palate, congenital heart defects, polycystic kidneys, orthopedic problems and anomalies of the genitalia, intestines and eyes. {ECO:0000269|PubMed:21378985, ECO:0000269|PubMed:21378989}. Note=The disease is caused by mutations affecting the gene represented in this entry. NOTCH2 mutations associated with Hajdu- Cheney syndrome cluster to the last coding exon of the gene. This suggests that the mutant mRNA products may escape nonsense- mediated decay and the resulting truncated NOTCH2 proteins act in a gain-of-function manner.	EDTA Blood Sample
Hajdu-Cheney Sendromu, 102500, Otozomal Dominant; HJCYS (Akroosteolitis Dominant Tip) (Prenatal) (NOTCH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hajdu-Cheney syndrome, 102500, Autosomal dominant; HJCYS (Acroosteolysis dominant type) (NOTCH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOTCH2	NOTCH2, AGS2, HJCYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hajdu-Cheney syndrome (HJCYS) [MIM:102500]: A rare skeletal disorder characterized by the association of facial anomalies, acro-osteolysis, general osteoporosis, insufficient ossification of the skull, and periodontal disease (premature loss of permanent teeth). Other features include cleft palate, congenital heart defects, polycystic kidneys, orthopedic problems and anomalies of the genitalia, intestines and eyes. {ECO:0000269|PubMed:21378985, ECO:0000269|PubMed:21378989}. Note=The disease is caused by mutations affecting the gene represented in this entry. NOTCH2 mutations associated with Hajdu- Cheney syndrome cluster to the last coding exon of the gene. This suggests that the mutant mRNA products may escape nonsense- mediated decay and the resulting truncated NOTCH2 proteins act in a gain-of-function manner.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hamamy Sendromu, 611174, Otozomal Resesif; HMMS (Kraniofasiyal Displazi-Osteopeni Sendromu) (IRX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hamamy syndrome, 611174, Autosomal recessive; HMMS (Craniofacial dysplasia-osteopenia syndrome) (IRX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRX5	IRX5, HMMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hamamy syndrome (HMMS) [MIM:611174]: A syndrome characterized by severe hypertelorism, upslanting palpebral fissures, brachycephaly, abnormal ears, sloping shoulders, enamel hypoplasia, and osteopenia with repeated fractures. Additional features include myopia, mild to moderate sensorineural hearing loss, gonadal anomalies and borderline intelligence. {ECO:0000269|PubMed:22581230}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hamamy Sendromu, 611174, Otozomal Resesif; HMMS (Kraniofasiyal Displazi-Osteopeni Sendromu) (Prenatal) (IRX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hamamy syndrome, 611174, Autosomal recessive; HMMS (Craniofacial dysplasia-osteopenia syndrome) (IRX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRX5	IRX5, HMMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hamamy syndrome (HMMS) [MIM:611174]: A syndrome characterized by severe hypertelorism, upslanting palpebral fissures, brachycephaly, abnormal ears, sloping shoulders, enamel hypoplasia, and osteopenia with repeated fractures. Additional features include myopia, mild to moderate sensorineural hearing loss, gonadal anomalies and borderline intelligence. {ECO:0000269|PubMed:22581230}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
El-Ayak-Uterus Sendromu, 140000, Otozomal Dominant; HFG (El Ayak Genital Sendromi) (HOXA13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hand-foot-uterus syndrome, 140000, Autosomal dominant; HFG (Hand-foot-genital syndrome) (HOXA13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA13	HOXA13, HOX1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hand-foot-genital syndrome (HFG) [MIM:140000]: A disorder characterized by limb and genitourinary anomalies. Clinical features include small feet with unusually short great toes and abnormal thumbs. Females with the disorder have duplication of the genital tract. {ECO:0000269|PubMed:10839976, ECO:0000269|PubMed:12073020}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hangover, Yatkınlık, 610251, Otozomal Dominant (ALDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hangover, susceptibility to, 610251, Autosomal dominant (ALDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH2	ALDH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Harderoporfiria, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (CPOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Harderoporphyria, 121300, Autosomal dominant (Hereditary coproporphyria) (CPOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPOX	CPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Harderoporfiria, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (MLPA)	Harderoporphyria, 121300, Autosomal dominant (Hereditary coproporphyria) (MLPA)	CPOX	CPOX	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Harderoporfiria, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (CPOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Harderoporphyria, 121300, Autosomal dominant (Hereditary coproporphyria) (CPOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPOX	CPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9888388}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Harderoporfiria, 121300, Otozomal Dominant (Kalıtsal Koproporfiri) (Prenatal) (MLPA)	Harderoporphyria, 121300, Autosomal dominant (Hereditary coproporphyria) (Prenatal) (MLPA)	CPOX	CPOX	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary coproporphyria (HCP) [MIM:121300]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Hereditary coproporphyria is an acute hepatic porphyria characterized by skin photosensitivity, attacks of abdominal pain, neurological disturbances, and psychiatric symptoms. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. Hereditary coproporphyria is biochemically characterized by overexcretion of coproporphyrin III in the urine and in the feces. {ECO:0000269|PubMed:12181641, ECO:0000269|PubMed:15896662, ECO:0000269|PubMed:16398658, ECO:0000269|PubMed:7757079, ECO:0000269|PubMed:7849704, ECO:0000269|PubMed:8012360, ECO:0000269|PubMed:8990017, ECO:0000269|PubMed:9048920, ECO:0000269|PubMed:9298818, ECO:0000269|PubMed:9883388}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Harel Yoon Sendromu, 617183, Otozomal Resesif, Otozomal Dominant; HAYOS (ATAD3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Harel-Yoon syndrome, 617183, Autosomal recessive, Autosomal dominant; HAYOS (ATAD3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATAD3A	ATAD3A, HAYOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Harel Yoon Sendromu, 617183, Otozomal Resesif, Otozomal Dominant; HAYOS (Prenatal) (ATAD3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Harel-Yoon syndrome, 617183, Autosomal recessive, Autosomal dominant; HAYOS (ATAD3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATAD3A	ATAD3A, HAYOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HARP Sendromu, 607236, Otozomal Resesif (ARP Sendromu) (PANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HARP syndrome, 607236, Autosomal recessive (HARP syndrome) (PANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PANK2	PANK2, NBIA1, PKAN, HARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269|PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
HARP Sendromu, 607236, Otozomal Resesif (ARP Sendromu) (MLPA)	HARP syndrome, 607236, Autosomal recessive (HARP syndrome) (MLPA)	PANK2	PANK2, NBIA1, PKAN, HARP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269|PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
HARP Sendromu, 607236, Otozomal Resesif (ARP Sendromu) (Prenatal) (PANK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	HARP syndrome, 607236, Autosomal recessive (HARP syndrome) (PANK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PANK2	PANK2, NBIA1, PKAN, HARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269|PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HARP Sendromu, 607236, Otozomal Resesif (ARP Sendromu) (Prenatal) (MLPA)	HARP syndrome, 607236, Autosomal recessive (HARP syndrome) (Prenatal) (MLPA)	PANK2	PANK2, NBIA1, PKAN, HARP	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269|PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hartnup Hastalığı, 234500, Otozomal Resesif, HND (Hartnup Hastalığı) (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hartnup disorder, 234500, Autosomal recessive; HND (Hartnup disease) (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hartnup Hastalığı, 234500, Otozomal Resesif, HND (Hartnup Hastalığı) (Prenatal) (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hartnup disorder, 234500, Autosomal recessive; HND (Hartnup disease) (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hartsfield Sendromu, 615465, Otozomal Dominant; HRTFDS (Hartsfield-Bixler-Demyer Sendromu) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hartsfield syndrome, 615465, Autosomal dominant; HRTFDS (Hartsfield-Bixler-Demyer syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hartsfield Sendromu, 615465, Otozomal Dominant; HRTFDS (Hartsfield-Bixler-Demyer Sendromu) (MLPA)	Hartsfield syndrome, 615465, Autosomal dominant; HRTFDS (Hartsfield-Bixler-Demyer syndrome) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hartsfield Sendromu, 615465, Otozomal Dominant; HRTFDS (Hartsfield-Bixler-Demyer Sendromu) (Prenatal) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hartsfield syndrome, 615465, Autosomal dominant; HRTFDS (Hartsfield-Bixler-Demyer syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hartsfield Sendromu, 615465, Otozomal Dominant; HRTFDS (Hartsfield-Bixler-Demyer Sendromu) (Prenatal) (MLPA)	Hartsfield syndrome, 615465, Autosomal dominant; HRTFDS (Hartsfield-Bixler-Demyer syndrome) (Prenatal) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hashimoto Tiroidit, 140300, Otozomal Dominant (CTLA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hashimoto thyroiditis, 140300, Autosomal dominant (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hawkinsinüri, 140350, Otozomal Dominant (Hawkinsinüri) (HPD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hawkinsinuria, 140350, Autosomal dominant (Hawkinsinuria) (HPD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPD	HPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hawkinsinuria (HAWK) [MIM:140350]: An inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin, in the urine. {ECO:0000269|PubMed:11073718}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Hawkinsinüri, 140350, Otozomal Dominant (Hawkinsinüri) (Prenatal) (HPD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hawkinsinuria, 140350, Autosomal dominant (Hawkinsinuria) (HPD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPD	HPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hawkinsinuria (HAWK) [MIM:140350]: An inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin, in the urine. {ECO:0000269|PubMed:11073718}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hay-Wells Sendromu, 106260, Otozomal Dominant (Ankiloblefaron-Ektodermal Defekt-Yarık Dudak / Damak Sendromu (Hay-Wells Sendromu)) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hay-Wells syndrome, 106260, Autosomal dominant (Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hay-Wells Sendromu, 106260, Otozomal Dominant (Ankiloblefaron-Ektodermal Defekt-Yarık Dudak / Damak Sendromu (Hay-Wells Sendromu)) (Prenatal) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hay-Wells syndrome, 106260, Autosomal dominant (Ankyloblepharon-ectodermal defects-cleft lip/palate syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HDL Eksikliği, Tip 2, 604091 (Apolipoprotein A-I Eksikliği (ABCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HDL deficiency, type 2, 604091 (Apolipoprotein A-I deficiency) (ABCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA1	ABCA1, ABC1, HDLDT1, TGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hormon Reziztansı, HDL Tepki, Artmış (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HDL response to hormone replacement, augmented (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İşitme kaybı paneli (GJB3 1p34.3 (konnexin 31), WFS1 4p16.1 (Wolframin)) (MLPA)	Hearing loss panel (GJB3 1p34.3 (connexin 31), WFS1 4p16.1 (Wolframin)) (MLPA)	GJB3 1p34.3 (connexin 31), WFS1 4p16.1 (Wolframin)	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kalp ve Beyin Malformasyon Sendromu, 616920, Otozomal Resesif; HBMS (SMG9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heart and brain malformation syndrome, 616920, Autosomal recessive; HBMS (SMG9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMG9	C19orf61, SMG9, HBMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalp ve Beyin Malformasyon Sendromu, 616920, Otozomal Resesif; HBMS (Prenatal) (SMG9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heart and brain malformation syndrome, 616920, Autosomal recessive; HBMS (SMG9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMG9	C19orf61, SMG9, HBMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kalp Blokları, İlerlemeyen, 113900, Otozomal Dominant (Ailesel Progresif Kardiyak İletim Defekti) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heart block, nonprogressive, 113900, Autosomal dominant (Familial progressive cardiac conduction defect) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VFL, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalp Bloktan, Progresif, Tip 1A, 113900, Otozomal Dominant (Ailesel Progresif Kardiyak İletim Defekti) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heart block, progressive, type IA, 113900, Autosomal dominant (Familial progressive cardiac conduction defect) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VFL, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalp Anomalisi, Konjenital ve Diğer Konjenital Anomaliler; Hdca (Pankreatik Hipoplazi-Diyabet-Konjenital Kalp Hastalığı Sendromu) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HEART DEFECTS, CONGENITAL, AND OTHER CONGENITAL ANOMALIES; HDCA (Pancreatic hypoplasia-diabetes-congenital heart disease syndrome) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kalp-El Sendromu, Slovenya Türü, 610140, Otozomal Dominant (Kalp-El Sendromu, Sloven Tip) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heart-hand syndrome, Slovenian type, 610140, Autosomal dominant (Heart-hand syndrome, Slovenian type) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalp-El Sendromu, Slovenya Türü, 610140, Otozomal Dominant (Kalp-El Sendromu, Sloven Tip) (MLPA)	Heart-hand syndrome, Slovenian type, 610140, Autosomal dominant (Heart-hand syndrome, Slovenian type) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kalp-El Sendromu, Slovenya Türü, 610140, Otozomal Dominant (Kalp-El Sendromu, Sloven Tip) (Prenatal) (LMNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heart-hand syndrome, Slovenian type, 610140, Autosomal dominant (Heart-hand syndrome, Slovenian type) (LMNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kalp-El Sendromu, Slovenya Türü, 610140, Otozomal Dominant (Kalp-El Sendromu, Sloven Tip) (Prenatal) (MLPA)	Heart-hand syndrome, Slovenian type, 610140, Autosomal dominant (Heart-hand syndrome, Slovenian type) (Prenatal) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heimler Sendromu 1, 234580, Otozomal Resesif; HMLR1 (Sağırlık-Mine Hipoplazisi-Tırnak Defekti Sendromu (Heimler Sendromu)) (PEX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heimler syndrome 1, 234580, Autosomal recessive; HMLR1 (Deafness-enamel hypoplasia-nail defects syndrome) (PEX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX1	PEX1, ZWS1, PBD1A, PBD1B, HMLR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heimler Sendromu 2, 616617, Otozomal Resesif; HMLR2 (Sağırlık-Mine Hipoplazisi-Tırnak Defekti Sendromu (Heimler Sendromu)) (PEX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heimler syndrome 2, 616617, Autosomal recessive; HMLR2 (Deafness-enamel hypoplasia-nail defects syndrome) (PEX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX6	PEX6, PXAAA1, PAF2, PBD4A, PDB4B, HMLR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heinz Body Anemi, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (MLPA)	Heinz body anemia, 140700, Autosomal dominant (Heinz body anemia) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Heinz Body Anemi, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (Prenatal) (MLPA)	Heinz body anemia, 140700, Autosomal dominant (Heinz body anemia) (Prenatal) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heinz Body Anemiler, Alfa-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (MLPA)	Heinz body anemias, alpha-, 140700, Autosomal dominant (Heinz body anemia) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Heinz Body Anemiler, Alfa-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (Prenatal) (MLPA)	Heinz body anemias, alpha-, 140700, Autosomal dominant (Heinz body anemia) (Prenatal) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heinz Body Anemiler, Beta-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heinz body anemias, beta-, 140700, Autosomal dominant (Heinz body anemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Heinz Body Anemiler, Beta-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (MLPA)	Heinz body anemias, beta-, 140700, Autosomal dominant (Heinz body anemia) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Heinz Body Anemiler, Beta-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (Prenatal) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heinz body anemias, beta-, 140700, Autosomal dominant (Heinz body anemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heinz Body Anemiler, Beta-, 140700, Otozomal Dominant (Heinz Cisimciği Anemisi) (Prenatal) (MLPA)	Heinz body anemias, beta-, 140700, Autosomal dominant (Heinz body anemia) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HELLP Sendromu, Maternal, Gebelik, 609016, Otozomal Resesif (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (HADHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	HELLP syndrome, maternal, of pregnancy, 609016, Autosomal recessive (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269	PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
HELLP Sendromu, Maternal, Gebelik, 609016, Otozomal Resesif (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (Prenatal) (HADHA geni) (Tüm Gen Dizi Analizi) (Prenatal)	HELLP syndrome, maternal, of pregnancy, 609016, Autosomal recessive (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269	PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Helsmoortel-Van Der Aa Sendromu, 615873, Otozomal Dominant; HVDAS (ADNP İlişkili Multipl Konjenital Anomaliler-Mental Retardasyon-Otizm Spektrum Bozukluğu) (ADNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Helsmoortel-van der Aa syndrome, 615873, Autosomal dominant; HVDAS (ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder) (ADNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADNP	ADNP, ADNP1, KIAA0784, HVDAS, MRD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Helsmoortel-Van Der Aa Sendromu, 615873, Otozomal Dominant; HVDAS (ADNP İlişkili Multipl Konjenital Anomaliler-Mental Retardasyon-Otizm Spektrum Bozukluğu) (Prenatal) (ADNP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Helsmoortel-van der Aa syndrome, 615873, Autosomal dominant; HVDAS (ADNP-related multiple congenital anomalies-intellectual disability-autism spectrum disorder) (ADNP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADNP	ADNP, ADNP1, KIAA0784, HVDAS, MRD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemanjyoblastom, Serebellar Somatik (VHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemangioblastoma, cerebellar, somatic (VHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VHL	VHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hemanjyom, Kapiller İnfantil, Somatik, 602089 (KDR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemangioma, capillary infantile, somatic, 602089 (KDR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDR	KDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Hemangioma, capillary infantile (HCI) [MIM:602089]: A condition characterized by dull red, firm, dome-shaped hemangiomas, sharply demarcated from surrounding skin, usually presenting at birth or occurring within the first two or three months of life. They result from highly proliferative, localized growth of capillary endothelium and generally undergo regression and involution without scarring. {ECO:0000269|PubMed:11807987, ECO:0000269|PubMed:18931684}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample/ Biopsy of affected site-transport media
Hemanjyom, Kapiller İnfantil, Somatik, 602089 (FLT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemangioma, capillary infantile, somatic, 602089 (FLT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLT4	FLT4, VEGFR3, PCL, LMPH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Hemanjyom, İnfantil Kapiler, Yatkınlık, 602089, Otozomal Dominant (KDR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemangioma, capillary infantile, susceptibility to, 602089, Autosomal dominant (KDR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDR	KDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemangioma, capillary infantile (HCI) [MIM:602089]: A condition characterized by dull red, firm, dome-shaped hemangiomas, sharply demarcated from surrounding skin, usually presenting at birth or occurring within the first two or three months of life. They result from highly proliferative, localized growth of capillary endothelium and generally undergo regression and involution without scarring. {ECO:0000269|PubMed:11807987, ECO:0000269|PubMed:18931684}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Hemanjyom, İnfantil Kapiler, Yatkınlık, 602089, Otozomal Dominant (GAPO Sendromu) (ANTXR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemangioma, capillary infantile, susceptibility to, 602089, Autosomal dominant (GAPO syndrome) (ANTXR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANTXR1	ANTXR1, TEM8, ATR, GAPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hematüri, Benign Ailesel, 141200, Otozomal Dominant (COL4A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hematuria, benign familial, 141200, Autosomal dominant (COL4A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A3	COL4A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hematüri, Benign Ailesel, 141200, Otozomal Dominant (MLPA)	Hematuria, benign familial, 141200, Autosomal dominant (MLPA)	COL4A3	COL4A3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Hematüri, Ailesel Benign (COL4A4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hematuria, familial benign (COL4A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A4	COL4A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hematuria, benign familial (BFH) [MIM:141200]: An autosomal dominant condition characterized by non-progressive isolated microscopic hematuria that does not result in renal failure. It is characterized pathologically by thinning of the glomerular basement membrane. (ECO:0000269|PubMed:11961012, ECO:0000269|PubMed:12631110, ECO:0000269|PubMed:8787673). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hematüri, Ailesel Benign (MLPA)	Hematuria, familial benign (MLPA)	COL4A4	COL4A4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hematuria, benign familial (BFH) [MIM:141200]: An autosomal dominant condition characterized by non-progressive isolated microscopic hematuria that does not result in renal failure. It is characterized pathologically by thinning of the glomerular basement membrane. (ECO:0000269|PubMed:11961012, ECO:0000269|PubMed:12631110, ECO:0000269|PubMed:8787673). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hem Oksijenaz-1 Eksikliği, 614034; HMOX1D (HMOX1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Heme oxygenase-1 deficiency, 614034; HMOX1D (HMOX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMOX1	HMOX1, HMOX1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heme oxygenase 1 deficiency (HMOX1D) [MIM:614034]: A disease characterized by impaired stress hematopoiesis, resulting in marked erythrocyte fragmentation and intravascular hemolysis, coagulation abnormalities, endothelial damage, and iron deposition in renal and hepatic tissues. Clinical features include persistent hemolytic anemia, asplenia, nephritis, generalized erythematous rash, growth retardation and hepatomegaly. (ECO:0000269	PubMed:9884342). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hem Oksijenaz-1 Eksikliği, 614034; HMOX1D (Prenatal) (HMOX1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Heme oxygenase-1 deficiency, 614034; HMOX1D (HMOX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HMOX1	HMOX1, HMOX1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heme oxygenase 1 deficiency (HMOX1D) [MIM:614034]: A disease characterized by impaired stress hematopoiesis, resulting in marked erythrocyte fragmentation and intravascular hemolysis, coagulation abnormalities, endothelial damage, and iron deposition in renal and hepatic tissues. Clinical features include persistent hemolytic anemia, asplenia, nephritis, generalized erythematous rash, growth retardation and hepatomegaly. (ECO:0000269	PubMed:9884342). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemokromatozis, 235200, Otozomal Resesif (Hemokromatoz Tip 1 Semptomatik Form) (HFE gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, 235200, Autosomal recessive (Symptomatic form of hemochromatosis type 1) (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemokromatozis, Tip 2A, 602390, Otozomal Resesif; HFE2A (Hemokromatozis Tip 2) (HJV (HFE2) gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, type 2A, 602390, Autosomal recessive; HFE2A (Hemochromatosis type 2) (HJV (HFE2) gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HJV	HFE2, HFE2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemochromatosis 2A (HFE2A) [MIM:602390]: A juvenile form of hemochromatosis, a disorder of iron metabolism with excess deposition of iron in a variety of organs leading to their failure, bronze skin pigmentation, hepatic cirrhosis, arthropathy and diabetes. The most common symptoms of juvenile hemochromatosis at presentation are hypogonadism and cardiomyopathy. (ECO:0000269|PubMed:14647275, ECO:0000269|PubMed:14982867, ECO:0000269|PubMed:14982873, ECO:0000269|PubMed:15461631). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemokromatozis, Tip 2B, 613313, Otozomal Resesif; HFE2B (Hemokromatozis Tip 3) (HAMP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, type 2B, 613313, Autosomal recessive; HFE2B (Hemochromatosis type 2) (HAMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HAMP	HAMP, LEAP1, HEPC, HFE2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemochromatosis 2B (HFE2B) [MIM:613313]: A juvenile form of hemochromatosis, a disorder of iron metabolism with excess deposition of iron in a variety of organs leading to their failure, bronze skin pigmentation, hepatic cirrhosis, arthropathy and diabetes. The most common symptoms of juvenile hemochromatosis at presentation are hypogonadism and cardiomyopathy. (ECO:0000269|PubMed:12915468, ECO:0000269|PubMed:14630809, ECO:0000269|PubMed:14633868, ECO:0000269|PubMed:14670915, ECO:0000269|PubMed:15099344). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemokromatozis, Tip 3, 604250, Otozomal Resesif (Hemokromatozis Tip 3) (TFR2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, type 3, 604250, Autosomal recessive (Hemochromatosis type 3) (TFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFR2	TFR2, HFE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemokromatozis, Tip 4, 606069, Otozomal Dominant; HFE4 (Hemokromatozis Tip 4) (SLC40A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, type 4, 606069, Autosomal dominant; HFE4 (Hemochromatosis type 4) (SLC40A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC40A1	SLC40A1, SLC11A3, FPN1, IREG1, HFE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemokromatozis, Tip 5, 615517, Otozomal Dominant; HFE5 (FTH1 İlişkili Aşırı Demir Yüklenmesi) (FTH1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemochromatosis, type 5, 615517, Autosomal dominant; HFE5 (FTH1-related iron overload) (FTH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTH1	FTH1, FTHL6, HFE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemoglobin H Hastalığı, Nondelesyonel, 613978 (Hemoglobin H Hastalığı) (MLPA)	Hemoglobin H disease, nondeletional, 613978 (Hemoglobin H disease) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hemoglobin H Hastalığı, Nondelesyonel, 613978 (Hemoglobin H Hastalığı) (Prenatal) (MLPA)	Hemoglobin H disease, nondeletional, 613978 (Hemoglobin H disease) (Prenatal) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemoglobin H Hastalığı, Nondelesyonel, 613978; HBH (Hemoglobin H Hastalığı) (MLPA)	Hemoglobin H disease, nondeletional, 613978; HBH (Hemoglobin H disease) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hemoglobin H Hastalığı, Nondelesyonel, 613978; HBH (Hemoglobin H Hastalığı) (Prenatal) (MLPA)	Hemoglobin H disease, nondeletional, 613978; HBH (Hemoglobin H disease) (Prenatal) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Adenilat Kinaz Eksikliğine Bağlı, 612631, Otozomal Resesif (Hemolitik Anemi, Adenilat Kinaz Eksikliğine Bağlı) (AK1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to adenylate kinase deficiency, 612631, Autosomal recessive (Hemolytic anemia due to adenylate kinase deficiency) (AK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AK1	AK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic anemia due to adenylate kinase deficiency (HAKD) [MIM:612631]: A disease characterized by hemolytic anemia and undetectable erythrocyte adenylate kinase activity. (ECO:0000269|PubMed:12649162, ECO:0000269|PubMed:2542324, ECO:0000269|PubMed:9432020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Adenilat Kinaz Eksikliğine Bağlı, 612631, Otozomal Resesif (Hemolitik Anemi, Adenilat Kinaz Eksikliğine Bağlı) (Prenatal) (AK1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to adenylate kinase deficiency, 612631, Autosomal recessive (Hemolytic anemia due to adenylate kinase deficiency) (AK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AK1	AK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic anemia due to adenylate kinase deficiency (HAKD) [MIM:612631]: A disease characterized by hemolytic anemia and undetectable erythrocyte adenylate kinase activity. (ECO:0000269|PubMed:12649162, ECO:0000269|PubMed:2542324, ECO:0000269|PubMed:9432020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, G6PD Eksikliğine Bağlı, 300908, X'e Bağlı Resesif (Sınıf 1 Glukoz-6-Fosfat Dehidrogenaz Eksikliği) (G6PD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to G6PD deficiency, 300908, X-linked recessive (Class I glucose-6-phosphate dehydrogenase deficiency) (G6PD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PD	G6PD, G6PD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, non-spherocytic hemolytic, due to G6PD deficiency (NSHA) [MIM:300908]: A disease characterized by G6PD deficiency, acute hemolytic anemia, fatigue, back pain, and jaundice. In most patients, the disease is triggered by an exogenous agent, such as some drugs, food, or infection. Increased unconjugated bilirubin, lactate dehydrogenase, and reticulocytosis are markers of the disorder. Although G6PD deficiency can be life-threatening, most patients are asymptomatic throughout their life. (ECO:0000269	PubMed:1611091). Note=The disease is caused by mutations affecting the gene represented in this entry. Deficiency of G6PD is associated with hemolytic anemia in two different situations. First, in areas in which malaria has been endemic, G6PD-deficiency alleles have reached high frequencies (1% to 50%) and deficient individuals, though essentially asymptomatic in the steady state, have a high risk of acute hemolytic attacks. Secondly, sporadic cases of G6PD deficiency occur at a very low frequencies, and they usually present a more severe phenotype. Several types of NSHA are recognized. Class-I variants are associated with severe NSHA; class-II have an activity <10% of normal; class-III have an activity of 10% to 60% of normal; class- IV have near normal activity.;	EDTA Blood Sample
Hemolitik Anemi, G6PD Eksikliğine Bağlı, 300908, X'e Bağlı Resesif (Sınıf 1 Glukoz-6-Fosfat Dehidrogenaz Eksikliği) (Prenatal) (G6PD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to G6PD deficiency, 300908, X-linked recessive (Class I glucose-6-phosphate dehydrogenase deficiency) (G6PD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	G6PD	G6PD, G6PD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, non-spherocytic hemolytic, due to G6PD deficiency (NSHA) [MIM:300908]: A disease characterized by G6PD deficiency, acute hemolytic anemia, fatigue, back pain, and jaundice. In most patients, the disease is triggered by an exogenous agent, such as some drugs, food, or infection. Increased unconjugated bilirubin, lactate dehydrogenase, and reticulocytosis are markers of the disorder. Although G6PD deficiency can be life-threatening, most patients are asymptomatic throughout their life. (ECO:0000269	PubMed:1611091). Note=The disease is caused by mutations affecting the gene represented in this entry. Deficiency of G6PD is associated with hemolytic anemia in two different situations. First, in areas in which malaria has been endemic, G6PD-deficiency alleles have reached high frequencies (1% to 50%) and deficient individuals, though essentially asymptomatic in the steady state, have a high risk of acute hemolytic attacks. Secondly, sporadic cases of G6PD deficiency occur at a very low frequencies, and they usually present a more severe phenotype. Several types of NSHA are recognized. Class-I variants are associated with severe NSHA; class-II have an activity <10% of normal; class-III have an activity of 10% to 60% of normal; class- IV have near normal activity.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Gama-Glutamil Sistein Sentetaz Eksikliğine Bağlı, 230450, Otozomal Resesif (Gama-Glutamilsistein Sentetaz Eksikliği) (GCLC gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450, Autosomal recessive (Gamma-glutamylcysteine synthetase deficiency) (GCLC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCLC	GCLC, GLCLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency (HAGGSD) [MIM:230450]: A disease characterized by hemolytic anemia, glutathione deficiency, myopathy, late-onset spinocerebellar degeneration, and peripheral neuropathy. (ECO:0000269|PubMed:10515893, ECO:0000269|PubMed:10733484, ECO:0000269|PubMed:12663448). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Gama-Glutamil Sistein Sentetaz Eksikliğine Bağlı, 230450, Otozomal Resesif (Gama-Glutamilsistein Sentetaz Eksikliği) (Prenatal) (GCLC gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450, Autosomal recessive (Gamma-glutamylcysteine synthetase deficiency) (GCLC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GCLC	GCLC, GLCLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency (HAGGSD) [MIM:230450]: A disease characterized by hemolytic anemia, glutathione deficiency, myopathy, late-onset spinocerebellar degeneration, and peripheral neuropathy. (ECO:0000269|PubMed:10515893, ECO:0000269|PubMed:10733484, ECO:0000269|PubMed:12663448). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Glutation Peroksidaz Eksikliğine Bağlı, 614164, Otozomal Resesif (GPX1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to glutathione peroxidase deficiency, 614164, Autosomal recessive (GPX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPX1	GPX1, GPXD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hemolitik Anemi, Glutation Peroksidaz Eksikliğine Bağlı, 614164, Otozomal Resesif (Prenatal) (GPX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to glutathione peroxidase deficiency, 614164, Autosomal recessive (GPX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPX1	GPX1, GPXD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Glutatyon Redüktaz Eksikliğine Bağlı (GSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to glutathione reductase deficiency (GSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSR	GSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemolitik Anemi, Glutatyon Redüktaz Eksikliğine Bağlı (GSR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to glutathione reductase deficiency (GSR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GSR	GSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Glutatyon Sentetaz Eksikliğine Bağlı, 231900, Otozomal Resesif; GSSDE (Glutatyon Sentetaz Eksikliği) (GSS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to glutathione synthetase deficiency, 231900, Autosomal recessive; GSSDE (Glutathione synthetase deficiency) (GSS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSS	GSS, GSHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glutathione synthetase deficiency of erythrocytes (GLUSYNDE) [MIM:231900]: Mild form causing hemolytic anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Glutatyon Sentetaz Eksikliğine Bağlı, 231900, Otozomal Resesif; GSSDE (Glutatyon Sentetaz Eksikliği) (Prenatal) (GSS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to glutathione synthetase deficiency, 231900, Autosomal recessive; GSSDE (Glutathione synthetase deficiency) (GSS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GSS	GSS, GSHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glutathione synthetase deficiency of erythrocytes (GLUSYNDE) [MIM:231900]: Mild form causing hemolytic anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı 235700, Otozomal Resesif (Non-Sferositik Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı) (HK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to hexokinase deficiency, 235700, Autosomal recessive (Non-spherocytic hemolytic anemia due to hexokinase deficiency) (HK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HK1	HK1, HKD, HMSNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hexokinase deficiency (HK deficiency) [MIM:235700]: Rare autosomal recessive disease with nonspherocytic hemolytic anemia as the predominant clinical feature. (ECO:0000269|PubMed:12393545, ECO:0000269|PubMed:7655856). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı 235700, Otozomal Resesif (Non-Sferositik Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı) (Prenatal) (HK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to hexokinase deficiency, 235700, Autosomal recessive (Non-spherocytic hemolytic anemia due to hexokinase deficiency) (HK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HK1	HK1, HKD, HMSNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hexokinase deficiency (HK deficiency) [MIM:235700]: Rare autosomal recessive disease with nonspherocytic hemolytic anemia as the predominant clinical feature. (ECO:0000269|PubMed:12393545, ECO:0000269|PubMed:7655856). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Fosfofruktokinaz Eksikliğine Bağlı (PFKL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to phosphofructokinase deficiency (PFKL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PFKL	PFKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemolitik Anemi, Fosfofruktokinaz Eksikliğine Bağlı (Prenatal) (PFKL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to phosphofructokinase deficiency (PFKL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PFKL	PFKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Triosefosfat İzomeraz Eksikliğine Bağlı, 615512, Otozomal Resesif; TPID (Trioz Fosfat İzomeraz Eksikliği) (TPI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512, Autosomal recessive; TPID (Triose phosphate-isomerase deficiency) (TPI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPI1	TPI1, TPID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Triosephosphate isomerase deficiency (TPID) [MIM:615512]: An autosomal recessive multisystem disorder characterized by congenital hemolytic anemia, progressive neuromuscular dysfunction, susceptibility to bacterial infection, and cardiomyopathy. (ECO:0000269|PubMed:2876430, ECO:0000269|PubMed:8503454, ECO:0000269|PubMed:8571957, ECO:0000269|PubMed:9338582, ECO:0000269|Ref.34). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Triosefosfat İzomeraz Eksikliğine Bağlı, 615512, Otozomal Resesif; TPID (Trioz Fosfat İzomeraz Eksikliği) (Prenatal) (TPI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia due to triosephosphate isomerase deficiency, 615512, Autosomal recessive; TPID (Triose phosphate-isomerase deficiency) (TPI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPI1	TPI1, TPID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Triosephosphate isomerase deficiency (TPID) [MIM:615512]: An autosomal recessive multisystem disorder characterized by congenital hemolytic anemia, progressive neuromuscular dysfunction, susceptibility to bacterial infection, and cardiomyopathy. (ECO:0000269|PubMed:2876430, ECO:0000269|PubMed:8503454, ECO:0000269|PubMed:8571957, ECO:0000269|PubMed:9338582, ECO:0000269|Ref.34). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, CD59 İlişkili,e İmmün İlişkili Polinöropati, 612300, Otozomal Resesif; HACD59 (Primer CD59 Eksikliği) (CD59 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300, Autosomal recessive; HACD59 (Primary CD59 deficiency) (CD59 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD59	CD59, MIC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic anemia, CD59-mediated, with or without polyneuropathy (HACD59) [MIM:612300]: An autosomal recessive disorder characterized by infantile onset of chronic hemolysis and a relapsing-remitting polyneuropathy, often exacerbated by infection, and manifested as hypotonia, limb muscle weakness, and hyporeflexia. (ECO:0000269|PubMed:1382994, ECO:0000269|PubMed:23149847). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, CD59 İlişkili,e İmmün İlişkili Polinöropati, 612300, Otozomal Resesif; HACD59 (Primer CD59 Eksikliği) (Prenatal) (CD59 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia, CD59-mediated, with or without immune-mediated polyneuropathy, 612300, Autosomal recessive; HACD59 (Primary CD59 deficiency) (CD59 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD59	CD59, MIC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic anemia, CD59-mediated, with or without polyneuropathy (HACD59) [MIM:612300]: An autosomal recessive disorder characterized by infantile onset of chronic hemolysis and a relapsing-remitting polyneuropathy, often exacerbated by infection, and manifested as hypotonia, limb muscle weakness, and hyporeflexia. (ECO:0000269|PubMed:1382994, ECO:0000269|PubMed:23149847). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Non-Sferositik, Glükoz Fosfat İzomeraz Eksikliğine Bağlı, 613470, Otozomal Resesif (Hemolitik Anemi, Glikofosfat İzomeraz Eksikliğine Bağlı) (GPI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470, Autosomal recessive (Hemolytic anemia due to glucophosphate isomerase deficiency) (GPI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPI	GPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic anemia, non-spherocytic, due to glucose phosphate isomerase deficiency (HA-GPID) [MIM:613470]: A form of anemia in which there is no abnormal hemoglobin or spherocytosis. It is caused by glucose phosphate isomerase deficiency. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Non-Sferositik, Glükoz Fosfat İzomeraz Eksikliğine Bağlı, 613470, Otozomal Resesif (Hemolitik Anemi, Glikofosfat İzomeraz Eksikliğine Bağlı) (Prenatal) (GPI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic anemia, nonspherocytic, due to glucose phosphate isomerase deficiency, 613470, Autosomal recessive (Hemolytic anemia due to glucophosphate isomerase deficiency) (GPI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPI	GPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic anemia, non-spherocytic, due to glucose phosphate isomerase deficiency (HA-GPID) [MIM:613470]: A form of anemia in which there is no abnormal hemoglobin or spherocytosis. It is caused by glucose phosphate isomerase deficiency. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Anemi, Nonsferositik, Heksokinaz Eksikliğine Bağlı (Non-Sferositik Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı) (HK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HEMOLYTIC ANEMIA, NONSPHEROCYTIC, DUE TO HEXOKINASE DEFICIENCY (Non-spherocytic hemolytic anemia due to hexokinase deficiency) (HK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HK1	HK1, HKD, HMSNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hexokinase deficiency (HK deficiency) [MIM:235700]: Rare autosomal recessive disease with nonspherocytic hemolytic anemia as the predominant clinical feature. (ECO:0000269|PubMed:12393545, ECO:0000269|PubMed:7655856). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hemolitik Anemi, Nonsferositik, Heksokinaz Eksikliğine Bağlı (Non-Sferositik Hemolitik Anemi, Heksokinaz Eksikliğine Bağlı) (Prenatal) (HK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	HEMOLYTIC ANEMIA, NONSPHEROCYTIC, DUE TO HEXOKINASE DEFICIENCY (Non-spherocytic hemolytic anemia due to hexokinase deficiency) (HK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HK1	HK1, HKD, HMSNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hexokinase deficiency (HK deficiency) [MIM:235700]: Rare autosomal recessive disease with nonspherocytic hemolytic anemia as the predominant clinical feature. (ECO:0000269|PubMed:12393545, ECO:0000269|PubMed:7655856). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 1, 235400, Otozomal Resesif, Otozomal Dominant (Atipik Hemolitik-Üremik Sendrom) (CFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 1, 235400, Autosomal recessive, Autosomal dominant (Atypical hemolytic-uremic syndrome) (CFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. (ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 1, 235400, Otozomal Resesif, Otozomal Dominant (Atipik Hemolitik-Üremik Sendrom) (MLPA)	Hemolytic uremic syndrome, atypical, susceptibility to, 1, 235400, Autosomal recessive, Autosomal dominant (Atypical hemolytic-uremic syndrome) (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. (ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 1, 235400, Otozomal Resesif, Otozomal Dominant (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (CFH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 1, 235400, Autosomal recessive, Autosomal dominant (Atypical hemolytic-uremic syndrome) (CFH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. (ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 1, 235400, Otozomal Resesif, Otozomal Dominant (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (MLPA)	Hemolytic uremic syndrome, atypical, susceptibility to, 1, 235400, Autosomal recessive, Autosomal dominant (Atypical hemolytic-uremic syndrome) (Prenatal) (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 2, 612922, Otozomal Resesif, Otozomal Dominant; AHUS2 (Atipik Hemolitik-Üremik Sendrom) (CD46 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 2, 612922, Autosomal recessive, Autosomal dominant; AHUS2 (Atypical hemolytic-uremic syndrome) (CD46 gen) (Sequence Analysis-All Coding Exons) (Postnatal)	CD46	MCP, CD46, AHUS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 2 (AHUS2) [MIM:612922]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:16386793, ECO:0000269|PubMed:16621965, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype. Patients with CD46 mutations seem to have an overall better prognosis compared to patients carrying CFH mutations.;	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 2, 612922, Otozomal Resesif, Otozomal Dominant; AHUS2 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (CD46 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 2, 612922, Autosomal recessive, Autosomal dominant; AHUS2 (Atypical hemolytic-uremic syndrome) (CD46 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD46	MCP, CD46, AHUS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 2 (AHUS2) [MIM:612922]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:16386793, ECO:0000269|PubMed:16621965, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype. Patients with CD46 mutations seem to have an overall better prognosis compared to patients carrying CFH mutations.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 235400, Otozomal Resesif, Otozomal Dominant; AHUS1 (Atipik Hemolitik-Üremik Sendrom) (MLPA)	Hemolytic uremic syndrome, atypical, susceptibility to, 235400, Autosomal recessive, Autosomal dominant; AHUS1 (Atypical hemolytic-uremic syndrome) (MLPA)	CFHR1	CFHR1, FHR1, HFL1, CFHL1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 235400, Otozomal Resesif, Otozomal Dominant; AHUS1 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (MLPA)	Hemolytic uremic syndrome, atypical, susceptibility to, 235400, Autosomal recessive, Autosomal dominant; AHUS1 (Atypical hemolytic-uremic syndrome) (Prenatal) (MLPA)	CFHR1	CFHR1, FHR1, HFL1, CFHL1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 3, 612923, Otozomal Dominant; AHUS3 (Atipik Hemolitik-Üremik Sendrom) (CFI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 3, 612923, Autosomal dominant; AHUS3 (Atypical hemolytic-uremic syndrome) (CFI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFI	CFI, FI, AHUS3, ARMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement factor I deficiency (CFI deficiency) [MIM:610984]: Autosomal recessive condition associated with a propensity to pyogenic infections. {ECO:0000269|PubMed:12562389, ECO:0000269|PubMed:17018561, ECO:0000269|PubMed:8613545}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 3, 612923, Otozomal Dominant; AHUS3 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (CFI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 3, 612923, Autosomal dominant; AHUS3 (Atypical hemolytic-uremic syndrome) (CFI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFI	CFI, FI, AHUS3, ARMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Complement factor I deficiency (CFI deficiency) [MIM:610984]: Autosomal recessive condition associated with a propensity to pyogenic infections. {ECO:0000269|PubMed:12562389, ECO:0000269|PubMed:17018561, ECO:0000269|PubMed:8613545}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 4, 612924, Otozomal Dominant; AHUS4 (Atipik Hemolitik-Üremik Sendrom) (CFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 4, 612924, Autosomal dominant; AHUS4 (Atypical hemolytic-uremic syndrome) (CFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFB	CFB, BF, GBG, AHUS4, ARMD14, CFBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 4 (AHUS4) [MIM:612924]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:17182750, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to the development of atypical hemolytic uremic syndrome can be conferred by mutations in various components of or regulatory factors in the complement cascade system. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 4, 612924, Otozomal Dominant; AHUS4 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (CFB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 4, 612924, Autosomal dominant; AHUS4 (Atypical hemolytic-uremic syndrome) (CFB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFB	CFB, BF, GBG, AHUS4, ARMD14, CFBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 4 (AHUS4) [MIM:612924]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:17182750, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to the development of atypical hemolytic uremic syndrome can be conferred by mutations in various components of or regulatory factors in the complement cascade system. Other genes may play a role in modifying the phenotype.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 5, 612925, Otozomal Dominant; AHUS5 (Atipik Hemolitik-Üremik Sendrom) (C3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 5, 612925, Autosomal dominant; AHUS5 (Atypical hemolytic-uremic syndrome) (C3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C3	C3, ARMD9, AHUS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 5, 612925, Otozomal Dominant; AHUS5 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (C3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 5, 612925, Autosomal dominant; AHUS5 (Atypical hemolytic-uremic syndrome) (C3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C3	C3, ARMD9, AHUS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 6, 612926, Otozomal Dominant; AHUS6 (Atipik Hemolitik-Üremik Sendrom) (THBD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 6, 612926, Autosomal dominant; AHUS6 (Atypical hemolytic-uremic syndrome) (THBD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THBD	THBD, THRM, AHUS6, THPH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 6 (AHUS6) [MIM:612926]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:19625716, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.;	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 6, 612926, Otozomal Dominant; AHUS6 (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (THBD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 6, 612926, Autosomal dominant; AHUS6 (Atypical hemolytic-uremic syndrome) (THBD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	THBD	THBD, THRM, AHUS6, THPH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 6 (AHUS6) [MIM:612926]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:19625716, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 7, 615008, Otozomal Resesif (Atipik Hemolitik-Üremik Sendrom) (DGKE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 7, 615008, Autosomal recessive (Atypical hemolytic-uremic syndrome) (DGKE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGKE	DGKE, NPHS7, AHUS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 7 (AHUS7) [MIM:615008]: An atypical form of hemolytic uremic syndrome characterized by acute onset in the first year of life of microangiopathic hemolytic anemia, thrombocytopenia, and renal failure. After the acute episode, most patients develop chronic renal insufficiency. Unlike other genetic forms of aHUS, AHUS7 is not related to abnormal activation of the complement system. {ECO:0000269	PubMed:23542698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Üremik Sendrom, Atipik, Yatkınlık, 7, 615008, Otozomal Resesif (Atipik Hemolitik-Üremik Sendrom) (Prenatal) (DGKE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemolytic uremic syndrome, atypical, susceptibility to, 7, 615008, Autosomal recessive (Atypical hemolytic-uremic syndrome) (DGKE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGKE	DGKE, NPHS7, AHUS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 7 (AHUS7) [MIM:615008]: An atypical form of hemolytic uremic syndrome characterized by acute onset in the first year of life of microangiopathic hemolytic anemia, thrombocytopenia, and renal failure. After the acute episode, most patients develop chronic renal insufficiency. Unlike other genetic forms of aHUS, AHUS7 is not related to abnormal activation of the complement system. {ECO:0000269	PubMed:23542698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel (Panel) (UNC13D 17q25.1, PRF1 10q22.1, STX11 6q24.2) (MLPA)	Hemophagocytic lymphohistiocytosis, familial (Panel) (UNC13D 17q25.1, PRF1 10q22.1, STX11 6q24.2) (MLPA)	UNC13D 17q25.1, PRF1 10q22.1, STX11 6q24.2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 2, 603553, Otozomal Resesif; FHL2 (Ailesel Hemofagositik Lenfohistiyositoz) (PRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophagocytic lymphohistiocytosis, familial, 2, 603553, Autosomal recessive; FHL2 (Familial hemophagocytic lymphohistiocytosis) (PRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRF1	PRF1, HPLH2, FLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. {ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Hemofagositik Lenfohistiyositozis, Ailesel, 2, 603553, Otozomal Resesif; FHL2 (Ailesel Hemofagositik Lenfohistiyositoz) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 2, 603553, Autosomal recessive; FHL2 (Familial hemophagocytic lymphohistiocytosis) (MLPA)	PRF1	PRF1, HPLH2, FLH2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 2, 603553, Otozomal Resesif; FHL2 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (PRF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophagocytic lymphohistiocytosis, familial, 2, 603553, Autosomal recessive; FHL2 (Familial hemophagocytic lymphohistiocytosis) (PRF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRF1	PRF1, HPLH2, FLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 2, 603553, Otozomal Resesif; FHL2 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 2, 603553, Autosomal recessive; FHL2 (Familial hemophagocytic lymphohistiocytosis) (Prenatal) (MLPA)	PRF1	PRF1, HPLH2, FLH2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 3, 608898; FHL3 (Ailesel Hemofagositik Lenfohistiyositoz) (UNC13D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophagocytic lymphohistiocytosis, familial, 3, 608898; FHL3 (Familial hemophagocytic lymphohistiocytosis) (UNC13D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNC13D	UNC13D, MUNC13-4, HPLH3, HLH3, FHL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 3 (FHL3) [MIM:608898]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:14622600). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 3, 608898; FHL3 (Ailesel Hemofagositik Lenfohistiyositoz) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 3, 608898; FHL3 (Familial hemophagocytic lymphohistiocytosis) (MLPA)	UNC13D	UNC13D, MUNC13-4, HPLH3, HLH3, FHL3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 3 (FHL3) [MIM:608898]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:14622600). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 3, 608898; FHL3 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (UNC13D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophagocytic lymphohistiocytosis, familial, 3, 608898; FHL3 (Familial hemophagocytic lymphohistiocytosis) (UNC13D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UNC13D	UNC13D, MUNC13-4, HPLH3, HLH3, FHL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 3 (FHL3) [MIM:608898]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:14622600). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 3, 608898; FHL3 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 3, 608898; FHL3 (Familial hemophagocytic lymphohistiocytosis) (Prenatal) (MLPA)	UNC13D	UNC13D, MUNC13-4, HPLH3, HLH3, FHL3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 3 (FHL3) [MIM:608898]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:14622600). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 4, 603552, Otozomal Resesif; FHL4 (Ailesel Hemofagositik Lenfohistiyositoz) (STX11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophagocytic lymphohistiocytosis, familial, 4, 603552, Autosomal recessive; FHL4 (Familial hemophagocytic lymphohistiocytosis) (STX11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STX11	STX11, FHL4, HPLH4, HLH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 4 (FHL4) [MIM:603552]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:15703195). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 4, 603552, Otozomal Resesif; FHL4 (Ailesel Hemofagositik Lenfohistiyositoz) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 4, 603552, Autosomal recessive; FHL4 (Familial hemophagocytic lymphohistiocytosis) (MLPA)	STX11	STX11, FHL4, HPLH4, HLH4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 4 (FHL4) [MIM:603552]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:15703195). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 4, 603552, Otozomal Resesif; FHL4 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (STX11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophagocytic lymphohistiocytosis, familial, 4, 603552, Autosomal recessive; FHL4 (Familial hemophagocytic lymphohistiocytosis) (STX11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STX11	STX11, FHL4, HPLH4, HLH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 4 (FHL4) [MIM:603552]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:15703195). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 4, 603552, Otozomal Resesif; FHL4 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (MLPA)	Hemophagocytic lymphohistiocytosis, familial, 4, 603552, Autosomal recessive; FHL4 (Familial hemophagocytic lymphohistiocytosis) (Prenatal) (MLPA)	STX11	STX11, FHL4, HPLH4, HLH4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 4 (FHL4) [MIM:603552]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. (ECO:0000269	PubMed:15703195). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 5, 613101; FHL5 (Ailesel Hemofagositik Lenfohistiyositoz) (STXBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophagocytic lymphohistiocytosis, familial, 5, 613101; FHL5 (Familial hemophagocytic lymphohistiocytosis) (STXBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STXBP2	STXBP2, UNC18B, FHL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemofagositik Lenfohistiyositozis, Ailesel, 5, 613101; FHL5 (Ailesel Hemofagositik Lenfohistiyositoz) (Prenatal) (STXBP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophagocytic lymphohistiocytosis, familial, 5, 613101; FHL5 (Familial hemophagocytic lymphohistiocytosis) (STXBP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STXBP2	STXBP2, UNC18B, FHL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofili A, 306700, X'e Bağlı; HEMA (Hemofili A) (F8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophilia A, 306700, X-linked recessive; HEMA (Hemophilia A) (F8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F8	F8, F8C, HEMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemophilia A (HEMA) [MIM:306700]: A disorder of blood coagulation characterized by a permanent tendency to hemorrhage. About 50% of patients have severe hemophilia, resulting in frequent spontaneous bleeding into joints, muscles and internal organs. Less severe forms are characterized by bleeding after trauma or surgery. (ECO:0000269|PubMed:10215414, ECO:0000269|PubMed:10338101, ECO:0000269|PubMed:10404764, ECO:0000269|PubMed:10408784, ECO:0000269|PubMed:10554831, ECO:0000269|PubMed:10612839, ECO:0000269|PubMed:10691849, ECO:0000269|PubMed:10800171, ECO:0000269|PubMed:10886198, ECO:0000269|PubMed:10896236, ECO:0000269|PubMed:10910910, ECO:0000269|PubMed:10910913, ECO:0000269|PubMed:11298607, ECO:0000269|PubMed:11341489, ECO:0000269|PubMed:11410838, ECO:0000269|PubMed:11442643, ECO:0000269|PubMed:11442647, ECO:0000269|PubMed:11554935, ECO:0000269|PubMed:11748850, ECO:0000269|PubMed:11857744, ECO:0000269|PubMed:11858487, ECO:0000269|PubMed:12195713, ECO:0000269|PubMed:12199686, ECO:0000269|PubMed:12203998, ECO:0000269|PubMed:12325022, ECO:0000269|PubMed:12351418,	EDTA Blood Sample

Hemofili A, 306700, X'e Bağlı; HEMA (Hemofili A) (MLPA)	Hemophilia A, 306700, X-linked recessive; HEMA (Hemophilia A) (MLPA)	F8	F8, F8C, HEMA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hemophilia A (HEMA) [MIM:306700]: A disorder of blood coagulation characterized by a permanent tendency to hemorrhage. About 50% of patients have severe hemophilia resulting in frequent spontaneous bleeding into joints, muscles and internal organs. Less severe forms are characterized by bleeding after trauma or surgery. {ECO:0000269|PubMed:10215414, ECO:0000269|PubMed:10338101, ECO:0000269|PubMed:10404764, ECO:0000269|PubMed:10408784, ECO:0000269|PubMed:10554831, ECO:0000269|PubMed:10612839, ECO:0000269|PubMed:10691849, ECO:0000269|PubMed:10800171, ECO:0000269|PubMed:10886198, ECO:0000269|PubMed:10896236, ECO:0000269|PubMed:10910910, ECO:0000269|PubMed:10910913, ECO:0000269|PubMed:11298607, ECO:0000269|PubMed:11341489, ECO:0000269|PubMed:11410838, ECO:0000269|PubMed:11442643, ECO:0000269|PubMed:11442647, ECO:0000269|PubMed:11554935, ECO:0000269|PubMed:11748850, ECO:0000269|PubMed:11857744, ECO:0000269|PubMed:11858487, ECO:0000269|PubMed:12195713, ECO:0000269|PubMed:12199686, ECO:0000269|PubMed:12203998, ECO:0000269|PubMed:12325022, ECO:0000269|PubMed:12351418	EDTA Blood Sample
Hemofili A, 306700, X'e Bağlı; HEMA (Hemofili A) (Prenatal) (F8 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophilia A, 306700, X-linked recessive; HEMA (Hemophilia A) (F8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	F8	F8, F8C, HEMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemophilia A (HEMA) [MIM:306700]: A disorder of blood coagulation characterized by a permanent tendency to hemorrhage. About 50% of patients have severe hemophilia resulting in frequent spontaneous bleeding into joints, muscles and internal organs. Less severe forms are characterized by bleeding after trauma or surgery. {ECO:0000269|PubMed:10215414, ECO:0000269|PubMed:10338101, ECO:0000269|PubMed:10404764, ECO:0000269|PubMed:10408784, ECO:0000269|PubMed:10554831, ECO:0000269|PubMed:10612839, ECO:0000269|PubMed:10691849, ECO:0000269|PubMed:10800171, ECO:0000269|PubMed:10886198, ECO:0000269|PubMed:10896236, ECO:0000269|PubMed:10910910, ECO:0000269|PubMed:10910913, ECO:0000269|PubMed:11298607, ECO:0000269|PubMed:11341489, ECO:0000269|PubMed:11410838, ECO:0000269|PubMed:11442643, ECO:0000269|PubMed:11442647, ECO:0000269|PubMed:11554935, ECO:0000269|PubMed:11748850, ECO:0000269|PubMed:11857744, ECO:0000269|PubMed:11858487, ECO:0000269|PubMed:12195713, ECO:0000269|PubMed:12199686, ECO:0000269|PubMed:12203998, ECO:0000269|PubMed:12325022, ECO:0000269|PubMed:12351418	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofili A, 306700, X'e Bağlı; HEMA (Hemofili A) (Prenatal) (MLPA)	Hemophilia A, 306700, X-linked recessive; HEMA (Hemophilia A) (Prenatal) (MLPA)	F8	F8, F8C, HEMA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemophilia A (HEMA) [MIM:306700]: A disorder of blood coagulation characterized by a permanent tendency to hemorrhage. About 50% of patients have severe hemophilia resulting in frequent spontaneous bleeding into joints, muscles and internal organs. Less severe forms are characterized by bleeding after trauma or surgery. {ECO:0000269|PubMed:10215414, ECO:0000269|PubMed:10338101, ECO:0000269|PubMed:10404764, ECO:0000269|PubMed:10408784, ECO:0000269|PubMed:10554831, ECO:0000269|PubMed:10612839, ECO:0000269|PubMed:10691849, ECO:0000269|PubMed:10800171, ECO:0000269|PubMed:10886198, ECO:0000269|PubMed:10896236, ECO:0000269|PubMed:10910910, ECO:0000269|PubMed:10910913, ECO:0000269|PubMed:11298607, ECO:0000269|PubMed:11341489, ECO:0000269|PubMed:11410838, ECO:0000269|PubMed:11442643, ECO:0000269|PubMed:11442647, ECO:0000269|PubMed:11554935, ECO:0000269|PubMed:11748850, ECO:0000269|PubMed:11857744, ECO:0000269|PubMed:11858487, ECO:0000269|PubMed:12195713, ECO:0000269|PubMed:12199686, ECO:0000269|PubMed:12203998, ECO:0000269|PubMed:12325022, ECO:0000269|PubMed:12351418	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofili B, 306900, X'e Bağlı Resesif; HEMB (Hemofili B) (F9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hemophilia B, 306900, X-linked recessive; HEMB (Hemophilia B) (F9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F9	F9, HEMB, THPHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemofili B, 306900, X'e Bağlı Resesif; HEMB (Hemofili B) (MLPA)	Hemophilia B, 306900, X-linked recessive; HEMB (Hemophilia B) (MLPA)	F9	F9, HEMB, THPHB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hemofili B, 306900, X'e Bağlı Resesif; HEMB (Hemofili B) (Prenatal) (F9 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hemophilia B, 306900, X-linked recessive; HEMB (Hemophilia B) (F9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	F9	F9, HEMB, THPHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemofili B, 306900, X'e Bağlı Resesif; HEMB (Hemofili B) (MLPA)	Hemophilia B, 306900, X-linked recessive; HEMB (Hemophilia B) (MLPA)	F9	F9, HEMB, THPHB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kanama, İntraserebral, Yatkınlık, 614519 (COL4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemorrhage, intracerebral, susceptibility to, 614519 (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kanama, İntraserebral, Yatkınlık, 614519; ICH (COL4A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemorrhage, intracerebral, susceptibility to, 614519; ICH (COL4A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A2	COL4A2, POREN2, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hemorajik Beyin Yıkımı, Subependimal Kalsifikasyon ve Katarakt, 613730, Otozomal Resesif; HDBSCC (Porensefali-Mikrosefali-Bilateral Konjenital Katarakt Sendromu) (JAM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730, Autosomal recessive; HDBSCC (Porencephaly-microcephaly-bilateral congenital cataract syndrome) (JAM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAM3	JAM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemorrhagic destruction of the brain with subependymal calcification and cataracts (HDBSCC) [MIM:613730]: A syndrome characterized by congenital cataracts and severe brain abnormalities apparently resulting from hemorrhagic destruction of the brain parenchyma, including the cerebral white matter and basal ganglia. Patients manifest profound developmental delay, and other neurologic features included seizures, spasticity, and hyperreflexia. The clinical course is very severe resulting in death in infancy. Brain imaging shows multifocal intraparenchymal hemorrhage with associated liquefaction and massive cystic degeneration, and calcification in the subependymal region and in brain tissue. {ECO:0000269|PubMed:21109224, ECO:0000269|PubMed:23255094}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemorajik Beyin Yıkımı, Subependimal Kalsifikasyon ve Katarakt, 613730, Otozomal Resesif; HDBSCC (Porensefali-Mikrosefali-Bilateral Konjenital Katarakt Sendromu) (Prenatal) (JAM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hemorrhagic destruction of the brain, subependymal calcification, and cataracts, 613730, Autosomal recessive; HDBSCC (Porencephaly-microcephaly-bilateral congenital cataract syndrome) (JAM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JAM3	JAM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemorrhagic destruction of the brain with subependymal calcification and cataracts (HDBSCC) [MIM:613730]: A syndrome characterized by congenital cataracts and severe brain abnormalities apparently resulting from hemorrhagic destruction of the brain parenchyma, including the cerebral white matter and basal ganglia. Patients manifest profound developmental delay, and other neurologic features included seizures, spasticity, and hyperreflexia. The clinical course is very severe resulting in death in infancy. Brain imaging shows multifocal intraparenchymal hemorrhage with associated liquefaction and massive cystic degeneration, and calcification in the subependymal region and in brain tissue. {ECO:0000269|PubMed:21109224, ECO:0000269|PubMed:23255094}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hemorajik Diyatez, Antitrombin Pittsburgh Bağlı, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (SERPINA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (SERPINA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA1	SERPINA1, PI, AAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemorajik Diyatez, Antitrombin Pittsburgh Bağlı, 613490, Otozomal Resesif (Alfa-1-Antitripsin Eksikliği) (MLPA)	Hemorrhagic diathesis due to antithrombin Pittsburgh, 613490, Autosomal recessive (Alpha-1-antitrypsin deficiency) (MLPA)	SERPINA1	SERPINA1, PI, AAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (A1ATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemosiderosis, Sistemik, Aceruloplazminemiye Bağlı Olarak, 604290, Otozomal Resesif (Aseruloplazminemi) (CP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hemosiderosis, systemic, due to aceruloplasminemia, 604290, Autosomal recessive (Aceruloplasminemia) (CP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CP	CP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hennekam Lenfanjiektazi Lenfödem Sendromu 1, 235510, Otozomal Resesif; HKLLS1 (Hennekam Sendromu) (CCBE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510, Autosomal recessive; HKLLS1 (Hennekam syndrome) (CCBE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCBE1	CCBE1, KIAA1983, HKLLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hennekam Lenfanjiektazi Lenfdem Sendromu 1, 235510, Otozomal Resesif; HKLLS1 (Hennekam Sendromu) (Prenatal) (CCBE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510, Autosomal recessive; HKLLS1 (Hennekam syndrome) (CCBE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCBE1	CCBE1, KIAA1983, HKLLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hennekam Lenfanjiektazi Lenfdem Sendromu 2, 616006, Otozomal Resesif; HKLLS2 (Hennekam Sendromu) (FAT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006, Autosomal recessive; HKLLS2 (Hennekam syndrome) (FAT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAT4	FAT4, VMLDS2, HKLLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hennekam Lenfanjiektazi Lenfdem Sendromu 2, 616006, Otozomal Resesif; HKLLS2 (Hennekam Sendromu) (Prenatal) (FAT4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hennekam lymphangiectasia-lymphedema syndrome 2, 616006, Autosomal recessive; HKLLS2 (Hennekam syndrome) (FAT4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAT4	FAT4, VMLDS2, HKLLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hepatik Adenom, Somatik, 142330 (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatic adenoma, somatic, 142330 (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hepatic adenomas familial (HEPAF) [MIM:142330]: Rare benign liver tumors of presumable epithelial origin that develop in an otherwise normal liver. Hepatic adenomas may be single or multiple. They consist of sheets of well-differentiated hepatocytes that contain fat and glycogen and can produce bile. Bile ducts or portal areas are absent. Kupffer cells, if present, are reduced in number and are non-functional. Conditions associated with adenomas are insulin-dependent diabetes mellitus and glycogen storage diseases (types 1 and 3). Note=The disease is caused by mutations affecting the gene represented in this entry. Bi-allelic inactivation of HNF1A, whether sporadic or associated with MODY3, may be an early step in the development of some hepatocellular carcinomas.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatik Lipaz Eksikliği, 614025, Otozomal Resesif (Hiperlipidemi, Hepatik Trigliserid Lipaz Eksikliğine Bağlı) (LIPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatic lipase deficiency, 614025, Autosomal recessive (Hyperlipidemia due to hepatic triglyceride lipase deficiency) (LIPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPC	LIPC, HL, LIPH, HDLCQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic lipase deficiency (HL deficiency) [MIM:614025]: A disorder characterized by elevated levels of beta-migrating very low density lipoproteins, and abnormally triglyceride-rich low and high density lipoproteins. {ECO:0000269|PubMed:10660332, ECO:0000269|PubMed:13019391}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hepatik Venookluisif Hastalık,İmmün Yetmezlik, 235550, Otozomal Resesif; VODI (Hepatik Veno-Oklüzif Hastalık-İmmun Yetersizlik Sendromu) (SP110 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatic venocclusive disease with immunodeficiency, 235550, Autosomal recessive; VODI (Hepatic veno-occlusive disease-immunodeficiency syndrome) (SP110 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SP110	SP110, IFI41, IFI75, VODI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic venocclusive disease with immunodeficiency (VODI) [MIM:235550]: Autosomal recessive primary immunodeficiency associated with hepatic vascular occlusion and fibrosis. The immunodeficiency is characterized by severe hypogammaglobulinemia, combined T and B-cell immunodeficiency, absent lymph node germinal centers, and absent tissue plasma cells. {ECO:0000269|PubMed:16648851}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hepatit C Virüsü, Yatkınlık, 609532 (Hepatit C Antiviral Tedaviye Yanıt) (PTPRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitic C virus, susceptibility to, 609532 (Response to antiviral treatment in hepatitis C) (PTPRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRC	PTPRC, CD45, LCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit B Virüsü Enfeksiyonu, Yatkınlık, 610424 (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis B virus infection, susceptibility to, 610424 (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit B Virüsü, Yatkınlık, 610424 (IFNAR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis B virus, susceptibility to, 610424 (IFNAR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNAR2	IFNAR2, IMD45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit B Virüsü, Yatkınlık, 610424 (IL10RB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis B virus, susceptibility to, 610424 (IL10RB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10RB	CRFB4, IBD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit C Virüsü Enfeksiyonu, Tedaviye Yanıt, 609532 (Hepatit C Antiviral Tedaviye Yanıt) (IFNL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis C virus infection, response to therapy of, 609532 (Response to antiviral treatment in hepatitis C) (IFNL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNL3	IFNL3, IL28B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit C Virüsü, Direnç, 609532 (Hepatit C Antiviral Tedaviye Yanıt) (CCR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis C virus, resistance to, 609532 (Response to antiviral treatment in hepatitis C) (CCR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCR5	CCR5, CMKBR5, CCCKR5, IDDM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatit C Virüsü, ve Tedaviye Yanıt, 609532 (Hepatit C Antiviral Tedaviye Yanıt) (IFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatitis C virus, response to therapy of, 609532 (Response to antiviral treatment in hepatitis C) (IFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hepatoblastom, Somatik, 114550 (Hepatoblastom) (APC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatoblastoma, somatic, 114550 (Hepatoblastoma) (APC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC	APC, GS, FPC, BTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hepatocellular carcinoma (HCC) [MIM:114550]: A primary malignant neoplasm of epithelial liver cells. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. Note=The gene represented in this entry may be involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Kanser, Somatik, 114550 (PDGFRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular cancer, somatic, 114550 (PDGFRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRL	PDGFRL, PDGRL, PRLTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinoma, 114550, Somatik Mutasyon (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, 114550, Somatic mutation (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinoma, 114550, Somatik Mutasyon (MLPA)	Hepatocellular carcinoma, 114550, Somatic mutation (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		20	60	.	.	30	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Çocukluk Çağı, Somatik, 114550 (MET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, childhood type, somatic, 114550 (MET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MET	MET, DFNB97, OSFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hepatocellular carcinoma (HCC) [MIM:114550]: A primary malignant neoplasm of epithelial liver cells. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. {ECO:0000269|PubMed:9927037}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Somatik, 114550 (Hepatoblastom) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, somatic, 114550 (Hepatoblastoma) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Somatik, 114550 (Hepatoblastom) (IGF2R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, somatic, 114550 (Hepatoblastoma) (IGF2R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGF2R	IGF2R, MPRI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Somatik, 114550 (Hepatoblastom) (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, somatic, 114550 (Hepatoblastoma) (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Somatik, 114550 (Hepatoblastom) (CASP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, somatic, 114550 (Hepatoblastoma) (CASP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP8	CASP8, MCH5, ALPS2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hepatoselüler Karsinom, Somatik, 114550 (Hepatoblastom) (AXIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hepatocellular carcinoma, somatic, 114550 (Hepatoblastoma) (AXIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AXIN1	AXIN1, AXIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hepatocellular carcinoma (HCC) [MIM:114550]: A primary malignant neoplasm of epithelial liver cells. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. {ECO:0000269|PubMed:17010426}. Note=The gene represented in this entry is involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Her2 (ERBB2-NEU) (FISH)	HER2 Amplification (ERBB2 or NEU gene) (FISH)	17q12	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Herediter hemorajik telenjiektazi/ İdiopatik ve / veya ailesel pulmoner arteriyel hipertansiyon (ENG 9q34.11, ACVRL1 12q13.3, BMPR2 2q33.1-2) (MLPA)	Hereditary hemorrhagic telangiectasia / idiopathic and / or familial pulmonary arterial hypertension (ENG 9q34.11, ACVRL1 12q13.3, BMPR2 2q33.1-2) (MLPA)	ENG 9q34.11, ACVRL1 12q13.3, BMPR2 2q33.1-2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kalıtsal Motor ve Duyusal Nöropati, 6A, 601152, Otozomal Dominant; HMSN6A (Kalıtsal Motor ve Duyusal Nöropati Tip 6) (MFN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hereditary motor and sensory neuropathy VIA, 601152, Autosomal dominant; HMSN6A (Hereditary motor and sensory neuropathy type 6) (MFN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary motor and sensory, 6A (HMSN6A) [MIM:601152]: An autosomal dominant neurologic disorder characterized by optic atrophy and peripheral sensorimotor neuropathy manifesting as axonal Charcot-Marie-Tooth disease. Charcot-Marie-Tooth disease is a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. It is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies and primary peripheral axonal neuropathies. Peripheral axonal neuropathies are characterized by signs of axonal regeneration in the absence of obvious myelin alterations, and normal or slightly reduced nerve conduction velocities. {ECO:0000269|PubMed:16437557}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kalıtsal Motor ve Duyusal Nöropati, 6A, 601152, Otozomal Dominant; HMSN6A (Kalıtsal Motor ve Duyusal Nöropati Tip 6) (MLPA)	Hereditary motor and sensory neuropathy VIA, 601152, Autosomal dominant; HMSN6A (Hereditary motor and sensory neuropathy type 6) (MLPA)	MFN2	MFN2, KIAA0214, CMT2A2A, HMSN6A, CMT2A2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neuropathy, hereditary motor and sensory, 6A (HMSN6A) [MIM:601152]: An autosomal dominant neurologic disorder characterized by optic atrophy and peripheral sensorimotor neuropathy manifesting as axonal Charcot-Marie-Tooth disease. Charcot-Marie-Tooth disease is a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. It is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies and primary peripheral axonal neuropathies. Peripheral axonal neuropathies are characterized by signs of axonal regeneration in the absence of obvious myelin alterations, and normal or slightly reduced nerve conduction velocities. {ECO:0000269|PubMed:16437557}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kalıtsal Motor ve Duyusal Nöropati, Okinawa Tipi, 604484, Otozomal Dominant; HMSN6O (Kalıtsal Motor ve Duyusal Nöropati, Okinawa Tipi) (TFG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hereditary motor and sensory neuropathy, Okinawa type, 604484, Autosomal dominant; HMSN6O (Hereditary motor and sensory neuropathy, Okinawa type) (TFG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFG	TFG, HMSNP, SPG57	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary motor and sensory, Okinawa type (HMSN6O) [MIM:604484]: A neurodegenerative disorder characterized by young adult onset of proximal muscle weakness and atrophy, muscle cramps, and fasciculations, with later onset of distal sensory impairment. The disorder is slowly progressive and clinically resembles amyotrophic lateral sclerosis. {ECO:0000269|PubMed:22883144}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kalıtsal Motor ve Duyusal Nöropati, Tip 2c, 606071, Otozomal Dominant; HMSN2C (Charcot-Marie-Tooth Hastalığı Tip 2C) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hereditary motor and sensory neuropathy, type IIc, 606071, Autosomal dominant; HMSN2C (Autosomal dominant Charcot-Marie-Tooth disease type 2C) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Herediter Fetal Hemoglobin Persistansı, 141749, Otozomal Dominant (Fetal Hemoglobin Persistansı-Orak Hücre Hastalığı Sendromu) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hereditary persistence of fetal hemoglobin, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Herediter Fetal Hemoglobin Persistansı, 141749, Otozomal Dominant (Fetal Hemoglobin Persistansı-Orak Hücre Hastalığı Sendromu) (MLPA)	Hereditary persistence of fetal hemoglobin, 141749, Autosomal dominant (Hereditary persistence of fetal hemoglobin-sickle cell disease syndrome) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Hermansky-Pudlak Sendromu 1, 203300, Otozomal Resesif; HPS1 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis) (HPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 1, 203300, Autosomal recessive; HPS1 (Hermansky-Pudlak syndrome with pulmonary fibrosis) (HPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPS1	HPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 1, 203300, Otozomal Resesif; HPS1 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis) (Prenatal) (HPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 1, 203300, Autosomal recessive; HPS1 (Hermansky-Pudlak syndrome with pulmonary fibrosis) (HPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPS1	HPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 10, 617050, Otozomal Resesif; HPS10 (AP3D1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 10, 617050, Autosomal recessive; HPS10 (AP3D1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP3D1	AP3D1, HPS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 10, 617050, Otozomal Resesif; HPS10 (Prenatal) (AP3D1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 10, 617050, Autosomal recessive; HPS10 (AP3D1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP3D1	AP3D1, HPS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 2, 608233, Otozomal Resesif; HPS2 (Hermansky-Pudlak Sendromu-Nötropeni) (AP3B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 2, 608233, Autosomal recessive; HPS2 (Hermansky-Pudlak syndrome with neutropenia) (AP3B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP3B1	AP3B1, ADTB3A, HPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hermansky-Pudlak syndrome 2 (HPS2) [MIM:608233]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. HPS2 differs from the other forms of HPS in that it includes immunodeficiency in its phenotype and patients with HPS2 have an increased susceptibility to infections. {ECO:0000269	PubMed:10024875}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hermansky-Pudlak Sendromu 2, 608233, Otozomal Resesif; HPS2 (Hermansky-Pudlak Sendromu-Nötropeni) (Prenatal) (AP3B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 2, 608233, Autosomal recessive; HPS2 (Hermansky-Pudlak syndrome with neutropenia) (AP3B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP3B1	AP3B1, ADTB3A, HPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hermansky-Pudlak syndrome 2 (HPS2) [MIM:608233]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. HPS2 differs from the other forms of HPS in that it includes immunodeficiency in its phenotype and patients with HPS2 have an increased susceptibility to infections. {ECO:0000269	PubMed:10024875}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 3, 614072; HPS3 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (HPS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 3, 614072; HPS3 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPS3	HPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 3, 614072; HPS3 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (Prenatal) (HPS3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 3, 614072; HPS3 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPS3	HPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 4, 614073; HPS4 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis) (HPS4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 4, 614073; HPS4 (Hermansky-Pudlak syndrome with pulmonary fibrosis) (HPS4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPS4	HPS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hermansky-Pudlak syndrome 4 (HPS4) [MIM:614073]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:11836498}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hermansky-Pudlak Sendromu 4, 614073; HPS4 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis) (Prenatal) (HPS4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 4, 614073; HPS4 (Hermansky-Pudlak syndrome with pulmonary fibrosis) (HPS4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPS4	HPS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hermansky-Pudlak syndrome 4 (HPS4) [MIM:614073]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:11836498}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 5, 614074; HPS5 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (HPS5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 5, 614074; HPS5 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPS5	HPS5, RU2, KIAA1017	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 5, 614074; HPS5 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (Prenatal) (HPS5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 5, 614074; HPS5 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPS5	HPS5, RU2, KIAA1017	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 6, 614075; HPS6 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (HPS6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 6, 614075; HPS6 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPS6	HPS6, RU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 6, 614075; HPS6 (Hermansky-Pudlak Sendromu-Pulmoner Fibrozis Olmayan) (Prenatal) (HPS6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 6, 614075; HPS6 (Hermansky-Pudlak syndrome without pulmonary fibrosis) (HPS6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPS6	HPS6, RU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 7, 614076, Otozomal Resesif; HPS7 (Hermansky-Pudlak Sendromu Tip 7) (DTNBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 7, 614076, Autosomal recessive; HPS7 (Hermansky-Pudlak syndrome type 7) (DTNBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DTNBP1	DTNBP1, HPS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hermansky-Pudlak syndrome 7 (HPS7) [MIM:614076]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:12923531}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hermansky-Pudlak Sendromu 7, 614076, Otozomal Resesif; HPS7 (Hermansky-Pudlak Sendromu Tip 7) (Prenatal) (DTNBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 7, 614076, Autosomal recessive; HPS7 (Hermansky-Pudlak syndrome type 7) (DTNBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DTNBP1	DTNBP1, HPS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hermansky-Pudlak syndrome 7 (HPS7) [MIM:614076]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:12923531}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 8, 614077; HPS8 (Hermansky-Pudlak Sendromu Tip 8) (BLOC1S3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-Pudlak syndrome 8, 614077; HPS8 (Hermansky-Pudlak syndrome type 8) (BLOC1S3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLOC1S3	BLOC1S3, BLOS3, HPS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hermansky-Pudlak syndrome 8 (HPS8) [MIM:614077]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:16385460}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hermansky-Pudlak Sendromu 8, 614077; HPS8 (Hermansky-Pudlak Sendromu Tip 8) (Prenatal) (BLOC1S3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-Pudlak syndrome 8, 614077; HPS8 (Hermansky-Pudlak syndrome type 8) (BLOC1S3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BLOC1S3	BLOC1S3, BLOS3, HPS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hermansky-Pudlak syndrome 8 (HPS8) [MIM:614077]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:16385460}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hermansky-Pudlak Sendromu 9, 614171, Otozomal Resesif; HPS9 (Hermansky-Pudlak Sendromu Tip 9) (BLOC1S6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hermansky-pudlak syndrome 9, 614171, Autosomal recessive; HPS9 (Hermansky-Pudlak syndrome type 9) (BLOC1S6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLOC1S6	BLOC1S6, BLOS6, PLDN, PA, HPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hermansky-Pudlak syndrome 9 (HPS9) [MIM:614171]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:21665000}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hermansky-Pudlak Sendromu 9, 614171, Otozomal Resesif; HPS9 (Hermansky-Pudlak Sendromu Tip 9) (Prenatal) (BLOC1S6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hermansky-pudlak syndrome 9, 614171, Autosomal recessive; HPS9 (Hermansky-Pudlak syndrome type 9) (BLOC1S6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BLOC1S6	BLOC1S6, BLOS6, PLDN, PA, HPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hermansky-Pudlak syndrome 9 (HPS9) [MIM:614171]: A form of Hermansky-Pudlak syndrome, a genetically heterogeneous autosomal recessive disorder characterized by oculocutaneous albinism, bleeding due to platelet storage pool deficiency, and lysosomal storage defects. This syndrome results from defects of diverse cytoplasmic organelles including melanosomes, platelet dense granules and lysosomes. Ceroid storage in the lungs is associated with pulmonary fibrosis, a common cause of premature death in individuals with HPS. {ECO:0000269	PubMed:21665000}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Herpes Simpleks Ensefaliti, Yatkınlık, 6, 614850 (Herpes Simpleks Ensefaliti) (TICAM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Herpes simplex encephalitic, susceptibility to, 6, 614850 (Herpes simplex encephalitis) (TICAM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TICAM1	TICAM1, TRIF, IIAE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Herpes simplex encephalitis 4 (HSE4) [MIM:614850]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:22105173}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 6, 614850 (Herpes Simpleks Ensefaliti) (Prenatal) (TICAM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Herpes simplex encephalitic, susceptibility to, 6, 614850 (Herpes simplex encephalitis) (TICAM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TICAM1	TICAM1, TRIF, IIAE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Herpes simplex encephalitis 4 (HSE4) [MIM:614850]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:22105173}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 2, 613002 (Herpes Simpleks Ensefaliti) (TLR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Herpes simplex encephalitis, susceptibility to, 2, 613002 (Herpes simplex encephalitis) (TLR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR3	TLR3, IIAE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Herpes simplex encephalitis 2 (HSE2) [MIM:613002]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:17872438}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. TLR3 mutations predispose otherwise healthy individuals to isolated herpes simplex encephalitis through a mechanism that involves impaired IFNs production and reduced immune defense against viral infection in the central nervous system.	EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 2, 613002 (Herpes Simpleks Ensefaliti) (Prenatal) (TLR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Herpes simplex encephalitis, susceptibility to, 2, 613002 (Herpes simplex encephalitis) (TLR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TLR3	TLR3, IIAE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Herpes simplex encephalitis 2 (HSE2) [MIM:613002]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:17872438}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. TLR3 mutations predispose otherwise healthy individuals to isolated herpes simplex encephalitis through a mechanism that involves impaired IFNs production and reduced immune defense against viral infection in the central nervous system.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 3, 614849 (Herpes Simpleks Ensefaliti) (TRAF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Herpes simplex encephalitis, susceptibility to, 3, 614849 (Herpes simplex encephalitis) (TRAF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAF3	TRAF3, CD40BP, LAP1, CAP1, CRAF1, IIAE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Herpes simplex encephalitis 3 (HSE3) [MIM:614849]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:20832341}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 3, 614849 (Herpes Simpleks Ensefaliti) (Prenatal) (TRAF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Herpes simplex encephalitis, susceptibility to, 3, 614849 (Herpes simplex encephalitis) (TRAF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAF3	TRAF3, CD40BP, LAP1, CAP1, CRAF1, IIAE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Herpes simplex encephalitis 3 (HSE3) [MIM:614849]: A rare complication of human herpesvirus 1 (HHV-1) infection, occurring in only a small minority of HHV-1 infected individuals. HSE is characterized by hemorrhagic necrosis of parts of the temporal and frontal lobes. Onset is over several days and involves fever, headache, seizures, stupor, and often coma, frequently with a fatal outcome. {ECO:0000269|PubMed:20832341}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 7, 616532, Otozomal Dominant (Herpes Simpleks Ensefaliti) (IRF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Herpes simplex encephalitis, susceptibility to, 7, 616532, Autosomal dominant (Herpes simplex encephalitis) (IRF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF3	IRF3, IIAE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Herpes Simpleks Ensefaliti, Yatkınlık, 7, 616532, Otozomal Dominant (Herpes Simpleks Ensefaliti) (Prenatal) (IRF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Herpes simplex encephalitis, susceptibility to, 7, 616532, Autosomal dominant (Herpes simplex encephalitis) (IRF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF3	IRF3, IIAE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 1, X'e Bağlı, 306955, X'e Bağlı Reseslf, HTX1 (Heterotaksi) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 1, X-linked, 306955, X-linked recessive; HTX1 (Heterotaxia) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Heterotaksi, Visseral, 1, X'e Bağlı, 306955, X'e Bağlı Reseslf, HTX1 (Heterotaksi) (Prenatal) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 1, X-linked, 306955, X-linked recessive; HTX1 (Heterotaxia) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 2, Otozomal, 605376, Otozomal Dominant; HTX2 (Heterotaksi) (CFC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 2, autosomal, 605376, Autosomal dominant; HTX2 (Heterotaxia) (CFC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFC1	CFC1, CRYPTIC, HTX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 2, autosomal (HTX2) [MIM:605376]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:11062482, ECO:0000269|PubMed:11799476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Heterotaksi, Visseral, 2, Otozomal, 605376, Otozomal Dominant; HTX2 (Heterotaksi) (Prenatal) (CFC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 2, autosomal, 605376, Autosomal dominant; HTX2 (Heterotaxia) (CFC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFC1	CFC1, CRYPTIC, HTX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 2, autosomal (HTX2) [MIM:605376]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:11062482, ECO:0000269|PubMed:11799476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 4, Otozomal, 613751; HTX4 (Heterotaksi) (ACVR2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 4, autosomal, 613751; HTX4 (Heterotaxia) (ACVR2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACVR2B	ACVR2B, ACTRIIB, HTX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 4, autosomal (HTX4) [MIM:613751]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. HTX4 clinical features include dextrocardia, right aortic arch and a right-sided spleen, anomalies of the inferior and the superior vena cava, atrial ventricular canal defect with dextro-transposed great arteries, pulmonary stenosis, polysplenia and midline liver. {ECO:0000269|PubMed:9916847}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Heterotaksi, Visseral, 4, Otozomal, 613751; HTX4 (Heterotaksi) (Prenatal) (ACVR2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 4, autosomal, 613751; HTX4 (Heterotaxia) (ACVR2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACVR2B	ACVR2B, ACTRIIB, HTX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 4, autosomal (HTX4) [MIM:613751]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. HTX4 clinical features include dextrocardia, right aortic arch and a right-sided spleen, anomalies of the inferior and the superior vena cava, atrial ventricular canal defect with dextro-transposed great arteries, pulmonary stenosis, polysplenia and midline liver. {ECO:0000269|PubMed:9916847}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 5, 270100, Otozomal Dominant; HTX5 (Heterotaksi) (NODAL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 5, 270100, Autosomal dominant; HTX5 (Heterotaxia) (NODAL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NODAL	NODAL, HTX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 5, autosomal (HTX5) [MIM:270100]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. HTX5 clinical features include situs inversus viscerum or situs ambiguus, congenital heart defect, transposition of the great vessels ventricular septal defect, atrial septal defect, truncus communis, and dextrocardia. {ECO:0000269|PubMed:19064609, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Heterotaksi, Visseral, 5, 270100, Otozomal Dominant; HTX5 (Heterotaksi) (Prenatal) (NODAL gen) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 5, 270100, Autosomal dominant; HTX5 (Heterotaxia) (NODAL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NODAL	NODAL, HTX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 5, autosomal (HTX5) [MIM:270100]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. HTX5 clinical features include situs inversus viscerum or situs ambiguus, congenital heart defect, transposition of the great vessels ventricular septal defect, atrial septal defect, truncus communis, and dextrocardia. (ECO:0000269|PubMed:19064609, ECO:0000269|PubMed:9354794). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 6, Otozomal Resesif, 614779, Otozomal Resesif; HTX6 (Heterotaksi) (CFAP53 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 6, autosomal recessive, 614779, Autosomal recessive; HTX6 (Heterotaxia) (CFAP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFAP53	CCDC11, HTX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heterotaksi, Visseral, 6, Otozomal Resesif, 614779, Otozomal Resesif; HTX6 (Heterotaksi) (Prenatal) (CFAP53 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 6, autosomal recessive, 614779, Autosomal recessive; HTX6 (Heterotaxia) (CFAP53 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFAP53	CCDC11, HTX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 7, Otozomal, 616749, Otozomal Resesif; HTX7 (Heterotaksi) (MMP21 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 7, autosomal, 616749, Autosomal recessive; HTX7 (Heterotaxia) (MMP21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP21	MMP21, HTX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heterotaksi, Visseral, 7, Otozomal, 616749, Otozomal Resesif; HTX7 (Heterotaksi) (Prenatal) (MMP21 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 7, autosomal, 616749, Autosomal recessive; HTX7 (Heterotaxia) (MMP21 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP21	MMP21, HTX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotaksi, Visseral, 8, Otozomal, 617205, Otozomal Resesif; HTX8 (Heterotaksi) (PKD1L1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotaxy, visceral, 8, autosomal, 617205, Autosomal recessive; HTX8 (Heterotaxia) (PKD1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKD1L1	PKD1L1, HTX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heterotaksi, Visseral, 8, Otozomal, 617205, Otozomal Resesif; HTX8 (Heterotaksi) (Prenatal) (PKD1L1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotaxy, visceral, 8, autosomal, 617205, Autosomal recessive; HTX8 (Heterotaxia) (PKD1L1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PKD1L1	PKD1L1, HTX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotopi, Periventriküler 300049, X'e Bağlı Dominant (Nodüler Nöronal Heterotopi) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Heterotopia, periventricular, 300049, X-linked dominant (Nodular neuronal heterotopia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Heterotopi, Periventriküler 300049, X'e Bağlı Dominant (Nodüler Nöronal Heterotopi) (MLPA)	Heterotopia, periventricular, 300049, X-linked dominant (Nodular neuronal heterotopia) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Heterotopi, Periventriküler 300049, X'e Bağlı Dominant (Nodüler Nöronal Heterotopi) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Heterotopia, periventricular, 300049, X-linked dominant (Nodular neuronal heterotopia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Heterotopi, Periventriküler 300049, X'e Bağlı Dominant (Nodüler Nöronal Heterotopi) (Prenatal) (MLPA)	Heterotopia, periventricular, 300049, X-linked dominant (Nodular neuronal heterotopia) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HFE Hemokromatozis, Modifiye Edici, 235200, Otozomal Resesif (Hemokromatoz Tip 1 Semptomatik Form) (BMP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HFE hemochromatosis, modifier of, 235200, Autosomal recessive (Symptomatic form of hemochromatosis type 1) (BMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP2	BMP2, BMP2A, BDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yüksek Dansiteli Lipoprotein Kolesterol Seviyesi QTL 7 (EDN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	High density lipoprotein cholesterol level QTL 7 (EDN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN1	EDN1, ARCND3, QME, HDLCQ7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kalça Displazisi, Beukes Tipi, 142669, Otozomal Dominant; BHD (Kalça Displazisi, Beukes Tipi) (UFSP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hip dysplasia, Beukes type, 142669, Autosomal dominant; BHD (Hip dysplasia, Beukes type) (UFSP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UFSP2	UFSP2, C4orf20, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beukes familial hip dysplasia (BFHD) [MIM:142669]: A severe progressive degenerative osteoarthritis of the hip joint with underlying dysplasia confined to that region. Affected individuals are of normal stature and have no associated health problems. (ECO:0000269	PubMed:26428751). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hirschsprung hastalığı veya aganglionik megakolon (RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2) (MLPA)	Hirschsprung disease or aganglionic megacolon (RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2) (MLPA)	RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hirschsprung hastalığı veya aganglionik megakolon (RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2) (MLPA) (Prenatal)	Hirschsprung disease or aganglionic megacolon (RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2) (MLPA) (Prenatal)	RET 10q11.2, ZFHX1B 2q22.3, EDN3 20q13.3, GDNF 5q13.2		MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Kardiyak Defektler ve Otonomikdisfonksiyon, 613870, Otozomal Dominant; HCAD (ECE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870, Autosomal dominant; HCAD (ECE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ECE1	ECE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Kardiyak Defektler ve Otonomikdisfonksiyon, 613870, Otozomal Dominant; HCAD (Prenatal) (ECE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hirschsprung disease, cardiac defects, and autonomic dysfunction, 613870, Autosomal dominant; HCAD (ECE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ECE1	ECE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 1, 142623, Otozomal Dominant; HSCR1 (Hirschsprung Hastalığı) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hirschsprung disease, susceptibility to, 1, 142623, Autosomal dominant; HSCR1 (Hirschsprung disease) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 1, 142623, Otozomal Dominant; HSCR1 (Hirschsprung Hastalığı) (MLPA)	Hirschsprung disease, susceptibility to, 1, 142623, Autosomal dominant; HSCR1 (Hirschsprung disease) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 1, 142623, Otozomal Dominant; HSCR1 (Hirschsprung Hastalığı) (Prenatal) (RET geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hirschsprung disease, susceptibility to, 1, 142623, Autosomal dominant; HSCR1 (Hirschsprung disease) (RET gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 1, 142623, Otozomal Dominant; HSCR1 (Hirschsprung Hastalığı) (Prenatal) (MLPA)	Hirschsprung disease, susceptibility to, 1, 142623, Autosomal dominant; HSCR1 (Hirschsprung disease) (Prenatal) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 2, 600155, Otozomal Resesif; HSCR2 (Hirschsprung Hastalığı) (EDNRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hirschsprung disease, susceptibility to, 2, 600155, Autosomal recessive; HSCR2 (Hirschsprung disease) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 2, 600155, Otozomal Resesif; HSCR2 (Hirschsprung Hastalığı) (Prenatal) (EDNRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hirschsprung disease, susceptibility to, 2, 600155, Autosomal recessive; HSCR2 (Hirschsprung disease) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 3, 613711, HSCR3 (Hirschsprung Hastalığı) (GDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hirschsprung disease, susceptibility to, 3, 613711, HSCR3 (Hirschsprung disease) (GDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDNF	GDNF, HSCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 3, 613711, HSCR3 (Hirschsprung Hastalığı) (MLPA)	Hirschsprung disease, susceptibility to, 3, 613711, HSCR3 (Hirschsprung disease) (MLPA)	GDNF	GDNF, HSCR3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 3, 613711, HSCR3 (Hirschsprung Hastalığı) (Prenatal) (GDNF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hirschsprung disease, susceptibility to, 3, 613711, HSCR3 (Hirschsprung disease) (GDNF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDNF	GDNF, HSCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 3, 613711, HSCR3 (Hirschsprung Hastalığı) (Prenatal) (MLPA)	Hirschsprung disease, susceptibility to, 3, 613711, HSCR3 (Hirschsprung disease) (Prenatal) (MLPA)	GDNF	GDNF, HSCR3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 4, 613712, HSCR4 (Hirschsprung Hastalığı) (EDN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hirschsprung disease, susceptibility to, 4, 613712, HSCR4 (Hirschsprung disease) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 4, 613712, HSCR4 (Hirschsprung Hastalığı) (MLPA)	Hirschsprung disease, susceptibility to, 4, 613712, HSCR4 (Hirschsprung disease) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 4, 613712, HSCR4 (Hirschsprung Hastalığı) (Prenatal) (EDN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hirschsprung disease, susceptibility to, 4, 613712, HSCR4 (Hirschsprung disease) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hirschsprung Hastalığı, Yatkınlık, 4, 613712, HSCR4 (Hirschsprung Hastalığı) (Prenatal) (MLPA)	Hirschsprung disease, susceptibility to, 4, 613712, HSCR4 (Hirschsprung disease) (Prenatal) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Histiositom, Anjiyomatoid Fibröz, Somatik, 612160 (CREB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Histiocytoma, angiomatoid fibrous, somatic, 612160 (CREB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CREB1	CREB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Angiomatoid fibrous histiocytoma (AFH) [MIM:612160]: A distinct variant of malignant fibrous histiocytoma that typically occurs in children and adolescents and is manifest by nodular subcutaneous growth. Characteristic microscopic features include lobulated sheets of histiocyte-like cells intimately associated with areas of hemorrhage and cystic pseudovascular spaces, as well as a striking cuffing of inflammatory cells, mimicking a lymph node metastasis. Note=The gene represented in this entry may be involved in disease pathogenesis. A chromosomal aberration involving CREB1 is found in a patient with angiomatoid fibrous histiocytoma. Translocation t(2;22)(q33;q12) with CREB1 generates a EWSR1/CREB1 fusion gene that is most common genetic abnormality in this tumor type.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Histiyositozis-Lenfadenopati Artı Sendromu, 602782, Otozomal Resesif (H Sendromu) (SLC29A3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Histiocytosis-lymphadenopathy plus syndrome, 602782, Autosomal recessive (H syndrome) (SLC29A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC29A3	SLC29A3, ENT3, PHID, HCLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Histiyositozis-Lenfadenopati Artı Sendromu, 602782, Otozomal Resesif (H Sendromu) (Prenatal) (SLC29A3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Histiocytosis-lymphadenopathy plus syndrome, 602782, Autosomal recessive (H syndrome) (SLC29A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC29A3	SLC29A3, ENT3, PHID, HCLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HIV Enfeksiyonu, Direnç, 609423 (CCL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV infection, resistance to, 609423 (CCL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL3	CCL3, SCYA3, MIP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV Enfeksiyonu, Yatkınlık / Direnç (CCR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV infection, susceptibility/resistance to (CCR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCR2	CCR2, CMKBR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV Enfeksiyonu, Yatkınlık / Direnç (Hepatit C Antiviral Tedaviye Yanıt) (CCR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV infection, susceptibility/resistance to (Response to antiviral treatment in hepatitis C) (CCR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCR5	CCR5, CMKBR5, CCCKR5, IDDM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV Tip 1, Yatkınlık, 609423 (CD209 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV type 1, susceptibility to, 609423 (CD209 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD209	CD209, CDSIGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1 Hastalığı, Geciktirmeli Progresyon (CCL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV-1 disease, delayed progression of (CCL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL5	CCL5, SCYA5, D17S136E, TCP228	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1 Hastalığı, Hızlı İlerlemesi (CCL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HIV-1 disease, rapid progression of (CCL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL5	CCL5, SCYA5, D17S136E, TCP228	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

HIV-1 Viremi, Yatkınlık, 609423 (HLA-C gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV-1 viremia, susceptibility to, 609423 (HLA-C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-C	HLA-C, PSORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1, Direnç, 609423 (CCL2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV-1, resistance to, 609423 (CCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL2	CCL2, SCYA2, MCP1, MCAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1, Yatkınlık, 609423 (IL10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV-1, susceptibility to, 609423 (IL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10	IL10, CSIF, GVHDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV / AIDS, Yatkınlık, 609423 (CCL3L1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV/AIDS, susceptibility to, 609423 (CCL3L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL3L1	CCL3L1, SCYA3L1, LD78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1 Enfeksiyonu, Direnç, 609423 (TLR3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV1 infection, resistance to, 609423 (TLR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR3	TLR3, IIAE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIV-1, Direnç, 609423 (CCL11 gen) (Tüm Gen Dizi Analizi) (Postnatal)	HIV1, resistance to, 609423 (CCL11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL11	CCL11, SCYA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA B27 (HLA-B gen) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA B27 (HLA-B gene) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA-B	HLA-B, SPDA1	HLA Analizi/ HLA Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HLA B27 (HLA-B gen) (Real Time-PCR) (Postnatal)	HLA B27 (HLA-B gene) (Real Time-PCR) (Postnatal)	HLA-B	HLA-B, SPDA1	Real Time-PCR	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HLA B5 (HLA-B gen) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA B5 (HLA-B gene) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA-B	HLA-B, SPDA1	HLA Analizi/ HLA Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HLA B51 (HLA-B gen) (Real Time-PCR) (Postnatal)	HLA B51 (HLA-B gene) (Real Time-PCR) (Postnatal)	HLA-B	HLA-B, SPDA1	Real Time-PCR	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HLA B51 (HLA-B gen) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA B51 (HLA-B gene) (HLA Analizi/ HLA Analiysis) (Postnatal)	HLA-B	HLA-B, SPDA1	HLA Analizi/ HLA Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HMG-CoA Sentaz-2 Eksikliği, 605911; HMGCS2D (3-Hidroksi-3-Metilglutaril-CoA Sentaz 2 Eksikliği) (3-Hidroksi-3-Metilglutaril-CoA Sentaz Eksikliği) (HMGCS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HMG-CoA synthase-2 deficiency, 605911; HMGCS2D (3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency) (3-hydroxy-3-methylglutaryl-CoA synthase deficiency) (HMGCS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGCS2	HMGCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: 3-hydroxy-3-methylglutaryl-CoA synthase-2 deficiency (HMGCS2D) [MIM:605911]: A metabolic disorder characterized by severe hypoketotic hypoglycemia, encephalopathy, and hepatomegaly. {ECO:0000269|PubMed:11228257, ECO:0000269|PubMed:11479731, ECO:0000269|PubMed:12647205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
HMG-CoA Sentaz-2 Eksikliği, 605911; HMGCS2D (3-Hidroksi-3-Metilglutaril-CoA Sentaz 2 Eksikliği) (3-Hidroksi-3-Metilglutaril-CoA Sentaz Eksikliği) (Prenatal) (HMGCS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	HMG-CoA synthase-2 deficiency, 605911; HMGCS2D (3-hydroxy-3-methylglutaryl-CoA synthase 2 deficiency) (3-hydroxy-3-methylglutaryl-CoA synthase deficiency) (HMGCS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HMGCS2	HMGCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: 3-hydroxy-3-methylglutaryl-CoA synthase-2 deficiency (HMGCS2D) [MIM:605911]: A metabolic disorder characterized by severe hypoketotic hypoglycemia, encephalopathy, and hepatomegaly. {ECO:0000269|PubMed:11228257, ECO:0000269|PubMed:11479731, ECO:0000269|PubMed:12647205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hodgkin Lenfoma, Yatkınlık, 236000, Otozomal Resesif, CHL (Klasik Hodgkin Lenfoma) (KLHDC8B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hodgkin lymphoma, susceptibility to, 236000, Autosomal recessive, CHL (Classic Hodgkin lymphoma) (KLHDC8B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHDC8B	KLHDC8B, CHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphoma, Hodgkin, classic (CHL) [MIM:236000]: A malignant disease characterized by progressive enlargement of the lymph nodes, spleen and general lymphoid tissue, and the presence of large, usually multinucleate, cells (Reed-Sternberg cells). Reed-Sternberg cells compose only 1-2% of the total tumor cell mass. The remainder is composed of a variety of reactive, mixed inflammatory cells consisting of lymphocytes, plasma cells, neutrophils, eosinophils and histiocytes. {ECO:0000269|PubMed:19706467}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A variant in the 5'-UTR of KLHDC8B, responsible for decreasing its expression, is associated with classic Hodgkin lymphoma and segregates with the disease in some families (PubMed:19706467). {ECO:0000269|PubMed:19706467}.	EDTA Blood Sample
Holokarboksilaz Sentaz Eksikliği, 253270, Otozomal Resesif (Holokarboksilaz Sentetaz Eksikliği) (HLCS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holocarboxylase synthetase deficiency, 253270, Autosomal recessive (Holocarboxylase synthetase deficiency) (HLCS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLCS	HLCS, HCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Holokarboksilaz Sentaz Eksikliği, 253270, Otozomal Resesif (Holokarboksilaz Sentetaz Eksikliği) (Prenatal) (HLCS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holocarboxylase synthetase deficiency, 253270, Autosomal recessive (Holocarboxylase synthetase deficiency) (HLCS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HLCS	HLCS, HCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali paneli (PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11) (MLPA)	Holoprencephalic panel (PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11) (MLPA)	PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Holoprozensefali paneli (PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11) (MLPA) (Prenatal)	Holoprencephalic panel (PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11) (MLPA) (Prenatal)	PTCH, SHH, ZIC2, SIX3, TGIF TMEM1, FBXW11	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 11, 614226, Otozomal Dominant, Sporadik Vakalar; HPE11 (Holoprozensefali) (CDON geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 11, 614226, Autosomal dominant, Isolated cases; HPE11 (Holoprosencephaly) (CDON gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDON	CDON, CDO, HPE11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 11 (HPE11) [MIM:614226]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269	PubMed:21802063}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Holoprozensefali 11, 614226, Otozomal Dominant, Sporadik Vakalar; HPE11 (Holoprozensefali) (Prenatal) (CDON geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 11, 614226, Autosomal dominant, Isolated cases; HPE11 (Holoprosencephaly) (CDON gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDON	CDON, CDO, HPE11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 11 (HPE11) [MIM:614226]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269	PubMed:21802063}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 2, 157170, Otozomal Dominant, Sporadik Vakalar; HPE2 (Holoprozensefali) (SIX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 2, 157170, Autosomal dominant, Isolated cases; HPE2 (Holoprosencephaly) (SIX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX3	SIX3, HPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Holoprozensefali 2, 157170, Otozomal Dominant, Sporadik Vakalar; HPE2 (Holoprozensefali) (MLPA)	Holoprosencephaly 2, 157170, Autosomal dominant, Isolated cases; HPE2 (Holoprosencephaly) (MLPA)	SIX3	SIX3, HPE2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Holoprozensefali 2, 157170, Otozomal Dominant, Sporadik Vakalar; HPE2 (Holoprozensefali) (Prenatal) (SIX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 2, 157170, Autosomal dominant, Isolated cases; HPE2 (Holoprosencephaly) (SIX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIX3	SIX3, HPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 2, 157170, Otozomal Dominant, Sporadik Vakalar; HPE2 (Holoprozensefali) (Prenatal) (MLPA)	Holoprosencephaly 2, 157170, Autosomal dominant, Isolated cases; HPE2 (Holoprosencephaly) (Prenatal) (MLPA)	SIX3	SIX3, HPE2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 3, 142945, Otozomal Dominant, HPE3 (Holoprozensefali) (SHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 3, 142945, Autosomal dominant; HPE3 (Holoprosencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Holoprozensefali 3, 142945, Otozomal Dominant; HPE3 (Holoprozensefali) (MLPA)	Holoprosencephaly 3, 142945, Autosomal dominant; HPE3 (Holoprosencephaly) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Holoprozensefali 3, 142945, Otozomal Dominant; HPE3 (Holoprozensefali) (Prenatal) (SHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 3, 142945, Autosomal dominant; HPE3 (Holoprosencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 3, 142945, Otozomal Dominant; HPE3 (Holoprozensefali) (Prenatal) (MLPA)	Holoprosencephaly 3, 142945, Autosomal dominant; HPE3 (Holoprosencephaly) (Prenatal) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 4, 142946, Otozomal Dominant; HPE4 (Holoprozensefali) (TGIF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 4, 142946, Autosomal dominant; HPE4 (Holoprosencephaly) (TGIF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGIF1	TGIF1, HPE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 4 (HPE4) [MIM:142946]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10835638, ECO:0000269|PubMed:15221788}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Holoprozensefali 4, 142946, Otozomal Dominant; HPE4 (Holoprozensefali) (Prenatal) (TGIF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 4, 142946, Autosomal dominant; HPE4 (Holoprosencephaly) (TGIF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGIF1	TGIF1, HPE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 4 (HPE4) [MIM:142946]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:10835638, ECO:0000269|PubMed:15221788}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 5, 609637, Otozomal Dominant; HPE5 (Holoprozensefali) (ZIC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 5, 609637, Autosomal dominant; HPE5 (Holoprosencephaly) (ZIC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZIC2	ZIC2, HPE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 5 (HPE5) [MIM:609637]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:11285244, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:19177455}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Holoprozensefali 5, 609637, Otozomal Dominant; HPE5 (Holoprozensefali) (MLPA)	Holoprosencephaly 5, 609637, Autosomal dominant; HPE5 (Holoprosencephaly) (MLPA)	ZIC2	ZIC2, HPE5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Holoprosencephaly 5 (HPE5) [MIM:609637]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:11285244, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:19177455}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Holoprozensefali 5, 609637, Otozomal Dominant; HPE5 (Holoprozensefali) (Prenatal) (ZIC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 5, 609637, Autosomal dominant; HPE5 (Holoprosencephaly) (ZIC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZIC2	ZIC2, HPE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 5 (HPE5) [MIM:609637]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:11285244, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:19177455}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 5, 609637, Otozomal Dominant; HPE5 (Holoprozensefali) (Prenatal) (MLPA)	Holoprosencephaly 5, 609637, Autosomal dominant; HPE5 (Holoprosencephaly) (Prenatal) (MLPA)	ZIC2	ZIC2, HPE5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 5 (HPE5) [MIM:609637]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. {ECO:0000269|PubMed:11285244, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:19177455}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 7, 610828, Otozomal Dominant; HPE7 (Holoprozensefali) (PTCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 7, 610828, Autosomal dominant; HPE7 (Holoprosencephaly) (PTCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH1	PTCH1, NBCCS, BCNS, HPE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Holoprozensefali 7, 610828, Otozomal Dominant; HPE7 (Holoprozensefali) (Prenatal) (PTCH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 7, 610828, Autosomal dominant; HPE7 (Holoprosencephaly) (PTCH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTCH1	PTCH1, NBCCS, BCNS, HPE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holoprozensefali 9, 610829, Otozomal Dominant; HPE9 (Holoprozensefali) (GLI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holoprosencephaly 9, 610829, Autosomal dominant; HPE9 (Holoprosencephaly) (GLI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI2	GLI2, HPE9, CJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 9 (HPE9) [MIM:610829]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. Holoprosencephaly type 9 is characterized by defective anterior pituitary formation and pan-hypopituitarism, with or without overt forebrain cleavage abnormalities, and holoprosencephaly-like midfacial hypoplasia. {ECO:0000269|PubMed:14581620, ECO:0000269|PubMed:17096318, ECO:0000269|PubMed:20685856}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Holoprozensefali 9, 610829, Otozomal Dominant; HPE9 (Holoprozensefali) (Prenatal) (GLI2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holoprosencephaly 9, 610829, Autosomal dominant; HPE9 (Holoprosencephaly) (GLI2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLI2	GLI2, HPE9, CJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 9 (HPE9) [MIM:610829]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. Holoprosencephaly type 9 is characterized by defective anterior pituitary formation and pan-hypopituitarism, with or without overt forebrain cleavage abnormalities, and holoprosencephaly-like midfacial hypoplasia. {ECO:0000269|PubMed:14581620, ECO:0000269|PubMed:17096318, ECO:0000269|PubMed:20685856}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Holt-Oram Sendromu, 142900, Otozomal Dominant; HOS (Holt-Oram Sendromu) (TBX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Holt-Oram syndrome, 142900, Autosomal dominant; HOS (Holt-Oram syndrome) (TBX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX5	TBX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holt-Oram syndrome (HOS) [MIM:142900]: Developmental disorder affecting the heart and upper limbs. It is characterized by thumb anomaly and atrial septal defects. {ECO:0000269|PubMed:10077612, ECO:0000269|PubMed:10842287, ECO:0000269|PubMed:12818525, ECO:0000269|PubMed:15735645, ECO:0000269|PubMed:20450920, ECO:0000269|PubMed:8988164, ECO:0000269|PubMed:8988165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Holt-Oram Sendromu, 142900, Otozomal Dominant; HOS (Holt-Oram Sendromu) (Prenatal) (TBX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Holt-Oram syndrome, 142900, Autosomal dominant; HOS (Holt-Oram syndrome) (TBX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX5	TBX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holt-Oram syndrome (HOS) [MIM:142900]: Developmental disorder affecting the heart and upper limbs. It is characterized by thumb anomaly and atrial septal defects. {ECO:0000269|PubMed:10077612, ECO:0000269|PubMed:10842287, ECO:0000269|PubMed:12818525, ECO:0000269|PubMed:15735645, ECO:0000269|PubMed:20450920, ECO:0000269|PubMed:8988164, ECO:0000269|PubMed:8988165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistein Plazma Seviyesi, 600008 (NNMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocysteine plasma level, 600008 (NNMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NNMT	NNMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistein, Total Plazma, Artmış (CTH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocysteine, total plasma, elevated (CTH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTH	CTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistinüri, Sistatiyonin Beta-Synthase Eksikliğine Bağlı (Klasik Homosistinüri) (CBS geni) (Tüm Gen Dizi Analizi) (Postnatal)	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY (Classic homocystinuria) (CBS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBS	CBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistinüri, Sistatiyonin Beta-Synthase Eksikliğine Bağlı (Klasik Homosistinüri) (Prenatal) (CBS geni) (Tüm Gen Dizi Analizi) (Prenatal)	HOMOCYSTINURIA DUE TO CYSTATHIONINE BETA-SYNTHASE DEFICIENCY (Classic homocystinuria) (CBS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CBS	CBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistinüri, MTHFR Eksikliğine Bağlı, 236250, Otozomal Resesif (Homosistinüri, Metilen Tetrahidrofolat Redüktaz Eksikliğine Bağlı) (MTHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocystinuria due to MTHFR deficiency, 236250, Autosomal recessive (Homocystinuria due to methylene tetrahydrofolate reductase deficiency) (MTHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Homosistinüri, MTHFR Eksikliğine Bağlı 236250, Otozomal Resesif (Homosistinüri, Metilen Tetrahidrofolat Redüktaz Eksikliğine Bağlı) (MLPA)	Homocystinuria due to MTHFR deficiency, 236250, Autosomal recessive (Homocystinuria due to methylene tetrahydrofolate reductase deficiency) (MLPA)	MTHFR	MTHFR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Homosistinüri, MTHFR Eksikliğine Bağlı 236250, Otozomal Resesif (Homosistinüri, Metilen Tetrahidrofolat Redüktaz Eksikliğine Bağlı) (Prenatal) (MTHFR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Homocystinuria due to MTHFR deficiency, 236250, Autosomal recessive (Homocystinuria due to methylene tetrahydrofolate reductase deficiency) (MTHFR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistinüri, MTHFR Eksikliğine Bağlı 236250, Otozomal Resesif (Homosistinüri, Metilen Tetrahidrofolat Redüktaz Eksikliğine Bağlı) (Prenatal) (MLPA)	Homocystinuria due to MTHFR deficiency, 236250, Autosomal recessive (Homocystinuria due to methylene tetrahydrofolate reductase deficiency) (Prenatal) (MLPA)	MTHFR	MTHFR	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Homosistinüri-Megaloblastik Anemi, Ctbl E Tipi, 236270, Otozomal Resesif; HMAE (Metilkobalamin Eksikliği Tip C'dek) (MTRR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocystinuria-megaloblastic anemia, cbl E type, 236270, Autosomal recessive; HMAE (Methylcobalamin deficiency type cblE) (MTRR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTRR	MTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistinüri-Megaloblastik Anemi, Ctbl E Tipi, 236270, Otozomal Resesif; HMAE (Metilkobalamin Eksikliği Tip C'dek) (Prenatal) (MTRR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Homocystinuria-megaloblastic anemia, cbl E type, 236270, Autosomal recessive; HMAE (Methylcobalamin deficiency type cblE) (MTRR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTRR	MTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistinüri-Megaloblastik Anemi, Cblg Komplementasyon Grubu, 250940, Otozomal Resesif; HMAG (Metilkobalamin Eksikliği Cblg Tipi) (MTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940, Autosomal recessive; HMAG (Methylcobalamin deficiency CblG type) (MTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTR	MTR, HMAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Homocystinuria-megaloblastic anemia, cblG complementation type (HMAG) [MIM:250940]: An autosomal recessive inborn error of metabolism resulting from defects in the cobalamin-dependent pathway that converts homocysteine to methionine. It causes delayed psychomotor development, megaloblastic anemia, homocystinuria, and hypomethioninemia. {ECO:0000269|PubMed:8968736, ECO:0000269|PubMed:8968737}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Homosistinüri-Megaloblastik Anemi, Cblg Komplementasyon Grubu, 250940, Otozomal Resesif; HMAG (Metilkobalamin Eksikliği Cblg Tipi) (Prenatal) (MTR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Homocystinuria-megaloblastic anemia, cblG complementation type, 250940, Autosomal recessive; HMAG (Methylcobalamin deficiency CblG type) (MTR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTR	MTR, HMAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Homocystinuria-megaloblastic anemia, cblG complementation type (HMAG) [MIM:250940]: An autosomal recessive inborn error of metabolism resulting from defects in the cobalamin-dependent pathway that converts homocysteine to methionine. It causes delayed psychomotor development, megaloblastic anemia, homocystinuria, and hypomethioninemia. {ECO:0000269|PubMed:8968736, ECO:0000269|PubMed:8968737}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistinüri,B6 Yanıtlı ve Yanıt Vermeyen Türleri, 236200, Otozomal Resesif (Klasik Homosistinüri) (CBS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocystinuria, B6-responsive and nonresponsive types, 236200, Autosomal recessive (Classic homocystinuria) (CBS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBS	CBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistinüri,B6 Yanıtlı ve Yanıt Vermeyen Türleri, 236200, Otozomal Resesif (Klasik Homosistinüri) (Prenatal) (CBS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Homocystinuria, B6-responsive and nonresponsive types, 236200, Autosomal recessive (Classic homocystinuria) (CBS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CBS	CBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Homosistinüri, Ctbl Tipi, varyant 1, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (MMADHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Homocystinuria, cblD type, variant 1, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Homosistinüri, Ctbl Tipi, varyant 1, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (MMADHC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Homocystinuria, cblD type, variant 1, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
HPRT İlişkili Gut, 300323, X'e Bağlı (Hipoksantin Guanin Fosforibositransferaz Parsiyel Eksikliği) (HPRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HPRT-related gout, 300323, X-linked recessive (Hypoxanthine guanine phosphoribosyltransferase partial deficiency) (HPRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPRT1	HPRT1, HPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPRT İlişkili Gut, 300323, X'e Bağlı (Hipoksantin Guanin Fosforibositransferaz Parsiyel Eksikliği) (MLPA)	HPRT-related gout, 300323, X-linked recessive (Hypoxanthine guanine phosphoribosyltransferase partial deficiency) (MLPA)	HPRT1	HPRT1, HPRT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
HSAN2D, Otozomal Resesif, 243000, Otozomal Resesif; CIP (Ailesel Duyusal ve Otonomik Nöropati Tip 2) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	HSAN2D, autosomal recessive, 243000, Autosomal recessive; CIP (Hereditary sensory and autonomic neuropathy type 2) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Human Platelet Antijen (HPA1, HPA2, HPA3, HPA4, HPA5 ve HPA15 genleri a ve b alelleri) (SnapShot analizi) (Dizi Analizi) (HER BİR BİREY İÇİN)	Human Platelet Antigen (HPA1, HPA2, HPA3, HPA4, HPA5 and HPA15 genes, a and b alleles) (Snapshot analysis)	.		Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal		EDTA Blood Sample
Maligniteye Bağlı Humoral Hiperkalsemi (Brakidaktili Tip E) (PTHLH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Humoral hypercalcemia of malignancy (Brachydactyly type E) (PTHLH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTHLH	PTHLH, BDE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Huntington Benzeri Hastalık 1, 603218, Otozomal Dominant (Huntington Hastalığı Benzeri 1) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Huntington disease-like 1, 603218, Autosomal dominant (Huntington disease-like 1) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Huntington Hastalığı, 143100, Otozomal Dominant; HD (Huntington Hastalığı) (Tekrar Sayısı)	Huntington disease, 143100, Autosomal dominant; HD (Huntington disease)(Repeat Analysis)	HTT	HTT, HD, IT15	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Huntington disease (HD) [MIM:143100]: A neurodegenerative disorder characterized by involuntary movements (chorea), general motor impairment, psychiatric disorders and dementia. Onset of the disease occurs usually in the third or fourth decade of life. Onset and clinical course depend on the degree of poly-Gln repeat expansion, longer expansions resulting in earlier onset and more severe clinical manifestations. Neuropathology of Huntington disease displays a distinctive pattern with loss of neurons, especially in the caudate and putamen. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hurler Sendromu (Mukopolisakaridozisis Tip 1), MPS1 (IDUA geni) (Tüm Gen Dizi Analizi) (Postnatal)	HURLER SYNDROME (Mucopolysaccharidosis type 1), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hurler Sendromu (Mukopolisakaridozisis Tip 1), MPS1 (Prenatal) (IDUA geni) (Tüm Gen Dizi Analizi) (Prenatal)	HURLER SYNDROME (Mucopolysaccharidosis type 1), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hurler-Scheie Sendromu (Mukopolisakaridozisis Tip 1h), MPS1 (IDUA geni) (Tüm Gen Dizi Analizi) (Postnatal)	HURLER-SCHEIE SYNDROME (Mucopolysaccharidosis type 1h), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Hurler-Scheie Sendromu (Mukopolisakaridozis Tip 1h), MPS1 (Prenatal) (IDUA geni) (Tüm Gen Dizi Analizi) (Prenatal)	HURLER-SCHEIE SYNDROME (Mucopolysaccharidosis type 1h), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300847, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7592232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:9401515, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hutchinson-Gilford Progeria, 176670, Otozomal Resesif, Otozomal Dominant; HGPS (Hutchinson-Gilford Progeria Sendromu) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hutchinson-Gilford progeria, 176670, Autosomal recessive, Autosomal dominant; HGPS (Hutchinson-Gilford progeria syndrome) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hutchinson-Gilford Progeria, 176670, Otozomal Resesif, Otozomal Dominant; HGPS (Hutchinson-Gilford Progeria Sendromu) (MLPA)	Hutchinson-Gilford progeria, 176670, Autosomal recessive, Autosomal dominant; HGPS (Hutchinson-Gilford progeria syndrome) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hutchinson-Gilford Progeria, 176670, Otozomal Resesif, Otozomal Dominant; HGPS (Hutchinson-Gilford Progeria Sendromu) (Prenatal) (LMNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hutchinson-Gilford progeria, 176670, Autosomal recessive, Autosomal dominant; HGPS (Hutchinson-Gilford progeria syndrome) (LMNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hutchinson-Gilford Progeria, 176670, Otozomal Resesif, Otozomal Dominant; HGPS (Hutchinson-Gilford Progeria Sendromu) (Prenatal) (MLPA)	Hutchinson-Gilford progeria, 176670, Autosomal recessive, Autosomal dominant; HGPS (Hutchinson-Gilford progeria syndrome) (Prenatal) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hyalin Fibromatozis Sendromu, 228600, Otozomal Resesif; HFS (Juvenil Hyalin Fibromatozis) (ANTXR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyaline fibromatosis syndrome, 228600, Autosomal recessive; HFS (Juvenile hyaline fibromatosis) (ANTXR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANTXR2	ANTXR2, CMG2, HFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyaline fibromatosis syndrome (HFS) [MIM:228600]: An autosomal recessive syndrome characterized by abnormal growth of hyalinized fibrous tissue usually affecting subcutaneous regions on the scalp, ears, neck, face, hands, and feet. The lesions appear as pearly papules or fleshy nodules. Additional features include gingival hypertrophy, progressive joint contractures resulting in severe limitation of mobility, osteopenia, and osteoporosis. Disease severity is variable. Some individuals manifest symptoms in infancy and have additional visceral or systemic involvement. Hyaline deposits in multiple organs, recurrent infections and intractable diarrhea often lead to early death. Surviving children may suffer from severely reduced mobility due to joint contractures. Other patients have later onset of a milder disorder affecting only the face and digits. {ECO:0000269|PubMed:12973667, ECO:0000269|PubMed:14508707, ECO:0000269|PubMed:15725249). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidatidiform Mol, Tekrarlayan, 1, 231090, Otozomal Resesif; HYDM1 (Komplet Hidatidiform Mol) (NLRP7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydatidiform mole, recurrent, 1, 231090, Autosomal recessive; HYDM1 (Complete hydatidiform mole) (NLRP7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP7	NALP7, NOD12, PYPAF3, HYDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydatidiform mole, recurrent, 1 (HYDM1) [MIM:231090]: A disorder characterized by excessive trophoblast development that produces a growing mass of tissue inside the uterus at the beginning of a pregnancy. It leads to abnormal pregnancies with no embryo, and cystic degeneration of the chorionic villi. {ECO:0000269|PubMed:16462743, ECO:0000269|PubMed:19246479). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidatidiform Mol, Tekrarlayan, 2, 614293, Otozomal Resesif; HYDM2 (Komplet Hidatidiform Mol) (KHDC3L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydatidiform mole, recurrent, 2, 614293, Autosomal recessive; HYDM2 (Complete hydatidiform mole) (KHDC3L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KHDC3L	KHDC3L, C6orf221, ECAT1, HYDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydatidiform mole, recurrent, 2 (HYDM2) [MIM:614293]: A disorder characterized by excessive trophoblast development that produces a growing mass of tissue inside the uterus at the beginning of a pregnancy. It leads to abnormal pregnancies with no embryo, and cystic degeneration of the chorionic villi. {ECO:0000269	PubMed:21885028). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidranensefali-Anormal Genitalya, 300215, X'e Bağlı (X'e Bağlı Lizensefali-Anormal Genitalya) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydranencephaly with abnormal genitalia, 300215, X-linked (X-linked lissencephaly with abnormal genitalia) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hidranensefali-Anormal Genitalya, 300215, X'e Bağlı (X'e Bağlı Lizensefali-Anormal Genitalya) (MLPA)	Hydranencephaly with abnormal genitalia, 300215, X-linked (X-linked lissencephaly with abnormal genitalia) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hidranensefali-Anormal Genitalya, 300215, X'e Bağlı (X'e Bağlı Lizensefali-Anormal Genitalya) (Prenatal) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydranencephaly with abnormal genitalia, 300215, X-linked (X-linked lissencephaly with abnormal genitalia) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidranensefali-Anormal Genitalya, 300215, X'e Bağlı (X'e Bağlı Lizensefali-Anormal Genitalya) (Prenatal) (MLPA)	Hydranencephaly with abnormal genitalia, 300215, X-linked (X-linked lissencephaly with abnormal genitalia) (Prenatal) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akueduktal Stenoz Nedeniyle Hidrosefali, 307000, X'e Bağlı Resesif; HSAS (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrocephalus due to aqueductal stenosis, 307000, X-linked recessive; HSAS (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akueduktal Stenoz Nedeniyle Hidrosefali, 307000, X'e Bağlı Resesif; HSAS (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrocephalus due to aqueductal stenosis, 307000, X-linked recessive; HSAS (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrosefali, Konjenital İdyopatik İntestinal Psödoobstrüksiyon, 307000, X'e Bağlı Resesif (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000, X-linked recessive (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Hidrosefali, Konjenital İyopatik İntestinal Psödoobstrüksiyon, 307000, X'e Bağlı Resesif (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000, X-linked recessive (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrosefali-Hirschsprung Hastalığı, 307000, X'e Bağlı Resesif (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (L1 Sendromu) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrocephalus with Hirschsprung disease, 307000, X-linked recessive (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1 syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hidrosefali-Hirschsprung Hastalığı, 307000, X'e Bağlı Resesif (Akueduktus Sylvius Stenozuna Bağlı Hidrosefali) (L1 Sendromu) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrocephalus with Hirschsprung disease, 307000, X-linked recessive (Hydrocephalus with stenosis of the aqueduct of Sylvius) (L1 syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrosefali, Sendromik Olmayan Otozomal 2, 615219, Otozomal Resesif; HYC2 (Konjenital Hidrosefali) (MPDZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219, Autosomal recessive; HYC2 (Congenital hydrocephalus) (MPDZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPDZ	MPDZ, MUPP1, HYC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydrocephalus, non-syndromic, autosomal recessive 2 (HYC2) [MIM:615219]: A disease characterized by a disturbance of cerebrospinal fluid circulation causing accumulation of ventricular cerebrospinal fluid, which results in progressive ventricular dilatation with onset in utero. Affected individuals may have neurologic impairment. {ECO:0000269|PubMed:23240096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidrosefali, Sendromik Olmayan Otozomal 2, 615219, Otozomal Resesif; HYC2 (Konjenital Hidrosefali) (Prenatal) (MPDZ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrocephalus, nonsyndromic, autosomal recessive 2, 615219, Autosomal recessive; HYC2 (Congenital hydrocephalus) (MPDZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPDZ	MPDZ, MUPP1, HYC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hydrocephalus, non-syndromic, autosomal recessive 2 (HYC2) [MIM:615219]: A disease characterized by a disturbance of cerebrospinal fluid circulation causing accumulation of ventricular cerebrospinal fluid, which results in progressive ventricular dilatation with onset in utero. Affected individuals may have neurologic impairment. {ECO:0000269|PubMed:23240096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrosefali, Sendromik Olmayan, Otozomal Resesif, 236600, Otozomal Resesif; HYC1 (Konjenital Hidrosefali) (CCDC88C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrocephalus, nonsyndromic, autosomal recessive, 236600, Autosomal recessive; HYC1 (Congenital hydrocephalus) (CCDC88C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC88C	CCDC88C, HKRP2, DAPLE, KIAA1509, HYC, SCA40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydrocephalus, non-syndromic, autosomal recessive 1 (HYC1) [MIM:236600]: A disease characterized by a disturbance of cerebrospinal fluid circulation causing accumulation of ventricular cerebrospinal fluid, which results in progressive ventricular dilatation with onset in utero. Affected individuals may have neurologic impairment. {ECO:0000269|PubMed:21031079, ECO:0000269|PubMed:23042809}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hidrosefali, Sendromik Olmayan, Otozomal Resesif, 236600, Otozomal Resesif; HYC1 (Konjenital Hidrosefali) (Prenatal) (CCDC88C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrocephalus, nonsyndromic, autosomal recessive, 236600, Autosomal recessive; HYC1 (Congenital hydrocephalus) (CCDC88C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC88C	CCDC88C, HKRP2, DAPLE, KIAA1509, HYC, SCA40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hydrocephalus, non-syndromic, autosomal recessive 1 (HYC1) [MIM:236600]: A disease characterized by a disturbance of cerebrospinal fluid circulation causing accumulation of ventricular cerebrospinal fluid, which results in progressive ventricular dilatation with onset in utero. Affected individuals may have neurologic impairment. {ECO:0000269|PubMed:21031079, ECO:0000269|PubMed:23042809}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidroletalus Sendromu 2, 614120, Otozomal Resesif; HLS2 (Hidroletalus) (KIF7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrolethalus syndrome 2, 614120, Autosomal recessive; HLS2 (Hydrolethalus) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hidroletalus Sendromu 2, 614120, Otozomal Resesif; HLS2 (Hidroletalus) (Prenatal) (KIF7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrolethalus syndrome 2, 614120, Autosomal recessive; HLS2 (Hydrolethalus) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidroletalus Sendromu, 236680, Otozomal Resesif; HLS1 (Hidroletalus) (HYLS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrolethalus syndrome, 236680, Autosomal recessive; HLS1 (Hydroletalus) (HYLS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HYLS1	HYLS1, FLJ32915	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydrolethalus syndrome 1 (HLS1) [MIM:236680]: A lethal syndrome characterized by polydactyly, central nervous system malformation, and hydrocephalus. The polydactyly is postaxial in the hands and preaxial in the feet. A highly characteristic hallux duplex is seen in almost no other situation. In half of the cases, a large atrioventricular communis defect of the heart is found. The pregnancy is characterized by hydramnios, which is often massive, and by preterm delivery. {ECO:0000269|PubMed:15843405}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidroletalus Sendromu, 236680, Otozomal Resesif; HLS1 (Hidroletalus) (Prenatal) (HYLS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrolethalus syndrome, 236680, Autosomal recessive; HLS1 (Hydroletlalus) (HYLS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HYLS1	HYLS1, FLJ32915	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hydrolethalus syndrome 1 (HLS1) [MIM:236680]: A lethal syndrome characterized by polydactyly, central nervous system malformation, and hydrocephalus. The polydactyly is postaxial in the hands and preaxial in the feet. A highly characteristic hallux duplex is seen in almost no other situation. In half of the cases, a large atrioventricular communis defect of the heart is found. The pregnancy is characterized by hydramnios, which is often massive, and by preterm delivery. {ECO:0000269|PubMed:15843405}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrops Fetalis, Non-İmmün ve / Veya Atriyal Septal Defekt, 617300, Otozomal Dominant (EPHB4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrops fetalis, nonimmune, and/or atrial septal defect, 617300, Autosomal dominant (EPHB4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHB4	EPHB4, HTK, MYK1, HFASD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hidrops Fetalis, Non-İmmün ve / Veya Atriyal Septal Defekt, 617300, Otozomal Dominant (Prenatal) (EPHB4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrops fetalis, nonimmune, and/or atrial septal defect, 617300, Autosomal dominant (EPHB4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPHB4	EPHB4, HTK, MYK1, HFASD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrops, Laktik Asidoz ve Sideroblastik Anemi, 617021, Otozomal Resesif; HLASA (LARS2 gene) (Tüm Gen Dizi Analizi) (Postnatal)	Hydrops, lactic acidosis, and sideroblastic anemia, 617021, Autosomal recessive; HLASA (LARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARS2	LARS2, PRLTS4, HLASA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hidrops, Laktik Asidoz ve Sideroblastik Anemi, 617021, Otozomal Resesif; HLASA (Prenatal) (LARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydrops, lactic acidosis, and sideroblastic anemia, 617021, Autosomal recessive; HLASA (LARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARS2	LARS2, PRLTS4, HLASA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hidrokinürenüri, 236800, Otozomal Resesif (Ensefalopati, Hidrokinürenüri/arye Bağlı) (KYNU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hydroxykynureninuria, 236800, Autosomal recessive (Encephalopathy due to hydroxykynureninuria) (KYNU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KYNU	KYNU, KYNUU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydroxykynureninuria (HYXKY) [MIM:236800]: An inborn error of amino acid metabolism characterized by massive urinary excretion of large amounts of kynurenine, 3-hydroxykynurenine and xanthurenic acid. Affected individuals manifest renal tubular dysfunction, metabolic acidosis, psychomotor retardation, non- progressive encephalopathy, and muscular hypertonia. {ECO:0000269|PubMed:17334708}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hidrokinürenüri, 236800, Otozomal Resesif (Ensefalopati, Hidrokinürenüri/arye Bağlı) (Prenatal) (KYNU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hydroxykynureninuria, 236800, Autosomal recessive (Encephalopathy due to hydroxykynureninuria) (KYNU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KYNU	KYNU, KYNUU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hydroxykynureninuria (HYXKY) [MIM:236800]: An inborn error of amino acid metabolism characterized by massive urinary excretion of large amounts of kynurenine, 3-hydroxykynurenine and xanthurenic acid. Affected individuals manifest renal tubular dysfunction, metabolic acidosis, psychomotor retardation, non- progressive encephalopathy, and muscular hypertonia. {ECO:0000269|PubMed:17334708}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper IgE sendromu, otozomal resesif (9p24.3) (MLPA)	Hyper IgE syndrome, autosomal recessive (9p24.3) (MLPA)	9p24.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hiper IgE sendromu, otozomal resesif (9p24.3) (MLPA) (Prenatal)	Hyper IgE syndrome, autosomal recessive (9p24.3) (MLPA) (Prenatal)	9p24.3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper IgE sendromu, otozomal resesif/ Hiper-IgE sendromu-Otozomal dominant (17q21.2, 9p24.3) (MLPA)	Hyper IgE syndrome, autosomal recessive / hyper-IgE syndrome-Autosomal dominant (17q21.2, 9p24.3) (MLPA)	17q21.2, 9p24.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hiper IgE sendromu, otozomal resesif/ Hiper-IgE sendromu-Otozomal dominant (17q21.2, 9p24.3) (MLPA) (Prenatal)	Hyper IgE syndrome, autosomal recessive / hyper-IgE syndrome-Autosomal dominant (17q21.2, 9p24.3) (MLPA) (Prenatal)	17q21.2, 9p24.3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hiper-Igd Sendromu, 260920, Otozomal Resesif; HIDS (Periyodik Ateşli (Febril) Hiperimmunoglobulinemi D) (MVK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyper-IgD syndrome, 260920, Autosomal recessive; HIDS (Hyperimmunoglobulinemia D with periodic fever) (MVK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MVK	MVK, MVLK, POROK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiper-Igd Sendromu, 260920, Otozomal Resesif; HIDS (Periyodik Ateşli (Febril) Hiperimmunoglobulinemi D) (Prenatal) (MVK geni) (Sequence Analysis-All Coding Exons) (Prenatal)	Hyper-IgD syndrome, 260920, Autosomal recessive; HIDS (Hyperimmunoglobulinemia D with periodic fever) (MVK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MVK	MVK, MVLK, POROK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper-IgE Tekrarlayan Enfeksiyon Sendromu, 147060, Otozomal Dominant (Hiper-Ige Sendromu-Otozomal Dominant) (STAT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyper-IgE recurrent infection syndrome, 147060, Autosomal dominant (Autosomal dominant hyper-IgE syndrome) (STAT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT3	STAT3, APRF, HIES, ADMIO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperimmunoglobulin E recurrent infection syndrome, autosomal dominant (AD-HIES) [MIM:147060]: A rare disorder of immunity and connective tissue characterized by immunodeficiency, chronic eczema, recurrent Staphylococcal infections, increased serum IgE, eosinophilia, distinctive coarse facial appearance, abnormal dentition, hyperextensibility of the joints, and bone fractures. {ECO:0000269|PubMed:17676033, ECO:0000269|PubMed:17881745, ECO:0000269|PubMed:23342295}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiper-IgE Tekrarlayan Enfeksiyon Sendromu, 147060, Otozomal Dominant (Hiper-Ige Sendromu-Otozomal Dominant) (Prenatal) (STAT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyper-IgE recurrent infection syndrome, 147060, Autosomal dominant (Autosomal dominant hyper-IgE syndrome) (STAT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAT3	STAT3, APRF, HIES, ADMIO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperimmunoglobulin E recurrent infection syndrome, autosomal dominant (AD-HIES) [MIM:147060]: A rare disorder of immunity and connective tissue characterized by immunodeficiency, chronic eczema, recurrent Staphylococcal infections, increased serum IgE, eosinophilia, distinctive coarse facial appearance, abnormal dentition, hyperextensibility of the joints, and bone fractures. {ECO:0000269|PubMed:17676033, ECO:0000269|PubMed:17881745, ECO:0000269|PubMed:23342295}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper-Ige Tekrarlanan Enfeksiyon Sendromu, Otozomal Resesif, 243700, Otozomal Resesif (Kombine İmmün Yetmezlik, DOCK8 Eksikliğine Bağlı) (DOCK8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700, Autosomal recessive (Combined immunodeficiency due to DOCK8 deficiency) (DOCK8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOCK8	DOCK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperimmunoglobulin E recurrent infection syndrome, autosomal recessive (AR-HIES) [MIM:243700]: A rare disorder characterized by immunodeficiency, recurrent infections, eczema, increased serum IgE, eosinophilia and lack of connective tissue and skeletal involvement. {ECO:0000269	PubMed:19776401}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiper-Ige Tekrarlanan Enfeksiyon Sendromu, Otozomal Resesif, 243700, Otozomal Resesif (Kombine İmmün Yetmezlik, DOCK8 Eksikliğine Bağlı) (Prenatal) (DOCK8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyper-IgE recurrent infection syndrome, autosomal recessive, 243700, Autosomal recessive (Combined immunodeficiency due to DOCK8 deficiency) (DOCK8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOCK8	DOCK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperimmunoglobulin E recurrent infection syndrome, autosomal recessive (AR-HIES) [MIM:243700]: A rare disorder characterized by immunodeficiency, recurrent infections, eczema, increased serum IgE, eosinophilia and lack of connective tissue and skeletal involvement. {ECO:0000269	PubMed:19776401}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper-IgE Tekrarlanan Enfeksiyon Sendromu (DOCK8 geni) (MLPA)	Hyper-IgE recurrent infection syndrome (DOCK8 gene) (MLPA)	DOCK8	DOCK8	MLPA	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperimmunoglobulin E recurrent infection syndrome, autosomal recessive (AR-HIES) [MIM:243700]: A rare disorder characterized by immunodeficiency, recurrent infections, eczema, increased serum IgE, eosinophilia and lack of connective tissue and skeletal involvement. {ECO:0000269	PubMed:19776401}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperaldosteronizm, Ailesel, Tip 3, 613677, Otozomal Dominant; HALD3 (Ailesel Hiperaldosteronizm Tip 3) (KCNJ5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperaldosteronism, familial, type III, 613677, Autosomal dominant; HALD3 (Familial hyperaldosteronism type III) (KCNJ5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ5	KCNJ5, GIRK4, KATP1, LQT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 3 (FH3) [MIM:613677]: A form of hyperaldosteronism characterized by hypertension secondary to massive adrenal mineralocorticoid production. Like patients with familial hyperaldosteronism type 1 (glucocorticoid-remediable aldosteronism), patients with FH3 present with childhood hypertension, elevated aldosteronism levels, and high levels of the hybrid steroids 18-oxocortisol and 18-hydroxycortisol. However, hypertension and aldosteronism are not reversed by administration of exogenous glucocorticoids and patients require adrenalectomy to control hypertension. {ECO:0000269|PubMed:21311022, ECO:0000269|PubMed:22203740, ECO:0000269|PubMed:22275527, ECO:0000269|PubMed:22308486, ECO:0000269|PubMed:22848660}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperaldosteronizm, Ailesel, Tip 4, 617027, Otozomal Dominant; HALD4 (CACNA1H geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperaldosteronism, familial, type IV, 617027, Autosomal dominant; HALD4 (CACNA1H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1H	CACNA1H, EIG6, ECA6, HALD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 3 (FH3) [MIM:613677]: A form of hyperaldosteronism characterized by hypertension secondary to massive adrenal mineralocorticoid production. Like patients with familial hyperaldosteronism type 1 (glucocorticoid-remediable aldosteronism), patients with FH3 present with childhood hypertension, elevated aldosteronism levels, and high levels of the hybrid steroids 18-oxocortisol and 18-hydroxycortisol. However, hypertension and aldosteronism are not reversed by administration of exogenous glucocorticoids and patients require adrenalectomy to control hypertension. {ECO:0000269|PubMed:21311022, ECO:0000269|PubMed:22203740, ECO:0000269|PubMed:22275527, ECO:0000269|PubMed:22308486, ECO:0000269|PubMed:22848660}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperalfalipoproteinemi, 143470, Otozomal Dominant; HALP1 (Kolesterol Ester Transfer Proteini Eksikliği) (CETP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperalphalipoproteinemia, 143470, Autosomal dominant; HALP1 (Cholesterol-ester transfer protein deficiency) (CETP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CETP	CETP, HDLCQ10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperalphalipoproteinemia 1 (HALP1) [MIM:143470]: A condition characterized by high levels of high density lipoprotein (HDL) and increased HDL cholesterol levels. {ECO:0000269|PubMed:12091484, ECO:0000269|PubMed:2215607, ECO:0000269|PubMed:8408659}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperalfalipoproteinemi, 143470, Otozomal Dominant; HALP1 (Kolesterol Ester Transfer Proteini Eksikliği) (Prenatal) (CETP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperalphalipoproteinemia, 143470, Autosomal dominant; HALP1 (Cholesterol-ester transfer protein deficiency) (CETP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CETP	CETP, HDLCQ10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperalphalipoproteinemia 1 (HALP1) [MIM:143470]: A condition characterized by high levels of high density lipoprotein (HDL) and increased HDL cholesterol levels. {ECO:0000269|PubMed:12091484, ECO:0000269|PubMed:2215607, ECO:0000269|PubMed:8408659}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperamonemi, Karbonik Anhidraz VA Eksikliğine Bağlı, 615751, Otozomal Resesif (Hiperamonemik Ensefalopati, Karbonik Anhidraz VA Eksikliğine Bağlı) (CA5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751, Autosomal recessive (Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency) (CA5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CA5A	CA5A, CA5AD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperammonemia due to carbonic anhydrase VA deficiency (CA5AD) [MIM:615751]: An autosomal recessive inborn error of metabolism, clinically characterized by infantile hyperammonemic encephalopathy. Metabolic abnormalities include hypoglycemia, hyperlactatemia, metabolic acidosis and respiratory alkalosis. {ECO:0000269	PubMed:24530203}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperamonemi, Karbonik Anhidraz VA Eksikliğine Bağlı, 615751, Otozomal Resesif (Hiperamonemik Ensefalopati, Karbonik Anhidraz VA Eksikliğine Bağlı) (Prenatal) (CA5A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperammonemia due to carbonic anhydrase VA deficiency, 615751, Autosomal recessive (Hyperammonemic encephalopathy due to carbonic anhydrase VA deficiency) (CA5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CA5A	CA5A, CA5AD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperammonemia due to carbonic anhydrase VA deficiency (CA5AD) [MIM:615751]: An autosomal recessive inborn error of metabolism, clinically characterized by infantile hyperammonemic encephalopathy. Metabolic abnormalities include hypoglycemia, hyperlactatemia, metabolic acidosis and respiratory alkalosis. {ECO:0000269	PubMed:24530203}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperandrojenizm, Klassik Olmayan Tip, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (CYP21A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (CYP21A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP21A2	CYP21A2, CYP21, CA21H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOH)and 'cryptic' (asymptomatic). {ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:1406699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14675460	EDTA Blood Sample

Hiperandrojenizm, Klasik Olmayan Tip, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (MLPA)	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (MLPA)	CYP21A2	CYP21A2, CYP21, CA21H	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:1406699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	EDTA Blood Sample
Hiperandrojenizm, Klassik Olmayan Tip, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (Prenatal) (CYP21A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (CYP21A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP21A2	CYP21A2, CYP21, CA21H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:1406699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperandrojenizm, Klassik Olmayan Tip, 21-Hidroksilaz Eksikliğine Bağlı, 201910, Otozomal Resesif (Konjenital Adrenal Hiperplazi) (Prenatal) (MLPA)	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910, Autosomal recessive (Congenital adrenal hyperplasia) (Prenatal) (MLPA)	CYP21A2	CYP21A2, CYP21, CA21H	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adrenal hyperplasia 3 (AH3) [MIM:201910]: A form of congenital adrenal hyperplasia, a common recessive disease due to defective synthesis of cortisol. Congenital adrenal hyperplasia is characterized by androgen excess leading to ambiguous genitalia in affected females, rapid somatic growth during childhood in both sexes with premature closure of the epiphyses and short adult stature. Four clinical types: 'salt wasting' (SW, the most severe type), 'simple virilizing' (SV, less severely affected patients), with normal aldosterone biosynthesis, 'non-classic form' or late- onset (NC or LOH)and 'cryptic' (asymptomatic). (ECO:0000269|PubMed:10051010, ECO:0000269|PubMed:10094562, ECO:0000269|PubMed:10198222, ECO:0000269|PubMed:10364682, ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:10408778, ECO:0000269|PubMed:10408786, ECO:0000269|PubMed:10443693, ECO:0000269|PubMed:10496074, ECO:0000269|PubMed:10720040, ECO:0000269|PubMed:11232002, ECO:0000269|PubMed:11598371, ECO:0000269|PubMed:11600539, ECO:0000269|PubMed:11746135, ECO:0000269|PubMed:12213891, ECO:0000269|PubMed:12222711, ECO:0000269|PubMed:12788866, ECO:0000269|PubMed:12887291, ECO:0000269|PubMed:12915679, ECO:0000269|PubMed:1406699, ECO:0000269|PubMed:1406709, ECO:0000269|PubMed:14676460.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperapobetalipoproteinemi, Yatkınlık (PPARA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperapobetalipoproteinemia, susceptibility to (PPARA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARA	PPARA, PPAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperbilirubinemi, Ailesel Neonatal Geçici, 237900, Otozomal Resesif; HBLRTFN (Geçici Ailesel Neonatal Hiperbilirubinemi) (UGT1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperbilirubinemia, familial transient neonatal, 237900, Autosomal recessive; HBLRTFN (Transient familial neonatal hyperbilirubinemia) (UGT1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UGT1A1	UGT1A1, UGT1, GNT1, BILIQTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gilbert syndrome (GILBS) [MIM:143500]: Occurs as a consequence of reduced bilirubin transferase activity and is often detected in young adults with vague non-specific complaints. (ECO:0000269|PubMed:11013440, ECO:0000269|PubMed:12139570, ECO:0000269|PubMed:7715297. ECO:0000269|PubMed:9627603). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperbilirubinemi, Rotor Tipi, Dijenik, 237450, Dijenik Resesif (Rotor Sendromu) (SLCO1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperbilirubinemia, Rotor type, digenic, 237450, Digenic recessive (Rotor syndrome) (SLCO1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLCO1B1	SLCO1B1, LST1, OATP2, OATPC, OATP1B1, HBLRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperbilirubinemi, Rotor Tipi, Dijenik, 237450, Dijenik Resesif (Rotor Sendromu) (Prenatal) (SLCO1B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperbilirubinemia, Rotor type, digenic, 237450, Digenic recessive (Rotor syndrome) (SLCO1B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLCO1B1	SLCO1B1, LST1, OATP2, OATPC, OATP1B1, HBLRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperbilirubinemi, Rotor Tipi, Dijenik, 237450, Dijenik Resesif; HBLRR (Rotor Sendromu) (SLCO1B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperbilirubinemia, Rotor type, digenic, 237450, Digenic recessive; HBLRR (Rotor syndrome) (SLCO1B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLCO1B3	SLCO1B3, OATP8, OATP1B3, SLC21A8, HBLRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperbilirubinemia, Rotor type (HBLRR) [MIM:237450]: An autosomal recessive form of primary conjugated hyperbilirubinemia. Affected individuals develop mild jaundice not associated with hemolysis shortly after birth or in childhood. They have delayed plasma clearance of the unconjugated anionic dye bromsulphthalein and prominent urinary excretion of coproporphyrin I. Hepatic pigmentation is normal. (ECO:0000269	PubMed:22232210). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperbilirubinemi, Rotor Tipi, Dijenik, 237450, Dijenik Resesif; HBLRR (Rotor Sendromu) (Prenatal) (SLCO1B3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperbilirubinemia, Rotor type, digenic, 237450, Digenic recessive; HBLRR (Rotor syndrome) (SLCO1B3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLCO1B3	SLCO1B3, OATP8, OATP1B3, SLC21A8, HBLRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperbilirubinemia, Rotor type (HBLRR) [MIM:237450]: An autosomal recessive form of primary conjugated hyperbilirubinemia. Affected individuals develop mild jaundice not associated with hemolysis shortly after birth or in childhood. They have delayed plasma clearance of the unconjugated anionic dye bromsulphthalein and prominent urinary excretion of coproporphyrin I. Hepatic pigmentation is normal. (ECO:0000269	PubMed:22232210). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperbilverdinemi, 614156, Otozomal Resesif, Otozomal Dominant; HBLVD (Hiperbilverdinemi) (BLVRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperbilverdinemia, 614156, Autosomal recessive, Autosomal dominant; HBLVD (Hyperbilverdinemia) (BLVRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLVRA	BLVRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperbilverdinemi, 614156, Otozomal Resesif, Otozomal Dominant; HBLVD (Hiperbilverdinemi) (Prenatal) (BLVRA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperbilverdinemia, 614156, Autosomal recessive, Autosomal dominant; HBLVD (Hyperbilverdinemia) (BLVRA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BLVRA	BLVRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperkalsemi, İnfantil, 1, 143880, Otozomal Resesif; HCINF1 (Otozomal Resesif İnfantil Hiperkalsemi) (CYP24A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercalcemia, infantile, 1, 143880, Autosomal recessive; HCINF1 (Autosomal recessive infantile hypercalcemia) (CYP24A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP24A1	CYP24A1, CYP24, HCINF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypercalcemia infantile (HCAI) [MIM:143880]: A disorder characterized by abnormally high level of calcium in the blood, failure to thrive, vomiting, dehydration, and nephrocalcinosis. (ECO:0000269	PubMed:21675912). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkalsemi, İnfantil, 1, 143880, Otozomal Resesif; HCINF1 (Otozomal Resesif İnfantil Hiperkalsemi) (Prenatal) (CYP24A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypercalcemia, infantile, 1, 143880, Autosomal recessive; HCINF1 (Autosomal recessive infantile hypercalcemia) (CYP24A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP24A1	CYP24A1, CYP24, HCINF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypercalcemia infantile (HCAI) [MIM:143880]: A disorder characterized by abnormally high level of calcium in the blood, failure to thrive, vomiting, dehydration, and nephrocalcinosis. (ECO:0000269	PubMed:21675912). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperkalsemi, İnfantil, 2, 616963, Otozomal Resesif; HCINF2 (Otozomal Resesif İnfantil Hiperkalsemi) (SLC34A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercalcemia, infantile, 2, 616963, Autosomal recessive; HCINF2 (Autosomal recessive infantile hypercalcemia) (SLC34A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC34A1	SLC34A1, SLC17A2, NPT2, NPHLOP1, FRTS2, HCINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperkalsemi, İnfantil, 2, 616963, Otozomal Resesif; HCINF2 (Otozomal Resesif İnfantil Hiperkalsemi) (Prenatal) (SLC34A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypercalcemia, infantile, 2, 616963, Autosomal recessive; HCINF2 (Autosomal recessive infantile hypercalcemia) (SLC34A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC34A1	SLC34A1, SLC17A2, NPT2, NPHLOP1, FRTS2, HCINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hiperkalsiürik Hiperkalsemi (CASR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercalciuric hypercalcemia (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkalsiürik Hiperkalsemi (MLPA)	Hypercalciuric hypercalcemia (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkarotenemi ve A Vitamini Eksikliği, Otozomal Dominant, 115300, Otozomal Dominant (Kalıtsal Hiperkarotenemi ve A Vitamini Eksikliği) (BCO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300, Autosomal dominant (Hereditary hypercarotenemia and vitamin A deficiency) (BCO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCO1	BCMO1, BCDO, BCO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypercarotenemia and vitamin A deficiency, autosomal dominant (ADHVAD) [MIM:115300]: A disorder characterized by increased serum beta-carotene, decreased conversion of beta- carotene to vitamin A and decreased serum vitamin A. {ECO:0000269|PubMed:17951468}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkarotenemi ve A Vitamini Eksikliği, Otozomal Dominant, 115300, Otozomal Dominant (Kalıtsal Hiperkarotenemi ve A Vitamini Eksikliği) (BCO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypercarotenemia and vitamin A deficiency, autosomal dominant, 115300, Autosomal dominant (Hereditary hypercarotenemia and vitamin A deficiency) (BCO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCO1	BCMO1, BCDO, BCO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypercarotenemia and vitamin A deficiency, autosomal dominant (ADHVAD) [MIM:115300]: A disorder characterized by increased serum beta-carotene, decreased conversion of beta- carotene to vitamin A and decreased serum vitamin A. {ECO:0000269|PubMed:17951468}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperklorhidrosis, İzole, 143860, Otozomal Resesif (CA12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperchlorhidrosis, isolated, 143860, Autosomal recessive (CA12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CA12	CA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperkolanemi, Ailesel, 607748 (Ailesel Hiperkolanemi) (EPHX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholanemia, familial, 607748 (Familial hypercholanemia) (EPHX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHX1	EPHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hypercholanemia (FHCA) [MIM:607748]: A disorder characterized by elevated serum bile acid concentrations, itching, and fat malabsorption. {ECO:0000269|PubMed:12878321}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkolanemi, Ailesel, 607748 (Ailesel Hiperkolanemi) (BAAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholanemia, familial, 607748 (Familial hypercholanemia) (BAAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAAT	BAAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperkolanemi, Ailesel, 607748; FHCA (Ailesel Hiperkolanemi) (TJP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholanemia, familial, 607748; FHCA (Familial hypercholanemia) (TJP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TJP2	TJP2, ZO2, PFIC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hypercholanemia (FHCA) [MIM:607748]: A disorder characterized by elevated serum bile acid concentrations, itching, and fat malabsorption. {ECO:0000269|PubMed:12878321}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkolesterolemi, Ligand Kusurlu Apo B'ye Bağlı, 144010, Otozomal Dominant (APOB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, due to ligand-defective apo B, 144010, Autosomal dominant (APOB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOB	APOB, FLDB, LDLCQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial ligand-defective apolipoprotein B-100 (FDB) [MIM:144010]: Dominantly inherited disorder of lipoprotein metabolism leading to hypercholesterolemia and increased proneness to coronary artery disease (CAD). The plasma cholesterol levels are dramatically elevated due to impaired clearance of LDL particles by defective APOB/E receptors. {ECO:0000269|PubMed:2138299, ECO:0000269|PubMed:2563166, ECO:0000269|PubMed:7883971, ECO:0000269|PubMed:9259199). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperkolesterolemi, Ailesel, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (LDLR geni) (Tüm Geni Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (LDLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDLR	LDLR, FHC, FH, LDLCQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hypercholesterolemia (FH) [MIM:143890]: Common autosomal semi-dominant disease that affects about 1 in 500 individuals. The receptor defect impairs the catabolism of LDL, and the resultant elevation in plasma LDL-cholesterol promotes deposition of cholesterol in the skin (xanthelasma), tendons (xanthomas), and coronary arteries (atherosclerosis). {ECO:0000269|PubMed:10090484, ECO:0000269|PubMed:10206683, ECO:0000269|PubMed:10422803, ECO:0000269|PubMed:10447263, ECO:0000269|PubMed:10532089, ECO:0000269|PubMed:10660340, ECO:0000269|PubMed:10882754, ECO:0000269|PubMed:10978268, ECO:0000269|PubMed:10980548, ECO:0000269|PubMed:11298688, ECO:0000269|PubMed:11462246, ECO:0000269|PubMed:1446662, ECO:0000269|PubMed:1464748, ECO:0000269|PubMed:17142622, ECO:0000269|PubMed:17347910, ECO:0000269|PubMed:1867200, ECO:0000269|PubMed:19318025, ECO:0000269|PubMed:19319977, ECO:0000269|PubMed:22160468, ECO:0000269|PubMed:2318961, ECO:0000269|PubMed:24529145, ECO:0000269|PubMed:2569482, ECO:0000269|PubMed:2726768, ECO:0000269|PubMed:3263645, ECO:0000269|PubMed:3955657, ECO:0000269|PubMed:7502039.	EDTA Blood Sample
Hiperkolesterolemi, Ailesel, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (MLPA)	Hypercholesterolemia, familial, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (MLPA)	LDLR	LDLR, FHC, FH, LDLCQ2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hypercholesterolemia (FH) [MIM:143890]: Common autosomal semi-dominant disease that affects about 1 in 500 individuals. The receptor defect impairs the catabolism of LDL, and the resultant elevation in plasma LDL-cholesterol promotes deposition of cholesterol in the skin (xanthelasma), tendons (xanthomas), and coronary arteries (atherosclerosis). {ECO:0000269|PubMed:10090484, ECO:0000269|PubMed:10206683, ECO:0000269|PubMed:10422803, ECO:0000269|PubMed:10447263, ECO:0000269|PubMed:10532089, ECO:0000269|PubMed:10660340, ECO:0000269|PubMed:10882754, ECO:0000269|PubMed:10978268, ECO:0000269|PubMed:10980548, ECO:0000269|PubMed:11298688, ECO:0000269|PubMed:11462246, ECO:0000269|PubMed:1446662, ECO:0000269|PubMed:1464748, ECO:0000269|PubMed:17142622, ECO:0000269|PubMed:17347910, ECO:0000269|PubMed:1867200, ECO:0000269|PubMed:19318025, ECO:0000269|PubMed:19319977, ECO:0000269|PubMed:22160468, ECO:0000269|PubMed:2318961, ECO:0000269|PubMed:24529145, ECO:0000269|PubMed:2569482, ECO:0000269|PubMed:2726768, ECO:0000269|PubMed:3263645, ECO:0000269|PubMed:3955657, ECO:0000269|PubMed:7502039.	EDTA Blood Sample
Hiperkolesterolemi, Ailesel, 3, 603776 (PCSK9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, 3, 603776 (PCSK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCSK9	PCSK9, NARC1, HCHOLA3, FH3, LDLCQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypercholesterolemia, autosomal dominant, 3 (HCHOLA3) [MIM:603776]: A familial condition characterized by elevated circulating cholesterol contained in either low-density lipoproteins alone or also in very-low-density lipoproteins. {ECO:0000269|PubMed:12730697, ECO:0000269|PubMed:18799458, ECO:0000269|PubMed:24808179}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperkolesterolemi, Ailesel, Otozomal Resesif, 603813 (Homozigot Ailesel Hiperkolesterolemi) (LDLRAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, autosomal recessive, 603813 (Homozygous familial hypercholesterolemia) (LDLRAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDLRAP1	LDLRAP1, ARH, FHCB2, FHCB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypercholesterolemia, autosomal recessive (ARH) [MIM:603813]: A familial condition characterized by elevated circulating cholesterol contained in either low-density lipoproteins alone or also in very-low-density lipoproteins. {ECO:0000269|PubMed:11326085}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
LDLR Defekti Nedeniyle Ailesel Hiperkolesterolemi, Modifiye Edici, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (EPHX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (EPHX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHX2	EPHX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample

Hiperkolesterolemi, Ailesel,Modifiye Edici, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (GHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, modifier of, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (GHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHR	GHR, GHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperkolesterolemi, Ailesel,Modifiye Edici, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (APOA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, familial, modifier of, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (APOA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA2	APOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Hiperkolesterolemi, Yatkınlık, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (PPP1R17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, susceptibility to, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (PPP1R17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP1R17	GSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Hiperkolesterolemi, Yatkınlık, 143890, Otozomal Dominant (Homozigot Ailesel Hiperkolesterolemi) (ITIH4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypercholesterolemia, susceptibility to, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (ITIH4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITIH4	ITIH4, PK120, ITIHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Hiperşilomikronemi, Geç Başlangıçlı, 144650, Otozomal Dominant (Hiperlipoproteinemi Tip 5) (APOA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperchylomicronemia, late-onset, 144650, Autosomal dominant (Hyperlipoproteinemia type 5) (APOA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA5	APOA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperekplexia 2, Otozomal Resesif, 614619; HKPX2 (Kalıtsal Hiperekpleksia) (GLRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperekplexia 2, autosomal recessive, 614619; HKPX2 (Hereditary hyperekplexia) (GLRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLRB	GLRB, HKPX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperekplexia 2 (HKPX2) [MIM:614619]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:11929858, ECO:0000269|PubMed:21391991). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperekplexia 2, Otozomal Resesif, 614619; HKPX2 (Kalıtsal Hiperekpleksia) (MLPA)	Hyperekplexia 2, autosomal recessive, 614619; HKPX2 (Hereditary hyperekplexia) (MLPA)	GLRB	GLRB, HKPX2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hyperekplexia 2 (HKPX2) [MIM:614619]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:11929858, ECO:0000269|PubMed:21391991). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperekplexia 2, Otozomal Resesif, 614619; HKPX2 (Kalıtsal Hiperekpleksia) (Prenatal) (GLRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperekplexia 2, autosomal recessive, 614619; HKPX2 (Hereditary hyperekplexia) (GLRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLRB	GLRB, HKPX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperekplexia 2 (HKPX2) [MIM:614619]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:11929858, ECO:0000269|PubMed:21391991). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperekplexia 2, Otozomal Resesif, 614619; HKPX2 (Kalıtsal Hiperekpleksia) (Prenatal) (MLPA)	Hyperekplexia 2, autosomal recessive, 614619; HKPX2 (Hereditary hyperekplexia) (Prenatal) (MLPA)	GLRB	GLRB, HKPX2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperekplexia 2 (HKPX2) [MIM:614619]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:11929858, ECO:0000269|PubMed:21391991). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperekplexia 3, 614618, Otozomal Resesif, Otozomal Dominant; HKPX3 (Kalıtsal Hiperekpleksia) (SLC6A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperekplexia 3, 614618, Autosomal recessive, Autosomal dominant; HKPX3 (Hereditary hyperekplexia) (SLC6A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A5	SLC6A5, GLYT2, HKPX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperekplexia 3, 614618, Otozomal Resesif, Otozomal Dominant; HKPX3 (Kalıtsal Hiperekpleksia)	Hyperekplexia 3, 614618, Autosomal recessive, Autosomal dominant; HKPX3 (Hereditary hyperekplexia)	SLC6A5	SLC6A5, GLYT2, HKPX3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hiperekplexia 3, 614618, Otozomal Resesif, Otozomal Dominant; HKPX3 (Kalıtsal Hiperekpleksia) (Prenatal) (SLC6A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperekplexia 3, 614618, Autosomal recessive, Autosomal dominant; HKPX3 (Hereditary hyperekplexia) (SLC6A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A5	SLC6A5, GLYT2, HKPX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperekplexia 3, 614618, Otozomal Resesif, Otozomal Dominant; HKPX3 (Kalıtsal Hiperekpleksia) (Prenatal) (MLPA)	Hyperekplexia 3, 614618, Autosomal recessive, Autosomal dominant; HKPX3 (Hereditary hyperekplexia) (Prenatal) (MLPA)	SLC6A5	SLC6A5, GLYT2, HKPX3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperekplekia, Kalıtsal 1, Otozomal Dominant Veya Resesif 149400, Otozomal Resesif, Otozomal Dominant; HKPX1 (Kalıtsal Hiperekpleksia) (GLRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400, Autosomal recessive, Autosomal dominant; HKPX1 (Hereditary hyperekplexia) (GLRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLRA1	GLRA1, STHE, HKPX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperekplexia 1 (HKPX1) [MIM:149400]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:10514101, ECO:0000269|PubMed:7611730, ECO:0000269|PubMed:7881416, ECO:0000269|PubMed:7981700, ECO:0000269|PubMed:8298642, ECO:0000269|PubMed:8571969, ECO:0000269|PubMed:8733061, ECO:0000269|PubMed:9067762, ECO:0000269|PubMed:9920650, ECO:0000269|Ref.10). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperekplekia, Kalıtsal 1, Otozomal Dominant Veya Resesif 149400, Otozomal Resesif, Otozomal Dominant; HKPX1 (Kalıtsal Hiperekpleksia) (MLPA)	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400, Autosomal recessive, Autosomal dominant; HKPX1 (Hereditary hyperekplexia) (MLPA)	GLRA1	GLRA1, STHE, HKPX1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hyperekplexia 1 (HKPX1) [MIM:149400]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:10514101, ECO:0000269|PubMed:7611730, ECO:0000269|PubMed:7881416, ECO:0000269|PubMed:7981700, ECO:0000269|PubMed:8298642, ECO:0000269|PubMed:8571969, ECO:0000269|PubMed:8733061, ECO:0000269|PubMed:9067762, ECO:0000269|PubMed:9920650, ECO:0000269|Ref.10). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperekplekia, Kalıtsal 1, Otozomal Dominant Veya Resesif 149400, Otozomal Resesif, Otozomal Dominant; HKPX1 (Kalıtsal Hiperekpleksia) (Prenatal) (GLRA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400, Autosomal recessive, Autosomal dominant; HKPX1 (Hereditary hyperekplexia) (GLRA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLRA1	GLRA1, STHE, HKPX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperekplexia 1 (HKPX1) [MIM:149400]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:10514101, ECO:0000269|PubMed:7611730, ECO:0000269|PubMed:7881416, ECO:0000269|PubMed:7981700, ECO:0000269|PubMed:8298642, ECO:0000269|PubMed:8571969, ECO:0000269|PubMed:8733061, ECO:0000269|PubMed:9067762, ECO:0000269|PubMed:9920650, ECO:0000269|Ref.10). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperekplekia, Kalıtsal 1, Otozomal Dominant Veya Resesif 149400, Otozomal Resesif, Otozomal Dominant; HKPX1 (Kalıtsal Hiperekpleksia) (Prenatal) (MLPA)	Hyperekplexia, hereditary 1, autosomal dominant or recessive, 149400, Autosomal recessive, Autosomal dominant; HKPX1 (Hereditary hyperekplexia) (Prenatal) (MLPA)	GLRA1	GLRA1, STHE, HKPX1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperekplexia 1 (HKPX1) [MIM:149400]: A neurologic disorder characterized by muscular rigidity of central nervous system origin, particularly in the neonatal period, and by an exaggerated startle response to unexpected acoustic or tactile stimuli. (ECO:0000269|PubMed:10514101, ECO:0000269|PubMed:7611730, ECO:0000269|PubMed:7881416, ECO:0000269|PubMed:7981700, ECO:0000269|PubMed:8298642, ECO:0000269|PubMed:8571969, ECO:0000269|PubMed:8733061, ECO:0000269|PubMed:9067762, ECO:0000269|PubMed:9920650, ECO:0000269|Ref.10). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
PDGFRA (Ekzon 12 ve18) (PDGFRA geni) (Dizi Analizi) (Postnatal)	(PDGFRA gene (Exon12,18) (Sequence analysis) (PDGFRA gene) (Sequence Analysis) (Postnatal)	PDGFRA	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Gastrointestinal stromal tumor (GIST) [MIM:606764]: Common mesenchymal neoplasms arising in the gastrointestinal tract, most often in the stomach. They are histologically, immunohistochemically, and genetically different from typical leiomyomas, leiomyosarcomas, and schwannomas. Most GISTs are composed of a fairly uniform population of spindle-shaped cells. Some tumors are dominated by epithelioid cells or contain a mixture of spindle and epithelioid morphologies. Primary GISTs in the gastrointestinal tract commonly metastasize in the omentum and mesenteries, often as multiple nodules. However, primary tumors may also occur outside of the gastrointestinal tract, in other intra-abdominal locations, especially in the omentum and mesentery. (ECO:0000269|PubMed:12522257, ECO:0000269|PubMed:15928335). Note=The gene represented in this entry may be involved in disease pathogenesis. Mutations causing PDGFRA constitutive activation have been found in gastrointestinal stromal tumors lacking KIT mutations (PubMed:12522257). (ECO:0000269|PubMed:12522257).;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Hipereozinofilik Sendrom, İdyopatik, İmatinib Dirençli, 607685, Sporadik Vakalar, Somatik Mutasyon; HES (İdyopatik Hipereozinofilik Sendrom) (PDGFRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685, Isolated cases, Somatic mutation; HES (Idiopathic hypereosinophilic syndrome) (PDGFRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRA	PDGFRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperferitinemi-Katarakt Sendromu, 600886, Otozomal Dominant (Kalıtsal Hiperferitinemi-Konjenital Katarakt) (FTL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperferritinemia-cataract syndrome, 600886, Autosomal dominant (Hereditary hyperferritinemia with congenital cataracts) (FTL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTL	FTL, NBIA3, LFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. (ECO:0000269|PubMed:19176363). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperferitinemi-Katarakt Sendromu, 600886, Otozomal Dominant (Kalıtsal Hiperferitinemi-Konjenital Katarakt) (Prenatal) (FTL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperferritinemia-cataract syndrome, 600886, Autosomal dominant (Hereditary hyperferritinemia with congenital cataracts) (FTL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FTL	FTL, NBIA3, LFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. (ECO:0000269|PubMed:19176363). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hiperfibrinoliz, Allesel, PLAT Salgılanmasında Artış, 612348 (PLAT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hiperfibrinolysis, familial, due to increased release of PLAT, 612348 (PLAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAT	PLAT, TPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Increased activity of TPA results in increased fibrinolysis of fibrin blood clots that is associated with excessive bleeding. Defective release of TPA results in hypofibrinolysis that can lead to thrombosis or embolism. [ECO:0000269|PubMed:17671464]	EDTA Blood Sample
Hiperglisinemi, Laktik Asidoz ve Nöbetler, 614462, Otozomal Resesif; HGCLAS (Lipoik Asit Sentaz Eksikliği) (LIAS gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperglycinemia, lactic acidosis, and seizures, 614462, Autosomal recessive; HGCLAS (Lipoic acid synthetase deficiency) (LIAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIAS	LIAS, PDHLD, HGCLAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase lipoic acid synthetase deficiency (PDHLD) [MIM:614462]: An enzymatic defect resulting in an autosomal recessive disorder of mitochondrial metabolism. It is characterized by early-onset lactic acidosis, severe encephalomyopathy, and a pyruvate oxidation defect. Affected individuals have neonatal-onset epilepsy, poor growth, psychomotor retardation, muscular hypotonia, lactic acidosis, and elevated glycine concentration in plasma and urine. {ECO:0000269|PubMed:22152680}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperglisinemi, Laktik Asidoz ve Nöbetler, 614462, Otozomal Resesif; HGCLAS (Lipoik Asit Sentaz Eksikliği) (Prenatal) (LIAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperglycinemia, lactic acidosis, and seizures, 614462, Autosomal recessive; HGCLAS (Lipoic acid synthetase deficiency) (LIAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIAS	LIAS, PDHLD, HGCLAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase lipoic acid synthetase deficiency (PDHLD) [MIM:614462]: An enzymatic defect resulting in an autosomal recessive disorder of mitochondrial metabolism. It is characterized by early-onset lactic acidosis, severe encephalomyopathy, and a pyruvate oxidation defect. Affected individuals have neonatal-onset epilepsy, poor growth, psychomotor retardation, muscular hypotonia, lactic acidosis, and elevated glycine concentration in plasma and urine. {ECO:0000269|PubMed:22152680}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (SLC6A20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC6A20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A20	SLC6A20, XT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (SLC36A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC36A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC36A2	SLC36A2, PAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (Prenatal) (SLC6A20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC6A20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A20	SLC6A20, XT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (Prenatal) (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperglisinüri, 138500, Otozomal Dominant (Prenatal) (SLC36A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperglycinuria, 138500, Autosomal dominant (SLC36A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC36A2	SLC36A2, PAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269|PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 1, 256450, Otozomal Resesif, Otozomal Dominant; HHF1(Otozomal Dominant Hiperinsülinemik Hipoglisemi, SUR1 Eksikliğine Bağlı) (ABCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 1, 256450, Autosomal recessive, Autosomal dominant; HHF1(Autosomal dominant hyperinsulinism due to SUR1 deficiency) (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC8	ABCC8, SUR, PHHI, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 1, 256450, Otozomal Resesif, Otozomal Dominant; HHF1(Otozomal Dominant Hiperinsülinemik Hipoglisemi, SUR1 Eksikliğine Bağlı) (Prenatal) (ABCC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 1, 256450, Autosomal recessive, Autosomal dominant; HHF1(Autosomal dominant hyperinsulinism due to SUR1 deficiency) (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCC8	ABCC8, SUR, PHHI, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 2, 601820, Otozomal Resesif (Otozomal Dominant Hiperinsülinemik Hipoglisemi, Kir6.2 Eksikliğine Bağlı) (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 2, 601820, Autosomal recessive (Autosomal dominant hyperinsulinism due to Kir6.2 deficiency) (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 2 (HHF2) [MIM:601820]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:10204114, ECO:0000269|PubMed:12364426, ECO:0000269|PubMed:15562009, ECO:0000269|PubMed:15579781, ECO:0000269|PubMed:15807877, ECO:0000269|PubMed:15998776, ECO:0000269|PubMed:16332676, ECO:0000269|PubMed:16357843, ECO:0000269|PubMed:18596924, ECO:0000269|PubMed:19357197, ECO:0000269|PubMed:7847376, ECO:0000269|PubMed:8923010}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 2, 601820, Otozomal Resesif (Otozomal Dominant Hiperinsülinemik Hipoglisemi, Kir6.2 Eksikliğine Bağlı) (Prenatal) (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 2, 601820, Autosomal recessive (Autosomal dominant hyperinsulinism due to Kir6.2 deficiency) (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 2 (HHF2) [MIM:601820]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:10204114, ECO:0000269|PubMed:12364426, ECO:0000269|PubMed:15562009, ECO:0000269|PubMed:15579781, ECO:0000269|PubMed:15807877, ECO:0000269|PubMed:15998776, ECO:0000269|PubMed:16332676, ECO:0000269|PubMed:16357843, ECO:0000269|PubMed:18596924, ECO:0000269|PubMed:19357197, ECO:0000269|PubMed:7847376, ECO:0000269|PubMed:8923010}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 3, 602485, Otozomal Dominant; HHF3 (Hiperinsülinizm, Glukokinaz Eksikliğine Bağlı) (GCK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 3, 602485, Autosomal dominant; HHF3 (Hyperinsulinism due to glucokinase deficiency) (GCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCK	GCK, HHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 3, 602485, Otozomal Dominant; HHF3 (Hiperinsülinizm, Glukokinaz Eksikliğine Bağlı) (MLPA)	Hyperinsulinemic hypoglycemia, familial, 3, 602485, Autosomal dominant; HHF3 (Hyperinsulinism due to glucokinase deficiency) (MLPA)	GCK	GCK, HHF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 3, 602485, Otozomal Dominant; HHF3 (Hiperinsülinizm, Glukokinaz Eksikliğine Bağlı) (Prenatal) (GCK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 3, 602485, Autosomal dominant; HHF3 (Hyperinsulinism due to glucokinase deficiency) (GCK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GCK	GCK, HHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 3, 602485, Otozomal Dominant; HHF3 (Hiperinsülinizm, Glukokinaz Eksikliğine Bağlı) (Prenatal) (MLPA)	Hyperinsulinemic hypoglycemia, familial, 3, 602485, Autosomal dominant; HHF3 (Hyperinsulinism due to glucokinase deficiency) (Prenatal) (MLPA)	GCK	GCK, HHF3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269|PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 4, 609975, Otozomal Resesif; HHF4 (3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (Hiperinsülinizm, Kısa Zincirli 3-Hydroxylacyl-CoA Dehidrojenaz Yetmezliğine Bağlı) (HADH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 4, 609975, Autosomal recessive; HHF4 (3-hydroxyacyl dehydrogenase deficiency) (Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency) (HADH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADH	HADHSC, SCHAD, HHF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 4, 609975, Otozomal Resesif; HHF4 (3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (Hiperinsülinizm, Kısa Zincirli 3-Hydroxylacyl-CoA Dehidrojenaz Yetmezliğine Bağlı) (Prenatal) (HADH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 4, 609975, Autosomal recessive; HHF4 (3-hydroxyacyl dehydrogenase deficiency) (Hyperinsulinism due to short chain 3-hydroxylacyl-CoA dehydrogenase deficiency) (HADH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADH	HADHSC, SCHAD, HHF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Allesel, 5, 609968, Otozomal Dominant; HHF5 (Hiperinsülinizm INSR Eksikliğine Bağlı) (INSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 5, 609968, Autosomal dominant; HHF5 (Hyperinsulinism due to INSR deficiency) (INSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INSR	INSR, HHF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hiperinsülinemik Hipoglisemi, Ailesel, 5, 609968, Otozomal Dominant; HHF5 (Hiperinsülinizm İNSR Eksikliğine Bağlı (Prenatal) (INSR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 5, 609968, Autosomal dominant; HHF5 (Hyperinsulinism due to INSR deficiency) (INSR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INSR	INSR, HHF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Ailesel, 7, 610021, Otozomal Dominant; HHF7 (Egzersize Uyarılan Hiperinsülinizm) (SLC16A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinemic hypoglycemia, familial, 7, 610021, Autosomal dominant; HHF7 (Exercise-induced hyperinsulinism) (SLC16A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC16A1	SLC16A1, MCT1, HHF7, MCT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 7 (HHF7) [MIM:610021]: Dominantly inherited hypoglycemic disorder characterized by inappropriate insulin secretion during anaerobic exercise or on pyruvate load. {ECO:0000269	PubMed:17701893}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperinsülinemik Hipoglisemi, Ailesel, 7, 610021, Otozomal Dominant; HHF7 (Egzersize Uyarılan Hiperinsülinizm) (Prenatal) (SLC16A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinemic hypoglycemia, familial, 7, 610021, Autosomal dominant; HHF7 (Exercise-induced hyperinsulinism) (SLC16A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC16A1	SLC16A1, MCT1, HHF7, MCT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 7 (HHF7) [MIM:610021]: Dominantly inherited hypoglycemic disorder characterized by inappropriate insulin secretion during anaerobic exercise or on pyruvate load. {ECO:0000269	PubMed:17701893}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperinsülinizm-Hiperamonyemi Sendromu, 606762, Otozomal Dominant; HHF6 (Hiperinsülinizm-Hiperamonemi Sendromu) (GLUD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperinsulinism-hyperammonemia syndrome, 606762, Autosomal dominant; HHF6 (Hyperinsulinism-hyperammonemia syndrome) (GLUD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLUD1	GLUD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 6 (HHF6) [MIM:606762]; Familial hyperinsulinemic hypoglycemia [MIM:256450], also referred to as congenital hyperinsulinism, nesidioblastosis, or persistent hyperinsulinemic hypoglycemia of infancy (PHHI), is the most common cause of persistent hypoglycemia in infancy and is due to defective negative feedback regulation of insulin secretion by low glucose levels. In HHF6 elevated oxidation rate of glutamate to alpha-ketoglutarate stimulates insulin secretion in the pancreatic beta cells, while they impair detoxification of ammonium in the liver. {ECO:0000269|PubMed:10636977, ECO:0000269|PubMed:11214910, ECO:0000269|PubMed:11297618, ECO:0000269|PubMed:9571255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperinsülinizm-Hiperamonyemi Sendromu, 606762, Otozomal Dominant; HHF6 (Hiperinsülinizm-Hiperamonemi Sendromu) (Prenatal) (GLUD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperinsulinism-hyperammonemia syndrome, 606762, Autosomal dominant; HHF6 (Hyperinsulinism-hyperammonemia syndrome) (GLUD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLUD1	GLUD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 6 (HHF6) [MIM:606762]; Familial hyperinsulinemic hypoglycemia [MIM:256450], also referred to as congenital hyperinsulinism, nesidioblastosis, or persistent hyperinsulinemic hypoglycemia of infancy (PHHI), is the most common cause of persistent hypoglycemia in infancy and is due to defective negative feedback regulation of insulin secretion by low glucose levels. In HHF6 elevated oxidation rate of glutamate to alpha-ketoglutarate stimulates insulin secretion in the pancreatic beta cells, while they impair detoxification of ammonium in the liver. {ECO:0000269|PubMed:10636977, ECO:0000269|PubMed:11214910, ECO:0000269|PubMed:11297618, ECO:0000269|PubMed:9571255}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperkalemik Periodik Paralizi, Tip 2, Otozomal Dominant, 170500, Otozomal Dominant; HYPP (Hipokalemik Periyodik Paraliz Tip 2) (SCN4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperkalemic periodic paralysis, type 2, 170500, Autosomal dominant; HYPP (Hyperkalemic periodic paralysis) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperkalemik Periodik Paralizi, Tip 2, Otozomal Dominant, 170500, Otozomal Dominant; HYPP (Hipokalemik Periyodik Paralizi Tip 2) (MLPA)	Hyperkalemic periodic paralysis, type 2, 170500, Autosomal dominant; HYPP (Hyperkalemic periodic paralysis) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hiperkalemik Periodik Paralizi, Tip 2, Otozomal Dominant, 170500, Otozomal Dominant; HYPP (Hipokalemik Periyodik Paralizi Tip 2) (Prenatal) (SCN4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperkalemic periodic paralysis, type 2, 170500, Autosomal dominant; HYPP (Hyperkalemic periodic paralysis) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperkalemik Periodik Paralizi, Tip 2, Otozomal Dominant, 170500, Otozomal Dominant; HYPP (Hipokalemik Periyodik Paralizi Tip 2i (Prenatal) (MLPA)	Hyperkalemic periodic paralysis, type 2, 170500, Autosomal dominant; HYPP (Hyperkalemic periodic paralysis) (Prenatal) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperkeratotik Kutanöz Kapillerler- Venöz Malformasyonlar-Serebral Kapiller Malformasyonlar, 116860, Otozomal Dominant (Ailesel Serebral Kavernöz Malformasyon) (KRIT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860, Autosomal dominant (Familial cerebral cavernous malformation) (KRIT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRIT1	CCM1, CAM, KRIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral cavernous malformations 1 (CCM1) [MIM:116860]: A congenital vascular anomaly of the central nervous system that can result in hemorrhagic stroke, seizures, recurrent headaches, and focal neurologic deficits. The lesions are characterized by grossly enlarged blood vessels consisting of a single layer of endothelium and without any intervening neural tissue, ranging in diameter from a few millimeters to several centimeters. {ECO:0000269	PubMed:12172936}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperlizinemi-İzobisinemi Veya Hipervalinemi (Prenatal) (BCAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperleucinemia-isoleucinemia or hypervalinemia (BCAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCAT1	BCAT1, BCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperlizinemi-İzobisinemi Veya Hipervalinemi (Prenatal) (BCAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperleucinemia-isoleucinemia or hypervalinemia (BCAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCAT1	BCAT1, BCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperlipidemi, Ailesel Kombine, Yatkınlık, 602491 (USF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperlipidemia, familial combined, susceptibility to, 602491 (USF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USF1	USF1, HYPLIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlipidemia combined 1 (HYPLIP1) [MIM:602491]: A disorder characterized by a variable pattern of elevated levels of serum total cholesterol, triglycerides or both. It is observed in a percentage of individuals with premature coronary heart disease. {ECO:0000269	PubMed:14991056}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperlipoproteinemi, Tip 1D, 615947, Otozomal Resesif (Ailesel Şilomikronemi Sendromu) (GPIHBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperlipoproteinemia, type 1D, 615947, Autosomal recessive (Familial chylomicronemia syndrome) (GPIHBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPIHBP1	GPIHBP1, HYPL1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlipoproteinemia 1D (HLPP1D) [MIM:615947]: An autosomal recessive disorder characterized by hyperlipoproteinemia, decreased plasma LPL levels in some patients, high plasma triglyceride levels, and refractory fasting chylomicronemia. {ECO:0000269|PubMed:19304573, ECO:0000269|PubMed:20026666, ECO:0000269|PubMed:21816778}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperlipoproteinemi, Tip 1b, 207750, Otozomal Resesif (Ailesel Apolipoprotein C-II Eksikliği) (APOC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperlipoproteinemia, type Ib, 207750, Autosomal recessive (Familial apolipoprotein C-II deficiency) (APOC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOC2	APOC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlipoproteinemia 1B (HLPP1B) [MIM:207750]: Autosomal recessive trait characterized by hypertriglyceridemia, xanthomas, and increased risk of pancreatitis and early atherosclerosis. {ECO:0000269	PubMed:8323539}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperlipoproteinemi, Tip 3, 617347 (Hiperlipoproteinemi Tip 3) (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperlipoproteinemia, type III, 617347 (Hyperlipoproteinemia type 3) (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperlipoproteinemi, Tip 5 (Hiperlipoproteinemi Tip 5) (APOA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPERLIPOPROTEINEMIA, TYPE V (Hyperlipoproteinemia type 5) (APOA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA5	APOA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlipoproteinemia 5 (HLPP5) [MIM:144650]: Characterized by increased amounts of chylomicrons and very low density lipoprotein (VLDL) and decreased low density lipoprotein (LDL) and high density lipoprotein (HDL) in the plasma after a fast. Numerous conditions cause this phenotype, including insulin- dependent diabetes mellitus, contraceptive steroids, alcohol abuse, and glycogen storage disease type 1A (GSD1A). {ECO:0000269	PubMed:16200213}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperlizinemi, Tip 1, 238700, Otozomal Resesif (Hiperlizinemi) (AASS geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPERLYSINEMIA, TYPE I, 238700, Autosomal recessive (Hyperlysinemia) (AASS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AASS	AASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlysinemia, 1 (HYPLYS1) [MIM:238700]: An autosomal recessive metabolic condition with variable clinical features. Some patients present with non-specific seizures, hypotonia, or mildly delayed psychomotor development, and increased serum lysine and pipecolic acid on laboratory analysis. However, about half of the probands are reported to be asymptomatic, and hyperlysinemia is generally considered to be a benign metabolic variant. {ECO:0000269	PubMed:10775527}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperlizinemi, Tip 1, 238700, Otozomal Resesif (Hiperlizinemi) (Prenatal) (AASS geni) (Tüm Gen Dizi Analizi) (Prenatal)	HYPERLYSINEMIA, TYPE I, 238700, Autosomal recessive (Hyperlysinemia) (AASS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AASS	AASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperlysinemia, 1 (HYPLYS1) [MIM:238700]: An autosomal recessive metabolic condition with variable clinical feautures. Some patients present with non-specific seizures, hypotonia, or mildly delayed psychomotor development, and increased serum lysine and pipecolic acid on laboratory analysis. However, about half of the probands are reported to be asymptomatic, and hyperlysinemia is generally considered to be a benign metabolic variant. {ECO:0000269	PubMed:10775527}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipermanganezemi, Distoni 1, 613280, Otozomal Resesif; HMNDYT1 (Siroz-Distoni-Polisitemi-Hipermanganesemi Sendromu) (SLC30A10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypermanganesemia with dystonia 1, 613280, Autosomal recessive; HMNDYT1 (Cirrhosis-dystonia-polycythemia-hypermanganesemia syndrome) (SLC30A10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC30A10	SLC30A10, ZNT10, HMNDYT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypermanganesemia with dystonia, polycythemia, and cirrhosis (HMDPC) [MIM:613280]: A metabolic autosomal recessive disorder characterized by dystonia, parkinsonism, extrapyramidal signs, severe hypermanganesemia, polycythemia, and chronic hepatic disease, including steatosis and cirrhosis. {ECO:0000269|PubMed:22341971, ECO:0000269|PubMed:22341972}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipermanganezemi, Distoni 2, 617013, Otozomal Resesif; HMNDYT2 (SLC39A14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypermanganesemia with dystonia 2, 617013, Autosomal recessive; HMNDYT2 (SLC39A14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC39A14	SLC39A14, ZIP14, KIAA0062, HMNDYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adenosin Kinaz Eksikliğine Bağlı Hipermetioninemi, 614300, Otozomal Resesif (Hipermethioninemi Ensefalopati, Adenosin Kinaz Eksikliğine Bağlı) (ADK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypermethioninemia due to adenosine kinase deficiency, 614300, Autosomal recessive (Hypermethioninemia encephalopathy due to adenosine kinase deficiency) (ADK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADK	ADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Adenosin Kinaz Eksikliğine Bağlı Hipermetioninemi, 614300, Otozomal Resesif (Hipermethioninemi Ensefalopati, Adenosin Kinaz Eksikliğine Bağlı) (Prenatal) (ADK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypermethioninemia due to adenosine kinase deficiency, 614300, Autosomal recessive (Hypermethioninemia encephalopathy due to adenosine kinase deficiency) (ADK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADK	ADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hipermetioninemi, S-Adenozilhomosistein Hidrolaz Eksikliğine Bağlı, 613752, Otozomal Resesif (Psikomotor Retardasyon, S-Adenozilhomosistein Hidrolaz Eksikliğine Bağlı) (AHCY geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752, Autosomal recessive (Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency) (AHCY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AHCY	AHCY, SAHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency (HMAHCHD) [MIM:613752]: A metabolic disorder characterized by hypermethioninemia associated with failure to thrive, mental and motor retardation, facial dysmorphism with abnormal hair and teeth, and myocardiopathy. {ECO:0000269|PubMed:15024124, ECO:0000269|PubMed:16736098, ECO:0000269|PubMed:19177456, ECO:0000269|PubMed:20852937}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipermetioninemi, S-Adenozilhomosistein Hidrolaz Eksikliğine Bağlı, 613752, Otozomal Resesif (Psikomotor Retardasyon, S-Adenozilhomosistein Hidrolaz Eksikliğine Bağlı) (Prenatal) (AHCY geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase, 613752, Autosomal recessive (Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency) (AHCY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AHCY	AHCY, SAHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypermethioninemia with S-adenosylhomocysteine hydrolase deficiency (HMAHCHD) [MIM:613752]: A metabolic disorder characterized by hypermethioninemia associated with failure to thrive, mental and motor retardation, facial dysmorphism with abnormal hair and teeth, and myocardiopathy. {ECO:0000269|PubMed:15024124, ECO:0000269|PubMed:16736098, ECO:0000269|PubMed:19177456, ECO:0000269|PubMed:20852937}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipermetioninemi, Persistan, Otozomal Dominant, Metionin Adenosiltransferaz 1 / 3 Eksikliği. 250850, Otozomal Resesif, Otozomal Dominant (Beyin Demiyelinizasyonu, Metionin Adenoziltransferaz Eksikliğine Bağlı) (MAT1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850, Autosomal recessive, Autosomal dominant (Brain demyelination due to methionine adenosyltransferase deficiency) (MAT1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAT1A	MAT1A, MATA1, SAMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methionine adenosyltransferase deficiency (MATD) [MIM:250850]: An inborn error of metabolism resulting in isolated hypermethioninemia. Most patients have no clinical abnormalities, although some neurologic symptoms may be present in rare cases with severe loss of methionine adenosyltransferase activity. {ECO:0000269|PubMed:10677294, ECO:0000269|PubMed:7560086, ECO:0000269|PubMed:8770875, ECO:0000269|PubMed:9042912}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipermetioninemi, Persistan, Otozomal Dominant, Metionin Adenosiltransferaz 1 / 3 Eksikliği. 250850, Otozomal Resesif, Otozomal Dominant (Beyin Demiyelinizasyonu, Metionin Adenoziltransferaz Eksikliğine Bağlı) (Prenatal) (MAT1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency, 250850, Autosomal recessive, Autosomal dominant (Brain demyelination due to methionine adenosyltransferase deficiency) (MAT1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAT1A	MAT1A, MATA1, SAMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methionine adenosyltransferase deficiency (MATD) [MIM:250850]: An inborn error of metabolism resulting in isolated hypermethioninemia. Most patients have no clinical abnormalities, although some neurologic symptoms may be present in rare cases with severe loss of methionine adenosyltransferase activity. {ECO:0000269|PubMed:10677294, ECO:0000269|PubMed:7560086, ECO:0000269|PubMed:8770875, ECO:0000269|PubMed:9042912}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperornitinemi-Hiperamonemi-Homositrullinemia Sendromu, 238970, Otozomal Resesif (Hiperornitinemi-Hiperammonemi-Homositrülinüri Sendromu) (SLC25A15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970, Autosomal recessive (Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome) (SLC25A15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A15	SLC25A15, ORNT1, HHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (HHH syndrome) [MIM:238970]: Autosomal recessive disorder resulting in various neurologic symptoms, including mental retardation, spastic paraparesis with pyramidal signs, cerebellar ataxia, and episodic disturbance of consciousness or coma caused by hyperammonemia. It causes a functional impairment of the urea cycle. {ECO:0000269|PubMed:10369256, ECO:0000269|PubMed:10805333, ECO:0000269|PubMed:11552031, ECO:0000269|PubMed:11668643, ECO:0000269|PubMed:11814739, ECO:0000269|PubMed:16601889, ECO:0000269|PubMed:19242930}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperornitinemi-Hiperamonemi-Homocitrullinemia Sendromu, 238970, Otozomal Resesif (Hiperornitinemi-Hiperammonemi-Homositrülinüri Sendromu) (Prenatal) (SLC25A15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970, Autosomal recessive (Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome) (SLC25A15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A15	SLC25A15, ORNT1, HHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome (HHH syndrome) [MIM:238970]: Autosomal recessive disorder resulting in various neurologic symptoms, including mental retardation, spastic paraparesis with pyramidal signs, cerebellar ataxia, and episodic disturbance of consciousness or coma caused by hyperammonemia. It causes a functional impairment of the urea cycle. {ECO:0000269|PubMed:10369256, ECO:0000269|PubMed:10805333, ECO:0000269|PubMed:11552031, ECO:0000269|PubMed:11668643, ECO:0000269|PubMed:11814739, ECO:0000269|PubMed:16601889, ECO:0000269|PubMed:19242930}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperostozis Kortikalis Jeneralizata (Hiperostozis Kortikalis Jeneralizata (Van Buchem Hastalığı)) (SOST geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPEROSTOSIS CORTICALIS GENERALISATA (Hyperostosis corticalis generalisata) (SOST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of craniofacial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269	PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	EDTA Blood Sample
Hiperostozis Kortikalis Jeneralizata (Hiperostozis Kortikalis Jeneralizata (Van Buchem Hastalığı)) (Prenatal) (SOST geni) (Tüm Gen Dizi Analizi) (Prenatal)	HYPEROSTOSIS CORTICALIS GENERALISATA (Hyperostosis corticalis generalisata) (SOST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of craniofacial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269	PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperostozis, Endosteal, 144750, Otozomal Dominant (Otozomal Dominant Osteosklerоz, Worth Tip) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperostosis, endosteal, 144750, Autosomal dominant (Autosomal dominant osteosclerosis, Worth type) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperostozis, Endosteal, 144750, Otozomal Dominant (Otozomal Dominant Osteosklerоz, Worth Tip) (Prenatal) (LRP5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperostosis, endosteal, 144750, Autosomal dominant (Autosomal dominant osteosclerosis, Worth type) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperoksalüri, Primer, Tip 1, 259900, Otozomal Resesif; HP1 (Primer Hiperoksalüri) (AGXT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperoxaluria, primary, type 1, 259900, Autosomal recessive; HP1 (Primary hyperoxaluria) (AGXT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGXT	AGXT, SPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperoxaluria primary 1 (HP1) [MIM:259900]: An inborn error of glyoxylate metabolism characterized by increased excretion of oxalate and glycolate, and progressive tissue accumulation of insoluble calcium oxalate. Affected individuals are at risk for nephrolithiasis, nephrocalcinosis and early onset end-stage renal disease. {ECO:0000269|PubMed:10394939, ECO:0000269|PubMed:10453743, ECO:0000269|PubMed:10541294, ECO:0000269|PubMed:10862087, ECO:0000269|PubMed:12559847, ECO:0000269|PubMed:1301173, ECO:0000269|PubMed:1349575, ECO:0000269|PubMed:15253729, ECO:0000269|PubMed:15849466, ECO:0000269|PubMed:15961946, ECO:0000269|PubMed:15963748, ECO:0000269|PubMed:16971151, ECO:0000269|PubMed:170353S, ECO:0000269|PubMed:17495019, ECO:0000269|PubMed:2039493, ECO:0000269|PubMed:23229545, ECO:0000269|PubMed:24055001, ECO:0000269|PubMed:24934730, ECO:0000269|PubMed:26149463, ECO:0000269|PubMed:8101040, ECO:0000269|PubMed:9102270, ECO:0000269|PubMed:9604803}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Hiperoksalüri, Primer, Tip 1, 259900, Otozomal Resesif; HP1 (Primer Hiperoksalüri) (MLPA)	Hyperoxaluria, primary, type 1, 259900, Autosomal recessive; HP1 (Primary hyperoxaluria) (MLPA)	AGXT	AGXT, SPAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hyperoxaluria primary 1 (HP1) [MIM:259900]: An inborn error of glyoxylate metabolism characterized by increased excretion of oxalate and glycolate, and progressive tissue accumulation of insoluble calcium oxalate. Affected individuals are at risk for nephrolithiasis, nephrocalcinosis and early onset end-stage renal disease. {ECO:0000269|PubMed:10394939, ECO:0000269|PubMed:10453743, ECO:0000269|PubMed:10541294, ECO:0000269|PubMed:10862087, ECO:0000269|PubMed:12559847, ECO:0000269|PubMed:1301173, ECO:0000269|PubMed:1349575, ECO:0000269|PubMed:15253729, ECO:0000269|PubMed:15849466, ECO:0000269|PubMed:15961946, ECO:0000269|PubMed:15963748, ECO:0000269|PubMed:16971151, ECO:0000269|PubMed:1703535, ECO:0000269|PubMed:17495019, ECO:0000269|PubMed:2039493, ECO:0000269|PubMed:2329545, ECO:0000269|PubMed:24055001, ECO:0000269|PubMed:24934730, ECO:0000269|PubMed:26149463, ECO:0000269|PubMed:8101040, ECO:0000269|PubMed:9192270, ECO:0000269|PubMed:9604803}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperoksalüri, Primer, Tip 1, 259900, Otozomal Resesif; HP1 (Primer Hiperoksalüri) (Prenatal) (AGXT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperoxaluria, primary, type 1, 259900, Autosomal recessive; HP1 (Primary hyperoxaluria) (AGXT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGXT	AGXT, SPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperoxaluria primary 1 (HP1) [MIM:259900]: An inborn error of glyoxylate metabolism characterized by increased excretion of oxalate and glycolate, and progressive tissue accumulation of insoluble calcium oxalate. Affected individuals are at risk for nephrolithiasis, nephrocalcinosis and early onset end-stage renal disease. {ECO:0000269|PubMed:10394939, ECO:0000269|PubMed:10453743, ECO:0000269|PubMed:10541294, ECO:0000269|PubMed:10862087, ECO:0000269|PubMed:12559847, ECO:0000269|PubMed:1301173, ECO:0000269|PubMed:1349575, ECO:0000269|PubMed:15253729, ECO:0000269|PubMed:15849466, ECO:0000269|PubMed:15961946, ECO:0000269|PubMed:15963748, ECO:0000269|PubMed:16971151, ECO:0000269|PubMed:1703535, ECO:0000269|PubMed:17495019, ECO:0000269|PubMed:2039493, ECO:0000269|PubMed:2329545, ECO:0000269|PubMed:24055001, ECO:0000269|PubMed:24934730, ECO:0000269|PubMed:26149463, ECO:0000269|PubMed:8101040, ECO:0000269|PubMed:9192270, ECO:0000269|PubMed:9604803}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperoksalüri, Primer, Tip 1, 259900, Otozomal Resesif; HP1 (Primer Hiperoksalüri) (Prenatal) (MLPA)	Hyperoxaluria, primary, type 1, 259900, Autosomal recessive; HP1 (Primary hyperoxaluria) (Prenatal) (MLPA)	AGXT	AGXT, SPAT	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperoxaluria primary 1 (HP1) [MIM:259900]: An inborn error of glyoxylate metabolism characterized by increased excretion of oxalate and glycolate, and progressive tissue accumulation of insoluble calcium oxalate. Affected individuals are at risk for nephrolithiasis, nephrocalcinosis and early onset end-stage renal disease. {ECO:0000269|PubMed:10394939, ECO:0000269|PubMed:10453743, ECO:0000269|PubMed:10541294, ECO:0000269|PubMed:10862087, ECO:0000269|PubMed:12559847, ECO:0000269|PubMed:1301173, ECO:0000269|PubMed:1349575, ECO:0000269|PubMed:15253729, ECO:0000269|PubMed:15849466, ECO:0000269|PubMed:15961946, ECO:0000269|PubMed:15963748, ECO:0000269|PubMed:16971151, ECO:0000269|PubMed:1703535, ECO:0000269|PubMed:17495019, ECO:0000269|PubMed:2039493, ECO:0000269|PubMed:2329545, ECO:0000269|PubMed:24055001, ECO:0000269|PubMed:24934730, ECO:0000269|PubMed:26149463, ECO:0000269|PubMed:8101040, ECO:0000269|PubMed:9192270, ECO:0000269|PubMed:9604803}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperoksalüri, Primer, Tip 2, 260000, Otozomal Resesif; HP2 (Primer Hiperoksalüri) (GRHPR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperoxaluria, primary, type II, 260000, Autosomal recessive; HP2 (Primary hyperoxaluria) (GRHPR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRHPR	GRHPR, GLXR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperoksalüri, Primer, Tip 2, 260000, Otozomal Resesif; HP2 (Primer Hiperoksalüri) (MLPA)	Hyperoxaluria, primary, type II, 260000, Autosomal recessive; HP2 (Primary hyperoxaluria) (MLPA)	GRHPR	GRHPR, GLXR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hiperoksalüri, Primer, Tip 2, 260000, Otozomal Resesif; HP2 (Primer Hiperoksalüri) (Prenatal) (GRHPR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperoxaluria, primary, type II, 260000, Autosomal recessive; HP2 (Primary hyperoxaluria) (GRHPR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRHPR	GRHPR, GLXR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperoksalüri, Primer, Tip 2, 260000, Otozomal Resesif; HP2 (Primer Hiperoksalüri) (Prenatal) (MLPA)	Hyperoxaluria, primary, type II, 260000, Autosomal recessive; HP2 (Primary hyperoxaluria) (Prenatal) (MLPA)	GRHPR	GRHPR, GLXR	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hperoksalüri, Primer, Tip 3, 613616; HP3 (Primer Hiperoksalüri) (HOGA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperoxaluria, primary, type III, 613616; HP3 (Primary hyperoxaluria) (HOGA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOGA1	HOGA1, DHDPSL, HP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperoxaluria primary 3 (HP3) [MIM:613616]: A disorder phenotypically similar to hyperoxaluria type 1 and type 2. It is characterized by increase in urinary oxalate excretion and mild glycolic aciduria. Clinical manifestations include calcium oxalate urolithiasis, hematuria, pain, and/or urinary tract infection. {ECO:0000269|PubMed:20797690}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hperoksalüri, Primer, Tip 3, 613616; HP3 (Primer Hiperoksalüri) (Prenatal) (HOGA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperoxaluria, primary, type III, 613616; HP3 (Primary hyperoxaluria) (HOGA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOGA1	HOGA1, DHDPSL, HP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperoxaluria primary 3 (HP3) [MIM:613616]: A disorder phenotypically similar to hyperoxaluria type 1 and type 2. It is characterized by increase in urinary oxalate excretion and mild glycolic aciduria. Clinical manifestations include calcium oxalate urolithiasis, hematuria, pain, and/or urinary tract infection. {ECO:0000269|PubMed:20797690}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperparatiroidizm 1; Hrpt1 (Allesel İzole Hiperparatiroidizm) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPERPARATHYROIDISM 1; HRPT1 (Familial isolated hyperparathyroidism) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. {ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperparatiroidizm 2-Gene Kanserleri; Hrpt2 (Allesel İzole Hiperparatiroidizm) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPERPARATHYROIDISM 2 WITH JAW TUMORS; HRPT2 (Hyperparathyroidism-jaw tumor syndrome) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. {ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperparatiroidizm 4, 617343, Otozomal Dominant; HRPT4 (Allesel İzole Hiperparatiroidizm) (GCM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperparathyroidism 4, 617343, Autosomal dominant; HRPT4 (Familial isolated hyperparathyroidism) (GCM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCM2	GCM2, GCMB, HRPT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism, familial isolated (FIH) [MIM:146200]: A disorder characterized by hypocalcemia and hyperphosphatemia due to inadequate secretion of parathyroid hormone. Clinical features include seizures, tetany and cramps. {ECO:0000269|PubMed:15728199, ECO:0000269|PubMed:15863676, ECO:0000269|PubMed:20190276, ECO:0000269|PubMed:20463099, ECO:0000269|Ref.3}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperparatiroidizm-Çene Tümör Sendromu, 145001, Otozomal Dominant (Allesel İzole Hiperparatiroidizm) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperparathyroidism-jaw tumor syndrome, 145001, Autosomal dominant (Hyperparathyroidism-jaw tumor syndrome) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. {ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Hiperparatiroidizm, Ailesel Primer, 145000, Otozomal Dominant (Ailesel Izole Hiperparatiroidizm) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperparathyroidism, familial primary, 145000, Autosomal dominant (Familial isolated hyperparathyroidism) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. {ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperparatiroidizm, Neonatal, 239200, Otozomal Resesif, Otozomal Dominant; NSHPT (Neonatal Ağır Primer Hiperparatiroidizm) (CASR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperparathyroidism, neonatal, 239200, Autosomal recessive, Autosomal dominant; NSHPT (Neonatal severe primary hyperparathyroidism) (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperparatiroidizm, Neonatal, 239200, Otozomal Resesif, Otozomal Dominant; NSHPT (Neonatal Ağır Primer Hiperparatiroidizm) (MLPA)	Hyperparathyroidism, neonatal, 239200, Autosomal recessive, Autosomal dominant; NSHPT (Neonatal severe primary hyperparathyroidism) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, A, 261640, Otozomal Resesif; HPABHA (6-Piruvil-Tetrahidropterin Sentaz Eksikliği) (PTS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphenylalaninemia, BH4-deficient, A, 261640, Autosomal recessive; HPABHA (6-pyruvoyl-tetrahydropterin synthase deficiency) (PTS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTS	PTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, A (HPABH4A) [MIM:261640]: An autosomal recessive disorder characterized by hyperphenylalaninemia, depletion of the neurotransmitters dopamine and serotonin, and progressive cognitive and motor deficits. Neurological symptoms are unresponsive to the classic phenylalanine-low diet. {ECO:0000269|PubMed:10220141, ECO:0000269|PubMed:10585341, ECO:0000269|PubMed:10874306, ECO:0000269|PubMed:11388593, ECO:0000269|PubMed:7698774, ECO:0000269|PubMed:8178819, ECO:0000269|PubMed:8707300, ECO:0000269|PubMed:9159737, ECO:0000269|PubMed:9222757, ECO:0000269|PubMed:9450907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, A, 261640, Otozomal Resesif; HPABHA (6-Piruvil-Tetrahidropterin Sentaz Eksikliği) (Prenatal) (PTS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphenylalaninemia, BH4-deficient, A, 261640, Autosomal recessive; HPABHA (6-pyruvoyl-tetrahydropterin synthase deficiency) (PTS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTS	PTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, A (HPABH4A) [MIM:261640]: An autosomal recessive disorder characterized by hyperphenylalaninemia, depletion of the neurotransmitters dopamine and serotonin, and progressive cognitive and motor deficits. Neurological symptoms are unresponsive to the classic phenylalanine-low diet. {ECO:0000269|PubMed:10220141, ECO:0000269|PubMed:10585341, ECO:0000269|PubMed:10874306, ECO:0000269|PubMed:11388593, ECO:0000269|PubMed:7698774, ECO:0000269|PubMed:8178819, ECO:0000269|PubMed:8707300, ECO:0000269|PubMed:9159737, ECO:0000269|PubMed:9222757, ECO:0000269|PubMed:9450907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, B, 233910, Otozomal Resesif; HPABHB (GTP Siklohidrolaz I Eksikliği) (GCH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphenylalaninemia, BH4-deficient, B, 233910, Autosomal recessive; HPABHB (GTP cyclohydrolase I deficiency) (GCH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCH1	GCH1, DYT5, HPABH4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7874165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, B, 233910, Otozomal Resesif; HPABHB (GTP Siklohidrolaz I Eksikliği) (MLPA)	Hyperphenylalaninemia, BH4-deficient, B, 233910, Autosomal recessive; HPABHB (GTP cyclohydrolase I deficiency) (MLPA)	GCH1	GCH1, DYT5, HPABH4B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7874165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, B, 233910, Otozomal Resesif; HPABHB (GTP Siklohidrolaz I Eksikliği) (Prenatal) (GCH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphenylalaninemia, BH4-deficient, B, 233910, Autosomal recessive; HPABHB (GTP cyclohydrolase I deficiency) (GCH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GCH1	GCH1, DYT5, HPABH4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7874165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hiperfenilalaninemi, BH4 Eksikliği, B, 233910, Otozomal Resesif; HPABH4B (GTP Siklohidrolaz 1 Eksikliği) (Prenatal) (MLPA)	Hyperphenylalaninemia, BH4-deficient, B, 233910, Autosomal recessive; HPABH4B (GTP cyclohydrolase I deficiency) (Prenatal) (MLPA)	GCH1	GCH1, DYT5, HPABH4B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dystonia, dopa-responsive (DRD) [MIM:128230]: A form of dystonia that responds to L-DOPA treatment without side effects. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DRD typically presents in childhood with walking problems due to dystonia of the lower limbs and worsening of the dystonia towards the evening. It is characterized by postural and motor disturbances showing marked diurnal fluctuation. Torsion of the trunk is unusual. Symptoms are alleviated after sleep and aggravated by fatigue and exercise. {ECO:0000269|PubMed:10076897, ECO:0000269|PubMed:10208576, ECO:0000269|PubMed:10582612, ECO:0000269|PubMed:10825351, ECO:0000269|PubMed:10987649, ECO:0000269|PubMed:11113234, ECO:0000269|PubMed:12391354, ECO:0000269|PubMed:17101830, ECO:0000269|PubMed:7501255, ECO:0000269|PubMed:7874165, ECO:0000269|PubMed:8852666, ECO:0000269|PubMed:8957022, ECO:0000269|PubMed:9120469, ECO:0000269|PubMed:9328244, ECO:0000269|PubMed:9778264}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, C\, 261630, Otozomal Resesif; HPABH4C (Dihidropteridin Redüktaz Eksikliği) (QDPR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphenylalaninemia, BH4-deficient, C, 261630, Autosomal recessive; HPABH4C (Dihydropteridine reductase deficiency) (QDPR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	QDPR	QDPR, DHPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, C (HPABH4C) [MIM:261630]: Rare autosomal recessive disorder characterized by hyperphenylalaninemia and severe neurologic symptoms (malignant hyperphenylalaninemia) including axial hypotonia and truncal hypertonia, abnormal thermogenesis, and microcephaly. These signs are attributable to depletion of the neurotransmitters dopamine and serotonin, whose syntheses are controlled by tryptophan and tyrosine hydroxylases that use BH-4 as cofactor. Patients do not respond to phenylalanine-restricted diet. HPABH4C is lethal if untreated. {ECO:0000269|PubMed:10408783, ECO:0000269|PubMed:11153907, ECO:0000269|PubMed:2116088, ECO:0000269|PubMed:8326489, ECO:0000269|PubMed:9744478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, C\, 261630, Otozomal Resesif; HPABH4C (Dihidropteridin Redüktaz Eksikliği) (Prenatal) (QDPR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphenylalaninemia, BH4-deficient, C, 261630, Autosomal recessive; HPABH4C (Dihydropteridine reductase deficiency) (QDPR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	QDPR	QDPR, DHPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, C (HPABH4C) [MIM:261630]: Rare autosomal recessive disorder characterized by hyperphenylalaninemia and severe neurologic symptoms (malignant hyperphenylalaninemia) including axial hypotonia and truncal hypertonia, abnormal thermogenesis, and microcephaly. These signs are attributable to depletion of the neurotransmitters dopamine and serotonin, whose syntheses are controlled by tryptophan and tyrosine hydroxylases that use BH-4 as cofactor. Patients do not respond to phenylalanine-restricted diet. HPABH4C is lethal if untreated. {ECO:0000269|PubMed:10408783, ECO:0000269|PubMed:11153907, ECO:0000269|PubMed:2116088, ECO:0000269|PubMed:8326489, ECO:0000269|PubMed:9744478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, D, 264070, Otozomal Resesif; HPABH4D (Dehidrataz Eksikliği) (PCBD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphenylalaninemia, BH4-deficient, D, 264070, Autosomal recessive; HPABH4D (Dehydratase deficiency) (PCBD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCBD1	PCBD1, DCOH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, D (HPABH4D) [MIM:264070]: An autosomal recessive disease characterized by primapterinuria, a variant form of hyperphenylalaninemia defined by increased excretion of 7-substituted pterins in the urine. Patients with primapterinuria show an increased ratio of neopterin to biopterin in the urine, excretion of subnormal levels of biopterins, and normal levels of biogenic amines in cerebrospinal fluid. Neurologic signs are mild, present in the neonatal period only, and include hypotonia, delayed motor development and tremor. {ECO:0000269|PubMed:8352282, ECO:0000269|PubMed:9760199}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfenilalaninemi, BH4 Eksikliği, D, 264070, Otozomal Resesif; HPABH4D (Dehidrataz Eksikliği) (Prenatal) (PCBD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphenylalaninemia, BH4-deficient, D, 264070, Autosomal recessive; HPABH4D (Dehydratase deficiency) (PCBD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCBD1	PCBD1, DCOH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphenylalaninemia, BH4-deficient, D (HPABH4D) [MIM:264070]: An autosomal recessive disease characterized by primapterinuria, a variant form of hyperphenylalaninemia defined by increased excretion of 7-substituted pterins in the urine. Patients with primapterinuria show an increased ratio of neopterin to biopterin in the urine, excretion of subnormal levels of biopterins, and normal levels of biogenic amines in cerebrospinal fluid. Neurologic signs are mild, present in the neonatal period only, and include hypotonia, delayed motor development and tremor. {ECO:0000269|PubMed:8352282, ECO:0000269|PubMed:9760199}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfenilalaninemi, Hafif, Non-BH4 Eksikliği, 617384; HPANBH4 (DNAJC12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384; HPANBH4 (DNAJC12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC12	DNAJC12, JDP1, HPANBH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperfenilalaninemi, Hafif, Non-BH4 Eksikliği, 617384; HPANBH4 (Prenatal) (DNAJC12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphenylalaninemia, mild, non-BH4-deficient, 617384; HPANBH4 (DNAJC12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNAJC12	DNAJC12, JDP1, HPANBH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 1, 239300, Otozomal Resesif; HPMRS1 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PIGV geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 1, 239300, Autosomal recessive; HPMRS1 (Hyperphosphatasia-intellectual disability syndrome) (PIGV gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGV	PIGV, HPMRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 1 (HPMRS1) [MIM:239300]: A severe syndrome characterized by elevated serum alkaline phosphatase, severe mental retardation, seizures, hypotonia, facial dysmorphism, and hypoplastic terminal phalanges. {ECO:0000269|PubMed:20802478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 1, 239300, Otozomal Resesif; HPMRS1 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PIGV geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 1, 239300, Autosomal recessive; HPMRS1 (Hyperphosphatasia-intellectual disability syndrome) (PIGV gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGV	PIGV, HPMRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 1 (HPMRS1) [MIM:239300]: A severe syndrome characterized by elevated serum alkaline phosphatase, severe mental retardation, seizures, hypotonia, facial dysmorphism, and hypoplastic terminal phalanges. {ECO:0000269|PubMed:20802478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 2, 614749, Otozomal Resesif; HPMRS2 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PIGO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 2, 614749, Autosomal recessive; HPMRS2 (Hyperphosphatasia-intellectual disability syndrome) (PIGO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGO	PIGO, HPMRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 2 (HPMRS2) [MIM:614749]: An autosomal recessive form of intellectual disability characterized by facial dysmorphism, brachytelephalangy, and persistent elevated serum alkaline phosphatase (hyperphosphatasia). Some patients may have additional features, such as cardiac septal defects or seizures. {ECO:0000269|PubMed:22683086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 2, 614749, Otozomal Resesif; HPMRS2 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PIGO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 2, 614749, Autosomal recessive; HPMRS2 (Hyperphosphatasia-intellectual disability syndrome) (PIGO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGO	PIGO, HPMRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 2 (HPMRS2) [MIM:614749]: An autosomal recessive form of intellectual disability characterized by facial dysmorphism, brachytelephalangy, and persistent elevated serum alkaline phosphatase (hyperphosphatasia). Some patients may have additional features, such as cardiac septal defects or seizures. {ECO:0000269|PubMed:22683086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 3, 614207, Otozomal Resesif; HPMRS3 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PGAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 3, 614207, Autosomal recessive; HPMRS3 (Hyperphosphatasia-intellectual disability syndrome) (PGAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGAP2	PGAP2, FRAG1, HPMRS3, MRT17, MRT21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 3, 614207, Otozomal Resesif; HPMRS3 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PGAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 3, 614207, Autosomal recessive; HPMRS3 (Hyperphosphatasia-intellectual disability syndrome) (PGAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGAP2	PGAP2, FRAG1, HPMRS3, MRT17, MRT21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 4, 615716, Otozomal Resesif; HPMRS4 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PGAP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 4, 615716, Autosomal recessive; HPMRS4 (Hyperphosphatasia-intellectual disability syndrome) (PGAP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGAP3	PGAP3, PERLD1, CAB2, MGC9753, HPMRS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 4, 615716, Otozomal Resesif; HPMRS4 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PGAP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 4, 615716, Autosomal recessive; HPMRS4 (Hyperphosphatasia-intellectual disability syndrome) (PGAP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGAP3	PGAP3, PERLD1, CAB2, MGC9753, HPMRS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 5, 616025, Otozomal Resesif; HPMRS5 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PIGW geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 5, 616025, Autosomal recessive; HPMRS5 (Hyperphosphatasia-intellectual disability syndrome) (PIGW gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGW	PIGW, HPMRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 5 (HPMRS5) [MIM:616025]: An autosomal recessive neurologic disorder characterized by developmental delay, mental retardation, tonic seizures associated with hypsarrhythmia, dysmorphic facial features, and elevated serum alkaline phosphatase. {ECO:0000269|PubMed:24367057}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 5, 616025, Otozomal Resesif; HPMRS5 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PIGW geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 5, 616025, Autosomal recessive; HPMRS5 (Hyperphosphatasia-intellectual disability syndrome) (PIGW gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGW	PIGW, HPMRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperphosphatasia with mental retardation syndrome 5 (HPMRS5) [MIM:616025]: An autosomal recessive neurologic disorder characterized by developmental delay, mental retardation, tonic seizures associated with hypsarrhythmia, dysmorphic facial features, and elevated serum alkaline phosphatase. {ECO:0000269|PubMed:24367057}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperfosfatazia, Mental Retardasyon Sendromu 6, 616809, Otozomal Resesif; HPMRS6 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (PIGY geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperphosphatasia with mental retardation syndrome 6, 616809, Autosomal recessive; HPMRS6 (Hyperphosphatasia-intellectual disability syndrome) (PIGY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGY	PIGY, HPMRS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hiperfosfatazia, Mental Retardasyon Sendromu 6, 616809, Otozomal Resesif; HPMRS6 (Hiperfosfatazemi-Mental Retardasyon Sendromu) (Prenatal) (PIGY gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperphosphatasia with mental retardation syndrome 6, 616809, Autosomal recessive; HPMRS6 (Hyperphosphatasia-intellectual disability syndrome) (PIGY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGY	PIGY, HPMRS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperpigmentasyon ve Hipopigmentasyon, 145250, Otozomal Dominant; FPHH (Ailesel Progresif Hiper ve Hipopigmentasyon) (KITLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperpigmentation with or without hypopigmentation, 145250, Autosomal dominant; FPHH (Familial progressive hyper- and hypopigmentation) (KITLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KITLG	KITLG, MGF, SF, SCF, SHEP7, FPHH, DCUA, DFNA69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperpigmentation with or without hypopigmentation, familial progressive (FPHH) [MIM:145250]: A disorder characterized by hyperpigmented patches in the skin, present in early infancy and increasing in size and number with age. Hyperpigmentation has variable intensity, and sometimes is associated with cafe-au-lait macules and larger hypopigmented ash-leaf macules. (ECO:0000269	PubMed:19375057). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hiperproinsülinemi, 616214, Otozomal Dominant (INS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperproinsulinemia, 616214, Autosomal dominant (INS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INS	INS, MODY10, IDDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 2 (IDDM2) [MIM:125852]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269	PubMed:18192540). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperprolaktinemi, 615555, Otozomal Dominant; HPRL (Ailesel Hiperprolaktinemi) (PRLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperprolactinemia, 615555, Autosomal dominant; HPRL (Familial hyperprolactinemia) (PRLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRLR	PRLR, MFAB, HPRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple fibroadenomas of the breast (MFAB) [MIM:615554]: A benign breast disease marked by lobuloalveolar growth with abnormally high proliferation of the epithelium, and characterized by the presence of more than 3 fibroadenomas in one breast. Fibroadenomas are adenomas containing fibrous tissue. (ECO:0000269	PubMed:18779581). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperprolinemi, Tip 1, 239500, Otozomal Resesif; HYRPRO1 (Hiperprolinemi Tip 1) (PRODH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperprolinemia, type I, 239500, Autosomal recessive; HYRPRO1 (Hyperprolinemia type 1) (PRODH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRODH	PRODH, PRODH2, SCZD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperprolinemia 1 (HYRPRO1) [MIM:239500]: An inborn error of proline metabolism resulting in elevated levels of proline in the plasma and urine. The disorder is generally benign and most affected individuals are clinically asymptomatic. Some patients, however, have neurologic manifestations, including epilepsy and mental retardation. Association with certain forms of schizophrenia have been reported. (ECO:0000269|PubMed:12217952, ECO:0000269|PubMed:15662599, ECO:0000269|PubMed:17135275). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperprolinemi, Tip 2, 239510, Otozomal Resesif; HYRPRO2 (Hiperprolinemi Tip 2) (ALDH4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperprolinemia, type II, 239510, Autosomal recessive; HYRPRO2 (Hyperprolinemia type 2) (ALDH4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH4A1	ALDH4A1, ALDH4, P5CDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiperprolinemi, Tip 2, 239510, Otozomal Resesif; HYRPRO2 (Hiperprolinemi Tip 2) (Prenatal) (ALDH4A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperprolinemia, type II, 239510, Autosomal recessive; HYRPRO2 (Hyperprolinemia type 2) (ALDH4A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH4A1	ALDH4A1, ALDH4, P5CDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipersensitivite Sendromu, Karbamazepin İle İndüklenen, Yatkınlık, 608579 (Stevens-Johnson Sendromu) (HLA-A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypersensitivity syndrome, carbamazepine-induced, susceptibility to, 608579 (Stevens-Johnson syndrome) (HLA-A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-A	HLA-A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipersensitivite Sendromu, Karbamazepin İle İndüklenen, Yatkınlık, 608579 (Stevens-Johnson Sendromu) (Prenatal) (HLA-A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypersensitivity syndrome, carbamazepine-induced, susceptibility to, 608579 (Stevens-Johnson syndrome) (HLA-A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HLA-A	HLA-A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipertansiyon ve Brakidaktili Sendromu, 112410, Otozomal Dominant; HTNB (Brakidaktili-Ateriyel Hipertansiyon Sendromu) (PDE3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension and brachydactyly syndrome, 112410, Autosomal dominant; HTNB (Brachydactyly-arterial hypertension syndrome) (PDE3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE3A	PDE3A, HTNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypertension and brachydactyly syndrome (HTNB) [MIM:112410]: A syndrome characterized by brachydactyly type E, severe salt-independent but age-dependent hypertension, an increased fibroblast growth rate, neurovascular contact at the rostral-ventrolateral medulla, and altered baroreflex blood pressure regulation. It results in death from stroke before age 50 years when untreated. Brachydactyly type E is characterized by shortening of the fingers mainly in the metacarpals and metatarsals. (ECO:0000269	PubMed:25961942). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertansiyon, Diyastolik, Direnç, 608622, Otozomal Dominant (KCNMB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, diastolic, resistance to, 608622, Autosomal dominant (KCNMB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNMB1	KCNMB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Erken Başlangıçlı, Hamilelikte Ataklı Yapan, Otozomal Dominant, 605115 (Pseudohiperaldosteronizm Tip 2) (NR3C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (Pseudohyperaldosteronism type 2) (NR3C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR3C2	NR3C2, MLR, MCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudohypoaldosteronism 1, autosomal dominant (PHA1A) [MIM:177735]: A salt wasting disease resulting from target organ unresponsiveness to mineralocorticoids. PHA1A is a mild form characterized by target organ defects confined to kidney. Patients may present with neonatal renal salt wasting with hyperkalaemic acidosis despite high aldosterone levels. These patients improve with age and usually become asymptomatic without treatment. (ECO:0000269	PubMed:16972228). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipertansiyon, Esansiyel (ACSM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential (ACSM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACSM3	ACSM3, SAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Esansiyel, 145500, Multifaktöryel (PTGIS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, 145500, Multifactorial (PTGIS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTGIS	PTGIS, CYP8A1, PGIS, CYP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Esansiyel, 145500, Multifaktöryel (AGTR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, 145500, Multifactorial (AGTR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGTR1	AGTR1, AGTR1A, AT2R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). (ECO:0000269	PubMed:16116425). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertansiyon, Esansiyel, Tuza Duyarlı, 145500, Multifaktöryel (ADD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, salt-sensitive, 145500, Multifactorial (ADD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADD1	ADD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Esansiyel, Yatkınlık, 145500, Multifaktöryel (GNB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, susceptibility to, 145500, Multifactorial (GNB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB3	GNB3, CSNB1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Esansiyel, Yatkınlık, 145500, Multifaktöryel (ECE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, susceptibility to, 145500, Multifactorial (ECE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ECE1	ECE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Esansiyel, Yatkınlık, 145500, Multifaktöryel (AGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, essential, susceptibility to, 145500, Multifactorial (AGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGT	AGT, SERPINA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Essential hypertension (EHT) [MIM:145500]: A condition in which blood pressure is consistently higher than normal with no identifiable cause. (ECO:0000269|PubMed:1394429, ECO:0000269|PubMed:8513325). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Hipertansiyon, İnsülin Direnci İle İlgili, Yatkınlık, 125853, Otozomal Dominant (RETN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, insulin resistance-related, susceptibility to, 125853, Autosomal dominant (RETN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RETN	RETN, RSTN, FIZ73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hypertension, Pregnancy Induced, 189800, Otozomal Dominant (Preeklampsi) (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, pregnancy-induced, 189800, Autosomal dominant (Preeclampsia) (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Variation in NOS3 seem to be associated with susceptibility to coronary spasm. (ECO:0000269|PubMed:11740345, ECO:0000269|PubMed:9737779).	EDTA Blood Sample
Hipertansiyon, Tuza Dirençli (NPR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, salt-resistant (NPR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPR3	NPR3, ANPRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Tuza Karşı Duyarlı Esansiyel Yatkınlık, 145500, Multifaktöryel (CYP3A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, salt-sensitive essential, susceptibility to, 145500, Multifactorial (CYP3A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP3A5	CYP3A5, P450PCN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Tuza Karşı Duyarlı Esansiyel Yatkınlık, 145500, Multifaktöryel (MLPA)	Hypertension, salt-sensitive essential, susceptibility to, 145500, Multifactorial (MLPA)	CYP3A5	CYP3A5, P450PCN3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipertansiyon, Yatkınlık, 145500, Multifaktöryel (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, susceptibility to, 145500, Multifactorial (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Variation in NOS3 seem to be associated with susceptibility to coronary spasm. (ECO:0000269|PubMed:11740345, ECO:0000269|PubMed:9737779).	EDTA Blood Sample
Hipertansiyon, Yatkınlık, 145500, Multifaktöryel (NOS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertension, susceptibility to, 145500, Multifactorial (NOS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS2	NOS2A, NOS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertiroidizm, Ailesel Gestasyonel, 603373 (Ailesel Gestasyonel Hipertiroidizm) (TSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperthyroidism, familial gestational, 603373 (Familial gestational hyperthyroidism) (TSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHR	TSHR, CHNG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. (ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Hipertiroidizm, Allesel Gestasyonel, 603373 (Allesel Gestasyonel Hipertiroidizm) (MLPA)	Hyperthyroidism, familial gestational, 603373 (Familial gestational hyperthyroidism) (MLPA)	TSHR	TSHR, CHNG1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipertiroidi, Nonotoimün, 609152, Otozomal Dominant, Sporadik Vakalar (Allesel Hipertiroidi, TSH Reseptörü Mutasyonlarına Bağlı) (TSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperthyroidism, nonautoimmune, 609152, Autosomal dominant, isolated cases (Familial hyperthyroidism due to mutations in TSH receptor) (TSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHR	TSHR, CHNG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipertiroidi, Nonotoimün, 609152, Otozomal Dominant, Sporadik Vakalar (Allesel Hipertiroidi, TSH Reseptörü Mutasyonlarına Bağlı) (MLPA)	Hyperthyroidism, nonautoimmune, 609152, Autosomal dominant, isolated cases (Familial hyperthyroidism due to mutations in TSH receptor) (MLPA)	TSHR	TSHR, CHNG1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipertrikozis Terminalis, Jeneralize, ± Gingival Hiperplazi, 135400, Otozomal Resesif (Dişeti Fibromatozu-Hipertrikozis Sendromu) (2200)	Hypertrichosis terminalis, generalized, with or without gingival hyperplasia, 135400, Autosomal recessive (Gingival fibromatosis-hypertrichosis syndrome) (440)	.	HTGH, DEL17q24	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Hipertrikoz, Konjenital Jeneralize, 307150, X'e Bağlı Dominant (Hipertrikozis Lanuginosa Konjenital (2200)	Hypertrichosis, congenital generalized, 307150, X-linked dominant (Hypertrichosis lanuginosa congenita) (440)	Array-CGH	HTC2, HCG, CGH, CXINSp27.1,	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Hipertrikotik Osteokondrodisplazi, 239850, Otozomal Dominant (Hipertrikotik Osteokondrodisplazi, Cantu Tipi) (ABCC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertrichotic osteochondrodysplasia, 239850, Autosomal dominant (Hypertrichotic osteochondrodysplasia, Cantu type) (ABCC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC9	ABCC9, SUR2, CMD1O, ATFB12, CANTU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypertrichotic osteochondrodysplasia (HTOCD) [MIM:239850]: A rare disorder characterized by congenital hypertrichosis, neonatal macrosomia, a distinct osteochondrodysplasia, and cardiomegaly. The hypertrichosis leads to thick scalp hair, which extends onto the forehead, and a general increase in body hair. In addition, macrocephaly and coarse facial features, including a broad nasal bridge, epicanthal folds, a wide mouth, and full lips, can be suggestive of a storage disorder. About half of affected individuals are macrosomic and edematous at birth, whereas in childhood they usually have a muscular appearance with little subcutaneous fat. Thickened calvarium, narrow thorax, wide ribs, flattened or ovoid vertebral bodies, coxa valga, osteopenia, enlarged medullary canals, and metaphyseal widening of long bones have been reported. Cardiac manifestations such as patent ductus arteriosus, ventricular hypertrophy, pulmonary hypertension, and pericardial effusions are present in approximately 80% of cases. Motor development is usually delayed due to hypotonia. Most patients have a mild speech delay, and a small percentage have learning difficulties or intellectual disability. {ECO:0000269|PubMed:22608503, ECO:0000269|PubMed:22610116}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertrikotik Osteokondrodisplazi, 239850, Otozomal Dominant (Hipertrikotik Osteokondrodisplazi, Cantu Tipi) (Prenatal) (ABCC9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypertrichotic osteochondrodysplasia, 239850, Autosomal dominant (Hypertrichotic osteochondrodysplasia, Cantu type) (ABCC9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCC9	ABCC9, SUR2, CMD1O, ATFB12, CANTU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypertrichotic osteochondrodysplasia (HTOCD) [MIM:239850]: A rare disorder characterized by congenital hypertrichosis, neonatal macrosomia, a distinct osteochondrodysplasia, and cardiomegaly. The hypertrichosis leads to thick scalp hair, which extends onto the forehead, and a general increase in body hair. In addition, macrocephaly and coarse facial features, including a broad nasal bridge, epicanthal folds, a wide mouth, and full lips, can be suggestive of a storage disorder. About half of affected individuals are macrosomic and edematous at birth, whereas in childhood they usually have a muscular appearance with little subcutaneous fat. Thickened calvarium, narrow thorax, wide ribs, flattened or ovoid vertebral bodies, coxa valga, osteopenia, enlarged medullary canals, and metaphyseal widening of long bones have been reported. Cardiac manifestations such as patent ductus arteriosus, ventricular hypertrophy, pulmonary hypertension, and pericardial effusions are present in approximately 80% of cases. Motor development is usually delayed due to hypotonia. Most patients have a mild speech delay, and a small percentage have learning difficulties or intellectual disability. {ECO:0000269|PubMed:22608503, ECO:0000269|PubMed:22610116}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipertrigliseridemi, Yatkınlık, 145750, Otozomal Dominant (LIPI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertriglyceridemia, susceptibility to, 145750, Autosomal dominant (LIPI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPI	LIPI, LPDL, PRED5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipertrigliseridemi, Yatkınlık, 145750, Otozomal Dominant (Hiperlipoproteinemi Tip 5) (APOA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertriglyceridemia, susceptibility to, 145750, Autosomal dominant (Hyperlipoproteinemia type 5) (APOA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA5	APOA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlipoproteinemia 5 (HLPP5) [MIM:144650]: Characterized by increased amounts of chylomicrons and very low density lipoprotein (VLDL) and decreased low density lipoprotein (LDL) and high density lipoprotein (HDL) in the plasma after a fast. Numerous conditions cause this phenotype, including insulin- dependent diabetes mellitus, contraceptive steroids, alcohol abuse, and glycogen storage disease type 1A (GSD1A). {ECO:0000269	PubMed:16200213}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertrigliseridemi, Geçici İnfantil, 614480, Otozomal Resesif; HTGTI (Geçici İnfantil Hipertrigliseridemi ve Hepatosteatoz) (GPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertriglyceridemia, transient infantile, 614480, Autosomal recessive; HTGTI (Transient infantile hypertriglyceridemia and hepatosteatosis) (GPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPD1	GPD1, HTGTI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypertriglyceridemia, transient infantile (HTGTI) [MIM:614480]: An autosomal recessive disorder characterized by onset of moderate to severe transient hypertriglyceridemia in infancy that normalizes with age. The hypertriglyceridemia is associated with hepatomegaly, moderately elevated transaminases, persistent fatty liver, and the development of hepatic fibrosis. {ECO:0000269|PubMed:22226083, ECO:0000269|PubMed:24549054}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertrofik Osteoartropati, Primer, Otozomal Resesif 1, 259100, Otozomal Resesif; PHOAR1 (Kraniyo-Osteoartropati) (HPGD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100, Autosomal recessive; PHOAR1 (Cranio-osteoarthropathy) (HPGD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPGD	HPGD, PGDH1, PHOAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Isolated congenital nail clubbing (ICNC) [MIM:119900]: A rare genodermatosis characterized by enlargement of the nail plate and terminal segments of the fingers and toes, resulting from proliferation of the connective tissues between the nail matrix and the distal phalanx. It is usually symmetrical and bilateral (in some cases unilateral). In nail clubbing usually the distal end of the nail matrix is relatively high compared to the proximal end, while the nail plate is complete but its dimensions and diameter more or less vary in comparison to normal. There may be different fingers and toes involved to varying degrees. Some fingers or toes are spared, but the thumbs are almost always involved. {ECO:0000269	PubMed:18805827}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipertrofik Osteoartropati, Primer, Otozomal Resesif 2, 614441, Otozomal Resesif; PHOAR2 (Paklidermoperiostozis) (SLCO2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertrophic osteoarthropathy, primary, autosomal recessive 2, 614441, Autosomal recessive; PHOAR2 (Pachydermoperiostosis) (SLCO2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLCO2A1	SLCO2A1, OATP2A1, PGT, SLC21A2, PHOAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hipertripsinemi Neonatal (Kistik Fibroz) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypertrypsinemia, neonatal (Cystic fibrosis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Hipertripsinemi Neonatal (Kistik Fibroz) (MLPA)	Hypertrypsinemia, neonatal (Cystic fibrosis) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Hipertripsinemi Neonatal (Kistik Fibroz) (Prenatal) (CFTR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypertrypsinemia, neonatal (Cystic fibrosis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipertripsinemi Neonatal (Kistik Fibroz) (Prenatal) (MLPA)	Hypertrypsinemia, neonatal (Cystic fibrosis) (Prenatal) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperürisemi, Pulmoner Hipertansiyon, Böbrek Yetmezliği, ve Alkaloz 613845, Otozomal Resesif (Hiperürisemi-Pulmoner Hipertansiyon-Böbrek Yetmezliği-Alkaloz Sendromu) (SARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845, Autosomal recessive (Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome) (SARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SARS2	SARS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome (HUPRAS) [MIM:613845]: A multisystem disorder characterized by onset in infancy of progressive renal failure leading to electrolyte imbalances, metabolic alkalosis, pulmonary hypertension, hypotonia, and delayed development. Affected individuals are born prematurely. {ECO:0000269|PubMed:21255763}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Hiperürisemi, Pulmoner Hipertansiyon, Böbrek Yetmezliği, ve Alkaloz, 613845, Otozomal Resesif (Hiperürisemi-Pulmoner Hipertansiyon-Böbrek Yetmezliği-Alkaloz Sendromu) (Prenatal) (SARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis, 613845, Autosomal recessive (Hyperuricemia-pulmonary hypertension-renal failure-alkalosis syndrome) (SARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SARS2	SARS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperuricemia, pulmonary hypertension, renal failure, and alkalosis syndrome (HUPRAS) [MIM:613845]: A multisystem disorder characterized by onset in infancy of progressive renal failure leading to electrolyte imbalances, metabolic alkalosis, pulmonary hypertension, hypotonia, and delayed development. Affected individuals are born prematurely. {ECO:0000269|PubMed:21255763}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiperürisemik Nefropati, Ailesel Jüvenil 1, 162000, Otozomal Dominant; HNFJ1 (Ailesel Jüvenil Hiperürisemik Nefropati Tip 1) (UMOD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperuricemic nephropathy, familial juvenile 1, 162000, Autosomal dominant; HNFJ1 (Familial juvenile hyperuricemic nephropathy type 1) (UMOD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UMOD	UMOD, HNFJ1, FJHN, MCKD2, ADMCKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial juvenile hyperuricemic nephropathy 1 (HNFJ1) [MIM:162000]: A renal disease characterized by juvenile onset of hyperuricemia, polyuria, progressive renal failure, and gout. The disease is associated with interstitial pathological changes resulting in fibrosis. {ECO:0000269|PubMed:12471200, ECO:0000269|PubMed:12629136, ECO:0000269|PubMed:12900848, ECO:0000269|PubMed:14569098, ECO:0000269|PubMed:14570709, ECO:0000269|PubMed:15086896, ECO:0000269|PubMed:15575003, ECO:0000269|PubMed:15983957, ECO:0000269|PubMed:17010121, ECO:0000269|PubMed:21060763, ECO:0000269|PubMed:22776760, ECO:0000269|PubMed:23197950, ECO:0000269|PubMed:23988501}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperürisemik Nefropati, Ailesel Jüvenil 2, 613092, Otozomal Dominant; HNFJ2 (Hiperürisemi-Anemi-Böbrek Yetmezliği Sendromu) (REN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperuricemic nephropathy, familial juvenile 2, 613092, Autosomal dominant; HNFJ2 (Hyperuricemia-anemia-renal failure syndrome) (REN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REN	REN, HNFJ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269|PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hiperürisemik Nefropati, Ailesel Jüvenil, 4, 617056, Otozomal Dominant (SEC61A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hyperuricemic nephropathy, familial juvenile, 4, 617056, Autosomal dominant (SEC61A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC61A1	SEC61A1, SEC61, HNFJ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipervalinemi Veya Hiperlözin-İzolösinemi (BCAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypervalinemia or hyperleucine-isoleucinemia (BCAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCAT2	BCAT2, BCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipervalinemi Veya Hiperlözin-İzolösinemi (Prenatal) (BCAT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypervalinemia or hyperleucine-isoleucinemia (BCAT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCAT2	BCAT2, BCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hipoaldosteronizm, Konjenital, CMO 1 Eksikliğine Bağlı, 203400, Otozomal Resesif (Alesel Hipoaldosteronizm) (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400, Autosomal recessive (Familial hypoaldosteronism) (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.	EDTA Blood Sample
Hipoaldosteronizm, Konjenital, CMO 2 Eksikliği Nedeniyle, 610600, Otozomal Resesif (Alesel Hiperreninemik Hipoaldosteronizm Tip 1) (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600, Autosomal recessive (Familial hyperreninemic hypoaldosteronism type 1) (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.	EDTA Blood Sample
Hipoalfalipoproteinemi, 604091 (Apolipoprotein A-I Eksikliği) (APOA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoalphalipoproteinemia, 604091 (Apolipoprotein A-I deficiency) (APOA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOA1	APOA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 6 (AMYL6) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269|PubMed:1502149, ECO:0000269|PubMed:2123470, ECO:0000269|PubMed:3142462, ECO:0000269|PubMed:8208902}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoalfalipoproteinemi, Primer (Apolipoprotein A-I Eksikliği) (ABCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	HYPOALPHALIPOPROTEINEMIA, PRIMARY (Apolipoprotein A-I deficiency) (ABCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA1	ABCA1, ABC1, HDLDT1, TGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: High density lipoprotein deficiency 1 (HDLD1) [MIM:205400]: Recessive disorder characterized by absence of high density lipoprotein (HDL) cholesterol from plasma, accumulation of cholesteryl esters, premature coronary artery disease (CAD), hepatosplenomegaly, recurrent peripheral neuropathy and progressive muscle wasting and weakness. {ECO:0000269|PubMed:10431236, ECO:0000269|PubMed:10431237, ECO:0000269|PubMed:10706591, ECO:0000269|PubMed:10938021, ECO:0000269|PubMed:11086027, ECO:0000269|PubMed:11257260, ECO:0000269|PubMed:11476961, ECO:0000269|PubMed:11476965, ECO:0000269|PubMed:11785958, ECO:0000269|PubMed:12111371, ECO:0000269|PubMed:12111381, ECO:0000269|PubMed:12407001, ECO:0000269|PubMed:14576201, ECO:0000269|PubMed:15019541, ECO:0000269|PubMed:15158913, ECO:0000269|PubMed:15262183, ECO:0000269|PubMed:15297675, ECO:0000269|PubMed:15520867}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipobetalipoproteinemi, 615558, Otozomal Resesif (APOB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypobetalipoproteinemia, 615558, Autosomal recessive (APOB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOB	APOB, FLDB, LDLCQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial ligand-defective apolipoprotein B-100 (FDB) [MIM:144010]: Dominantly inherited disorder of lipoprotein metabolism leading to hypercholesterolemia and increased proneness to coronary artery disease (CAD). The plasma cholesterol levels are dramatically elevated due to impaired clearance of LDL particles by defective APOB/E receptors. {ECO:0000269|PubMed:21382890, ECO:0000269|PubMed:2563166, ECO:0000269|PubMed:7883971, ECO:0000269|PubMed:9259190}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipobetalipoproteinemi, Allesel, 2, 605019, Otozomal Resesif; FHBL2 (ANGPTL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypobetalipoproteinemia, familial, 2, 605019, Autosomal recessive; FHBL2 (ANGPTL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANGPTL3	ANGPTL3, ANGPTS, FHBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipokalsemi, Otozomal Dominant 2, 615361, Otozomal Dominant Hipokalsemi) (GNA11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypocalcemia, autosomal dominant 2, 615361, Autosomal dominant; HYPOC2 (Autosomal dominant hypocalcemia) (GNA11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNA11	GNA11, HHC2, HYPOC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 2 (HHC2) [MIM:145981]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:23802516}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsemi, Otozomal Dominant, Bartter Sendromu, 601198, Otozomal Dominant; HYPOC1 (Bartter Sendromu-Hipokalsemi) (CASR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198, Autosomal dominant; HYPOC1 (Bartter syndrome with hypocalcemia) (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsemi, Otozomal Dominant, Bartter Sendromu, 601198, Otozomal Dominant; HYPOC1 (Bartter Sendromu-Hipokalsemi) (MLPA)	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198, Autosomal dominant; HYPOC1 (Bartter syndrome with hypocalcemia) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsemi, Otozomal Dominant, Bartter Sendromu, 601198, Otozomal Dominant; HYPOC1 (Bartter Sendromu-Hipokalsemi) (Prenatal) (CASR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198, Autosomal dominant; HYPOC1 (Bartter syndrome with hypocalcemia) (CASR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hipokalsemi, Otozomal Dominant, Bartter Sendromu, 601198, Otozomal Dominant; HYPOC1 (Bartter Sendromu-Hipokalsemi) (Prenatal) (MLPA)	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198, Autosomal dominant; HYPOC1 (Bartter syndrome with hypocalcemia) (Prenatal) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipokalsiürik Hiperkalsemi, Tip 1, 145980, Otozomal Dominant; HHC1 (Allesel Hipokalsiürik Hiperkalsemi) (CASR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hypocalciuric hypercalcemia, type I, 145980, Autosomal dominant; HHC1 (Familial hypocalciuric hypercalcemia) (CASR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsiürik Hiperkalsemi, Tip 1, 145980, Otozomal Dominant; HHC1 (Allesel Hipokalsiürik Hiperkalsemi) (MLPA)	Hypocalciuric hypercalcemia, type I, 145980, Autosomal dominant; HHC1 (Familial hypocalciuric hypercalcemia) (MLPA)	CASR	CASR, HHC1, PCAR1, FIH, EIG8, HYPOC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 1 (HHC1) [MIM:145980]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:11762699, ECO:0000269|PubMed:15572418, ECO:0000269|PubMed:15579740, ECO:0000269|PubMed:15879434, ECO:0000269|PubMed:16598859, ECO:0000269|PubMed:17473068, ECO:0000269|PubMed:17698911, ECO:0000269|PubMed:21643651, ECO:0000269|PubMed:7673400, ECO:0000269|PubMed:7726161, ECO:0000269|PubMed:7916660, ECO:0000269|PubMed:9298824}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsiürik Hiperkalsemi, Otozomal Dominant Tip 2, 145981; HHC2 (Allesel Hipokalsiürik Hiperkalsemi) (GNA11 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hypocalciuric hypercalcemia, type II, 145981, Autosomal dominant; HHC2 (Familial hypocalciuric hypercalcemia) (GNA11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNA11	GNA11, HHC2, HYPOC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 2 (HHC2) [MIM:145981]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269	PubMed:23802516}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalsiürik Hiperkalsemi, Otozomal Dominant Tip 3, 600740; HHC3 (Allesel Hipokalsiürik Hiperkalsemi) (AP2S1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hypocalciuric hypercalcemia, type III, 600740, Autosomal dominant; HHC3 (Familial hypocalciuric hypercalcemia) (AP2S1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP2S1	AP2S1, CLAPS2, AP17, HHC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypocalciuric hypercalcemia, familial 3 (HHC3) [MIM:600740]: A form of hypocalciuric hypercalcemia, a disorder of mineral homeostasis that is transmitted as an autosomal dominant trait with a high degree of penetrance. It is characterized biochemically by lifelong elevation of serum calcium concentrations and is associated with inappropriately low urinary calcium excretion and a normal or mildly elevated circulating parathyroid hormone level. Hypermagnesemia is typically present. Affected individuals are usually asymptomatic and the disorder is considered benign. However, chondrocalcinosis and pancreatitis occur in some adults. {ECO:0000269|PubMed:23222959, ECO:0000269|PubMed:24081735}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipokondroplazi, 146000, Otozomal Dominant; HCH (Hipokondroplazi) (FGFR3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hypochondroplasia, 146000, Autosomal dominant; HCH (Hypochondroplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokondroplazi, 146000, Otozomal Dominant; HCH (Hipokondroplazi) (MLPA)	Hypochondroplasia, 146000, Autosomal dominant; HCH (Hypochondroplasia) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokondroplazi, 146000, Otozomal Dominant; HCH (Hipokondroplazi) (Prenatal) (FGFR3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Hypochondroplasia, 146000, Autosomal dominant; HCH (Hypochondroplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipokondroplazi, 146000, Otozomal Dominant; HCH (Hipokondroplazi) (Prenatal) (MLPA)	Hypochondroplasia, 146000, Autosomal dominant; HCH (Hypochondroplasia) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipokrom Mikrositik Anemi (MLPA)	Hypochromic microcytic anemia (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipokrom Mikrositik Anemi (Prenatal) (MLPA)	Hypochromic microcytic anemia (Prenatal) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipodisfibrinojenemi, 616004 (Allesel Hipodisfibrinojenemi) (FGG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Hypodysfibrinogenemia, 616004 (Familial hypodysfibrinogenemia) (FGG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample

Hipodisfibrinojenemi, 616004 (Allesel Hipodisfibrinojenemi) (Prenatal) (FGG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypodysfibrinogenemia, 616004 (Familial hypodysfibrinogenemia) (FGG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipodisfibrinojenemi, Konjenital, 616004 (Allesel Hipodisfibrinojenemi) (FGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypodysfibrinogenemia, congenital, 616004 (Familial hypodysfibrinogenemia) (FGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269	PubMed:8097946}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipodisfibrinojenemi, Konjenital, 616004 (Allesel Hipodisfibrinojenemi) (Prenatal) (FGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypodysfibrinogenemia, congenital, 616004 (Familial hypodysfibrinogenemia) (FGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGA	FGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Amyloidosis 8 (AMYL8) [MIM:105200]: A hereditary generalized amyloidosis due to deposition of apolipoprotein A1, fibrinogen and lysozyme amyloids. Viscera are particularly affected. There is no involvement of the nervous system. Clinical features include renal amyloidosis resulting in nephrotic syndrome, arterial hypertension, hepatosplenomegaly, cholestasis, petechial skin rash. {ECO:0000269	PubMed:8097946}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (FGG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypofibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Hipofibrinojenemi, Konjenital, 202400, Otozomal Resesif (Konjenital Fibrinojen Eksikliği) (Prenatal) (FGG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypofibrinogenemia, congenital, 202400, Autosomal recessive (Congenital fibrinogen deficiency) (FGG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGG	FGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofibrinojenemi, Konjenital, 202400, Otozomal Resesif (Allesel Hipodisfibrinojenemi) (FGB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypofibrinogenemia, congenital, 202400, Autosomal recessive (Familial hypodysfibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269	PubMed:10666208, ECO:0000269	PubMed:11468164, ECO:0000269	PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	EDTA Blood Sample
Hipofibrinojenemi, Konjenital, 202400, Otozomal Resesif (Allesel Hipodisfibrinojenemi) (Prenatal) (FGB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypofibrinogenemia, congenital, 202400, Autosomal recessive (Familial hypodysfibrinogenemia) (FGB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGB	FGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital afibrinogenemia (CAFBN) [MIM:202400]: Rare autosomal recessive disorder is characterized by bleeding that varies from mild to severe and by complete absence or extremely low levels of plasma and platelet fibrinogen. {ECO:0000269|PubMed:10666208, ECO:0000269|PubMed:11468164, ECO:0000269|PubMed:25427968}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients with congenital fibrinogen abnormalities can manifest different clinical pictures. Some cases are clinically silent, some show a tendency toward bleeding and some show a predisposition for thrombosis with or without bleeding.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İrritanlı Hipoglisemi, Lösin Duyarlı, 240800, Otozomal Dominant (ABCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoglycemia of infancy, leucine-sensitive, 240800, Autosomal dominant (ABCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC8	ABCC8, SUR, PHHI, SUR1, HHF1, TNDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Leucine-induced hypoglycemia (LIH) [MIM:240800]: Rare cause of hypoglycemia and is described as a condition in which symptomatic hypoglycemia is provoked by high protein feedings. Hypoglycemia is also elicited by administration of oral or intravenous infusions of a single amino acid, leucine. {ECO:0000269	PubMed:15356046}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 1, ± Anozmi (Kallmann Sendromu 1) 308700, X'e Bağlı; HH1 (Kallmann Sendromu) (ANOS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700, X-linked; HH1 (Kallmann syndrome) (ANOS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANOS1	KAL1, KMS, ADMLX, HH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 1 with or without anosmia (HH1) [MIM:308700]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nHH). {ECO:0000269|PubMed:11297579, ECO:0000269|PubMed:15001591, ECO:0000269|PubMed:15605412, ECO:0000269|PubMed:17054399, ECO:0000269|PubMed:17213338, ECO:0000269|PubMed:17223984, ECO:0000269|PubMed:20530987, ECO:0000269|PubMed:21168128, ECO:0000269|PubMed:23643382, ECO:0000269|PubMed:25077900, ECO:0000269|PubMed:8504298, ECO:0000269|PubMed:8989261, ECO:0000269|PubMed:9589672}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 10, ± Anozmi, 614839, Otozomal Resesif; HH10 (Normozmik Konjenital Hipogonadotropik Hipogonadizm) (TAC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839, Autosomal recessive; HH10 (Normosmic congenital hypogonadotropic hypogonadism) (TAC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAC3	TAC3, NKNB, HH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 10 with or without anosmia (HH10) [MIM:614839]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nHH). {ECO:0000269	PubMed:19079066, ECO:0000269	PubMed:23643382}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 11, ± Anozmi, 614840, Otozomal Resesif; HH11 (Kallmann Sendromu) (TACR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840, Autosomal recessive; HH11 (Kallmann syndrome) (TACR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TACR3	TACR3, NK3R, HH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 11 with or without anosmia (HH11) [MIM:614840]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nHH). {ECO:0000269|PubMed:19079066, ECO:0000269|PubMed:23643382, ECO:0000269|PubMed:25077900}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in TACR3 as well as in other HH-associated genes including FGFR1, SPRY4 and KAL1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}..	EDTA Blood Sample

Hipogonadotropik Hipogonadizm 12,± Anozmi, 614841, Otozomal Resesif; HH12 (Normozmik Konjenital Hipogonadotropik Hipogonadizmi) (GNRH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841, Autosomal recessive; HH12 (Normosmic congenital hypogonadotropic hypogonadism) (GNRH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNRH1	GNRH1, LNRH, HH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 12 with or without anosmia (HH12) [MIM:614841]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:19535795, ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in GNRH1 as well as in other HH- associated genes including PROKR2 and FGFR1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 12,± Anozmi, 614841, Otozomal Resesif; HH12 (Normozmik Konjenital Hipogonadotropik Hipogonadizmi) (MLPA)	Hypogonadotropic hypogonadism 12 with or without anosmia, 614841, Autosomal recessive; HH12 (Normosmic congenital hypogonadotropic hypogonadism) (MLPA)	GNRH1	GNRH1, LNRH, HH12	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypogonadotropic hypogonadism 12 with or without anosmia (HH12) [MIM:614841]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:19535795, ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in GNRH1 as well as in other HH- associated genes including PROKR2 and FGFR1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 13 ± Anozmi, 614842, Otozomal Resesif; HH13 (Normozmik Konjenital Hipogonadotropik Hipogonadizmi (KISS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 13 with or without anosmia, 614842, Autosomal recessive; HH13 (Normosmic congenital hypogonadotropic hypogonadism) (KISS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KISS1	KISS1, HH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 13 with or without anosmia (HH13) [MIM:614842]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269	PubMed:22335740}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 14 ± Anozmi, 614858, Otozomal Dominant; HH14 (Kallmann Sendromu) (WDR11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858, Autosomal dominant; HH14 (Kallmann syndrome) (WDR11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR11	WDR11, DR11, KIAA1351, BRWD2, HH14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 14 with or without anosmia (HH14) [MIM:614858]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269	PubMed:20887964}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 15 ± Anozmi, 614880, Otozomal Dominant; HH15 (Kallmann Sendromu) (HS6ST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 15 with or without anosmia, 614880, Autosomal dominant; HH15 (Kallmann syndrome) (HS6ST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HS6ST1	HS6ST1, HS6ST, HH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 15 with or without anosmia (HH15) [MIM:614880]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:21700882, ECO:0000269|PubMed:25077900}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 16 ± Anozmi, 614897, Otozomal Dominant; HH16 (Kallmann Sendromu) (SEMA3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 16 with or without anosmia, 614897, Autosomal dominant; HH16 (Kallmann syndrome) (SEMA3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEMA3A	SEMA3A, SEMAD, COLL1, HH16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 16 with or without anosmia (HH16) [MIM:614897]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:22416012, ECO:0000269|PubMed:22927827, ECO:0000269|PubMed:25077900}. Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 17 ± Anozmi, 615266, Otozomal Dominant; HH17 (Kallmann Sendromu) (SPRY4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266, Autosomal dominant; HH17 (Kallmann syndrome) (SPRY4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPRY4	SPRY4, HH17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 17 with or without anosmia (HH17) [MIM:615266]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Some patients carrying mutations in SPRY4 also have a heterozygous mutation in another HH-associated gene including DUSP6 and FGFR1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 18 ± Anozmi, 615267, Otozomal Dominant; HH18 (Kallmann Sendromu (IL17RD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267, Autosomal dominant; HH18 (Kallmann syndrome) (IL17RD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL17RD	IL17RD, SEF, HH18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hipogonadotropik Hipogonadizm 19 ± Anozmi, 615269, Otozomal Dominant; HH19 (Kallmann Sendromu) (DUSP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269, Autosomal dominant; HH19 (Kallmann syndrome) (DUSP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DUSP6	DUSP6, MKP3, PYST1, HH19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 19 with or without anosmia (HH19) [MIM:615269]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Some patients carrying mutations in DUSP6 also have a heterozygous mutation in another HH-associated gene including FGFR1 and SPRY4 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 2 ± Anozmi, 147950, Otozomal Dominant; HH2 (Kallmann Sendromu) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 2 with or without anosmia, 147950, Autosomal dominant; HH2 (Kallmann syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 2 ± Anozmi, 147950, Otozomal Dominant; HH2 (Kallmann Sendromu) (MLPA)	Hypogonadotropic hypogonadism 2 with or without anosmia, 147950, Autosomal dominant; HH2 (Kallmann syndrome) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 20 ± Anozmi, 615270, Otozomal Dominant; HH20 (Kallmann Sendromu) (FGF17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270, Autosomal dominant; HH20 (Kallmann syndrome) (FGF17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF17	FGF17, HH20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 20 with or without anosmia (HH20) [MIM:615270]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Some patients carrying mutations in FGF17 also have a mutation in another HH-associated gene including FGFR1, HS6ST1 and FLRT3 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 21 Anozmi İle, 615271, Otozomal Dominant; HH21 (Kallmann Sendromu) (FLRT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 21 with anosmia, 615271, Autosomal dominant; HH21 (Kallmann syndrome) (FLRT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLRT3	FLRT3, HH21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 21 with or without anosmia (HH21) [MIM:615271]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Some patients carrying mutations in FLRT3 also have a mutation in another HH-associated gene including FGFR1, HS6ST1 and FGF17 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 22, ± Anozmi, 616030, Otozomal Resesif; HH22 (Kallmann Sendromu) (FEZF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030, Autosomal recessive; HH22 (Kallmann syndrome) (FEZF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FEZF1	FEZF1, FEZ, ZNF312B, HH22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 23, ± Anozmi, 228300, Otozomal Resesif; HH23 (Leydig Hücre Hipoplazisi, LHB Eksikliğine Bağlı) (LHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300, Autosomal recessive; HH23 (Leydig cell hypoplasia due to LHB deficiency) (LHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHB	LHB, HH23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 24, Anozmi Olmadan, 229070, Otozomal Resesif; HH24 (İzole Folikül Uyarıcı Hormon (FSH) Eksikliği) (FSHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 24 without anosmia, 229070, Autosomal recessive; HH24 (Isolated follicle stimulating hormone deficiency) (FSHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FSHB	FSHB, HH24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 24 without anosmia (HH24) [MIM:229070]: A form of hypogonadotropic hypogonadism, a group of disorders characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. HH24 is characterized by primary amenorrhea in women, oligo or azoospermia with low to normal testosterone levels in men, and infertility. {ECO:0000269|PubMed:8220432, ECO:0000269|PubMed:9271483, ECO:0000269|PubMed:9280841}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 3 ± Anozmi, 244200, Otozomal Dominant; HH3 (Kallmann Sendromu) (PROKR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200, Autosomal dominant; HH3 (Kallmann syndrome) (PROKR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROKR2	PROKR2, PKR2, GPR73L1, HH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 4 ± Anozmi, 610628, Otozomal Dominant; HH4 (Kallmann Sendromu) (PROK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628, Autosomal dominant; HH4 (Kallmann syndrome) (PROK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROK2	PROK2, PK2, BV8, HH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 4 with or without anosmia (HH4) [MIM:610628]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nlHH). {ECO:0000269|PubMed:17054399, ECO:0000269|PubMed:18559922, ECO:0000269|PubMed:23643382, ECO:0000269|PubMed:25077900}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in PROK2 as well as in other HH- associated genes including PROKR2 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 5, ± Anozmi, 612370; HH5 (Kallmann Sendromu) (CHD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370; HH5 (Kallmann syndrome) (CHD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHD7	CHD7, HH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Idiopathic scoliosis 3 (IS3) [MIM:608765]: An abnormality of the vertebral column in which patients develop lateral curvature of the spine of at least 10 degrees. {ECO:0000269	PubMed:17436250}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 5, ± Anozmi, 612370; HH5 (Kallmann Sendromu) (MLPA)	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370; HH5 (Kallmann syndrome) (MLPA)	CHD7	CHD7, HH5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Idiopathic scoliosis 3 (IS3) [MIM:608765]: An abnormality of the vertebral column in which patients develop lateral curvature of the spine of at least 10 degrees. {ECO:0000269	PubMed:17436250}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

Hipogonadotropik Hipogonadizm 6 ± Anozmi, 612702, HH6 (Kallmann Sendromu) (FGF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702, HH6 (Kallmann syndrome) (FGF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF8	FGF8, HH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 6 with or without anosmia (HH6) [MIM:612702]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nIHH). {ECO:0000269|PubMed:18596921, ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in FGF8 as well as in other HH- associated genes including FGFR1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 7, Anozmi Olmadan, 146110, Otozomal Resesif; HH7 (Normozmik Konjenital Hipogonadotropik Hipogonadizm) (GNRHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 7 without anosmia, 146110, Autosomal recessive; HH7 (Normosmic congenital hypogonadotropic hypogonadism) (GNRHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNRHR	GNRHR, LHRHR, HH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 7, Anozmi Olmadan, 146110, Otozomal Resesif; HH7 (Normozmik Konjenital Hipogonadotropik Hipogonadizm (MLPA)	Hypogonadotropic hypogonadism 7 without anosmia, 146110, Autosomal recessive; HH7 (Normosmic congenital hypogonadotropic hypogonadism) (MLPA)	GNRHR	GNRHR, LHRHR, HH7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 8 ± Anozmi, 614837, Otozomal Resesif; HH8 (Kallmann Sendromu) (KISS1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837, Autosomal recessive; HH8 (Kallmann syndrome) (KISS1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KISS1R	KISS1R, GPR54, HH8, CPPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Precocious puberty, central 1 (CPPB1) [MIM:176400]: A condition defined as the development of secondary sexual characteristics in boys and girls at a chronological age that is 2.5 standard deviations below the mean age at onset of puberty in the population. Central precocious puberty results from premature activation of the hypothalamic-pituitary-gonadal axis. {ECO:0000269	PubMed:18272894}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipogonadotropik Hipogonadizm 9 ± Anozmi, 614838; HH9 (Kallmann Sendromu) (NSMF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838; HH9 (Kallmann syndrome) (NSMF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSMF	NSMF, NELF, HH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypogonadotropic hypogonadism 9 with or without anosmia (HH9) [MIM:614838]: A disorder characterized by absent or incomplete sexual maturation by the age of 18 years, in conjunction with low levels of circulating gonadotropins and testosterone and no other abnormalities of the hypothalamic-pituitary axis. In some cases, it is associated with non-reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss. Anosmia or hyposmia is related to the absence or hypoplasia of the olfactory bulbs and tracts. Hypogonadism is due to deficiency in gonadotropin-releasing hormone and probably results from a failure of embryonic migration of gonadotropin-releasing hormone-synthesizing neurons. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is referred to as Kallmann syndrome, whereas in the presence of a normal sense of smell, it has been termed normosmic idiopathic hypogonadotropic hypogonadism (nIHH). {ECO:0000269|PubMed:15362570, ECO:0000269|PubMed:21700882, ECO:0000269|PubMed:23643382}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. The genetics of hypogonadotropic hypogonadism involves various modes of transmission. Oligogenic inheritance has been reported in some patients carrying mutations in NSMF as well as in other HH-associated genes including FGFR1 (PubMed:23643382). {ECO:0000269|PubMed:23643382}.;	EDTA Blood Sample
Hipoinsülinemik Hipoglisemi-Hemihipertrofi, 240900, Otozomal Dominant; HIHGHH (Hipoinsülinemik Hipoglisemi ve Cisimciği Hemihipertrofi) (AKT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoinsulinemic hypoglycemia with hemihypertrophy, 240900, Autosomal dominant; HIHGHH (Hypoinsulinemic hypoglycemia and body hemihypertrophy) (AKT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT2	AKT2, HHGHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:15146380, ECO:0000269|PubMed:19164855}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalemik periyodik paralizi tip 1 ve tip 2 genleri (CACNA1S, SCN4A) (MLPA)	Hypokalemic periodic paralysis with type 1 and type 2 genes (CACNA1S, SCN4A) (MLPA)	CACNA1S, SCN4A	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipokalemik Periyodik Paralizi, Tip 1, 170400, Otozomal Dominant; HOKPP1 (Hipokalemik Periyodik Paralizi) (CACNA1S geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypokalemic periodic paralysis, type 1, 170400, Autosomal dominant; HOKPP1 (Hypokalemic periodic paralysis) (CACNA1S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalemik Periyodik Paralizi, Tip 1, 170400, Otozomal Dominant; HOKPP1 (Hipokalemik Periyodik Paralizi) (MLPA)	Hypokalemic periodic paralysis, type 1, 170400, Autosomal dominant; HOKPP1 (Hypokalemic periodic paralysis) (MLPA)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalemik Periyodik Paralizi, Tip 2, 613345, Otozomal Dominant; HOKPP2 (Hipokalemik Periyodik Paralizi) (SCN4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypokalemic periodic paralysis, type 2, 613345, Autosomal dominant; HOKPP2 (Hypokalemic periodic paralysis) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipokalemik Periyodik Paralizi, Tip 2, 613345, Otozomal Dominant; HOKPP2 (Hipokalemik Periyodik Paralizi) (MLPA)	Hypokalemic periodic paralysis, type 2, 613345, Autosomal dominant; HOKPP2 (Hypokalemic periodic paralysis) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipomagnezemi 1, Barsak, 602014, Otozomal Resesif; HOMG1 (Primer Hipomagnezemi-Sekonder Hipokalsemi) (TRPM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 1, intestinal, 602014, Autosomal recessive; HOMG1 (Primary hypomagnesemia with secondary hypocalcemia) (TRPM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPM6	TRPM6, CHAK2, HOMG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipomagnezemi 2, Böbrek, 154020, Otozomal Dominant; HOMG2 (Otozomal Dominant Primer Hipomagnezemi-Hipokalsüri) (FXYD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 2, renal, 154020, Autosomal dominant; HOMG2 (Autosomal dominant primary hypomagnesemia with hypocalciuria) (FXYD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FXYD2	FXYD2, ATP1G1, HOMG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipomagnezemi 3, Böbrek, 248250, Otozomal Resesif; HOMG3 (Ailesel Primer Hipomagnezemi-Nefrokalsinozis-Hiperkalsüri -Ağır Oküler Tutulum Olmadan) (CLDN16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 3, renal, 248250, Autosomal recessive; HOMG3 (Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis without severe ocular involvement) (CLDN16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLDN16	CLDN16, PCLN1, HOMG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Hipomagnezemi 4, Böbrek, 611718; HOMG4 (Ailesel Primer Hipomagnezemi - Normokalsiüri ve Normokalsemi) (EGF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 4, renal, 611718; HOMG4 (Familial primary hypomagnesemia with normocalciuria and normocalcemia) (EGF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGF	EGF, URG, HOMG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypomagnesemia 4 (HOMG4) [MIM:611718]: A disorder characterized by massive renal hypomagnesemia and normal levels of serum calcium and calcium excretion. Clinical features include seizures, mild-to moderate psychomotor retardation, and brisk tendon reflexes. [ECO:0000269	PubMed:17671655]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipomagnezemi 5, Renal, Göz Tutulumu, 248190, Otozomal Resesif; HOMG5 (Ailesel Primer Hipomagnezemi, Hiperkalsiüri ve Ağır Okiller Tutulumu Olan Nefrokalsinozis) (CLDN19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 5, renal, with ocular involvement, 248190, Autosomal recessive; HOMG5 (Familial primary hypomagnesemia with hypercalciuria and nephrocalcinosis with severe ocular involvement) (CLDN19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLDN19	CLDN19, HOMG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypomagnesemia 5 (HOMG5) [MIM:248190]: A progressive renal disease characterized by primary renal magnesium wasting with hypomagnesemia, hypercalciuria and nephrocalcinosis associated with severe ocular abnormalities such as bilateral chorioretinal scars, macular coloboma, significant myopia and nystagmus. The renal phenotype is virtually undistinguishable from that of patients with HOMG3. [ECO:0000269	PubMed:17033971]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipomagnezemi 6, Renal, 613882, Otozomal Dominant; HOMG6 (Ailesel Primer Hipomagnezemi - Normokalsiüri ve Normokalsemi) (CNNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia 6, renal, 613882, Autosomal dominant; HOMG6 (Familial primary hypomagnesemia with normocalciuria and normocalcemia) (CNNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNNM2	CNNM2, ACDP2, HOMG6, HOMGSMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypomagnesemia, seizures, and mental retardation (HOMGSMR) [MIM:616418]: A disease characterized by renal wasting of magnesium, low serum magnesium, seizures, and variable degrees of delayed psychomotor development. [ECO:0000269	PubMed:24699222]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipomagnezemi, Nöbetler ve Mental Retardasyon, 616418, Otozomal Resesif, Otozomal Dominant; HOMGSMR (Ailesel Primer Hipomagnezemi - Normokalsiüri ve Normokalsemi) (CNNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomagnesemia, seizures, and mental retardation, 616418, Autosomal recessive, Autosomal dominant; HOMGSMR (Familial primary hypomagnesemia with normocalciuria and normocalcemia) (CNNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNNM2	CNNM2, ACDP2, HOMG6, HOMGSMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypomagnesemia, seizures, and mental retardation (HOMGSMR) [MIM:616418]: A disease characterized by renal wasting of magnesium, low serum magnesium, seizures, and variable degrees of delayed psychomotor development. [ECO:0000269	PubMed:24699222]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipomagnezemi, Nöbetler ve Mental Retardasyon, 616418, Otozomal Resesif, Otozomal Dominant; HOMGSMR (Ailesel Primer Hipomagnezemi - Normokalsiüri ve Normokalsemi) (Prenatal) (CNNM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypomagnesemia, seizures, and mental retardation, 616418, Autosomal recessive, Autosomal dominant; HOMGSMR (Familial primary hypomagnesemia with normocalciuria and normocalcemia) (CNNM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNNM2	CNNM2, ACDP2, HOMG6, HOMGSMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypomagnesemia, seizures, and mental retardation (HOMGSMR) [MIM:616418]: A disease characterized by renal wasting of magnesium, low serum magnesium, seizures, and variable degrees of delayed psychomotor development. [ECO:0000269	PubMed:24699222]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipomyelinizasyon, Beyin Sapı ve Omurilik Tutulumu ve Bacak Spastisitesi, 615281, Otozomal Resesif; HBSL (Hipomiyelinizasyon-Beyin Sapı ve Omurilik Tutulumu ve Bacak Spastisitesi) (DARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281, Autosomal recessive; HBSL (Hypomyelination with brain stem and spinal cord involvement and leg spasticity) (DARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DARS	DARS, HBSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipomyelinizasyon, Beyin Sapı ve Omurilik Tutulumu ve Bacak Spastisitesi, 615281, Otozomal Resesif; HBSL (Hipomiyelinizasyon-Beyin Sapı ve Omurilik Tutulumu ve Bacak Spastisitesi) (Prenatal) (DARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypomyelination with brainstem and spinal cord involvement and leg spasticity, 615281, Autosomal recessive; HBSL (Hypomyelination with brain stem and spinal cord involvement and leg spasticity) (DARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DARS	DARS, HBSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoparatiroidizm-Gerilik-Dismorfizm Sendromu, 241410, Otozomal Resesif; HRDS (Sanjad-Sakati Sendromu) (TBCE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410, Autosomal recessive; HRDS (Sanjad-Sakati syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism-retardation-dysmorphism syndrome (HRD) [MIM:241410]: Autosomal recessive disorder reported almost exclusively in Middle Eastern populations. [ECO:0000269	PubMed:12389028]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipoparatiroidizm-Gerilik-Dismorfizm Sendromu, 241410, Otozomal Resesif; HRDS (Sanjad-Sakati Sendromu) (Prenatal) (TBCE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410, Autosomal recessive; HRDS (Sanjad-Sakati syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoparathyroidism-retardation-dysmorphism syndrome (HRD) [MIM:241410]: Autosomal recessive disorder reported almost exclusively in Middle Eastern populations. [ECO:0000269	PubMed:12389028]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoparatiroidizm, Otozomal Dominant, 146200, Otozomal Dominant (Hipoparatiroidizm, Otozomal Resesif) (PTH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoparathyroidism, autosomal dominant, 146200, Autosomal dominant (Familial isolated hypoparathyroidism due to impaired PTH secretion) (PTH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH	PTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism, familial isolated (FIH) [MIM:146200]: A disorder characterized by hypocalcemia and hyperphosphatemia due to inadequate secretion of parathyroid hormone. Clinical features include seizures, tetany and cramps. (ECO:0000269|PubMed:10523031, ECO:0000269|PubMed:2212001). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipoparatiroidizm, Otozomal Resesif, 146200, Otozomal Dominant (Hipoparatiroidizm, Otozomal Resesif) (PTH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoparathyroidism, autosomal recessive, 146200, Autosomal dominant (Familial isolated hypoparathyroidism due to impaired PTH secretion) (PTH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH	PTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism, familial isolated (FIH) [MIM:146200]: A disorder characterized by hypocalcemia and hyperphosphatemia due to inadequate secretion of parathyroid hormone. Clinical features include seizures, tetany and cramps. (ECO:0000269|PubMed:10523031, ECO:0000269|PubMed:22120001). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoparatiroidizm, Ailesel İzole 146200, Otozomal Dominant; FIH (Ailesel İzole Hipoparatiroidizm) (GCM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoparathyroidism, familial isolated, 146200, Autosomal dominant; FIH (Familial isolated hypoparathyroidism) (GCM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCM2	GCM2, GCMB, HRPT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism, familial isolated (FIH) [MIM:146200]: A disorder characterized by hypocalcemia and hyperphosphatemia due to inadequate secretion of parathyroid hormone. Clinical features include seizures, tetany and cramps. (ECO:0000269|PubMed:15728199, ECO:0000269|PubMed:15863676, ECO:0000269|PubMed:20190276, ECO:0000269|PubMed:20463099, ECO:0000269|Ref.3). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipoparatiroidizm,Sensorinöral İşitme Kaybı, ve Renal Displazi, 146255, Otozomal Dominant; HDR (Hipoparatiroidizm-Sağırlık-Böbrek Hastalığı Sendromu) (GATA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255, Autosomal dominant; HDR (Hypoparathyroidism-deafness-renal disease syndrome) (GATA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA3	GATA3, HDR, HDRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism, sensorineural deafness, and renal disease (HDR) [MIM:146255]: A disease characterized by steroid- resistant nephrosis with progressive renal failure, hypoparathyroidism, sensorineural deafness, and renal dysplasia. (ECO:0000269|PubMed:10935639, ECO:0000269|PubMed:11389161). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoparatiroidizm,Sensorinöral İşitme Kaybı, ve Renal Displazi, 146255, Otozomal Dominant; HDR (Hipoparatiroidizm-Sağırlık-Böbrek Hastalığı Sendromu) (MLPA)	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255, Autosomal dominant; HDR (Hypoparathyroidism-deafness-renal disease syndrome) (MLPA)	GATA3	GATA3, HDR, HDRS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypoparathyroidism, sensorineural deafness, and renal disease (HDR) [MIM:146255]: A disease characterized by steroid- resistant nephrosis with progressive renal failure, hypoparathyroidism, sensorineural deafness, and renal dysplasia. (ECO:0000269|PubMed:10935639, ECO:0000269|PubMed:11389161). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoparatiroidizm,Sensorinöral İşitme Kaybı, ve Renal Displazi, 146255, Otozomal Dominant; HDR (Hipoparatiroidizm-Sağırlık-Böbrek Hastalığı Sendromu) (Prenatal) (GATA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255, Autosomal dominant; HDR (Hypoparathyroidism-deafness-renal disease syndrome) (GATA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA3	GATA3, HDR, HDRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoparathyroidism, sensorineural deafness, and renal disease (HDR) [MIM:146255]: A disease characterized by steroid- resistant nephrosis with progressive renal failure, hypoparathyroidism, sensorineural deafness, and renal dysplasia. (ECO:0000269|PubMed:10935639, ECO:0000269|PubMed:11389161). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoparatiroidizm,Sensorinöral İşitme Kaybı, ve Renal Displazi, 146255, Otozomal Dominant; HDR (Hipoparatiroidizm-Sağırlık-Böbrek Hastalığı Sendromu) (Prenatal) (MLPA)	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255, Autosomal dominant; HDR (Hypoparathyroidism-deafness-renal disease syndrome) (Prenatal) (MLPA)	GATA3	GATA3, HDR, HDRS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoparathyroidism, sensorineural deafness, and renal disease (HDR) [MIM:146255]: A disease characterized by steroid- resistant nephrosis with progressive renal failure, hypoparathyroidism, sensorineural deafness, and renal dysplasia. (ECO:0000269|PubMed:10935639, ECO:0000269|PubMed:11389161). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofosfatazia, Erişkin, 146300, Otozomal Resesif, Otozomal Dominant (Hipofosfatazia) (ALPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatasia, adult, 146300, Autosomal recessive, Autosomal dominant (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). (ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10332035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760947, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027,	EDTA Blood Sample

Hipofosfatazia, Erişkin, 146300, Otozomal Resesif, Otozomal Dominant (Hipofosfatazia) (Prenatal) (ALPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypophosphatasia, adult, 146300, Autosomal recessive, Autosomal dominant (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10320035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofosfatazia, Çocukluk Çağı, 241510, Otozomal Resesif (Hipofosfataz) (ALPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatasia, childhood, 241510, Autosomal recessive (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10320035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	EDTA Blood Sample
Hipofosfatazia, Çocukluk Çağı, 241510, Otozomal Resesif (Hipofosfatazia) (Prenatal) (ALPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypophosphatasia, childhood, 241510, Autosomal recessive (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10320035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofosfatazia, İnfantil, 241500, Otozomal Resesif (Hipofosfataz) (ALPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatasia, infantile, 241500, Autosomal recessive (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10320035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	EDTA Blood Sample
Hipofosfatazia, İnfantil, 241500, Otozomal Resesif (Hipofosfatazia) (Prenatal) (ALPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypophosphatasia, infantile, 241500, Autosomal recessive (Hypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10320035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1409720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofosfatemi, X'e bağlı Paneli (PHEX Xp22.2, FGF23 12p13) (MLPA) (Prenatal)	Hypophosphatemia, X-Linked (PHEX Xp22.2, FGF23 12p13) (MLPA) (Prenatal)	PHEX Xp22.2, FGF23 12p13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipofosfatemi, X'e bağlı Paneli (PHEX Xp22.2, FGF23 12p13) (MLPA) (Prenatal)	Hypophosphatemia, X-Linked (PHEX Xp22.2, FGF23 12p13) (MLPA) (Prenatal)	PHEX Xp22.2, FGF23 12p13	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofosfatemik Raşitizm, Hiperkalsiüri, 241530, Otozomal Resesif; HHRH (Kalsitsal Hipofosfatemik Raşitizm-Hiperkalsiüri) (SLC34A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets with hypercalciuria, 241530, Autosomal recessive; HHRH (Hereditary hypophosphatemic rickets with hypercalciuria) (SLC34A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC34A3	SLC34A3, NPTIIC, HHRH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary hypophosphatemic rickets with hypercalciuria (HHRH) [MIM:241530]: Autosomal recessive form of hypophosphatemia characterized by reduced renal phosphate reabsorption and rickets. Increased serum levels of 1,25-dihydroxyvitamin D lead to increase in urinary calcium excretion. {ECO:0000269|PubMed:16358214, ECO:0000269|PubMed:16358215}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Hipofosfatemik Raşitizm, 300554, X'e Bağlı (Dent Hastalığı) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets, 300554, X-linked recessive (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. (ECO:0000269|PubMed:15986899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8559248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofosfatemik Raşitizm, AR, 241520, Otozomal Resesif (Otozomal Resesif Hipofosfatemik Raşitizm) (DMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets, AR, 241520, Autosomal recessive (Autosomal recessive hypophosphatemic rickets) (DMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMP1	DMP1, ARHR, ARHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal recessive, 1 (ARHR1) [MIM:241520]: A hereditary form of hypophosphatemic rickets, a disorder of proximal renal tubule function that causes phosphate loss, hypophosphatemia and skeletal deformities, including rickets and osteomalacia unresponsive to vitamin D. Symptoms are bone pain, fractures and growth abnormalities. (ECO:0000269|PubMed:17033621, ECO:0000269|PubMed:17033625). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipofosfatemik Raşitizm, Otozomal Dominant 193100, Otozomal Dominant; ADHR (Otozomal Dominant Hipofosfatemik Raşitizm) (FGF23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets, autosomal dominant, 193100, Autosomal dominant; ADHR (Autosomal dominant hypophosphatemic rickets) (FGF23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF23	FGF23, ADHR, HPDR2, PHPTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. (ECO:0000269	PubMed:11062477). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofosfatemik Raşitizm, Otozomal Dominant 193100, Otozomal Dominant; ADHR (Otozomal Dominant Hipofosfatemik Raşitizm) (MLPA)	Hypophosphatemic rickets, autosomal dominant, 193100, Autosomal dominant; ADHR (Autosomal dominant hypophosphatemic rickets) (MLPA)	FGF23	FGF23, ADHR, HPDR2, PHPTC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. (ECO:0000269	PubMed:11062477). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofosfatemik Raşitizm, Otozomal Resesif, 2, 613312; ARHR2 (Otozomal Resesif Hipofosfatemik Raşitizm) (ENPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets, autosomal recessive, 2, 613312; ARHR2 (Autosomal recessive hypophosphatemic rickets) (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENPP1	ENPP1, PDNP1, NPPS, M6S1, PCA1, ARHR2, COLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269	PubMed:16186408). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofosfatemik Raşitizm, X'e Bağlı Dominant; 307800, X'e Bağlı Dominant; XLHR (X'e Bağlı Hipofosfatemi) (PHEX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypophosphatemic rickets, X-linked dominant, 307800, X-linked dominant; XLHR (X-linked hypophosphatemia) (PHEX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHEX	PHEX, HYP, HPDR1, LXHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipofosfatemik Raşitizm, X'e Bağlı Dominant; 307800, X'e Bağlı Dominant; XLHR (X'e Bağlı Hipofosfatemi) (MLPA)	Hypophosphatemic rickets, X-linked dominant, 307800, X-linked dominant; XLHR (X-linked hypophosphatemia) (MLPA)	PHEX	PHEX, HYP, HPDR1, LXHR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipoplastik Sol Kalp Sendromu 1, 241550, Otozomal Resesif; HLHS1 (Hipoplastik Sol Kalp Sendromu) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoplastic left heart syndrome 1, 241550, Autosomal recessive; HLHS1 (Hypoplastic left heart syndrome) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. (ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23505041, ECO:0000269|PubMed:24508941). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipoplastik Sol Kalp Sendromu 1, 241550, Otozomal Resesif; HLHS1 (Hipoplastik Sol Kalp Sendromu) (Prenatal) (GJA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypoplastic left heart syndrome 1, 241550, Autosomal recessive; HLHS1 (Hypoplastic left heart syndrome) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. (ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23505041, ECO:0000269|PubMed:24508941). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoplastik Sol Kalp Sendromu 2, 614435, Otozomal Dominant; HLHS2 (Hipoplastik Sol Kalp Sendromu) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoplastic left heart syndrome 2, 614435, Autosomal dominant; HLHS2 (Hypoplastic left heart syndrome) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. (ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipoplastik Sol Kalp Sendromu 2, 614435, Otozomal Dominant; HLHS2 (Hipoplastik Sol Kalp Sendromu) (Prenatal) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypoplastic left heart syndrome 2, 614435, Autosomal dominant; HLHS2 (Hypoplastic left heart syndrome) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. (ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoplastik Veya Aplastik Tibia-Poldaktili, 188740, Otozomal Dominant (Tibia Yokluğu-Poldaktili Sendromu) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoplastic or aplastic tibia with polydactyly, 188740, Autosomal dominant (Absent tibia-polydactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269|PubMed:24456159). Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. (ECO:0000269|PubMed:24456159).;	EDTA Blood Sample

Hipoplastik Veya Aplastik Tibia-Polidaktili, 188740, Otozomal Dominant (Tibia Yokluğu-Polidaktili Sendromu) (Prenatal) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypoplastic or aplastic tibia with polydactyly, 188740, Autosomal dominant (Absent tibia-polydactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159}..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipoprotrombinemi, 613679, Otozomal Resesif (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypoprothrombinemia, 613679, Autosomal recessive (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to thrombin defect (THPH1) [MIM:188050]: A multifactorial disorder of hemostasis characterized by abnormal platelet aggregation in response to various agents and recurrent thrombi formation. {ECO:0000269|PubMed:2825773}. Note=The disease is caused by mutations affecting the gene represented in this entry. A common genetic variation in the 3-prime untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increased risk of venous thrombosis.	EDTA Blood Sample
Hipospadias 1, X'e Bağlı, 300633, X'e Bağlı Resesif; HYSP1 (Ailesel Hipospadias) (AR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypospadias 1, X-linked, 300633, X-linked recessive; HYSP1 (Familial hypospadias) (AR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398	EDTA Blood Sample
Hipospadias 1, X'e Bağlı, 300633, X'e Bağlı Resesif; HYSP1 (Ailesel Hipospadias) (MLPA)	Hypospadias 1, X-linked, 300633, X-linked recessive; HYSP1 (Familial hypospadias) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398	EDTA Blood Sample
Hipospadias 1, X'e Bağlı, 300633, X'e Bağlı Resesif; HYSP1 (Ailesel Hipospadias) (Prenatal) (AR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypospadias 1, X-linked, 300633, X-linked recessive; HYSP1 (Familial hypospadias) (AR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipospadias 1, X'e Bağlı, 300633, X'e Bağlı Resesif; HYSP1 (Ailesel Hipospadias) (Prenatal) (MLPA)	Hypospadias 1, X-linked, 300633, X-linked recessive; HYSP1 (Familial hypospadias) (Prenatal) (MLPA)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipospadias 2, X'e Bağlı, 300758, X'e Bağlı Resesif; HYSP2 (Ailesel Hipospadias) (MAMLD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypospadias 2, X-linked, 300758, X-linked recessive; HYSP2 (Familial hypospadias) (MAMLD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAMLD1	MAMLD1, CXorf6, F18, HYSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypospadias 2, X-linked (HYSP2) [MIM:300758]: A common malformation in which the urethra opens on the ventral side of the penis, due to developmental arrest of urethral fusion. The opening can be located glandular, penile, or even more posterior in the scrotum or perineum. Hypospadias is a feature of several syndromic disorders, including the androgen insensitivity syndrome and Opitz syndrome. {ECO:0000269|PubMed:17086185}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Hipospadias 2, X'e Bağlı, 300758, X'e Bağlı Resesif; HYSP2 (Ailesel Hipospadias) (Prenatal) (MAMLD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypospadias 2, X-linked, 300758, X-linked recessive; HYSP2 (Familial hypospadias) (MAMLD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAMLD1	MAMLD1, CXorf6, F18, HYSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypospadias 2, X-linked (HYSP2) [MIM:300758]: A common malformation in which the urethra opens on the ventral side of the penis, due to developmental arrest of urethral fusion. The opening can be located glandular, penile, or even more posterior in the scrotum or perineum. Hypospadias is a feature of several syndromic disorders, including the androgen insensitivity syndrome and Opitz syndrome. {ECO:0000269|PubMed:17086185}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotalamik Hamartomlar, Somatik, 241800 (Konjenital Hipotalamik Hamartom Sendromu) (GLI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothalamic hamartomas, somatic, 241800 (Congenital hypothalamic hamartoma syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Hipotalamik Hamartomlar, Somatik, 241800 (Konjenital Hipotalamik Hamartom Sendromu) (Prenatal) (GLI3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypothalamic hamartomas, somatic, 241800 (Congenital hypothalamic hamartoma syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotiroidizm, Konjenital, Guatrsız 4, 275100, Otozomal Resesif; CHNG4 (Izole Tiroid Uyarıcı Hormon (TSH) Eksikliği) (TSHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, congenital, nongoitrous 4, 275100, Autosomal recessive; CHNG4 (isolated thyroid-stimulating hormone deficiency) (TSHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHB	TSHB, CHNG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Hipotiroidizm, Merkezi ve Testis Büyümesi, 300888, X'e Bağlı Resesif; CHTE (X'e Bağlı Merkezi Konjenital Hipotiroidizm-Geç Başlangıçlı Testiküler Büyüme) (IGSF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, central, and testicular enlargement, 300888, X-linked recessive; CHTE (X-linked central congenital hypothyroidism with late-onset testicular enlargement) (IGSF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGSF1	IGSF1, IGDC1, CHTE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, central, and testicular enlargement (CHTE) [MIM:300888]: A disorder characterized by insufficient thyroid gland stimulation by thyroid stimulating hormone (TSH), resulting from hypothalamic and/or pituitary dysfunction. CHTE patients have delayed testosterone increase at puberty with normal testosterone levels in adulthood, normal testicular volume in childhood and enlarged testicles in adulthood. {ECO:0000269	PubMed:23143598}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotiroidizm, Konjenital Guatrsız, 5, 225250, Otozomal Dominant; CHNG5 (Atireozis) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, congenital nongoitrous, 5, 225250, Autosomal dominant; CHNG5 (Athyreosis) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotiroidizm, Konjenital, Tiroid Disgenezisi Veya Hipoplazliye Bağlı, 218700, Otozomal Dominant; CHNG2 (Atireozis) (PAX8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700, Autosomal dominant; CHNG2 (Athyreosis) (PAX8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX8	PAX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 2 (CHNG2) [MIM:218700]: A disease characterized by thyroid dysgenesis, the most frequent cause of congenital hypothyroidism, accounting for 85% of case. The thyroid gland can be completely absent (athyreosis), ectopically located and/or severely hypoplastic. Ectopic thyroid gland is the most frequent malformation, with thyroid tissue being found most often at the base of the tongue. {ECO:0000269|PubMed:11232006, ECO:0000269|PubMed:11502839, ECO:0000269|PubMed:9590296}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotiroidizm, Konjenital, Tiroid Disgenezisi Veya Hipoplazliye Bağlı, 218700, Otozomal Dominant; CHNG2 (Atireozis) (MLPA)	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700, Autosomal dominant; CHNG2 (Athyreosis) (MLPA)	PAX8	PAX8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 2 (CHNG2) [MIM:218700]: A disease characterized by thyroid dysgenesis, the most frequent cause of congenital hypothyroidism, accounting for 85% of case. The thyroid gland can be completely absent (athyreosis), ectopically located and/or severely hypoplastic. Ectopic thyroid gland is the most frequent malformation, with thyroid tissue being found most often at the base of the tongue. {ECO:0000269|PubMed:11232006, ECO:0000269|PubMed:11502839, ECO:0000269|PubMed:9590296}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotiroidizm, Konjenital, Guatrsız, 1, 275200, Otozomal Resesif; CHNG1 (Hipotiroidi, TSH Reseptörü Mutasyonlarına Bağlı) (TSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, congenital, nongoitrous, 1, 275200, Autosomal recessive; CHNG1 (Hypothyroidism due to TSH receptor mutations) (TSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHR	TSHR, CHNG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15331543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotiroidizm, Konjenital, Guatrsız, 1, 275200, Otozomal Resesif; CHNG1 (Hipotiroidi, TSH Reseptörü Mutasyonlarına Bağlı) (MLPA)	Hypothyroidism, congenital, nongoitrous, 1, 275200, Autosomal recessive; CHNG1 (Hypothyroidism due to TSH receptor mutations) (MLPA)	TSHR	TSHR, CHNG1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15331543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotiroidizm, Konjenital, Guatrsız, 6, 614450, Otozomal Dominant; CHNG6 (Tiroid Hormonlarına Periferik Direnç) (THRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypothyroidism, congenital, nongoitrous, 6, 614450, Autosomal dominant; CHNG6 (Peripheral resistance to thyroid hormones) (THRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THRA	THRA, ERBA1, THRA1, CHNG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 6 (CHNG6) [MIM:614450]: A disease characterized by growth retardation, developmental retardation, skeletal dysplasia, borderline low thyroxine levels and high triiodothyronine levels. There is differential sensitivity to thyroid hormone action, with retention of hormone responsiveness in the hypothalamic pituitary axis and liver but skeletal, gastrointestinal, and myocardial resistance. {ECO:0000269|PubMed:22168587, ECO:0000269|PubMed:24969835, ECO:0000269|PubMed:25670821, ECO:0000269|PubMed:26037512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotoni-Sistinüri Sendromu, 606407, Otozomal Resesif (2p21 Mikrodelesyon Sendromu) (2200)	Hypotonia-cystinuria syndrome, 606407, Autosomal recessive (2p21 microdeletion syndrome) (440)	.	DEL2p21, C2DELp21	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Hipotoni-Sistinüri Sendromu, 606407, Otozomal Resesif (2p21 Mikrodelesyon Sendromu) (Prenatal)	Hypotonia-cystinuria syndrome, 606407, Autosomal recessive (2p21 microdeletion syndrome) (Prenatal)	.	DEL2p21, C2DELp21	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotoni, Ataksi, ve Gecikmiş Gelişti̇rme Sendromu, 617330, Otozomal Dominant; HADDS (EBF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotonia, ataxia, and delayed development syndrome, 617330, Autosomal dominant; HADDS (EBF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EBF3	EBF3, COE3, HADDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Hipotoni, Ataksi, ve Gecikmiş Gelişti̇rme Sendromu, 617330, Otozomal Dominant; HADDS (Prenatal) (EBF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, ataxia, and delayed development syndrome, 617330, Autosomal dominant; HADDS (EBF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EBF3	EBF3, COE3, HADDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 1, 615419, Otozomal Resesif; IHPRF1 (Hipotoni-Konuşma Bozukluğu–Ağır Bilişsel Gecikme Sendromu) (NALCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419, Autosomal recessive; IHPRF1 (Hypotonia-speech impairment-severe cognitive delay syndrome) (NALCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NALCN	NALCN, IHPRF1, CLIFAHDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotonia, infantile, with psychomotor retardation and characteristic facies (IHPRF) [MIM:615419]: A neurodegenerative disease characterized by variable degrees of hypotonia, speech impairment, intellectual disability, pyramidal signs, subtle facial dysmorphism, and chronic constipation. Some patients manifest neuroaxonal dystrophy, optic atrophy, unmyelinated axons and spheroid bodies in tissue biopsies. {ECO:0000269|PubMed:23749988, ECO:0000269|PubMed:24075186}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 1, 615419, Otozomal Resesif; IHPRF1 (Hipotoni-Konuşma Bozukluğu–Ağır Bilişsel Gecikme Sendromu) (Prenatal) (NALCN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 1, 615419, Autosomal recessive; IHPRF1 (Hypotonia-speech impairment-severe cognitive delay syndrome) (NALCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NALCN	NALCN, IHPRF1, CLIFAHDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypotonia, infantile, with psychomotor retardation and characteristic facies (IHPRF) [MIM:615419]: A neurodegenerative disease characterized by variable degrees of hypotonia, speech impairment, intellectual disability, pyramidal signs, subtle facial dysmorphism, and chronic constipation. Some patients manifest neuroaxonal dystrophy, optic atrophy, unmyelinated axons and spheroid bodies in tissue biopsies. {ECO:0000269|PubMed:23749988, ECO:0000269|PubMed:24075186}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 2, 616801, Otozomal Resesif; IHPRF2 (Hipotoni-Konuşma Bozukluğu–Ağır Bilişsel Gecikme Sendromu) (UNC80 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801, Autosomal recessive; IHPRF2 (Hypotonia-speech impairment-severe cognitive delay syndrome) (UNC80 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNC80	UNC80, C2orf21, KIAA1843	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 2, 616801, Otozomal Resesif; IHPRF2 (Hipotoni-Konuşma Bozukluğu–Ağır Bilişsel Gecikme Sendromu) (Prenatal) (UNC80 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 2, 616801, Autosomal recessive; IHPRF2 (Hypotonia-speech impairment-severe cognitive delay syndrome) (UNC80 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UNC80	UNC80, C2orf21, KIAA1843	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 3, 616900, Otozomal Resesif; IHPRF3 (TBCK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900, Autosomal recessive; IHPRF3 (TBCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBCK	TBCK, IHPRF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik ve Karakteristik Yüz 3, 616900, Otozomal Resesif; IHPRF3 (TBCK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, infantile, with psychomotor retardation and characteristic facies 3, 616900, Autosomal recessive; IHPRF3 (TBCK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBCK	TBCK, IHPRF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Hipotoni, İnfantil, Psikomotor Gerilik, 616816, Otozomal Resesif; İHPMR (Ağır Hipotoni-Psikomotor Gelişim Gecikmesi-Şaşılık-Kardak Septal Defekt Sendromu) (CCDC174 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, infantile, with psychomotor retardation, 616816, Autosomal recessive; IHPMR (Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome) (CCDC174 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC174	CCDC174, HSPC212, IHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipotoni, İnfantil, Psikomotor Gerilik, 616816, Otozomal Resesif; İHPMR (Ağır Hipotoni-Psikomotor Gelişim Gecikmesi-Şaşılık-Kardak Septal Defekt Sendromu) (CCDC174 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotonia, infantile, with psychomotor retardation, 616816, Autosomal recessive; IHPMR (Severe hypotonia-psychomotor developmental delay-strabismus-cardiac septal defect syndrome) (CCDC174 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC174	CCDC174, HSPC212, IHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipotrikozis 1, 605389; HYPT1 (Hipotrikozis Simpleks) (APCDD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 1, 605389; HYPT1 (Hypotrichosis simplex) (APCDD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APCDD1	APCDD1, HHS, HYPT1, HTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 1 (HYPT1) [MIM:605389]: A rare form of non- syndromic hereditary hypotrichosis without characteristic hair shaft anomalies. Affected individuals typically show normal hair at birth, but hair loss and thinning of the hair shaft start during early childhood and progress with age. {ECO:0000269	PubMed:20393562}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis 11, 615059, Otozomal Dominant; HYPT11 (Hipotrikozis Simpleks) (SNRPE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 11, 615059, Autosomal dominant; HYPT11 (Hypotrichosis simplex) (SNRPE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNRPE	SNRPE, HYPT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipotrikozis 12, 615885, Otozomal Dominant; HYPT12 (Hipotrikozis Simpleks) (RPL21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 12, 615885, Autosomal dominant; HYPT12 (Hypotrichosis simplex) (RPL21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPL21	RPL21, HYPT12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 12 (HYPT12) [MIM:615885]: A form of hypotrichosis, a condition characterized by the presence of less than the normal amount of hair and abnormal hair follicles and shafts, which are thin and atrophic. The extent of scalp and body hair involvement can be very variable, within as well as between families. HYPT12 patients have normal scalp hair density at birth. Hair loss begins during the first 6 months of life and gradually progresses to nearly complete loss of scalp hair. The remaining hairs grow slowly and are thin, sparse, dry, and fragile. Body hair, axillary and pubic hair, eyebrows and eyelashes are also sparse or absent. {ECO:0000269	PubMed:21412954}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis 13, 615896, Otozomal Dominant; HYPT13 (Yünsü Saçlar) (KRT71 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 13, 615896, Autosomal dominant; HYPT13 (Woolly hair) (KRT71 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT71	KRT71, K6IRS1, KRT6IRS1, HYPT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 13 (HYPT13) [MIM:615896]: A form of hypotrichosis, a condition characterized by the presence of less than the normal amount of hair and abnormal hair follicles and shafts, which are thin and atrophic. The extent of scalp and body hair involvement can be very variable, within as well as between families. HYPT13 is characterized by sparse woolly hair. {ECO:0000269	PubMed:22592156}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis 2, 146520, Otozomal Dominant; HYPT2 (Kafa Derisi Hipotrikozis Simpleks) (CDSN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 2, 146520, Autosomal dominant; HYPT2 (Hypotrichosis simplex of the scalp) (CDSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDSN	CDSN, HTSS1, HYPT2, PSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 2 (HYPT2) [MIM:146520]: A condition characterized by the presence of less than the normal amount of hair. Affected individuals have normal hair in early childhood but experience progressive hair loss limited to the scalp beginning in the middle of the first decade and almost complete baldness by the third decade. Body hair, beard, eyebrows, axillary hair, teeth, and nails develop normally. {ECO:0000269	PubMed:12754508}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotrikozis 3, 613981; HYPT3 (Kafa Derisi Hipotrikozis Simpleks) (KRT74 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 3, 613981; HYPT3 (Hypotrichosis simplex of the scalp) (KRT74 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT74	KRT74, K6IRS4, KRT6IRS4, HTSS2, HYPT3, ADWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 3 (HYPT3) [MIM:613981]: A condition characterized by the presence of less than the normal amount of hair. Affected individuals have normal hair in early childhood but experience progressive hair loss limited to the scalp beginning in the middle of the first decade and almost complete baldness by the third decade. Body hair, beard, eyebrows, axillary hair, teeth, and nails develop normally. {ECO:0000269	PubMed:21188418}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotrikozis 4, 146550, Otozomal Dominant; HYPT4 (Marie Unna Kalıtsal Hipotrikozis) (HR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 4, 146550, Autosomal dominant; HYPT4 (Marie Unna hereditary hypotrichosis) (HR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HR	HR, AU, MUHH1, HYPT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 4 (HYPT4) [MIM:146550]: An autosomal dominant condition characterized by reduced amount of hair, alopecia, little or no eyebrows, eyelashes or body hair, and coarse, wiry, twisted hair in early childhood. {ECO:0000269|PubMed:19122663, ECO:0000269|PubMed:24961381}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis 6, 607903, Otozomal Resesif; HYPT6 (Hipotrikozis Simpleks) (DSG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 6, 607903, Autosomal recessive; HYPT6 (Hypotrichosis simplex) (DSG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG4	DSG4, LAH, HYPT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 6 (HYPT6) [MIM:607903]: A condition characterized by the presence of less than the normal amount of hair and abnormal hair follicles and shafts, which are thin and atrophic. The disorder affects the trunk and extremities as well as the scalp, and the eyebrows and eyelashes may also be involved, whereas beard, pubic, and axillary hairs are largely spared. In addition, patients can develop hyperkeratotic follicular papules, erythema, and pruritus in affected areas. In some patients with congenital hypotrichosis, monilethrix-like hairs showing elliptical nodes have been observed. {ECO:0000269	PubMed:15191570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotrikozis 7, 604379, Otozomal Resesif; HYPT7 (Yünsü Saçlar) (LIPH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 7, 604379, Autosomal recessive; HYPT7 (Woolly hair) (LIPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPH	LIPH, LAH2, ARWH2, HYPT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Woolly hair autosomal recessive 2 (ARWH2) [MIM:604379]: A hair shaft disorder characterized by fine and tightly curled hair. Compared to normal curly hair that is observed in some populations, woolly hair grows slowly and stops growing after a few inches. Under light microscopy, woolly hair shows some structural anomalies, including trichorrhexis nodosa and tapered ends. Some individuals may present with hypotrichosis. {ECO:0000269	PubMed:18830268}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotrikozis 8, 278150, Otozomal Resesif; HYPT8 (Yünsü Saçlar) (LPAR6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis 8, 278150, Autosomal recessive; HYPT8 (Woolly hair) (LPAR6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPAR6	LPAR6, P2RY5, P2Y5, LAH3, ARWH1, HYPT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Woolly hair autosomal recessive 1 with or without hypotrichosis (ARWH1) [MIM:278150]: A hair shaft disorder characterized by fine and tightly curled hair. Compared to normal curly hair that is observed in some populations, woolly hair grows slowly and stops growing after a few inches. Under light microscopy, woolly hair shows some structural anomalies, including trichorrhexis nodosa and tapered ends. Some individuals exhibit features of hypotrichosis. {ECO:0000269	PubMed:18297072}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipotrikozis ve Rekürren Deri Veziküller, 613102, Otozomal Resesif (Kalıtsal Hipotrikoz-Tekrarlayan Cilt Veziküller) (DSC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis and recurrent skin vesicles, 613102, Autosomal recessive (Hereditary hypotrichosis with recurrent skin vesicles) (DSC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSC3	DSC3, DSC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis and recurrent skin vesicles (HRSV) [MIM:613102]: A disorder characterized by hypotrichosis and the appearance of recurrent skin vesicle formation. Affected individuals show sparse and fragile hair on scalp, as well as absent eyebrows and eyelashes. Vesicles filled with thin, watery fluid are observed on the scalp and skin of most of the body. Mucosal vesicles are absent. {ECO:0000269	PubMed:19765682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis-Lenfödem-Telenjiektazi Sendromu, 607823, Otozomal Resesif; HLTS (Hipotrikoz-Lenfödem-Telenjiektazi-Renal Defekt Sendromu) (SOX18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823, Autosomal recessive; HLTS (Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome) (SOX18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX18	SOX18, HLTS, HLTRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome (HLTRS) [MIM:137940]: A syndrome characterized by sparse hair, lymphatic edemas, peripheral vein anomalies, and renal disease. {ECO:0000269|PubMed:12740761, ECO:0000269|PubMed:24697860}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis Lenfödem-Telenjiektazi-Renal Defekt Sendromu, 137940, Otozomal Dominant; HLTRS (Hipotrikoz-Lenfödem-Telenjiektazi-Renal Defekt Sendromu) (SOX18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940, Autosomal dominant; HLTRS (Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome) (SOX18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX18	SOX18, HLTS, HLTRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome (HLTRS) [MIM:137940]: A syndrome characterized by sparse hair, lymphatic edemas, peripheral vein anomalies, and renal disease. {ECO:0000269|PubMed:12740761, ECO:0000269|PubMed:24697860}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipotrikozis Lenfödem-Telenjiektazi-Renal Defekt Sendromu, 137940, Otozomal Dominant; HLTRS (Hipotrikoz-Lenfödem-Telenjiektazi-Renal Defekt Sendromu) (Prenatal) (SOX18 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940, Autosomal dominant; HLTRS (Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome) (SOX18 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX18	SOX18, HLTS, HLTRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome (HLTRS) [MIM:137940]: A syndrome characterized by sparse hair, lymphatic edemas, peripheral vein anomalies, and renal disease. {ECO:0000269|PubMed:12740761, ECO:0000269|PubMed:24697860}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipertrikoz, Konjenital, Jüvenil Maküler Distrofi, 601553, Otozomal Resesif; HJMD (Jüvenil Maküler Dejenerasyonu-Hipotrikoz) (CDH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypotrichosis, congenital, with juvenile macular dystrophy, 601553, Autosomal recessive; HJMD (Hypotrichosis with juvenile macular degeneration) (CDH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH3	CDHS, CDHP, PCAD, HJMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome (EEMS) [MIM:225280]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is an autosomal recessive condition characterized by features of ectodermal dysplasia such as sparse eyebrows and scalp hair, and selective tooth agenesis associated with macular dystrophy and ectrodactyly. {ECO:0000269	PubMed:15805154}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoürisemi, Renal, 2, 612076, Otozomal Resesif, Otozomal Dominant; RHUC2 (Kalıtsal Renal Hipoürisemi) (SLC2A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypouricemia, renal, 2, 612076, Autosomal recessive, Autosomal dominant; RHUC2 (Hereditary renal hypouricemia) (SLC2A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A9	SLC2A9, GLUT9, UAQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypouricemia renal 2 (RHUC2) [MIM:612076]: A disorder characterized by impaired uric acid reabsorption at the apical membrane of proximal renal tubule cells, and high urinary urate excretion. Patients often appear asymptomatic, but may be subject to exercise-induced acute renal failure, chronic renal dysfunction and nephrolithiasis. {ECO:0000269|PubMed:19026395, ECO:0000269|PubMed:19926891, ECO:0000269|PubMed:21810765}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipoürisemi, Böbrek, 220150, Otozomal Resesif; RHUC1 (Kalıtsal Renal Hipoürisemi) (SLC22A12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hypouricemia, renal, 220150, Autosomal recessive; RHUC1 (Hereditary renal hypouricemia) (SLC22A12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A12	SLC22A12, OAT4L, URAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Histriks Benzeri İktyoz-Sağırlık, 602540, Otozomal Dominant (KID Sendromu) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Hystrix-like ichthyosis with deafness, 602540, Autosomal dominant (KID syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vohwinkel syndrome (VS) [MIM:124500]: VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on fingers and toes and congenital deafness. {ECO:0000269|PubMed:10369869, ECO:0000269|PubMed:15954104, ECO:0000269|PubMed:18688774}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Siemens Tipi İktiyozis Büllöza, 146800, Otozomal Dominant (Yüzeyel Epidermolitik İktiyoz) (KRT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis bullosa of Siemens, 146800, Autosomal dominant (Superficial epidermolytic ichthyosis) (KRT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT2	KRT2, KRT2A, KRT2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis bullosa of Siemens (IBS) [MIM:146800]: A rare autosomal dominant skin disorder displaying a type of epidermolytic hyperkeratosis characterized by generalized erythema and extensive blistering from birth. Large, dark gray hyperkeratoses are observed in later weeks. The skin of IBS patients is unusually fragile and has a tendency to shed the outer layers of the epidermis, producing localized denuded areas (molting effect). IBS usually improves with age so that in most middle-aged patients the hyperkeratosis and keratotic lichenification is limited to the flexural folds of the major joints. {ECO:0000269|PubMed:10084318, ECO:0000269|PubMed:10233323, ECO:0000269|PubMed:10564334, ECO:0000269|PubMed:10620137, ECO:0000269|PubMed:10688369, ECO:0000269|PubMed:11167982, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:15949009, ECO:0000269|PubMed:7521371, ECO:0000269|PubMed:7524919, ECO:0000269|PubMed:8077693, ECO:0000269|PubMed:9036938, ECO:0000269|PubMed:9204966, ECO:0000269|PubMed:9804344, ECO:0000269|PubMed:9833038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis Histriks, Curth-Macklin Tipi, 146590, Otozomal Dominant; IHCM (İktiyozis Histriks, Curth-Macklin Tipi) (KRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis histrix, Curth-Macklin type, 146590, Autosomal dominant; IHCM (Ichthyosis hystrix of Curth-Macklin) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10233403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:2127994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis Prematürite Sendromu, 608649; IPS (İktiyozis Prematürite Sendromu) (SLC27A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis prematurity syndrome, 608649; IPS (Ichthyosis prematurity syndrome) (SLC27A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC27A4	SLC27A4, FATP4, IPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis Vulgaris, 146700, Otozomal Dominant (FLG geni) (Tüm Gen Dizi Analizi) (Postnatal) K.E	Ichthyosis vulgaris, 146700, Autosomal dominant (FLG gene) (Sequence Analysis-All Coding Exons) (Postnatal) K.E	FLG	FLG, ATOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis vulgaris (Vi) [MIM:146700]: The most common form of ichthyosis inherited as an autosomal dominant trait. It is characterized by palmar hyperlinearity, keratosis pilaris and a fine scale that is most prominent over the lower abdomen, arms, and legs. Ichthyosis vulgaris is characterized histologically by absent or reduced keratohyalin granules in the epidermis and mild hyperkeratosis. The disease can be associated with frequent asthma, eczema or hay fever. {ECO:0000269|PubMed:16444271}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis Vulgaris, 146700, Otozomal Dominant, (c.2282delCAGT) ve (c.1501C>T (p.Arg501X)) mutasyonları (FLG geni) (Dizi Analizi) (Postnatal)	Ichthyosis vulgaris, 146700, Autosomal dominant, (c.2282delCAGT) ve (c.1501C>T (p.Arg501X)) mutations (FLG gene) (Sequence Analysis) (Postnatal)	FLG	FLG, ATOD2	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120			60	Postnatal	DISEASE: Ichthyosis vulgaris (Vi) [MIM:146700]: The most common form of ichthyosis inherited as an autosomal dominant trait. It is characterized by palmar hyperlinearity, keratosis pilaris and a fine scale that is most prominent over the lower abdomen, arms, and legs. Ichthyosis vulgaris is characterized histologically by absent or reduced keratohyalin granules in the epidermis and mild hyperkeratosis. The disease can be associated with frequent asthma, eczema or hay fever. {ECO:0000269|PubMed:16444271}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis-Konfeti, 609165, Otozomal Dominant (Konjenital Retiküler İktiyoziform Eritroderma) (KRT10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis with confetti, 609165, Autosomal dominant (Congenital reticular ichthyosiform erythroderma) (KRT10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT10	KRT10, EHK, BCIE, BIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10201536, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381287, ECO:0000269|PubMed:2127994, ECO:0000269|PubMed:7507150, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7508181, ECO:0000269|PubMed:7512983, ECO:0000269|Ref.7}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 1, 242300, Otozomal Resesif; ARCI1 (Lameller İktiyoz) (TGM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 1, 242300, Autosomal recessive; ARCI1 (Lamellar ichthyosis) (TGM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGM1	TGM1, ICR2, ARCI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 1 (ARCI1) [MIM:242300]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:11251583, ECO:0000269|PubMed:11511296, ECO:0000269|PubMed:19890349, ECO:0000269|PubMed:7773290, ECO:0000269|PubMed:7824952, ECO:0000269|PubMed:9326318}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 1, 242300, Otozomal Resesif; ARCI1 (Lameller İktiyoz) (Prenatal) (TGM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 1, 242300, Autosomal recessive; ARCI1 (Lamellar ichthyosis) (TGM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGM1	TGM1, ICR2, ARCI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 1 (ARCI1) [MIM:242300]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:11251583, ECO:0000269|PubMed:11511296, ECO:0000269|PubMed:19890349, ECO:0000269|PubMed:7773290, ECO:0000269|PubMed:7824952, ECO:0000269|PubMed:9326318}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 10, 615024, Otozomal Resesif; ARCI10 (Konjenital Büllöz Olmayan İktiyoziform Eritroderma) (PNPLA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 10, 615024, Autosomal recessive; ARCI10 (Congenital non-bullous ichthyosiform erythroderma) (PNPLA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA1	PNPLA1, ARCI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 10, 615024, Otozomal Resesif; ARCI10 (Konjenital Büllöz Olmayan İktiyoziform Eritroderma) (PNPLA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 10, 615024, Autosomal recessive; ARCI10 (Congenital non-bullous ichthyosiform erythroderma) (PNPLA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPLA1	PNPLA1, ARCI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 11, 602400, Otozomal Resesif; ARCI11 (İktiyozis-Hipotrikoz Sendromu) (ST14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 11, 602400, Autosomal recessive; ARCI11 (Ichthyosis-hypotrichosis syndrome) (ST14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ST14	ST14, MTSP1, ARCI11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 11 (ARCI11) [MIM:602400]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:17273967, ECO:0000269|PubMed:18843291}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

İktiyozis, Konjenital, Otozomal Resesif 11, 602400, Otozomal Resesif; ARCI11 (İktiyozis-Hipotirioz Sendromu) (Prenatal) (ST14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 11, 602400, Autosomal recessive; ARCI11 (Ichthyosis-hypotrichosis syndrome) (ST14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ST14	ST14, MTSP1, ARCI11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 11 (ARCI11) [MIM:602400]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. (ECO:0000269|PubMed:17273967, ECO:0000269|PubMed:18843291). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 12, 617320, Otozomal Resesif; ARCI12 (Otozomal Resesif Konjenital İktiyozis) (CASP14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 12, 617320, Autosomal recessive; ARCI12 (Autosomal recessive congenital ichthyosis) (CASP14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP14	CASP14, ARCI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 12, 617320, Otozomal Resesif; ARCI12 (Otozomal Resesif Konjenital İktiyozis) (Prenatal) (CASP14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 12, 617320, Autosomal recessive; ARCI12 (Autosomal recessive congenital ichthyosis) (CASP14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASP14	CASP14, ARCI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 2, 242100, Otozomal Resesif; ARCI2 (Kendiliğinden İyileşen Kolodyon Bebek) (ALOX12B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 2, 242100, Autosomal recessive; ARCI2 (Self-healing collodion baby) (ALOX12B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALOX12B	ALOX12B, ARCI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 2, 242100, Otozomal Resesif; ARCI2 (Kendiliğinden İyileşen Kolodyon Bebek) (Prenatal) (ALOX12B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 2, 242100, Autosomal recessive; ARCI2 (Self-healing collodion baby) (ALOX12B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALOX12B	ALOX12B, ARCI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 3, 606545, Otozomal Resesif; ARCI3 (Lameller İktiyoz) (ALOXE3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 3, 606545, Autosomal recessive; ARCI3 (Lamellar ichthyosis) (ALOXE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALOXE3	ALOXE3, ARCI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 3, 606545, Otozomal Resesif; ARCI3 (Lameller İktiyoz) (Prenatal) (ALOXE3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 3, 606545, Autosomal recessive; ARCI3 (Lamellar ichthyosis) (ALOXE3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALOXE3	ALOXE3, ARCI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 4A, 601277, Otozomal Resesif; ARCI4A (Lameller İktiyoz) (ABCA12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 4A, 601277, Autosomal recessive; ARCI4A (Lamellar ichthyosis) (ABCA12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA12	ABCA12, ARCI4A, ARCI4B, ICR2B, LI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 4B (ARCI4B) [MIM:242500]: A rare, very severe form of congenital ichthyosis, in which the neonate is born with a thick covering of armor-like scales. The skin dries out to form hard diamond-shaped plaques separated by fissures, resembling 'armor plating'. The normal facial features are severely affected, with distortion of the lips (ectropion), eyelids (ectropion), ears, and nostrils. Affected babies are often born prematurely and rarely survive the perinatal period. Babies who survive into infancy and beyond develop skin changes resembling severe non-bullous congenital ichthyosiform erythroderma. (ECO:0000269|PubMed:15756637, ECO:0000269|PubMed:16675967, ECO:0000269|PubMed:16902423). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 4A, 601277, Otozomal Resesif; ARCI4A (Lameller İktiyoz) (Prenatal) (ABCA12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 4A, 601277, Autosomal recessive; ARCI4A (Lamellar ichthyosis) (ABCA12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCA12	ABCA12, ARCI4A, ARCI4B, ICR2B, LI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 4B (ARCI4B) [MIM:242500]: A rare, very severe form of congenital ichthyosis, in which the neonate is born with a thick covering of armor-like scales. The skin dries out to form hard diamond-shaped plaques separated by fissures, resembling 'armor plating'. The normal facial features are severely affected, with distortion of the lips (ectropion), eyelids (ectropion), ears, and nostrils. Affected babies are often born prematurely and rarely survive the perinatal period. Babies who survive into infancy and beyond develop skin changes resembling severe non-bullous congenital ichthyosiform erythroderma. (ECO:0000269|PubMed:15756637, ECO:0000269|PubMed:16675967, ECO:0000269|PubMed:16902423). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 4B (Alacalı), 242500, Otozomal Resesif; ARCI4B (Harlequin İktiyoz) (ABCA12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500, Autosomal recessive; ARCI4B (Harlequin ichthyosis) (ABCA12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA12	ABCA12, ARCI4A, ARCI4B, ICR2B, LI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 4B (ARCI4B) [MIM:242500]: A rare, very severe form of congenital ichthyosis, in which the neonate is born with a thick covering of armor-like scales. The skin dries out to form hard diamond-shaped plaques separated by fissures, resembling 'armor plating'. The normal facial features are severely affected, with distortion of the lips (ectropion), eyelids (ectropion), ears, and nostrils. Affected babies are often born prematurely and rarely survive the perinatal period. Babies who survive into infancy and beyond develop skin changes resembling severe non-bullous congenital ichthyosiform erythroderma. (ECO:0000269|PubMed:15756637, ECO:0000269|PubMed:16675967, ECO:0000269|PubMed:16902423). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 4B (Alacalı), 242500, Otozomal Resesif; ARCI4B (Harlequin İktiyoz) (Prenatal) (ABCA12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500, Autosomal recessive; ARCI4B (Harlequin ichthyosis) (ABCA12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCA12	ABCA12, ARCI4A, ARCI4B, ICR2B, LI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 4B (ARCI4B) [MIM:242500]: A rare, very severe form of congenital ichthyosis, in which the neonate is born with a thick covering of armor-like scales. The skin dries out to form hard diamond-shaped plaques separated by fissures, resembling 'armor plating'. The normal facial features are severely affected, with distortion of the lips (ectropion), eyelids (ectropion), ears, and nostrils. Affected babies are often born prematurely and rarely survive the perinatal period. Babies who survive into infancy and beyond develop skin changes resembling severe non-bullous congenital ichthyosiform erythroderma. (ECO:0000269|PubMed:15756637, ECO:0000269|PubMed:16675967, ECO:0000269|PubMed:16902423). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 5, 604777, Otozomal Resesif; ARCI5 (Lameller İktiyoz) (CYP4F22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 5, 604777, Autosomal recessive; ARCI5 (Lamellar ichthyosis) (CYP4F22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP4F22	CYP4F22, ARCI5, LI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 5 (ARCI5) [MIM:604777]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. (ECO:0000269	PubMed:16436457). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 5, 604777, Otozomal Resesif; ARCI5 (Lameller İktiyoz) (Prenatal) (CYP4F22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 5, 604777, Autosomal recessive; ARCI5 (Lamellar ichthyosis) (CYP4F22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP4F22	CYP4F22, ARCI5, LI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 5 (ARCI5) [MIM:604777]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. (ECO:0000269	PubMed:16436457). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 6, 612281, Otozomal Resesif; ARCI6 (Lameller İktiyoz) (NIPAL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 6, 612281, Autosomal recessive; ARCI6 (Lamellar ichthyosis) (NIPAL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NIPAL4	NIPAL4, ICHYN, ARCI6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 6, 612281, Otozomal Resesif; ARCI6 (Lameller İktiyoz) (Prenatal) (NIPAL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 6, 612281, Autosomal recessive; ARCI6 (Lamellar ichthyosis) (NIPAL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NIPAL4	NIPAL4, ICHYN, ARCI6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 8, 613943, Otozomal Resesif; ARCI8 (Lameller İktiyoz) (LIPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 8, 613943, Autosomal recessive; ARCI8 (Lamellar ichthyosis) (LIPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPN	LIPN, LIPL4, ARCI8, LI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, congenital, autosomal recessive 8 (ARCI8) [MIM:613943]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. (ECO:0000269	PubMed:21439540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

İktiyozis, Konjenital, Otozomal Resesif 8, 613943, Otozomal Resesif; ARCI8 (Lameller İktiyoz) (Prenatal) (LIPN gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 8, 613943, Autosomal recessive; ARCI8 (Lamellar ichthyosis) (LIPN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIPN	LIPN, LIPL4, ARCI8, LI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, congenital, autosomal recessive 8 (ARCI8) [MIM:613943]: A form of autosomal recessive congenital ichthyosis, a disorder of keratinization with abnormal differentiation and desquamation of the epidermis, resulting in abnormal skin scaling over the whole body. The main skin phenotypes are lamellar ichthyosis (LI) and non-bullous congenital ichthyosiform erythroderma (NCIE), although phenotypic overlap within the same patient or among patients from the same family can occur. Lamellar ichthyosis is a condition often associated with an embedment in a collodion-like membrane at birth; skin scales later develop, covering the entire body surface. Non-bullous congenital ichthyosiform erythroderma characterized by fine whitish scaling on an erythrodermal background; larger brownish scales are present on the buttocks, neck and legs. {ECO:0000269|PubMed:21439540}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 9, 615023, Otozomal Resesif; ARCI9 (Konjenital Büllöz Olmayan İktiyozlu Eritroderma) (CERS3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, congenital, autosomal recessive 9, 615023, Autosomal recessive; ARCI9 (Congenital non-bullous ichthyosiform erythroderma) (CERS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CERS3	CERS3, LASS3, ARCI9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Konjenital, Otozomal Resesif 9, 615023, Otozomal Resesif; ARCI9 (Konjenital Büllöz Olmayan İktiyozlu Eritroderma) (Prenatal) (CERS3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, congenital, autosomal recessive 9, 615023, Autosomal recessive; ARCI9 (Congenital non-bullous ichthyosiform erythroderma) (CERS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CERS3	CERS3, LASS3, ARCI9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Siklik, Epidermolitik Hiperkeratozis, 607602, Otozomal Dominant (Epidermolitik İktiyoz) (KRT10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602, Autosomal dominant (Epidermolytic ichthyosis) (KRT10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT10	KRT10, EHK, BCIE, BIE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10201536, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381287, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507150, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7508181, ECO:0000269|PubMed:7512983, ECO:0000269|Ref.7}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis, Siklik, Epidermolitik Hiperkeratozis, 607602, Otozomal Dominant (Epidermolitik İktiyoz) (KRT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602, Autosomal dominant (Epidermolytic ichthyosis) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis, Lökosit Vakuolleri, Alopesi ve Sklerozan Kolanjit, 607626, Otozomal Resesif; ILVASC (İktiyozis-Hipotrikozis-Sklerozan Kolanjit Sendromu) (CLDN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626, Autosomal recessive; ILVASC (Ichthyosis-hypotrichosis-sclerosing cholangitis syndrome) (CLDN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLDN1	CLDN1, SEMP1, ILVASC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İktiyozis, Lökosit Vakuolleri, Alopesi ve Sklerozan Kolanjit, 607626, Otozomal Resesif; ILVASC (İktiyozis-Hipotrikozis-Sklerozan Kolanjit Sendromu) (Prenatal) (CLDN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626, Autosomal recessive; ILVASC (Ichthyosis-hypotrichosis-sclerosing cholangitis syndrome) (CLDN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLDN1	CLDN1, SEMP1, ILVASC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis, Spastik Kuadriplejі Vemental Retardasyon, 614457, Otozomal Resesif; ISQMR (Konjenital İktiyoz-Mental Retardasyon-Spastik Kuadriplejі Sendromu) (ELOVL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, spastic quadriplegia, and mental retardation, 614457, Autosomal recessive; ISQMR (Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome) (ELOVL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL4	ELOVL4, ADMD, STGD2, STGD3, ISQMR, SCA34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 34 (SCA34) [MIM:133190]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA34 is an autosomal dominant form characterized by the association of progressive cerebellar ataxia with erythrokeratodermia variabilis. {ECO:0000269|PubMed:24566826}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İktiyozis, Spastik Kuadriplejі Vemental Retardasyon, 614457, Otozomal Resesif; ISQMR (Konjenital İktiyoz-Mental Retardasyon-Spastik Kuadriplejі Sendromu) (Prenatal) (ELOVL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, spastic quadriplegia, and mental retardation, 614457, Autosomal recessive; ISQMR (Congenital ichthyosis-intellectual disability-spastic quadriplegia syndrome) (ELOVL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELOVL4	ELOVL4, ADMD, STGD2, STGD3, ISQMR, SCA34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinocerebellar ataxia 34 (SCA34) [MIM:133190]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA34 is an autosomal dominant form characterized by the association of progressive cerebellar ataxia with erythrokeratodermia variabilis. {ECO:0000269|PubMed:24566826}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis X'e Bağlı, 308100, X'e Bağlı; XLI (Resesif X'e Bağlı İktiyozis) (STS gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ichthyosis, X-linked, 308100, X-linked recessive; XLI (Recessive X-linked ichthyosis) (STS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STS	STS, ARSC1, ARSC, SSDD, XLI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ichthyosis, X-linked (IXL) [MIM:308100]: A keratinization disorder manifesting with mild erythroderma and generalized exfoliation of the skin within a few weeks after birth. Affected boys later develop large, polygonal, dark brown scales, especially on the neck, extremities, trunk, and buttocks. {ECO:0000269|PubMed:10679952, ECO:0000269|PubMed:10844566, ECO:0000269|PubMed:1539590, ECO:0000269|PubMed:9252398}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İktiyozis X'e Bağlı, 308100, X'e Bağlı; XLI (Resesif X'e Bağlı İktiyozis) (MLPA)	Ichthyosis, X-linked, 308100, X-linked recessive; XLI (Recessive X-linked ichthyosis) (MLPA)	STS	STS, ARSC1, ARSC, SSDD, XLI	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ichthyosis, X-linked (IXL) [MIM:308100]: A keratinization disorder manifesting with mild erythroderma and generalized exfoliation of the skin within a few weeks after birth. Affected boys later develop large, polygonal, dark brown scales, especially on the neck, extremities, trunk, and buttocks. {ECO:0000269|PubMed:10679952, ECO:0000269|PubMed:10844566, ECO:0000269|PubMed:1539590, ECO:0000269|PubMed:9252398}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
IDH1 ve IDH2 (IDH1 geni Kodon 132 ve IDH2 Kodon 172 Mutasyonları) (Dizi Analizi) (IDH1, IDH2 geni) (Dizi Analizi) (Postnatal)	IDH1 and IDH2 (IDH1 gene, codon 132 and IDH2 codon 172 mutations) (Sequence analysis) (IDH1, IDH2 gene) (Dizi Analizi) (Postnatal)	IDH1, IDH2	.	Dizi Analizi/ Sequence Analysis	Lösemilerde kemik iliği/ Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Lösemilerde kemik iliği/ Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
IFAP Sendromu ± BRESHECK Sendromu, 308205, X'e Bağlı (İktiyozis Follikularis-Alopesi-Fotofobi Sendromu) (MBTPS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	IFAP syndrome with or without BRESHECK syndrome, 308205, X-linked recessive (Ichthyosis follicularis-alopecia-photophobia syndrome) (MBTPS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBTPS2	MBTPS2, S2P, IFAP, KFSDX, OLMSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Olmsted syndrome, X-linked (OLMSX) [MIM:309918]: A rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma and periorificial keratotic plaques with severe itching at all lesions. Diffuse alopecia, constriction of digits, and onychodystrophy have also been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes. {ECO:0000269|PubMed:22931912}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Iga Nefropatisi, Yatkınlık, 3, 616818, Otozomal Dominant; IGAN3 (SPRY2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	IgA nephropathy, susceptibility to, 3, 616818, Autosomal dominant; IGAN3 (SPRY2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPRY2	SPRY2, IGAN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGH (14q32) Breakapart (FISH)	IGH Breakapart (FISH)	14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
IGH'FGFR3 t(4;14)(p16;q32) (FISH)	IGH'FGFR3 t(4;14)(p16;q32) (FISH)	4p14-14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
IGH'MAF t(14;16)(q32.3;q23) (FISH)	IGH'MAF t(14;16)(q32.3;q23) (FISH)	14q32-16q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
IMAGE Sendromu, 614732, Otozomal Dominant (IMAGE Sendromu) (MLPA)	IMAGE syndrome, 614732, Autosomal dominant (IMAGE syndrome) (MLPA)	CDKN1C	CDKN1C, KIP2, BWS, IMAGE	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:10424811}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
IMAGE Sendromu, 614732, Otozomal Dominant (IMAGE Sendromu) (Prenatal) (MLPA)	IMAGE syndrome, 614732, Autosomal dominant (IMAGE syndrome) (Prenatal) (MLPA)	CDKN1C	CDKN1C, KIP2, BWS, IMAGE	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:10424811}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Digenik Resesif (İminoglisinüri) (SLC6A20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Iminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC6A20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A20	SLC6A20, XT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iminoglycinuria (IG) [MIM:242600]: A disorder of renal tubular reabsorption of glycine and imino acids (proline and hydroxyproline), marked by excessive levels of all three substances in the urine. {ECO:0000269|PubMed:19033859}. Note=The disease is caused by mutations affecting the gene represented in this entry. Haploinsufficiency of SLC6A20 combined with deficiency of the neutral amino acid transporter SLC6A19 or partially inactivating mutations in SLC36A2, is responsible for iminoglycinuria. Additional polymorphisms and mutations in SLC6A18 can contribute to the IG phenotype in some families.;	EDTA Blood Sample

İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Dijenik Resesif (İminoglisinüri) (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	İminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269	PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	EDTA Blood Sample
İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Dijenik Resesif (İminoglisinüri) (SLC36A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	İminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC36A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC36A2	SLC36A2, PAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iminoglycinuria (IG) [MIM:242600]: A disorder of renal tubular reabsorption of glycine and imino acids (proline and hydroxyproline), marked by excessive levels of all three substances in the urine. {ECO:0000269	PubMed:19033659}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Mutations in SLC36A2 that retain residual transport activity result in the IG phenotype only when combined with haploinsufficiency of the imino acid transporter SLC6A20 or deficiency of the neutral amino acid transporter SLC6A19. Additional polymorphisms and mutations in SLC6A18 can contribute to iminoglycinuria in some families.	EDTA Blood Sample
İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Dijenik Resesif (İminoglisinüri) (Prenatal) (SLC6A20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	İminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC6A20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A20	SLC6A20, XT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Iminoglycinuria (IG) [MIM:242600]: A disorder of renal tubular reabsorption of glycine and imino acids (proline and hydroxyproline), marked by excessive levels of all three substances in the urine. {ECO:0000269	PubMed:19033659}. Note=The disease is caused by mutations affecting the gene represented in this entry. Haploinsufficiency of SLC6A20 combined with deficiency of the neutral amino acid transporter SLC6A19 or partially inactivating mutations in SLC36A2, is responsible for iminoglycinuria. Additional polymorphisms and mutations in SLC6A18 can contribute to the IG phenotype in some families.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Dijenik Resesif (İminoglisinüri) (Prenatal) (SLC6A19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	İminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC6A19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A19	SLC6A19, HND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hyperglycinuria (HG) [MIM:138500]: A condition characterized by excess of glycine in the urine. In some cases it is associated with renal colic and renal oxalate stones. {ECO:0000269	PubMed:19033659}. Note=The disease may be caused by mutations affecting the gene represented in this entry. SLC6A19 deficiency combined with haploinsufficiency of SLC6A20 or partially inactivating mutations in SLC36A2, can be responsible for hyperglycinuria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İminoglisinüri, Dijenik, 242600, Otozomal Resesif, Dijenik Resesif (İminoglisinüri) (Prenatal) (SLC36A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	İminoglycinuria, digenic, 242600, Autosomal recessive, Digenic recessive (Iminoglycinuria) (SLC36A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC36A2	SLC36A2, PAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Iminoglycinuria (IG) [MIM:242600]: A disorder of renal tubular reabsorption of glycine and imino acids (proline and hydroxyproline), marked by excessive levels of all three substances in the urine. {ECO:0000269	PubMed:19033659}. Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. Mutations in SLC36A2 that retain residual transport activity result in the IG phenotype only when combined with haploinsufficiency of the imino acid transporter SLC6A20 or deficiency of the neutral amino acid transporter SLC6A19. Additional polymorphisms and mutations in SLC6A18 can contribute to iminoglycinuria in some families.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, 10, 612783, Otozomal Resesif, IMD10 (Kombine İmmün Yetmezlik, CRAC Kanal Disfonksiyonuna Bağlı) (STIM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 10, 612783, Autosomal recessive, IMD10 (Combined immunodeficiency due to CRAC channel dysfunction) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. {ECO:0000269	PubMed:23323920}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik, 10, 612783, Otozomal Resesif, IMD10 (Kombine İmmün Yetmezlik, CRAC Kanal Disfonksiyonuna Bağlı) (Prenatal) (STIM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 10, 612783, Autosomal recessive, IMD10 (Combined immunodeficiency due to CRAC channel dysfunction) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. {ECO:0000269	PubMed:23323920}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 11, 615206, Otozomal Resesif, IMD11 (Ağır Kombine İmmün Yetmezlik, CARD11 Eksikliğine Bağlı) (CARD11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 11, 615206, Autosomal recessive, IMD11 (Severe combined immunodeficiency due to CARD11 deficiency) (CARD11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARD11	CARD11, CARMA1, BIMP3, PPBL, BENTA, IMD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 11 (IMD11) [MIM:615206]: An autosomal recessive primary immunodeficiency characterized by normal numbers of T and B-lymphocytes, but defective intracellular signaling. There is a block in B-cell differentiation with increased numbers of transitional B-cells and hypogammaglobulinemia, as well as decreased numbers of regulatory T-cells and defects in T-cell function. {ECO:0000269	PubMed:23374270}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 11, 615206, Otozomal Resesif, IMD11 (Ağır Kombine İmmün Yetmezlik, CARD11 Eksikliğine Bağlı) (Prenatal) (CARD11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 11, 615206, Autosomal recessive, IMD11 (Severe combined immunodeficiency due to CARD11 deficiency) (CARD11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CARD11	CARD11, CARMA1, BIMP3, PPBL, BENTA, IMD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 11 (IMD11) [MIM:615206]: An autosomal recessive primary immunodeficiency characterized by normal numbers of T and B-lymphocytes, but defective intracellular signaling. There is a block in B-cell differentiation with increased numbers of transitional B-cells and hypogammaglobulinemia, as well as decreased numbers of regulatory T-cells and defects in T-cell function. {ECO:0000269	PubMed:23374270}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 12, 615468, Otozomal Resesif, IMD12 (Kombine İmmün Yetmezlik, MALT1 Eksikliğine Bağlı) (MALT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 12, 615468, Autosomal recessive, IMD12 (Combined immunodeficiency due to MALT1 deficiency) (MALT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MALT1	MALT1, MLT, IMD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MALT1 is recurrent in low-grade mucosa-associated lymphoid tissue (MALT lymphoma). Translocation t(11;18)(q21;q21) with BIRC2. This translocation is found in approximately 50% of cytogenetically abnormal low-grade MALT lymphoma. {ECO:0000269|PubMed:10339464, ECO:0000269|PubMed:10523859, ECO:0000269|PubMed:10702396, ECO:0000269|PubMed:11092834}.	EDTA Blood Sample
İmmün Yetmezlik 12, 615468, Otozomal Resesif, IMD12 (Kombine İmmün Yetmezlik, MALT1 Eksikliğine Bağlı) (Prenatal) (MALT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 12, 615468, Autosomal recessive, IMD12 (Combined immunodeficiency due to MALT1 deficiency) (MALT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MALT1	MALT1, MLT, IMD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving MALT1 is recurrent in low-grade mucosa-associated lymphoid tissue (MALT lymphoma). Translocation t(11;18)(q21;q21) with BIRC2. This translocation is found in approximately 50% of cytogenetically abnormal low-grade MALT lymphoma. {ECO:0000269|PubMed:10339464, ECO:0000269|PubMed:10523859, ECO:0000269|PubMed:10702396, ECO:0000269|PubMed:11092834}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 13, 615518, Otozomal Dominant; IMD13 (İdiopatik CD4 Lenfositopeni) (UNC119 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 13, 615518, Autosomal dominant; IMD13 (Idiopathic CD4 lymphocytopenia) (UNC119 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNC119	UNC119, HRG4, IMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 13 (IMD13) [MIM:615518]: A rare and heterogeneous syndrome defined by a reproducible reduction in the CD4 T-lymphocyte count (less than 300 cells per microliter or less than 20% of total T-cells) in the absence of HIV infection or other known causes of immunodeficiency. IMD13 predisposes to infections and malignancy. {ECO:0000269|PubMed:22184408}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 13, 615518, Otozomal Dominant; IMD13 (İdiopatik CD4 Lenfositopeni) (Prenatal) (UNC119 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 13, 615518, Autosomal dominant; IMD13 (Idiopathic CD4 lymphocytopenia) (UNC119 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UNC119	UNC119, HRG4, IMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 13 (IMD13) [MIM:615518]: A rare and heterogeneous syndrome defined by a reproducible reduction in the CD4 T-lymphocyte count (less than 300 cells per microliter or less than 20% of total T-cells) in the absence of HIV infection or other known causes of immunodeficiency. IMD13 predisposes to infections and malignancy. {ECO:0000269|PubMed:22184408}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 14, 615513, Otozomal Dominant; IMD14 (Aktive PI3K Delta Sendromu; PIK3CD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 14, 615513, Autosomal dominant; IMD14 (Activated PI3K-delta syndrome) (PIK3CD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CD	PIK3CD, APDS, IMD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Activated PI3K-delta syndrome (APDS) [MIM:615513]: A disorder characterized by recurrent respiratory infections, progressive airway damage, lymphopenia, increased circulating transitional B cells, increased immunoglobulin M, reduced immunoglobulin G2 levels in serum, and impaired vaccine responses. {ECO:0000269|PubMed:24136356}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 14, 615513, Otozomal Dominant; IMD14 (Aktive PI3K Delta Sendromu) (Prenatal) (PIK3CD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 14, 615513, Autosomal dominant; IMD14 (Activated PI3K-delta syndrome) (PIK3CD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3CD	PIK3CD, APDS, IMD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Activated PI3K-delta syndrome (APDS) [MIM:615513]: A disorder characterized by recurrent respiratory infections, progressive airway damage, lymphopenia, increased circulating transitional B cells, increased immunoglobulin M, reduced immunoglobulin G2 levels in serum, and impaired vaccine responses. {ECO:0000269|PubMed:24136356}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 15, 615592, Otozomal Resesif, IMD15 (Ağır Kombine İmmün Yetmezlik, IKK2 Eksikliğine Bağlı) (IKBKB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 15, 615592, Autosomal recessive; IMD15 (Severe combined immunodeficiency due to IKK2 deficiency) (IKBKB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKB	IKBKB, NFKBIKB, IMD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 15 (IMD15) [MIM:615592]: An autosomal recessive primary immunodeficiency disorder characterized by onset in infancy of life-threatening bacterial, fungal, and viral infections and failure to thrive. Laboratory studies show hypo- or agammaglobulinemia with relatively normal numbers of B and T- cells, and impaired differentiation and activation of immune cells. {ECO:0000269|PubMed:24369075}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 15, 615592, Otozomal Resesif, IMD15 (Ağır Kombine İmmün Yetmezlik, IKK2 Eksikliğine Bağlı) (Prenatal) (IKBKB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 15, 615592, Autosomal recessive; IMD15 (Severe combined immunodeficiency due to IKK2 deficiency) (IKBKB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKB	IKBKB, NFKBIKB, IMD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 15 (IMD15) [MIM:615592]: An autosomal recessive primary immunodeficiency disorder characterized by onset in infancy of life-threatening bacterial, fungal, and viral infections and failure to thrive. Laboratory studies show hypo- or agammaglobulinemia with relatively normal numbers of B and T- cells, and impaired differentiation and activation of immune cells. {ECO:0000269|PubMed:24369075}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 16, 615593, Otozomal Resesif, IMD16 (Kombine İmmün Yetmezlik, OX40 Eksikliğine Bağlı) (TNFRSF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 16, 615593, Autosomal recessive; IMD16 (Combined immunodeficiency due to OX40 deficiency) (TNFRSF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF4	TNFRSF4, TXGP1L, OX40, ACT35, IMD16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 16 (IMD16) [MIM:615593]: An autosomal recessive primary immunodeficiency associated with classic Kaposi sarcoma of childhood and poor T-cell recall immune responses due to complete functional OX40 deficiency. {ECO:0000269|PubMed:23897980}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

İmmün Yetmezlik 16, 615593, Otozomal Resesif; IMD16 (Kombine İmmün Yetmezlik, OX40 Eksikliğine Bağlı) (Prenatal) (TNFRSF4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 16, 615593, Autosomal recessive; IMD16 (Combined immunodeficiency due to OX40 deficiency) (TNFRSF4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFRSF4	TNFRSF4, TXGP1L, OX40, ACT35, IMD16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 16 (IMD16) [MIM:615593]: An autosomal recessive primary immunodeficiency associated with classic Kaposi sarcoma of childhood and poor T-cell recall immune responses due to complete functional OX40 deficiency. {ECO:0000269	PubMed:23897980}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 17, CD3 Gama Eksikliği, 615607, Otozomal Resesif; IMD17 (Kombine Immun Yetmezlik, CD3 Gama Eksikliğine Bağlı) (CD3G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 17, CD3 gamma deficient, 615607, Autosomal recessive; IMD17 (Combined immunodeficiency due to CD3gamma deficiency) (CD3G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD3G	CD3G, IMD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 17 (IMD17) [MIM:615607]: An autosomal recessive primary immunodeficiency characterized by highly variable clinical severity. Some patients have onset of severe recurrent infections in early infancy that may be lethal, whereas others may be only mildly affected or essentially asymptomatic into young adulthood. More severely affected patients may have evidence of autoimmune disease or enteropathy. The immunologic pattern is similar among patients, showing partial T-cell lymphopenia, decreased amounts of the CD3 complex, and impaired proliferative responses to T-cell receptor dependent stimuli. The phenotype in some patients is reminiscent of severe combined immunodeficiency. {ECO:0000269|PubMed:16355567, ECO:0000269|PubMed:17277165}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 17, CD3 Gama Eksikliği, 615607, Otozomal Resesif; IMD17 (Kombine Immun Yetmezlik, CD3 Gama Eksikliğine Bağlı) (Prenatal) (CD3G geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 17, CD3 gamma deficient, 615607, Autosomal recessive; IMD17 (Combined immunodeficiency due to CD3gamma deficiency) (CD3G gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD3G	CD3G, IMD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 17 (IMD17) [MIM:615607]: An autosomal recessive primary immunodeficiency characterized by highly variable clinical severity. Some patients have onset of severe recurrent infections in early infancy that may be lethal, whereas others may be only mildly affected or essentially asymptomatic into young adulthood. More severely affected patients may have evidence of autoimmune disease or enteropathy. The immunologic pattern is similar among patients, showing partial T-cell lymphopenia, decreased amounts of the CD3 complex, and impaired proliferative responses to T-cell receptor dependent stimuli. The phenotype in some patients is reminiscent of severe combined immunodeficiency. {ECO:0000269|PubMed:16355567, ECO:0000269|PubMed:17277165}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 18, SCID Varyantı, 615615, Otozomal Resesif; IMD18 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (CD3E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 18, SCID variant, 615615, Autosomal recessive; IMD18 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD3E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD3E	CD3E, IMD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 18 (IMD18) [MIM:615615]: An autosomal recessive primary immunodeficiency characterized by onset in infancy or early childhood of recurrent infections. The severity is variable, encompassing both a mild immunodeficiency and severe combined immunodeficiency (SCID), resulting in early death without bone marrow transplantation in some patients. Immunologic work-up of the IMD18 SCID patients shows a T cell-negative, B cell- positive, natural killer (NK) cell-positive phenotype, whereas T- cell development is not impaired in the mild form of IMD18. {ECO:0000269|PubMed:15546002, ECO:0000269|PubMed:8490660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 18, SCID Varyantı, 615615, Otozomal Resesif; IMD18 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (Prenatal) (CD3E geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 18, SCID variant, 615615, Autosomal recessive; IMD18 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD3E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD3E	CD3E, IMD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 18 (IMD18) [MIM:615615]: An autosomal recessive primary immunodeficiency characterized by onset in infancy or early childhood of recurrent infections. The severity is variable, encompassing both a mild immunodeficiency and severe combined immunodeficiency (SCID), resulting in early death without bone marrow transplantation in some patients. Immunologic work-up of the IMD18 SCID patients shows a T cell-negative, B cell- positive, natural killer (NK) cell-positive phenotype, whereas T- cell development is not impaired in the mild form of IMD18. {ECO:0000269|PubMed:15546002, ECO:0000269|PubMed:8490660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 19, 615617, Otozomal Resesif; IMD19 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (CD3D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 19, 615617, Autosomal recessive; IMD19 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD3D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD3D	CD3D, T3D, IMD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-positive/NK-cell-positive (T(-)B(+)NK(+) SCID) [MIM:608971]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269	PubMed:14602880}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 19, 615617, Otozomal Resesif; IMD19 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (Prenatal) (CD3D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 19, 615617, Autosomal recessive; IMD19 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD3D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD3D	CD3D, T3D, IMD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-positive/NK-cell-positive (T(-)B(+)NK(+) SCID) [MIM:608971]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269	PubMed:14602880}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 20, 615707, Otozomal Resesif; IMD20 (Otozomal Resesif Primer İmmün Yetmezlik- Spontan NK Hücre Sitotoksisitesi Defekti) (FCGR3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 20, 615707, Autosomal recessive; IMD20 (Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity) (FCGR3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR3A	FCGR3A, CD16, IGFR3, IMD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 20 (IMD20) [MIM:615707]: A rare autosomal recessive primary immunodeficiency characterized by functional deficiency of NK cells. Affected individuals typically present with severe herpes viral infections, particularly Epstein Barr virus (EBV), and human papillomavirus (HPV). {ECO:0000269|PubMed:23006327, ECO:0000269|PubMed:8608639, ECO:0000269|PubMed:8609432, ECO:0000269|PubMed:8874200}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 20, 615707, Otozomal Resesif; IMD20 (Otozomal Resesif Primer İmmün Yetmezlik- Spontan NK Hücre Sitotoksisitesi Defekti) (Prenatal) (FCGR3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 20, 615707, Autosomal recessive; IMD20 (Autosomal recessive primary immunodeficiency with defective spontaneous natural killer cell cytotoxicity) (FCGR3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FCGR3A	FCGR3A, CD16, IGFR3, IMD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 20 (IMD20) [MIM:615707]: A rare autosomal recessive primary immunodeficiency characterized by functional deficiency of NK cells. Affected individuals typically present with severe herpes viral infections, particularly Epstein Barr virus (EBV), and human papillomavirus (HPV). {ECO:0000269|PubMed:23006327, ECO:0000269|PubMed:8608639, ECO:0000269|PubMed:8609432, ECO:0000269|PubMed:8874200}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 21, 614172, Otozomal Dominant; IMD21 (Monositopeni-Enfeksiyonlara Yatkınlık) (GATA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 21, 614172, Autosomal dominant; IMD21 (Monocytopenia with susceptibility to infections) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 21, 614172, Otozomal Dominant; IMD21 (Monositopeni-Enfeksiyonlara Yatkınlık) (Prenatal) (GATA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 21, 614172, Autosomal dominant; IMD21 (Monocytopenia with susceptibility to infections) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 22, 615758, Otozomal Resesif; IMD22 (Ağır Kombine İmmün Yetmezlik, LCK Eksikliğine Bağlı) (LCK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 22, 615758, Autosomal recessive; IMD22 (Severe combined immunodeficiency due to LCK deficiency) (LCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LCK	LCK, IMD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 22 (IMD22) [MIM:615758]: A primary immunodeficiency characterized by T-cell dysfunction. Affected individuals present with lymphopenia, recurrent infections, severe diarrhea, and failure to thrive. {ECO:0000269	PubMed:23985903}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 22, 615758, Otozomal Resesif; IMD22 (Ağır Kombine İmmün Yetmezlik, LCK Eksikliğine Bağlı) (Prenatal) (LCK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 22, 615758, Autosomal recessive; IMD22 (Severe combined immunodeficiency due to LCK deficiency) (LCK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LCK	LCK, IMD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 22 (IMD22) [MIM:615758]: A primary immunodeficiency characterized by T-cell dysfunction. Affected individuals present with lymphopenia, recurrent infections, severe diarrhea, and failure to thrive. {ECO:0000269	PubMed:23985903}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 23, 615816, Otozomal Resesif; IMD23 (PGM3-CDG) (PGM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 23, 615816, Autosomal recessive; IMD23 (PGM3-CDG) (PGM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGM3	PGM3, AGM1, IMD23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 23, 615816, Otozomal Resesif; IMD23 (PGM3-CDG) (Prenatal) (PGM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 23, 615816, Autosomal recessive; IMD23 (PGM3-CDG) (PGM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGM3	PGM3, AGM1, IMD23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 24, 615897, Otozomal Resesif; IMD24 (Immun Yetmezlik, Ağır Kombine, CTPS1 Eksikliğine Bağlı) (SCID) (CTPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 24, 615897, Autosomal recessive; IMD24 (Severe combined immunodeficiency due to CTPS1 deficiency) (SCID) (CTPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTPS1	CTPS1, CTPS, IMD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 24 (IMD24) [MIM:615897]: A life-threatening immunodeficiency, characterized by an impaired capacity of activated T and B cells to proliferate in response to antigen receptor-mediated activation. Patients have early onset of severe chronic viral infections, mostly caused by herpes viruses, including EBV and varicella zooster virus (VZV), and also suffer from recurrent encapsulated bacterial infections, a spectrum of infections typical of a combined deficiency of adaptive immunity. {ECO:0000269|PubMed:24870241}. Note=The disease is caused by mutations affecting the gene represented in this entry. A unique and recessive G to C mutation probably affecting a splice donor site at the junction of intron 17-18 and exon 18 has been identified in all patients. It results in expression of an abnormal transcript lacking exon 18 and a complete loss of the expression of the protein. {ECO:0000269|PubMed:24870241},.	EDTA Blood Sample

İmmün Yetmezlik 24, 615897, Otozomal Resesif; IMD24 (İmmun Yetmezlik, Ağır Kombine, CTPS1 Eksikliğine Bağlı) (SCID) (Prenatal) (CTPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 24, 615897, Autosomal recessive; IMD24 (Severe combined immunodeficiency due to CTPS1 deficiency) (SCID) (CTPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTPS1	CTPS1, CTPS, IMD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 24 (IMD24) [MIM:615897]: A life-threatening immunodeficiency, characterized by an impaired capacity of activated T and B cells to proliferate in response to antigen receptor-mediated activation. Patients have early onset of severe chronic viral infections, mostly caused by herpes viruses, including EBV and varicella zoster virus (VZV), and also suffer from recurrent encapsulated bacterial infections, a spectrum of infections typical of a combined deficiency of adaptive immunity. {ECO:0000269|PubMed:24870241}. Note=The disease is caused by mutations affecting the gene represented in this entry. A unique and recessive G to C mutation probably affecting a splice donor site at the junction of intron 17-18 and exon 18 has been identified in all patients. It results in expression of an abnormal transcript lacking exon 18 and a complete loss of the expression of the protein. {ECO:0000269|PubMed:24870241}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 25, 610163, Otozomal Resesif; IMD25 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (CD247 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 25, 610163, Autosomal recessive; IMD25 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD247 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD247	CD247, CD3Z, TCRZ, IMD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Immunodeficiency 25 (IMD25) [MIM:610163]: An immunological deficiency characterized by T-cells impaired immune response to alloantigens, tetanus toxoid and mitogens. {ECO:0000269	PubMed:16672702}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 25, 610163, Otozomal Resesif; IMD25 (T-B + Ağır Kombine İmmün Yetmezlik, CD3delta / CD3epsilon / CD3zeta'ya Bağlı) (Prenatal) (CD247 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 25, 610163, Autosomal recessive; IMD25 (T-B+ severe combined immunodeficiency due to CD3delta/CD3epsilon/CD3zeta) (CD247 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD247	CD247, CD3Z, TCRZ, IMD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 25 (IMD25) [MIM:610163]: An immunological deficiency characterized by T-cells impaired immune response to alloantigens, tetanus toxoid and mitogens. {ECO:0000269	PubMed:16672702}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 26, a Nörolojik Anormallikler, 615966, Otozomal Resesif; IMD26 (Ağır Kombine İmmün Yetmezlik, DNA-Pkcs Eksikliğine Bağlı) (PRKDC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 26, with or without neurologic abnormalities, 615966, Autosomal recessive; IMD26 (Severe combined immunodeficiency due to DNA-PKcs deficiency) (PRKDC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKDC	PRKDC, HYRC1, DNPK1, IMD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Immunodeficiency 26 with or without neurologic abnormalities (IMD26) [MIM:615966]: A form of severe combined immunodeficiency characterized by reduced or absent T and B cells, recurrent candidiasis, and lower respiratory tract infections. Some patients show dysmorphic features, severe growth failure, microcephaly, seizures, and impaired neurological functions. {ECO:0000269	PubMed:19075392, ECO:0000269	PubMed:23722905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 26, a Nörolojik Anormallikler, 615966, Otozomal Resesif; IMD26 (Ağır Kombine İmmün Yetmezlik, DNA-Pkcs Eksikliğine Bağlı) (Prenatal) (PRKDC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 26, with or without neurologic abnormalities, 615966, Autosomal recessive; IMD26 (Severe combined immunodeficiency due to DNA-PKcs deficiency) (PRKDC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKDC	PRKDC, HYRC1, DNPK1, IMD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 26 with or without neurologic abnormalities (IMD26) [MIM:615966]: A form of severe combined immunodeficiency characterized by reduced or absent T and B cells, recurrent candidiasis, and lower respiratory tract infections. Some patients show dysmorphic features, severe growth failure, microcephaly, seizures, and impaired neurological functions. {ECO:0000269|PubMed:19075392, ECO:0000269|PubMed:23722905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 27A, Mikobakteriyoz AR, 209950, Otozomal Resesif; IMD27A (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel İfngamar1 Eksikliğine Bağlı) (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 27A, mycobacteriosis, AR, 209950, Autosomal recessive; IMD27A (Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209950]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. {ECO:0000269	PubMed:10811850, ECO:0000269	PubMed:9389728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 27A, Mikobakteriyoz AR, 209950, Otozomal Resesif; IMD27A (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel İfngamar1 Eksikliğine Bağlı) (Prenatal) (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 27A, mycobacteriosis, AR, 209950, Autosomal recessive; IMD27A (Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209950]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. {ECO:0000269|PubMed:10811850, ECO:0000269|PubMed:9389728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 27B, Mikobakteriyoz, AD, 615978, Otozomal Dominant; IMD27B (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel İfngamar1 Eksikliğine Bağlı) (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 27B, mycobacteriosis, AD, 615978, Autosomal dominant; IMD27B (Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209950]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. {ECO:0000269	PubMed:10811850, ECO:0000269	PubMed:9389728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 27B, Mikobakteriyoz, AD, 615978, Otozomal Dominant; IMD27B (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel İfngamar1 Eksikliğine Bağlı) (Prenatal) (IFNGR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 27B, mycobacteriosis, AD, 615978, Autosomal dominant; IMD27B (Autosomal dominant mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209950]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. {ECO:0000269|PubMed:10811850, ECO:0000269|PubMed:9389728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 28, Mikobakteriyoz, 614889, Otozomal Resesif; IMD28 (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet İfngamar2 Eksikliğine Bağlı) (IFNGR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 28, mycobacteriosis, 614889, Autosomal recessive; IMD28 (Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency) (IFNGR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR2	IFNGR2, IFNGT1, IFGR2, IMD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Immunodeficiency 28 (IMD28) [MIM:614889]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. IMD28 manifests early in life, with severe, often fatal, infection. {ECO:0000269	PubMed:15924140}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 28, Mikobakteriyoz, 614889, Otozomal Resesif; IMD28 (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet İfngamar2 Eksikliğine Bağlı) (Prenatal) (IFNGR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 28, mycobacteriosis, 614889, Autosomal recessive; IMD28 (Mendelian susceptibility to mycobacterial diseases due to complete IFNgammaR2 deficiency) (IFNGR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNGR2	IFNGR2, IFNGT1, IFGR2, IMD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 28 (IMD28) [MIM:614889]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. IMD28 manifests early in life, with severe, often fatal, infection. {ECO:0000269	PubMed:15924140}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

İmmün Yetmezlik 29, Mikobakteriyozis, 614890, Otozomal Resesif; IMD29 (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet IL12RB1 Eksikliğine Bağlı IL12RB2) (IL12B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 29, mycobacteriosis, 614890, Autosomal recessive; IMD29 (Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency) (IL12B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL12B	IL12B, NKSF2, IMD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriasis 11 (PSORS11) [MIM:612599]: A common, chronic inflammatory disease of the skin with multifactorial etiology. It is characterized by red, scaly plaques usually found on the scalp, elbows and knees. These lesions are caused by abnormal keratinocyte proliferation and infiltration of inflammatory cells into the dermis and epidermis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 29, Mikobakteriyozis, 614890, Otozomal Resesif; IMD29 (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet IL12RB1 Eksikliğine Bağlı IL12RB2) (Prenatal) (IL12B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 29, mycobacteriosis, 614890, Autosomal recessive; IMD29 (Mendelian susceptibility to mycobacterial diseases due to complete IL12B deficiency) (IL12B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL12B	IL12B, NKSF2, IMD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Psoriasis 11 (PSORS11) [MIM:612599]: A common, chronic inflammatory disease of the skin with multifactorial etiology. It is characterized by red, scaly plaques usually found on the scalp, elbows and knees. These lesions are caused by abnormal keratinocyte proliferation and infiltration of inflammatory cells into the dermis and epidermis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 30, 614891, Otozomal Resesif; IMD30 (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet IL12RB1 Eksikliğine Bağlı IL12RB1) (IL12RB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 30, 614891, Autosomal recessive; IMD30 (Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency) (IL12RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL12RB1	IL12RB1, IMD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 30 (IMD30) [MIM:614891]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. IMD30 has low penetrance, and affected individuals have relatively mild disease and good prognosis. BCG disease and salmonellosis are the most frequent infections in IMD30 patients. {ECO:0000269	PubMed:11424023}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 30, 614891, Otozomal Resesif; IMD30 (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet IL12RB1 Eksikliğine Bağlı IL12RB1) (Prenatal) (IL12RB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 30, 614891, Autosomal recessive; IMD30 (Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency) (IL12RB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL12RB1	IL12RB1, IMD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 30 (IMD30) [MIM:614891]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. IMD30 has low penetrance, and affected individuals have relatively mild disease and good prognosis. BCG disease and salmonellosis are the most frequent infections in IMD30 patients. {ECO:0000269	PubMed:11424023}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 31A, Mikobakteriyozis, Otozomal Dominant, 614892, Otozomal Dominant; IMD31A (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet STAT1 Eksikliğine Bağlı) (STAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892, Autosomal dominant; IMD31A (Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 31A, Mikobakteriyozis, Otozomal Dominant, 614892, Otozomal Dominant; IMD31A (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet STAT1 Eksikliğine Bağlı) (Prenatal) (STAT1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 31A, mycobacteriosis, autosomal dominant, 614892, Autosomal dominant; IMD31A (Mendelian susceptibility to mycobacterial diseases due to partial STAT1 deficiency) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 31B, Mikobakteriyel ve Viral Enfeksiyonlar, Otozomal Resesif, 613796, Otozomal Resesif; IMD31B (Viral ve Mikobakteriyel Enfeksiyonlara Yatkınlık) (STAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796, Autosomal recessive; IMD31B (Susceptibility to viral and mycobacterial infections) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 31B, Mikobakteriyel ve Viral Enfeksiyonlar, Otozomal Resesif, 613796, Otozomal Resesif; IMD31B (Viral ve Mikobakteriyel Enfeksiyonlara Yatkınlık) (Prenatal) (STAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 31B, mycobacterial and viral infections, autosomal recessive, 613796, Autosomal recessive; IMD31B (Susceptibility to viral and mycobacterial infections) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 31C, Otozomal Dominant, 614162, Otozomal Dominant; IMD31C (Otoimmün Enteropati ve Endokrinopati-Kronik Enfeksiyon Sendromuna Yatkınlık) (STAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 31C, autosomal dominant, 614162, Autosomal dominant; IMD31C (Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 31C, Otozomal Dominant, 614162, Otozomal Dominant; IMD31C (Otoimmün Enteropati ve Endokrinopati-Kronik Enfeksiyon Sendromuna Yatkınlık) (Prenatal) (STAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 31C, autosomal dominant, 614162, Autosomal dominant; IMD31C (Autoimmune enteropathy and endocrinopathy-susceptibility to chronic infections syndrome) (STAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAT1	STAT1, CANDF7, IMD31A, IMD31B, IMD31C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 31B (IMD31B) [MIM:613796]: A disorder characterized by susceptibility to severe mycobacterial and viral infections. Affected individuals can develop disseminated infections and die of viral illness. {ECO:0000269|PubMed:12590259, ECO:0000269|PubMed:20841510}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 32A, Mikobakteriyoz, Otozomal Dominant, 614893, Otozomal Dominant; IMD32A (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel IRF8 Eksikliğine Bağlı) (IRF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893, Autosomal dominant; IMD32A (Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency) (IRF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF8	IRF8, ICSBP1, IMD32A, IMD32B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 32A (IMD32A) [MIM:614893]: An immunologic disorder characterized by abnormal peripheral blood myeloid phenotype with a marked loss of CD11C-positive/CD1C dendritic cells, resulting in selective susceptibility to mycobacterial infections. {ECO:0000269	PubMed:21524210}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 32A, Mikobakteriyoz, Otozomal Dominant, 614893, Otozomal Dominant; IMD32A (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel IRF8 Eksikliğine Bağlı) (Prenatal) (IRF8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 32A, mycobacteriosis, autosomal dominant, 614893, Autosomal dominant; IMD32A (Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency) (IRF8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF8	IRF8, ICSBP1, IMD32A, IMD32B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 32A (IMD32A) [MIM:614893]: An immunologic disorder characterized by abnormal peripheral blood myeloid phenotype with a marked loss of CD11C-positive/CD1C dendritic cells, resulting in selective susceptibility to mycobacterial infections. {ECO:0000269	PubMed:21524210}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 32B, Monosit ve Dendritik Hücre Eksikliği, Otozomal Resesif, 614894, Otozomal Resesif; IMD32B (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel IRF8 Eksikliğine Bağlı) (IRF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894, Autosomal recessive; IMD32B (Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency) (IRF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF8	IRF8, ICSBP1, IMD32A, IMD32B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 32A (IMD32A) [MIM:614893]: An immunologic disorder characterized by abnormal peripheral blood myeloid phenotype with a marked loss of CD11C-positive/CD1C dendritic cells, resulting in selective susceptibility to mycobacterial infections. {ECO:0000269	PubMed:21524210}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 32B, Monosit ve Dendritik Hücre Eksikliği, Otozomal Resesif, 614894, Otozomal Resesif; IMD32B (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel IRF8 Eksikliğine Bağlı) (Prenatal) (IRF8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 32B, monocyte and dendritic cell deficiency, autosomal recessive, 614894, Autosomal recessive; IMD32B (Mendelian susceptibility to mycobacterial diseases due to partial IRF8 deficiency) (IRF8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF8	IRF8, ICSBP1, IMD32A, IMD32B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 32A (IMD32A) [MIM:614893]: An immunologic disorder characterized by abnormal peripheral blood myeloid phenotype with a marked loss of CD11C-positive/CD1C dendritic cells, resulting in selective susceptibility to mycobacterial infections. {ECO:0000269	PubMed:21524210}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 33, 300636, X'e Bağlı Resesif; IMD33 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, IKBKG Eksikliğine Bağlı) (IKBKG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 33, 300636, X-linked recessive; IMD33 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPO2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAD) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
İmmün Yetmezlik 33, 300636, X'e Bağlı Resesif; IMD33 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, IKBKG Eksikliğine Bağlı) (MLPA)	Immunodeficiency 33, 300636, X-linked recessive; IMD33 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPO2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAD) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

İmmün Yetmezlik 33, 300636, X'e Bağlı Resesif; IMD33 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, IKBKG Eksikliğine Bağlı) (Prenatal) (IKBKG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 33, 300636, X-linked recessive; IMD33 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 33, 300636, X'e Bağlı Resesif; IMD33 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, IKBKG Eksikliğine Bağlı) (Prenatal) (MLPA)	Immunodeficiency 33, 300636, X-linked recessive; IMD33 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (Prenatal) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. (ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 34, Mikobakteriyoz, X'e Bağlı, 300645, X'e Bağlı Resesif; IMD34 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, CYBB Eksikliğine Bağlı) (CYBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 34, mycobacteriosis, X-linked, 300645, X-linked recessive; IMD34 (X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency) (CYBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. [ECO:0000269	PubMed:21278736]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 34, Mikobakteriyoz, X'e Bağlı, 300645, X'e Bağlı Resesif; IMD34 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, CYBB Eksikliğine Bağlı) (Prenatal) (CYBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 34, mycobacteriosis, X-linked, 300645, X-linked recessive; IMD34 (X-linked mendelian susceptibility to mycobacterial diseases due to CYBB deficiency) (CYBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYBB	CYBB, CGD, AMCBX2, IMD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 34 (IMD34) [MIM:300645]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. [ECO:0000269	PubMed:21278736]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 35, 611521, Otozomal Resesif; IMD35 (Enfeksiyona Yatkınlık, TYK2 Eksikliğine Bağlı) (TYK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 35, 611521, Autosomal recessive; IMD35 (Susceptibility to infection due to TYK2 deficiency) (TYK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYK2	TYK2, IMD35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 35 (IMD35) [MIM:611521]: A primary immunodeficiency characterized by recurrent skin abscesses, pneumonia, and highly elevated serum IgE. (ECO:0000269	PubMed:17088085). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 35, 611521, Otozomal Resesif; IMD35 (Enfeksiyona Yatkınlık, TYK2 Eksikliğine Bağlı) (Prenatal) (TYK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 35, 611521, Autosomal recessive; IMD35 (Susceptibility to infection due to TYK2 deficiency) (TYK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TYK2	TYK2, IMD35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 35 (IMD35) [MIM:611521]: A primary immunodeficiency characterized by recurrent skin abscesses, pneumonia, and highly elevated serum IgE. (ECO:0000269	PubMed:17088085). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 36, 616005, Otozomal Dominant; IMD36 (Aktive PI3K Delta Sendromu) (PIK3R1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 36, 616005, Autosomal dominant; IMD36 (Activated PI3K-delta syndrome) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: SHORT syndrome (SHORTS) [MIM:269880]: A rare, multisystem disease characterized by short stature, anomalies of the anterior chamber of the eye, characteristic facial features such as triangular facies, lack of facial fat, and hypoplastic nasal alae with overhanging columella, partial lipodystrophy, hernias, hyperextensibility, and delayed dentition. The clinical phenotype can include insulin resistance, nephrocalcinosis, and hearing deficits. Developmental milestones and cognition are normal. (ECO:0000269|PubMed:23810378, ECO:0000269|PubMed:23810379, ECO:0000269|PubMed:23810382). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 36, 616005, Otozomal Dominant; IMD36 (Aktive PI3K Delta Sendromu) (Prenatal) (PIK3R1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 36, 616005, Autosomal dominant; IMD36 (Activated PI3K-delta syndrome) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: SHORT syndrome (SHORTS) [MIM:269880]: A rare, multisystem disease characterized by short stature, anomalies of the anterior chamber of the eye, characteristic facial features such as triangular facies, lack of facial fat, and hypoplastic nasal alae with overhanging columella, partial lipodystrophy, hernias, hyperextensibility, and delayed dentition. The clinical phenotype can include insulin resistance, nephrocalcinosis, and hearing deficits. Developmental milestones and cognition are normal. (ECO:0000269|PubMed:23810378, ECO:0000269|PubMed:23810379, ECO:0000269|PubMed:23810382). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 37, 616098, Otozomal Resesif; IMD37 (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 37, 616098, Autosomal recessive; IMD37 (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 37 (IMD37) [MIM:616098]: A form of primary combined immunodeficiency, a group of disorders characterized by severe recurrent infections, with normal numbers or an absence of T and B lymphocytes, and impaired cellular and humoral immunity. IMD37 is characterized by hypogammaglobulinemia without lymphopenia, but with profoundly reduced memory B cells and memory T cells, and increased numbers of circulating naive lymphocytes. Inheritance is autosomal recessive. (ECO:0000269	PubMed:25365219). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 37, 616098, Otozomal Resesif; IMD37 (Prenatal) (BCL10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 37, 616098, Autosomal recessive; IMD37 (BCL10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 37 (IMD37) [MIM:616098]: A form of primary combined immunodeficiency, a group of disorders characterized by severe recurrent infections, with normal numbers or an absence of T and B lymphocytes, and impaired cellular and humoral immunity. IMD37 is characterized by hypogammaglobulinemia without lymphopenia, but with profoundly reduced memory B cells and memory T cells, and increased numbers of circulating naive lymphocytes. Inheritance is autosomal recessive. (ECO:0000269	PubMed:25365219). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 38, 616126, Otozomal Resesif; IMD38 (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet ISG15 Eksikliğine Bağlı) (ISG15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 38, 616126, Autosomal recessive; IMD38 (Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency) (ISG15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ISG15	ISG15, G1P2, IFI15, IMD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 38, with basal ganglia calcification (IMD38) [MIM:616126]: A primary immunodeficiency predisposing individuals to severe clinical disease upon infection with weakly virulent mycobacteria, including Mycobacterium bovis Bacille Calmette-Guerin (BCG) vaccines. Patients are also susceptible to Salmonella and Mycobacterium tuberculosis infections. Affected individuals have intracranial calcification. (ECO:0000269	PubMed:22859821). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 38, 616126, Otozomal Resesif; IMD38 (Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Komplet ISG15 Eksikliğine Bağlı) (Prenatal) (ISG15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 38, 616126, Autosomal recessive; IMD38 (Mendelian susceptibility to mycobacterial diseases due to complete ISG15 deficiency) (ISG15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ISG15	ISG15, G1P2, IFI15, IMD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 38, with basal ganglia calcification (IMD38) [MIM:616126]: A primary immunodeficiency predisposing individuals to severe clinical disease upon infection with weakly virulent mycobacteria, including Mycobacterium bovis Bacille Calmette-Guerin (BCG) vaccines. Patients are also susceptible to Salmonella and Mycobacterium tuberculosis infections. Affected individuals have intracranial calcification. (ECO:0000269	PubMed:22859821). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 39, 616345, Otozomal Resesif; IMD39 (IRF7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 39, 616345, Autosomal recessive; IMD39 (IRF7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF7	IRF7, IRF7A, IRF7B, IRF7C, IRF7H, IMD39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 39 (IMD39) [MIM:616345]: A primary immunodeficiency causing severe, life-threatening acute respiratory distress upon infection with H1N1 influenza A. (ECO:0000269	PubMed:25814066). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 39, 616345, Otozomal Resesif; IMD39 (Prenatal) (IRF7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 39, 616345, Autosomal recessive; IMD39 (IRF7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF7	IRF7, IRF7A, IRF7B, IRF7C, IRF7H, IMD39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 39 (IMD39) [MIM:616345]: A primary immunodeficiency causing severe, life-threatening acute respiratory distress upon infection with H1N1 influenza A. (ECO:0000269	PubMed:25814066). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 40, 616433, Otozomal Resesif; IMD40 (DOCK2 Eksikliği) (DOCK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 40, 616433, Autosomal recessive; IMD40 (DOCK2 deficiency) (DOCK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOCK2	DOCK2, IMD40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 40 (IMD40) [MIM:616433]: A form of combined immunodeficiency characterized by lymphopenia, and defective T-cell, B-cell, and NK-cell responses. Patients suffer from severe invasive bacterial and viral infections in early childhood and may die without bone marrow transplantation. (ECO:0000269	PubMed:26083206). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 40, 616433, Otozomal Resesif; IMD40 (DOCK2 Eksikliği) (Prenatal) (DOCK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 40, 616433, Autosomal recessive; IMD40 (DOCK2 deficiency) (DOCK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOCK2	DOCK2, IMD40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 40 (IMD40) [MIM:616433]: A form of combined immunodeficiency characterized by lymphopenia, and defective T-cell, B-cell, and NK-cell responses. Patients suffer from severe invasive bacterial and viral infections in early childhood and may die without bone marrow transplantation. (ECO:0000269	PubMed:26083206). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 41-Lenfoproliferasyon ve Otoimmünite, 606367, Otozomal Resesif; IMD41 (İmmün Yetmezlik, CD25 Eksikliğine Bağlı) (IL2RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367, Autosomal recessive; IMD41 (Immunodeficiency due to CD25 deficiency) (IL2RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL2RA	IL2RA, CD25, IL2R, IDDM10, IMD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 10 (IDDM10) [MIM:601942]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269	PubMed:17676041). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

İmmün Yetmezlik 41-Lenfoproliferasyon ve Otoimmünite, 606367, Otozomal Resesif; IMD41 (İmmün Yetmezlik, CD25 Eksikliğine Bağlı) (Prenatal) (IL2RA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 41 with lymphoproliferation and autoimmunity, 606367, Autosomal recessive; IMD41 (Immunodeficiency due to CD25 deficiency) (IL2RA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL2RA	IL2RA, CD25, IL2R, IDDM10, IMD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diabetes mellitus, insulin-dependent, 10 (IDDM10) [MIM:601942]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:17676041}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 42, 616622, Otozomal Resesif; IMD42 (Otozomal Resesif Mendelyen Mikobakteriyal Hastalık Yatkınlığı, Komplet Rorgama Reseptör Eksikliğine Bağlı) (RORC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 42, 616622, Autosomal recessive; IMD42 (Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency) (RORC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RORC	RORC, RORG, RZRG, IMD42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Biallelic RORC mutations are a cause of immunodeficiency resulting in concomitant candidiasis and mycobacteriosis. Candidiasis is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida. Mycobacteriosis is characterized by infections caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. {ECO:0000269	PubMed:26160376}.	EDTA Blood Sample
İmmün Yetmezlik 42, 616622, Otozomal Resesif; IMD42 (Otozomal Resesif Mendelyen Mikobakteriyal Hastalık Yatkınlığı, Komplet Rorgama Reseptör Eksikliğine Bağlı) (Prenatal) (RORC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 42, 616622, Autosomal recessive; IMD42 (Autosomal recessive mendelian susceptibility to mycobacterial diseases due to complete RORgamma receptor deficiency) (RORC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RORC	RORC, RORG, RZRG, IMD42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Biallelic RORC mutations are a cause of immunodeficiency resulting in concomitant candidiasis and mycobacteriosis. Candidiasis is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida. Mycobacteriosis is characterized by infections caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. {ECO:0000269	PubMed:26160376}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 43, 241600, Otozomal Resesif; IMD43 (İmmun Yetmezlik, HLA Sınıf 1 Defektif Ekspresyonuna Bağlı) (B2M geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 43, 241600, Autosomal recessive; IMD43 (Immunodeficiency by defective expression of HLA class 1) (B2M gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B2M	B2M, IMD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 43 (IMD43) [MIM:241600]: A disorder characterized by marked reduction in serum concentrations of immunoglobulins and albumin, and hyperproteinemia due to hypercatabolism. Patients may suffer from recurrent respiratory tract infections and severe skin disease. {ECO:0000269	PubMed:16549777}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 43, 241600, Otozomal Resesif; IMD43 (İmmun Yetmezlik, HLA Sınıf 1 Defektif Ekspresyonuna Bağlı) (Prenatal) (B2M geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 43, 241600, Autosomal recessive; IMD43 (Immunodeficiency by defective expression of HLA class 1) (B2M gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B2M	B2M, IMD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 43 (IMD43) [MIM:241600]: A disorder characterized by marked reduction in serum concentrations of immunoglobulins and albumin, and hyperproteinemia due to hypercatabolism. Patients may suffer from recurrent respiratory tract infections and severe skin disease. {ECO:0000269	PubMed:16549777}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 44, 616636, Otozomal Resesif; IMD44 (Primer İmmün Yetmezlik, STAT2 Eksikliğine Bağlı) (STAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 44, 616636, Autosomal recessive; IMD44 (Primary immunodeficiency due to STAT2 deficiency) (STAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT2	STAT2, IMD44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 44, 616636, Otozomal Resesif; IMD44 (Primer İmmün Yetmezlik, STAT2 Eksikliğine Bağlı) (Prenatal) (STAT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 44, 616636, Autosomal recessive; IMD44 (Primary immunodeficiency due to STAT2 deficiency) (STAT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAT2	STAT2, IMD44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 45, 616669, Otozomal Resesif; IMD45 (Primer İmmün Yetmezlik, STAT2 Eksikliğine Bağlı) (IFNAR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 45, 616669, Autosomal recessive; IMD45 (Primary immunodeficiency due to STAT2 deficiency) (IFNAR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNAR2	IFNAR2, IMD45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 45, 616669, Otozomal Resesif; IMD45 (Primer İmmün Yetmezlik, STAT2 Eksikliğine Bağlı) (Prenatal) (IFNAR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 45, 616669, Autosomal recessive; IMD45 (Primary immunodeficiency due to STAT2 deficiency) (IFNAR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNAR2	IFNAR2, IMD45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 46, 616740, Otozomal Resesif; IMD46 (TFRC İle İlişkili Kombine İmmün Yetmezlik) (TFRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 46, 616740, Autosomal recessive; IMD46 (TFRC-related combined immunodeficiency) (TFRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFRC	TFRC, TFR, CD71, IMD46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 46, 616740, Otozomal Resesif; IMD46 (TFRC İle İlişkili Kombine İmmün Yetmezlik) (Prenatal) (TFRC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 46, 616740, Autosomal recessive; IMD46 (TFRC-related combined immunodeficiency) (TFRC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TFRC	TFRC, TFR, CD71, IMD46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 47, 300972, X'e Bağlı Resesif (ATP6AP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 47, 300972, X-linked recessive; IMD47 (ATP6AP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6AP1	ATP6AP1, ATP6IP1, ATP6S1, VATPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 47, 300972, X'e Bağlı Resesif (Prenatal) (ATP6AP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 47, 300972, X-linked recessive; IMD47 (ATP6AP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP6AP1	ATP6AP1, ATP6IP1, ATP6S1, VATPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 48, 269840, Otozomal Resesif; IMD48 (Kombine İmmun Yetmezlik, ZAP70 Eksikliğine Bağlı) (ZAP70 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 48, 269840, Autosomal recessive; IMD48 (Combined immunodeficiency due to ZAP70 deficiency) (ZAP70 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZAP70	ZAP70, SRK, ADMIO2, IMD48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Selective T-cell defect (STCD) [MIM:269840]: A form of severe combined immunodeficiency characterized by a selective absence of CD8+ T-cells. {ECO:0000269|PubMed:11123350, ECO:0000269|PubMed:11412303, ECO:0000269|PubMed:18509675, ECO:0000269|PubMed:8124727, ECO:0000269|PubMed:8202713}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 48, 269840, Otozomal Resesif; IMD48 (Kombine İmmun Yetmezlik, ZAP70 Eksikliğine Bağlı) (Prenatal) (ZAP70 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 48, 269840, Autosomal recessive; IMD48 (Combined immunodeficiency due to ZAP70 deficiency) (ZAP70 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZAP70	ZAP70, SRK, ADMIO2, IMD48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Selective T-cell defect (STCD) [MIM:269840]: A form of severe combined immunodeficiency characterized by a selective absence of CD8+ T-cells. {ECO:0000269|PubMed:11123350, ECO:0000269|PubMed:11412303, ECO:0000269|PubMed:18509675, ECO:0000269|PubMed:8124727, ECO:0000269|PubMed:8202713}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 49, 617237, Otozomal Dominant; IMD49 (BCL11B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 49, 617237, Autosomal dominant; IMD49 (BCL11B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL11B	BCL11B, CTIP2, IMD49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 49, 617237, Otozomal Dominant; IMD49 (BCL11B geni) (Tüm Gen Dizi Analizi) (Prenatal) (BCL11B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 49, 617237, Autosomal dominant; IMD49 (BCL11B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCL11B	BCL11B, CTIP2, IMD49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 50, 300988, X'e Bağlı Resesif; IMD50 (MSN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 50, 300988, X-linked recessive; IMD50 (MSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSN	MSN, IMD50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik 50, 300988, X'e Bağlı Resesif; IMD50 (Prenatal) (MSN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 50, 300988, X-linked recessive; IMD50 (MSN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSN	MSN, IMD50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 51, 613953, Otozomal Resesif; IMD51 (Kronik Mukokutanöz Kandidiyaz) (IL17RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 51, 613953, Autosomal recessive; IMD51 (Chronic mucocutaneous candidiasis) (IL17RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL17RA	IL17RA, IL17R, IMD51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 5 (CANDF5) [MIM:613953]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. {ECO:0000269	PubMed:21350122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 51, 613953, Otozomal Resesif; IMD51 (Kronik Mukokutanöz Kandidiyaz) (Prenatal) (IL17RA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 51, 613953, Autosomal recessive; IMD51 (Chronic mucocutaneous candidiasis) (IL17RA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL17RA	IL17RA, IL17R, IMD51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Candidiasis, familial, 5 (CANDF5) [MIM:613953]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. {ECO:0000269	PubMed:21350122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 7, TCR-Alfa / Beta Eksikliği, 615387, Otozomal Resesif (TCR-Alfa-Beta-Pozitif T-Hücre Eksikliği) (TRAC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 7, TCR-alpha/beta deficient, 615387, Autosomal recessive (TCR-alpha-beta-positive T-cell deficiency) (TRAC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAC	TRAC, TRCA, TRA, IMD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 7 (IMD7) [MIM:615387]: A primary immunodeficiency disorder manifesting with recurrent respiratory infections, candidiasis, diarrhea, and failure to thrive. Patients show a clear predisposition to herpes viral infections, and features of immune dysregulation, including hypereosinophilia, vitiligo, and alopecia areata. Other features include lymphadenopathy and hepatosplenomegaly. CD3+ T-cells express TCR- gammadelta, but little or no TCR-alpha/beta. {ECO:0000269	PubMed:21206088}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 7, TCR-Alfa / Beta Eksikliği, 615387, Otozomal Resesif (TCR-Alfa-Beta-Pozitif T-Hücre Eksikliği) (TRAC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 7, TCR-alpha/beta deficient, 615387, Autosomal recessive (TCR-alpha-beta-positive T-cell deficiency) (TRAC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAC	TRAC, TRCA, TRA, IMD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 7 (IMD7) [MIM:615387]: A primary immunodeficiency disorder manifesting with recurrent respiratory infections, candidiasis, diarrhea, and failure to thrive. Patients show a clear predisposition to herpes viral infections, and features of immune dysregulation, including hypereosinophilia, vitiligo, and alopecia areata. Other features include lymphadenopathy and hepatosplenomegaly. CD3+ T-cells express TCR- gammadelta, but little or no TCR-alpha/beta. {ECO:0000269	PubMed:21206088}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik 8, 615401, Otozomal Resesif; IMD8 (Ağır Kombine İmmün Yetmezlik, CORO1A Eksikliğine Bağlı) (CORO1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 8, 615401, Autosomal recessive; IMD8 (Severe combined immunodeficiency due to CORO1A deficiency) (CORO1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CORO1A	CORO1A, TACO, CLIPINA, IMD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 8 (IMD8) [MIM:615401]: A disease of the immune system leading to recurrent infections, and characterized by CD4+ T-cells lymphopenia. Patients can develop B-cell lymphoproliferation associated with Epstein-Barr virus infection. {ECO:0000269|PubMed:19097825, ECO:0000269|PubMed:23522482}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik 8, 615401, Otozomal Resesif; IMD8 (Ağır Kombine İmmün Yetmezlik, CORO1A Eksikliğine Bağlı) (Prenatal) (CORO1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 8, 615401, Autosomal recessive; IMD8 (Severe combined immunodeficiency due to CORO1A deficiency) (CORO1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CORO1A	CORO1A, TACO, CLIPINA, IMD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 8 (IMD8) [MIM:615401]: A disease of the immune system leading to recurrent infections, and characterized by CD4+ T-cells lymphopenia. Patients can develop B-cell lymphoproliferation associated with Epstein-Barr virus infection. {ECO:0000269|PubMed:19097825, ECO:0000269|PubMed:23522482}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

İmmün Yetmezlik 9, 612782, Otozomal Resesif; IMD9 (Kombine İmmün Yetmezlik, CRAC Kanal Disfonksiyonuna Bağlı) (ORAI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency 9, 612782, Autosomal recessive; IMD9 (Combined immunodeficiency due to CRAC channel dysfunction) (ORAI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ORAI1	ORAI1, TMEM142A, CRACM1, IMD9, TAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 9 (IMD9) [MIM:612782]: An immune disorder characterized by recurrent infections, impaired activation and proliferative response of T-cells, decreased T-cell production of cytokines, and normal lymphocytes counts and serum immunoglobulin levels. In surviving patients ectodermal dysplasia with anhidrosis and non-progressive myopathy may be observed. (ECO:0000269|PubMed:16147976, ECO:0000269|PubMed:16582901). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik 9, 612782, Otozomal Resesif; IMD9 (Kombine İmmün Yetmezlik, CRAC Kanal Disfonksiyonuna Bağlı) (Prenatal) (ORAI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency 9, 612782, Autosomal recessive; IMD9 (Combined immunodeficiency due to CRAC channel dysfunction) (ORAI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ORAI1	ORAI1, TMEM142A, CRACM1, IMD9, TAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 9 (IMD9) [MIM:612782]: An immune disorder characterized by recurrent infections, impaired activation and proliferative response of T-cells, decreased T-cell production of cytokines, and normal lymphocytes counts and serum immunoglobulin levels. In surviving patients ectodermal dysplasia with anhidrosis and non-progressive myopathy may be observed. (ECO:0000269|PubMed:16147976, ECO:0000269|PubMed:16582901). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, MAPBP İle Etkileşen Protein Eksikliği, 610798, Otozomal Resesif (Primer İmmün Yetmezlik Sendromu, P14 Eksikliğine Bağlı) (LAMTOR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency due to defect in MAPBP-interacting protein, 610798, Autosomal recessive (Primary immunodeficiency syndrome due to p14 deficiency) (LAMTOR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMTOR2	LAMTOR2, MAPBPIP, p14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency due to defect in MAPBP-interacting protein (ID-MAPBPIP) [MIM:610798]: This form of primary immunodeficiency syndrome includes congenital neutropenia, partial albinism, short stature and B-cell and cytotoxic T-cell deficiency. (ECO:0000269	PubMed:17195838). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, MAPBP İle Etkileşen Protein Eksikliği, 610798, Otozomal Resesif (Primer İmmün Yetmezlik Sendromu, P14 Eksikliğine Bağlı) (Prenatal) (LAMTOR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency due to defect in MAPBP-interacting protein, 610798, Autosomal recessive (Primary immunodeficiency syndrome due to p14 deficiency) (LAMTOR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMTOR2	LAMTOR2, MAPBPIP, p14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency due to defect in MAPBP-interacting protein (ID-MAPBPIP) [MIM:610798]: This form of primary immunodeficiency syndrome includes congenital neutropenia, partial albinism, short stature and B-cell and cytotoxic T-cell deficiency. (ECO:0000269	PubMed:17195838). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Fikolin 3 Eksikliğine Bağlı, 613860, Otozomal Resesif (İmmün Yetmezlik, Ficolin3 Eksikliğine Bağlı) (FCN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency due to ficolin 3 deficiency, 613860, Autosomal recessive (Immunodeficiency due to ficolin3 deficiency) (FCN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCN3	FCN3, HAKA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ficolin 3 deficiency (FCN3D) [MIM:613860]: A disorder characterized by immunodeficiency, recurrent infections, brain abscesses and recurrent warts on the fingers. Affected individuals have normal levels of lymphocytes, normal T-cell responses, and normal antibodies, but a selective deficient antibody response to pneumococcal polysaccharide vaccine. (ECO:0000269	PubMed:19535802). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Fikolin 3 Eksikliğine Bağlı, 613860, Otozomal Resesif (İmmün Yetmezlik, Ficolin3 Eksikliğine Bağlı) (Prenatal) (FCN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency due to ficolin 3 deficiency, 613860, Autosomal recessive (Immunodeficiency due to ficolin3 deficiency) (FCN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FCN3	FCN3, HAKA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ficolin 3 deficiency (FCN3D) [MIM:613860]: A disorder characterized by immunodeficiency, recurrent infections, brain abscesses and recurrent warts on the fingers. Affected individuals have normal levels of lymphocytes, normal T-cell responses, and normal antibodies, but a selective deficient antibody response to pneumococcal polysaccharide vaccine. (ECO:0000269	PubMed:19535802). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Pürin Nükleozid Fosforilaz Eksikliğine Bağlı, 613179, Otozomal Resesif (Pürin Nükleozid Fosforilaz Eksikliği) (PNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179, Autosomal recessive (Purine nucleoside phosphorylase deficiency) (PNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNP	PNP, NP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik, Pürin Nükleozid Fosforilaz Eksikliğine Bağlı, 613179, Otozomal Resesif (Pürin Nükleozid Fosforilaz Eksikliği) (Prenatal) (PNP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency due to purine nucleoside phosphorylase deficiency, 613179, Autosomal recessive (Purine nucleoside phosphorylase deficiency) (PNP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNP	PNP, NP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Hiper-Igm, Tip 5, 608106, Otozomal Resesif; HGM5 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık Olmadanı) (UNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency with hyper IgM, type 5, 608106, Autosomal recessive; HGM5 (Hyper-IgM syndrome without susceptibility to opportunistic infections) (UNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UNG	UNG, DGU, HIGM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency with hyper-IgM 5 (HIGM5) [MIM:608106]: A rare immunodeficiency syndrome characterized by normal or elevated serum IgM levels with absence of IgG, IgA and IgE. It results in a profound susceptibility to bacterial infections. (ECO:0000269	PubMed:12958596). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Hiper-Igm, Tip 5, 608106, Otozomal Resesif; HGM5 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık Olmadanı) (Prenatal) (UNG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency with hyper IgM, type 5, 608106, Autosomal recessive; HGM5 (Hyper-IgM syndrome without susceptibility to opportunistic infections) (UNG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UNG	UNG, DGU, HIGM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency with hyper-IgM 5 (HIGM5) [MIM:608106]: A rare immunodeficiency syndrome characterized by normal or elevated serum IgM levels with absence of IgG, IgA and IgE. It results in a profound susceptibility to bacterial infections. (ECO:0000269	PubMed:12958596). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Hiper-IgM, Tip 2, 605258, Otozomal Resesif; HGM2 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık Olmadanı) (AICDA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency with hyper-IgM, type 2, 605258, Autosomal recessive; HGM2 (Hyper-IgM syndrome without susceptibility to opportunistic infections) (AICDA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AICDA	AICDA, AID, HIGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency with hyper-IgM 2 (HIGM2) [MIM:605258]: A rare immunodeficiency syndrome characterized by normal or elevated serum IgM levels with absence of IgG, IgA and IgE. It results in a profound susceptibility to bacterial infections. (ECO:0000269	PubMed:11007475). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Hiper-IgM, Tip 2, 605258, Otozomal Resesif; HGM2 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık Olmadanı) (Prenatal) (AICDA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency with hyper-IgM, type 2, 605258, Autosomal recessive; HGM2 (Hyper-IgM syndrome without susceptibility to opportunistic infections) (AICDA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AICDA	AICDA, AID, HIGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency with hyper-IgM 2 (HIGM2) [MIM:605258]: A rare immunodeficiency syndrome characterized by normal or elevated serum IgM levels with absence of IgG, IgA and IgE. It results in a profound susceptibility to bacterial infections. (ECO:0000269	PubMed:11007475). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hiper-Igm İle İmmün Yetmezlik, Tip 3, 606843, Otozomal Resesif; HIGM3 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık) (CD40 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency with hyper-IgM, type 3, 606843, Autosomal recessive; HIGM3 (Hyper-IgM syndrome with susceptibility to opportunistic infections) (CD40 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD40	CD40, TNFRSF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hiper-Igm İle İmmün Yetmezlik, Tip 3, 606843, Otozomal Resesif; HIGM3 (Hiper-Igm Sendromu-Füsatiç Enfeksiyonlara Yatkınlık) (Prenatal) (CD40 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency with hyper-IgM, type 3, 606843, Autosomal recessive; HIGM3 (Hyper-IgM syndrome with susceptibility to opportunistic infections) (CD40 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD40	CD40, TNFRSF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu (ICF Sendromu) 1, 242860, Otozomal Resesif; ICF1 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (DNMT3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860, Autosomal recessive; ICF1 (ICF syndrome) (DNMT3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNMT3B	DNMT3B, ICF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (ICF1) [MIM:242860]: A rare disorder characterized by a variable immunodeficiency resulting in recurrent infections, facial anomalies, and branching of chromosomes 1, 9, and 16. Other variable symptoms include growth retardation, failure to thrive, and psychomotor retardation. Laboratory studies show limited hypomethylation of DNA in a small fraction of the genome in some, but not all, patients. (ECO:0000269|PubMed:10555141, ECO:0000269|PubMed:10588719, ECO:0000269|PubMed:10647011, ECO:0000269|PubMed:11102980, ECO:0000269|PubMed:15580563). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu (ICF Sendromu) 1, 242860, Otozomal Resesif; ICF1 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (Prenatal) (DNMT3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 1, 242860, Autosomal recessive; ICF1 (ICF syndrome) (DNMT3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNMT3B	DNMT3B, ICF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency-centromeric instability-facial anomalies syndrome 1 (ICF1) [MIM:242860]: A rare disorder characterized by a variable immunodeficiency resulting in recurrent infections, facial anomalies, and branching of chromosomes 1, 9, and 16. Other variable symptoms include growth retardation, failure to thrive, and psychomotor retardation. Laboratory studies show limited hypomethylation of DNA in a small fraction of the genome in some, but not all, patients. (ECO:0000269|PubMed:10555141, ECO:0000269|PubMed:10588719, ECO:0000269|PubMed:10647011, ECO:0000269|PubMed:11102980, ECO:0000269|PubMed:15580563). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu (ICF Sendromu) 3, 616910; ICF3 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (CDCA7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910; ICF3 (ICF syndrome) (CDCA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDCA7	CDCA7, JPO1, ICF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu (ICF Sendromu) 3, 616910; ICF3 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (Prenatal) (CDCA7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 3, 616910; ICF3 (ICF syndrome) (CDCA7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDCA7	CDCA7, JPO1, ICF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu 4, 616911, Otozomal Resesif; ICF4 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu) (HELLS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911, Autosomal recessive; ICF4 (ICF syndrome) (HELLS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HELLS	HELLS, LSH, ICF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu 4, 616911, Otozomal Resesif; ICF4 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu) (Prenatal) (HELLS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome 4, 616911, Autosomal recessive; ICF4 (ICF syndrome) (HELLS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HELLS	HELLS, LSH, ICF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu-2, 614069, Otozomal Resesif; ICF2 (İmmün Yetmezlik-Sentromerik İnstabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (ZBTB24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069, Autosomal recessive; ICF2 (ICF syndrome) (ZBTB24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB24	ZBTB24, PATZ2, ZNF450, KIAA0441, ICF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (ICF2) [MIM:614069]: A rare disorder characterized by a variable immunodeficiency resulting in recurrent infections, facial anomalies, and branching of chromosomes 1, 9, and 16. Other variable symptoms include growth retardation, failure to thrive, and psychomotor retardation. Laboratory studies show limited hypomethylation of DNA in a small fraction of the genome in some, but not all, patients. (ECO:0000269	PubMed:21596365). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

İmmün Yetmezlik-Sentromerik İnstabilite-Yüz Anomalileri Sendromu-2, 614069, Otozomal Resesif; ICF2 (İmmün Yetmezlik-Sentromerik Instabilite-Fasiyal Anomalileri Sendromu (ICF Sendromu)) (Prenatal) (ZBTB24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency-centromeric instability-facial anomalies syndrome-2, 614069, Autosomal recessive; ICF2 (ICF syndrome) (ZBTB24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZBTB24	ZBTB24, PATZ2, ZNF450, KIAA0441, ICF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency-centromeric instability-facial anomalies syndrome 2 (ICF2) [MIM:614069]: A rare disorder characterized by a variable immunodeficiency resulting in recurrent infections, facial anomalies, and branching of chromosomes 1, 9, and 16. Other variable symptoms include growth retardation, failure to thrive, and psychomotor retardation. Laboratory studies show limited hypomethylation of DNA in a small fraction of the genome in some, but not all, patients. {ECO:0000269	PubMed:21596365}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 1, 607594, Otozomal Resesif; CVID1 (İmmün Yetmezlik, Yaygın Değişken) (ICOS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 1, 607594, Autosomal recessive; CVID1 (Common variable immunodeficiency) (ICOS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ICOS	ICOS, AILIM, CVID1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 1 (CVID1) [MIM:607594]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:12577056}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 1, 607594, Otozomal Resesif; CVID1 (İmmün Yetmezlik, Yaygın Değişken) (Prenatal) (ICOS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 1, 607594, Autosomal recessive; CVID1 (Common variable immunodeficiency) (ICOS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ICOS	ICOS, AILIM, CVID1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 1 (CVID1) [MIM:607594]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:12577056}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 10, 615577, Otozomal Dominant; CVID10 (İmmun Yetmezlik, Yaygın Değişken) (NFKB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 10, 615577, Autosomal dominant; CVID10 (Common variable immunodeficiency) (NFKB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFKB2	NFKB2, LYT10, CVID10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 10 (CVID10) [MIM:615577]: A primary immunodeficiency characterized by childhood-onset of recurrent infections, hypogammaglobulinemia, and decreased numbers of memory and marginal zone B-cells. Some patients may develop autoimmune features and have circulating autoantibodies. An unusual feature is central adrenal insufficiency. {ECO:0000269|PubMed:24140114, ECO:0000269|PubMed:25524009}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 10, 615577, Otozomal Dominant; CVID10 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (NFKB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 10, 615577, Autosomal dominant; CVID10 (Common variable immunodeficiency) (NFKB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFKB2	NFKB2, LYT10, CVID10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 10 (CVID10) [MIM:615577]: A primary immunodeficiency characterized by childhood-onset of recurrent infections, hypogammaglobulinemia, and decreased numbers of memory and marginal zone B-cells. Some patients may develop autoimmune features and have circulating autoantibodies. An unusual feature is central adrenal insufficiency. {ECO:0000269|PubMed:24140114, ECO:0000269|PubMed:25524009}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 11, 615767, Otozomal Resesif; CVID11 (Otozomal Resesif Erken Başlangıçlı İnflamatuar Bağırsak Hastalığı) (IL21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 11, 615767, Autosomal recessive; CVID11 (Autosomal recessive early-onset inflammatory bowel disease) (IL21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL21	IL21, CVID11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 11, 615767, Otozomal Resesif; CVID11 (Otozomal Resesif Erken Başlangıçlı İnflamatuar Bağırsak Hastalığı) (Prenatal) (IL21 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 11, 615767, Autosomal recessive; CVID11 (Autosomal recessive early-onset inflammatory bowel disease) (IL21 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL21	IL21, CVID11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 12, 616576, Otozomal Dominant; CVID12 (İmmun Yetmezlik, Yaygın Değişken) (NFKB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 12, 616576, Autosomal dominant; CVID12 (Common variable immunodeficiency) (NFKB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFKB1	NFKB1, CVID12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 12, 616576, Otozomal Dominant; CVID12 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (NFKB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 12, 616576, Autosomal dominant; CVID12 (Common variable immunodeficiency) (NFKB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFKB1	NFKB1, CVID12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 13, 616873, Otozomal Dominant; CVID13 (Pansitopeni, IKZF1 Mutasyonlarına Bağlı) (IKZF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 13, 616873, Autosomal dominant; CVID13 (Pancytopenia due to IKZF1 mutations) (IKZF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKZF1	IKZF1, ZNFN1A1, IK1, LYF1, CVID13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in IKZF1 are frequent occurrences (28.6%) in acute lymphoblastic leukemia (ALL). Such alterations or deletions lead to poor prognosis for ALL.	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 13, 616873, Otozomal Dominant; CVID13 (Pansitopeni, IKZF1 Mutasyonlarına Bağlı) (Prenatal) (IKZF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 13, 616873, Autosomal dominant; CVID13 (Pancytopenia due to IKZF1 mutations) (IKZF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKZF1	IKZF1, ZNFN1A1, IK1, LYF1, CVID13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Defects in IKZF1 are frequent occurrences (28.6%) in acute lymphoblastic leukemia (ALL). Such alterations or deletions lead to poor prognosis for ALL.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 2, 240500, Otozomal Resesif, Otozomal Dominant; CVID2 (İmmun Yetmezlik, Yaygın Değişken) (TNFRSF13B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 2, 240500, Autosomal recessive, Autosomal dominant; CVID2 (Common variable immunodeficiency) (TNFRSF13B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF13B	TNFRSF13B, TACI, CVID2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunoglobulin A deficiency 2 (IGAD2) [MIM:609529]: Selective deficiency of immunoglobulin A (IGAD) is the most common form of primary immunodeficiency, with an incidence of approximately 1 in 600 individuals in the western world. Individuals with symptomatic IGAD often have deficiency of IgG subclasses or decreased antibody response to carbohydrate antigens such as pneumococcal polysaccharide vaccine. Individuals with IGAD also suffer from recurrent sinopulmonary and gastrointestinal infections and have an increased incidence of autoimmune disorders and of lymphoid and non-lymphoid malignancies. In vitro studies have suggested that some individuals with IGAD have impaired isotype class switching to IgA and others may have a post-switch defect. IGAD and CVID have been known to coexist in families. Some individuals initially present with IGAD1 and then develop CVID. These observations suggest that some cases of IGAD and CVID may have a common etiology. {ECO:0000269	PubMed:16007086}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 2, 240500, Otozomal Resesif, Otozomal Dominant; CVID2 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (TNFRSF13B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 2, 240500, Autosomal recessive, Autosomal dominant; CVID2 (Common variable immunodeficiency) (TNFRSF13B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFRSF13B	TNFRSF13B, TACI, CVID2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunoglobulin A deficiency 2 (IGAD2) [MIM:609529]: Selective deficiency of immunoglobulin A (IGAD) is the most common form of primary immunodeficiency, with an incidence of approximately 1 in 600 individuals in the western world. Individuals with symptomatic IGAD often have deficiency of IgG subclasses or decreased antibody response to carbohydrate antigens such as pneumococcal polysaccharide vaccine. Individuals with IGAD also suffer from recurrent sinopulmonary and gastrointestinal infections and have an increased incidence of autoimmune disorders and of lymphoid and non-lymphoid malignancies. In vitro studies have suggested that some individuals with IGAD have impaired isotype class switching to IgA and others may have a post-switch defect. IGAD and CVID have been known to coexist in families. Some individuals initially present with IGAD1 and then develop CVID. These observations suggest that some cases of IGAD and CVID may have a common etiology. {ECO:0000269	PubMed:16007086}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 3, 613493, Otozomal Resesif; CVID3 (İmmun Yetmezlik, Yaygın Değişken) (CD19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 3, 613493, Autosomal recessive; CVID3 (Common variable immunodeficiency) (CD19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD19	CD19, CVID3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 3 (CVID3) [MIM:613493]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:16672701}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 3, 613493, Otozomal Resesif; CVID3 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (CD19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 3, 613493, Autosomal recessive; CVID3 (Common variable immunodeficiency) (CD19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD19	CD19, CVID3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 3 (CVID3) [MIM:613493]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:16672701}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 4, 613494, Otozomal Resesif; CVID4 (İmmun Yetmezlik, Yaygın Değişken) (TNFRSF13C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 4, 613494, Autosomal recessive; CVID4 (Common variable immunodeficiency) (TNFRSF13C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF13C	TNFRSF13C, BAFFR, CVID4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 4, 613494, Otozomal Resesif; CVID4 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (TNFRSF13C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 4, 613494, Autosomal recessive; CVID4 (Common variable immunodeficiency) (TNFRSF13C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFRSF13C	TNFRSF13C, BAFFR, CVID4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 5, 613495, Otozomal Resesif; CVID5 (İmmun Yetmezlik, Yaygın Değişken) (MS4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 5, 613495, Autosomal recessive; CVID5 (Common variable immunodeficiency) (MS4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MS4A1	MS4A1, CD20, CVID5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 5 (CVID5) [MIM:613495]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:20038800}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 5, 613495, Otozomal Resesif; CVID5 (İmmun Yetmezlik, Yaygın Değişken) (Prenatal) (MS4A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 5, 613495, Autosomal recessive; CVID5 (Common variable immunodeficiency) (MS4A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MS4A1	MS4A1, CD20, CVID5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 5 (CVID5) [MIM:613495]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:20038800}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

İmmün Yetmezlik, Yaygın Değişken, 6, 613496, Otozomal Resesif; CVID6 (Immun Yetmezlik, Yaygın Değişken) (CD81 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 6, 613496, Autosomal recessive; CVID6 (Common variable immunodeficiency) (CD81 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD81	CD81, TAPA1, CVID6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 6 (CVID6) [MIM:613496]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:20237408}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 6, 613496, Otozomal Resesif; CVID6 (Immun Yetmezlik, Yaygın Değişken) (Prenatal) (CD81 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 6, 613496, Autosomal recessive; CVID6 (Common variable immunodeficiency) (CD81 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD81	CD81, TAPA1, CVID6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 6 (CVID6) [MIM:613496]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:20237408}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 7, 614699, Otozomal Resesif; CVID7 (Immun Yetmezlik, Yaygın Değişken) (CR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 7, 614699, Autosomal recessive; CVID7 (Common variable immunodeficiency) (CR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR2	CR2, C3DR, SLEB9, CVID7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 7 (CVID7) [MIM:614699]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:22035880}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 7, 614699, Otozomal Resesif; CVID7 (Immun Yetmezlik, Yaygın Değişken) (Prenatal) (CR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 7, 614699, Autosomal recessive; CVID7 (Common variable immunodeficiency) (CR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CR2	CR2, C3DR, SLEB9, CVID7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 7 (CVID7) [MIM:614699]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:22035880}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 8, Otoimmünite İle, 614700, Otozomal Resesif; CVID8 (Kombine İmmün Yetmezlik, LRBA Eksikliğine Bağlı) (LRBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, common variable, 8, with autoimmunity, 614700, Autosomal recessive; CVID8 (Combined immunodeficiency due to LRBA deficiency) (LRBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRBA	LRBA, LBA, CDC4L, CVID8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 8, with autoimmunity (CVID8) [MIM:614700]: An autosomal recessive immunologic disorder associated with defective B-cell differentiation and decreased or absent antibody production. Affected individuals have early- childhood onset of recurrent infections, particularly respiratory infections, and also develop variable autoimmune disorders, including idiopathic thrombocytopenic purpura, autoimmune hemolytic anemia, and inflammatory bowel disease. {ECO:0000269	PubMed:22608502}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Yaygın Değişken, 8, Otoimmünite İle, 614700, Otozomal Resesif; CVID8 (Kombine İmmün Yetmezlik, LRBA Eksikliğine Bağlı) (Prenatal) (LRBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, common variable, 8, with autoimmunity, 614700, Autosomal recessive; CVID8 (Combined immunodeficiency due to LRBA deficiency) (LRBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRBA	LRBA, LBA, CDC4L, CVID8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, common variable, 8, with autoimmunity (CVID8) [MIM:614700]: An autosomal recessive immunologic disorder associated with defective B-cell differentiation and decreased or absent antibody production. Affected individuals have early- childhood onset of recurrent infections, particularly respiratory infections, and also develop variable autoimmune disorders, including idiopathic thrombocytopenic purpura, autoimmune hemolytic anemia, and inflammatory bowel disease. {ECO:0000269	PubMed:22608502}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, İzole, 300584 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, İKBKG Eksikliğine Bağlı) (İKBKG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, isolated, 300584 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
İmmün Yetmezlik, İzole, 300584 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, İKBKG Eksikliğine Bağlı) (MLPA)	Immunodeficiency, isolated, 300584 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik, İzole, 300584 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, İKBKG Eksikliğine Bağlı) (Prenatal) (İKBKG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, isolated, 300584 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, İzole, 300584 (X'e Bağlı Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, İKBKG Eksikliğine Bağlı) (Prenatal) (MLPA)	Immunodeficiency, isolated, 300584 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (Prenatal) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Primer, Otozomal Resesif, IL21R-İlgili 615207, Otozomal Resesif (Kriptosporidiosis-Kronik Kolanjit-Karaciğer Hastalığı Sendromu) (IL21R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207, Autosomal recessive (Cryptosporidiosis-chronic cholangitis-liver disease syndrome) (IL21R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL21R	IL21R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: IL21R immunodeficiency (IL21RD) [MIM:615207]: An autosomal recessive primary immunodeficiency characterized by B- and T-cell defects and variable dysfunction of NK cells. Patients tend to have normal numbers of lymphocytes, but show defective class-switched B-cells, low IgG, defective antibody response, and defective T-cell responses to certain antigens. {ECO:0000269	PubMed:23440042}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İmmün Yetmezlik, Primer, Otozomal Resesif, IL21R-İlgili 615207, Otozomal Resesif (Kriptosporidiosis-Kronik Kolanjit-Karaciğer Hastalığı Sendromu) (Prenatal) (IL21R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, primary, autosomal recessive, IL21R-related, 615207, Autosomal recessive (Cryptosporidiosis-chronic cholangitis-liver disease syndrome) (IL21R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL21R	IL21R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: IL21R immunodeficiency (IL21RD) [MIM:615207]: An autosomal recessive primary immunodeficiency characterized by B- and T-cell defects and variable dysfunction of NK cells. Patients tend to have normal numbers of lymphocytes, but show defective class-switched B-cells, low IgG, defective antibody response, and defective T-cell responses to certain antigens. {ECO:0000269	PubMed:23440042}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Hiper-Igm İle, X'e Bağlı, 308230, X'e Bağlı; HGM1 (Hiper-Igm Sendromu-Fmaatu Enfeksiyonlara Yatkınlık) (CD40LG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, X-linked, with hyper-IgM, 308230, X-linked recessive; HIGM1 (Hyper-IgM syndrome with susceptibility to opportunistic infections) (CD40LG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD40LG	TNFSF5, CD40LG, HIGM1, IGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked immunodeficiency with hyper-IgM 1 (HIGM1) [MIM:308230]: Immunoglobulin isotype switch defect characterized by elevated concentrations of serum IgM and decreased amounts of all other isotypes. Affected males present at an early age (usually within the first year of life) recurrent bacterial and opportunistic infections, including Pneumocystis carinii pneumonia and intractable diarrhea due to cryptosporidium infection. Despite substitution treatment with intravenous immunoglobulin, the overall prognosis is rather poor, with a death rate of about 10% before adolescence. {ECO:0000269|PubMed:7532185, ECO:0000269|PubMed:7678782, ECO:0000269|PubMed:7679206, ECO:0000269|PubMed:7679801, ECO:0000269|PubMed:7717401, ECO:0000269|PubMed:8094231, ECO:0000269|PubMed:8550833, ECO:0000269|PubMed:8889581, ECO:0000269|PubMed:9150729, ECO:0000269|PubMed:9746782}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

İmmün Yetmezlik, Hiper-Igm İle, X'e Bağlı, 308230, X'e Bağlı; HIGM1 (Hiper-Igm Sendromu-Füzatç Enfeksiyonlara Yatkınlık) (Prenatal) (CD40LG gen) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, X-linked, with hyper-IgM, 308230, X-linked recessive; HIGM1 (Hyper-IgM syndrome with susceptibility to opportunistic infections) (CD40LG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD40LG	TNFSF5, CD40LG, HIGM1, IGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: X-linked immunodeficiency with hyper-IgM 1 (HIGM1) [MIM:308230]: Immunoglobulin isotype switch defect characterized by elevated concentrations of serum IgM and decreased amounts of all other isotypes. Affected males present at an early age (usually within the first year of life) recurrent bacterial and opportunistic infections, including Pneumocystis carinii pneumonia and intractable diarrhea due to cryptosporidium infection. Despite substitution treatment with intravenous immunoglobulin, the overall prognosis is rather poor, with a death rate of about 10% before adolescence. {ECO:0000269|PubMed:7532185, ECO:0000269|PubMed:7678782, ECO:0000269|PubMed:7679206, ECO:0000269|PubMed:7679801, ECO:0000269|PubMed:7717401, ECO:0000269|PubMed:8094231, ECO:0000269|PubMed:8550833, ECO:0000269|PubMed:8889581, ECO:0000269|PubMed:9150729, ECO:0000269|PubMed:9746782}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Yetmezlik, Magnezyum Defekti, Epstein-Barr Virüsü Enfeksiyonu ve Neoplazi, 300853 İle, X'e Bağlı (X-Bağlı İmmun Yetmezlik-Magnezyum Defekti, Epstein-Barr Virüsü Enfeksiyonu ve Neoplazi) (MAGT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia) (MAGT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAGT1	MAGT1, IAP, XMEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia (XMEN) [MIM:300853]: A disease characterized by CD4 lymphopenia, severe chronic viral infections, and defective T-lymphocyte activation. {ECO:0000269	PubMed:21796205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Yetmezlik, Magnezyum Defekti, Epstein-Barr Virüsü Enfeksiyonu ve Neoplazi, 300853 İle, X'e Bağlı (X-Bağlı İmmun Yetmezlik-Magnezyum Defekti, Epstein-Barr Virüsü Enfeksiyonu ve Neoplazi) (Prenatal) (MAGT1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia, 300853 (X-linked immunodeficiency with magnesium defect, Epstein-Barr virus infection and neoplasia) (MAGT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAGT1	MAGT1, IAP, XMEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency, X-linked, with magnesium defect, Epstein-Barr virus infection and neoplasia (XMEN) [MIM:300853]: A disease characterized by CD4 lymphopenia, severe chronic viral infections, and defective T-lymphocyte activation. {ECO:0000269	PubMed:21796205}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmün Disregülasyon, Poliendokrinopati ve Enteropati, X'e Bağlı, 304790, X'e Bağlı Reseslf; IPEX (İmmun Disregülasyon-Poliendokrinopati-Enteropati Sendromu, X'e Bağlı) (FOXP3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790, X-linked recessive; IPEX (Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome) (FOXP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXP3	FOXP3, IPEX, AIID, XPID, PIDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) [MIM:304790]: Characterized by neonatal onset insulin-dependent diabetes mellitus, infections, secretory diarrhea, thrombocytopenia, anemia and eczema. It is usually lethal in infancy. {ECO:0000269|PubMed:11120765, ECO:0000269|PubMed:11137992, ECO:0000269|PubMed:11137993, ECO:0000269|PubMed:11768393}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmün Disregülasyon, Poliendokrinopati ve Enteropati, X'e Bağlı, 304790, X'e Bağlı Reseslf; IPEX (İmmun Disregülasyon-Poliendokrinopati-Enteropati Sendromu, X'e Bağlı) (Prenatal) (FOXP3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790, X-linked recessive; IPEX (Immune dysregulation-polyendocrinopathy-enteropathy-X-linked syndrome) (FOXP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXP3	FOXP3, IPEX, AIID, XPID, PIDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency polyendocrinopathy, enteropathy, X-linked syndrome (IPEX) [MIM:304790]: Characterized by neonatal onset insulin-dependent diabetes mellitus, infections, secretory diarrhea, thrombocytopenia, anemia and eczema. It is usually lethal in infancy. {ECO:0000269|PubMed:11120765, ECO:0000269|PubMed:11137992, ECO:0000269|PubMed:11137993, ECO:0000269|PubMed:11768393}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmmünoglobulin A Eksikliği 2, 609529; IGAD2 (TNFRSF13B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Immunoglobulin A deficiency 2, 609529; IGAD2 (TNFRSF13B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF13B	TNFRSF13B, TACI, CVID2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunoglobulin A deficiency 2 (IGAD2) [MIM:609529]: Selective deficiency of immunoglobulin A (IGAD) is the most common form of primary immunodeficiency, with an incidence of approximately 1 in 600 individuals in the western world. Individuals with symptomatic IGAD often have deficiency of IgG subclasses or decreased antibody response to carbohydrate antigens such as pneumococcal polysaccharide vaccine. Individuals with IGAD also suffer from recurrent sinopulmonary and gastrointestinal infections and have an increased incidence of autoimmune disorders and of lymphoid and non-lymphoid malignancies. In vitro studies have suggested that some individuals with IGAD have impaired isotype class switching to IgA and others may have a post-switch defect. IGAD and CVID have been known to coexist in families. Some individuals initially present with IGAD1 and then develop CVID. These observations suggest that some cases of IGAD and CVID may have a common etiology. {ECO:0000269	PubMed:16007086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İmmünoglobulin A Eksikliği 2, 609529; IGAD2 (Prenatal) (TNFRSF13B gen) (Tüm Gen Dizi Analizi) (Prenatal)	Immunoglobulin A deficiency 2, 609529; IGAD2 (TNFRSF13B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFRSF13B	TNFRSF13B, TACI, CVID2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunoglobulin A deficiency 2 (IGAD2) [MIM:609529]: Selective deficiency of immunoglobulin A (IGAD) is the most common form of primary immunodeficiency, with an incidence of approximately 1 in 600 individuals in the western world. Individuals with symptomatic IGAD often have deficiency of IgG subclasses or decreased antibody response to carbohydrate antigens such as pneumococcal polysaccharide vaccine. Individuals with IGAD also suffer from recurrent sinopulmonary and gastrointestinal infections and have an increased incidence of autoimmune disorders and of lymphoid and non-lymphoid malignancies. In vitro studies have suggested that some individuals with IGAD have impaired isotype class switching to IgA and others may have a post-switch defect. IGAD and CVID have been known to coexist in families. Some individuals initially present with IGAD1 and then develop CVID. These observations suggest that some cases of IGAD and CVID may have a common etiology. {ECO:0000269	PubMed:16007086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnklüzyon Cisimciği Miyopati-Erken Başlangıç Paget Hastalığı ve Frontotemporal Demans 1, 167320, Otozomal Dominant; IBMPFD1 (İnklüzyon Cisimciklı Miyopati-Kemiğin Paget Hastalığı-Frontotemporal Demans) (VCP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia 1, 167320, Autosomal dominant; IBMPFD1 (Inclusion body myopathy with Paget disease of bone and frontotemporal dementia) (VCP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCP	VCP, IBMPFD1, ALS14, CMT2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 1 (IBMPFD1) [MIM:167320]: An autosomal dominant disease characterized by disabling muscle weakness clinically resembling to limb girdle muscular dystrophy, osteolytic bone lesions consistent with Paget disease, and premature frontotemporal dementia. Clinical features show incomplete penetrance. {ECO:0000269|PubMed:15034582, ECO:0000269|PubMed:15732117, ECO:0000269|PubMed:16247064}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnklüzyon Cisimciklı Miyopati-Erken Başlangıçlı Paget Hastalığı ve Frontotemporal Demans 2, 615422; IBMPFD2 (İnklüzyon Cisimciklı Miyopati-Kemiğin Paget Hastalığı-Frontotemporal Demans) (HNRNPA2B1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2, 615422; IBMPFD2 (Inclusion body myopathy with Paget disease of bone and frontotemporal dementia) (HNRNPA2B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPA2B1	HNRPA2B1, IBMPFD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 2 (IBMPFD2) [MIM:615422]: An autosomal dominant disease characterized by disabling muscle weakness clinically resembling to limb girdle muscular dystrophy, osteolytic bone lesions consistent with Paget disease, and premature frontotemporal dementia. Clinical features show incomplete penetrance. {ECO:0000269	PubMed:23455423}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnklüzyon Cisimciklı Miyopati-Erken Başlangıçlı Paget Hastalığı, Frontotemporal Demans Olmadan 3, 615424, Otozomal Dominant; IBMPFD3 (İnklüzyon Cisimciklı Miyopati-Kemiğin Paget Hastalığı-Frontotemporal Demans) (HNRNPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inclusion body myopathy with early-onset Paget disease without frontotemporal dementia 3, 615424, Autosomal dominant; IBMPFD3 (Inclusion body myopathy with Paget disease of bone and frontotemporal dementia) (HNRNPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPA1	HNRNPA1, IBMPFD3, ALS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inclusion body myopathy with early-onset Paget disease with or without frontotemporal dementia 3 (IBMPFD3) [MIM:615424]: An autosomal dominant disease characterized by disabling muscle weakness clinically resembling to limb girdle muscular dystrophy, osteolytic bone lesions consistent with Paget disease, and premature frontotemporal dementia. Clinical features show incomplete penetrance. {ECO:0000269	PubMed:23455423}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnkontinentia Pigmenti, 308300, X'e Bağlı Dominant; IP (İnkontinentia Pigmenti) (IKBKG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Incontinentia pigmenti, 308300, X-linked dominant; IP (incontinentia pigmenti) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
İnkontinentia Pigmenti, 308300, X'e Bağlı Dominant; IP (İnkontinentia Pigmenti) (MLPA)	Incontinentia pigmenti, 308300, X-linked dominant; IP (incontinentia pigmenti) (MLPA)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAID) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Büyüme Hormonuna Yanıt Artması (GHR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Increased responsiveness to growth hormone (GHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHR	GHR, GHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laron syndrome (LARS) [MIM:262500]: A severe form of growth hormone insensitivity characterized by growth impairment, short stature, dysfunctional growth hormone receptor, and failure to generate insulin-like growth factor I in response to growth hormone. {ECO:0000269|PubMed:10670033, ECO:0000269|PubMed:14678285, ECO:0000269|PubMed:2779634, ECO:0000269|PubMed:8137822, ECO:0000269|PubMed:8421103, ECO:0000269|PubMed:8504296, ECO:0000269|PubMed:9024232, ECO:0000269|PubMed:9661642, ECO:0000269|PubMed:9851797}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

İnfantil Serebellar-Retinal Dejenerasyon, 614559, Otozomal Resesif, ICRD (İnfantil Serebellar-Retinal Dejenerasyon) (ACO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	İnfantile cerebellar-retinal degeneration, 614559, Autosomal recessive; ICRD (Infantile cerebellar-retinal degeneration) (ACO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACO2	ACO2, ICRD, OPA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile cerebellar-retinal degeneration (ICRD) [MIM:614559]: A severe autosomal recessive neurodegenerative disorder characterized by onset between ages 2 and 6 months of truncal hypotonia, athetosis, seizures, and ophthalmologic abnormalities, particularly optic atrophy and retinal degeneration. Affected individuals show profound psychomotor retardation, with only some achieving rolling, sitting, or recognition of family. Brain MRI shows progressive cerebral and cerebellar degeneration. {ECO:0000269|PubMed:22405087, ECO:0000269|PubMed:25351951}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnfantil Serebellar-Retinal Dejenerasyon, 614559, Otozomal Resesif, ICRD (İnfantil Serebellar-Retinal Dejenerasyon) (Prenatal) (ACO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Infantile cerebellar-retinal degeneration, 614559, Autosomal recessive; ICRD (Infantile cerebellar-retinal degeneration) (ACO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACO2	ACO2, ICRD, OPA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile cerebellar-retinal degeneration (ICRD) [MIM:614559]: A severe autosomal recessive neurodegenerative disorder characterized by onset between ages 2 and 6 months of truncal hypotonia, athetosis, seizures, and ophthalmologic abnormalities, particularly optic atrophy and retinal degeneration. Affected individuals show profound psychomotor retardation, with only some achieving rolling, sitting, or recognition of family. Brain MRI shows progressive cerebral and cerebellar degeneration. {ECO:0000269|PubMed:22405087, ECO:0000269|PubMed:25351951}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil Karaciğer Yetmezliği Sendromu 1, 615438, Otozomal Resesif; ILFS1 (Akut İnfantil Karaciğer Yetmezliği-Multisistemik Tutulum Sendromu) (LARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Infantile liver failure syndrome 1, 615438, Autosomal recessive; ILFS1 (Acute infantile liver failure-multisystemic involvement syndrome) (LARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARS	LARS, LFIS, ILFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile liver failure syndrome 1 (ILFS1) [MIM:615438]: A life-threatening disorder of hepatic function that manifests with acute liver failure in the first few months of life. Clinical features include anemia, renal tubulopathy, developmental delay, seizures, failure to thrive, and liver steatosis and fibrosis. {ECO:0000269|PubMed:22607940}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnfantil Karaciğer Yetmezliği Sendromu 1, 615438, Otozomal Resesif; ILFS1 (Akut İnfantil Karaciğer Yetmezliği-Multisistemik Tutulum Sendromu) (Prenatal) (LARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Infantile liver failure syndrome 1, 615438, Autosomal recessive; ILFS1 (Acute infantile liver failure-multisystemic involvement syndrome) (LARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARS	LARS, LFIS, ILFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile liver failure syndrome 1 (ILFS1) [MIM:615438]: A life-threatening disorder of hepatic function that manifests with acute liver failure in the first few months of life. Clinical features include anemia, renal tubulopathy, developmental delay, seizures, failure to thrive, and liver steatosis and fibrosis. {ECO:0000269|PubMed:22607940}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil Karaciğer Yetmezliği Sendromu 2, 616483, Otozomal Resesif; ILFS2 (Ateş İlişkili Akut İnfantil Karaciğer Yetmezliği Sendromu) (NBAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Infantile liver failure syndrome 2, 616483, Autosomal recessive; ILFS2 (Fever-associated acute infantile liver failure syndrome) (NBAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBAS	NBAS, NAG, SOPH, ILFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile liver failure syndrome 2 (ILFS2) [MIM:616483]: A form of infantile liver failure syndrome, a life-threatening disorder of hepatic function that manifests with acute liver failure in the first few months of life. Clinical features include anemia, renal tubulopathy, developmental delay, seizures, failure to thrive, and liver steatosis and fibrosis. {ECO:0000269|PubMed:26073778}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnfantil Karaciğer Yetmezliği Sendromu 2, 616483, Otozomal Resesif; ILFS2 (Ateş İlişkili Akut İnfantil Karaciğer Yetmezliği Sendromu) (Prenatal) (NBAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Infantile liver failure syndrome 2, 616483, Autosomal recessive; ILFS2 (Fever-associated acute infantile liver failure syndrome) (NBAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NBAS	NBAS, NAG, SOPH, ILFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile liver failure syndrome 2 (ILFS2) [MIM:616483]: A form of infantile liver failure syndrome, a life-threatening disorder of hepatic function that manifests with acute liver failure in the first few months of life. Clinical features include anemia, renal tubulopathy, developmental delay, seizures, failure to thrive, and liver steatosis and fibrosis. {ECO:0000269|PubMed:26073778}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil nöroaksonal distrofi (Beyin demir birikmesi-nörodejenerasyon) paneli (PANK2 20q13, PLA2G6 22q13) (MLPA)	İnfantile neuroaxonal dystrophy (Neurodegeneration with brain iron accumulation (NBIA) (PANK2 20q13, PLA2G6 22q13) (MLPA)	PANK2 20q13, PLA2G6 22q13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
İnfantil nöroaksonal distrofi (Beyin demir birikmesi-nörodejenerasyon) paneli (PANK2 20q13, PLA2G6 22q13) (MLPA) (Prenatal)	İnfantile neuroaxonal dystrophy (Neurodegeneration with brain iron accumulation (NBIA) (PANK2 20q13, PLA2G6 22q13) (MLPA) (Prenatal)	PANK2 20q13, PLA2G6 22q13	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil Nöroaksonal Distrofi 1, 256600, Otozomal Resesif; NBIA2A (İnfantil Nöroaksonal Distrofi) (PLA2G6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Infantile neuroaxonal dystrophy 1, 256600, Autosomal recessive; NBIA2A (Infantile neuroaxonal dystrophy) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnfantil Nöroaksonal Distrofi 1, 256600, Otozomal Resesif; NBIA2A (İnfantil Nöroaksonal Distrofi) (MLPA)	İnfantile neuroaxonal dystrophy 1, 256600, Autosomal recessive; NBIA2A (Infantile neuroaxonal dystrophy) (MLPA)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnfantil Nöroaksonal Distrofi 1, 256600, Otozomal Resesif; NBIA2A (İnfantil Nöroaksonal Distrofi) (Prenatal) (PLA2G6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	İnfantile neuroaxonal dystrophy 1, 256600, Autosomal recessive; NBIA2A (Infantile neuroaxonal dystrophy) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil Nöroaksonal Distrofi 1, 256600, Otozomal Resesif; NBIA2A (İnfantil Nöroaksonal Distrofi) (PLA2G6) (Prenatal) (MLPA)	İnfantile neuroaxonal dystrophy 1, 256600, Autosomal recessive; NBIA2A (Infantile neuroaxonal dystrophy) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnfantil Başlangıçlı Multisistem Nörolojik, Endokrin ve Pankreatik Hastalıklar, 616263, Otozomal Resesif (İnfantil Multisistem Nörolojik-Endokrin-Pankreatik Hastalık) (PTRH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263, Autosomal recessive (Infantile multisystem neurologic-endocrine-pancreatic disease) (PTRH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTRH2	PTRH2, BIT1, IMNEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset (IMNEPD) [MIM:616263]: A progressive multisystem disease characterized by a variety of neurologic, endocrine, and, in some patients, pancreatic features. Variable clinical symptoms include global developmental delay, hypotonia, hearing loss, ataxia, hyporeflexia, facial dysmorphism, hypothyroidism, and pancreatic insufficiency. {ECO:0000269|PubMed:25558065, ECO:0000269|PubMed:25574476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnfantil Başlangıçlı Multisistem Nörolojik, Endokrin ve Pankreatik Hastalıklar, 616263, Otozomal Resesif (İnfantil Multisistem Nörolojik-Endokrin-Pankreatik Hastalık) (Prenatal) (PTRH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Infantile-onset multisystem neurologic, endocrine, and pancreatic disease, 616263, Autosomal recessive (Infantile multisystem neurologic-endocrine-pancreatic disease) (PTRH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTRH2	PTRH2, BIT1, IMNEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurologic, endocrine, and pancreatic disease, multisystem, infantile-onset (IMNEPD) [MIM:616263]: A progressive multisystem disease characterized by a variety of neurologic, endocrine, and, in some patients, pancreatic features. Variable clinical symptoms include global developmental delay, hypotonia, hearing loss, ataxia, hyporeflexia, facial dysmorphism, hypothyroidism, and pancreatic insufficiency. {ECO:0000269|PubMed:25558065, ECO:0000269|PubMed:25574476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Enfeksiyonlar, Tekrarlayan, Ensefalopati, Hepatik Disfonksiyon ve Kardiyovasküler Malformasyonlar, 613759, Otozomal Resesif (FADD İle İlişkili İmmun Yetmezlik) (FADD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759, Autosomal recessive (FADD-related immunodeficiency) (FADD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FADD	FADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infections, recurrent, associated with encephalopathy, hepatic dysfunction and cardiovascular malformations (IEHDCM) [MIM:613759]: A condition with biological features of autoimmune lymphoproliferative syndrome such as high-circulating CD4(-)CD8(-)TCR-alpha-beta(+) T-cell counts, and elevated IL10 and FASL levels. Affected individuals suffer from recurrent, stereotypical episodes of fever, encephalopathy, and mild liver dysfunction sometimes accompanied by generalized seizures. The episodes can be triggered by varicella zoster virus (VZV), measles mumps rubella (MMR) attenuated vaccine, parainfluenza virus, and Epstein-Barr virus (EBV). {ECO:0000269|PubMed:21109225}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Enfeksiyonlar, Tekrarlayan, Ensefalopati, Hepatik Disfonksiyon ve Kardiyovasküler Malformasyonlar, 613759, Otozomal Resesif (FADD İle İlişkili İmmun Yetmezlik) (Prenatal) (FADD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Infections, recurrent, with encephalopathy, hepatic dysfunction, and cardiovascular malformations, 613759, Autosomal recessive (FADD-related immunodeficiency) (FADD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FADD	FADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infections, recurrent, associated with encephalopathy, hepatic dysfunction and cardiovascular malformations (IEHDCM) [MIM:613759]: A condition with biological features of autoimmune lymphoproliferative syndrome such as high-circulating CD4(-)CD8(-)TCR-alpha-beta(+) T-cell counts, and elevated IL10 and FASL levels. Affected individuals suffer from recurrent, stereotypical episodes of fever, encephalopathy, and mild liver dysfunction sometimes accompanied by generalized seizures. The episodes can be triggered by varicella zoster virus (VZV), measles mumps rubella (MMR) attenuated vaccine, parainfluenza virus, and Epstein-Barr virus (EBV). {ECO:0000269|PubMed:21109225}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnflamatuvar Barsak Hastalığı (Crohn Hastalığı), 10, 611081; IBD10 (ATG16L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease (Crohn disease) 10, 611081; IBD10 (ATG16L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATG16L1	ATG16L1, APG16L, IBD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 10 (IBD10) [MIM:611081]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistules. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:17200669, ECO:0000269|PubMed:17435756}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample

İnflamatuvar Barsak Hastalığı (Crohn Hastalığı) 19, 612278; IBD19 (IRGM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease (Crohn disease) 19, 612278; IBD19 (IRGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRGM	IRGM, LRG47, IFI1, IBD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 19 (IBD19) [MIM:612278]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:17554261, ECO:0000269|PubMed:19174780, ECO:0000269|PubMed:21278745}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı 1, Crohn Hastalığı, 266600, Multifaktöryel (NOD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 1, Crohn disease, 266600, Multifactorial (NOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOD2	NOD2, CARD15, IBD1, CD, YAOS, BLAUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blau syndrome (BLAUS) [MIM:186580]: A rare autosomal dominant disorder characterized by early-onset granulomatous arthritis, uveitis and skin rash. {ECO:0000269|PubMed:11528384, ECO:0000269|PubMed:15812565, ECO:0000269|PubMed:19116920, ECO:0000269|PubMed:19169908, ECO:0000269|PubMed:19479837, ECO:0000269|PubMed:20199415, ECO:0000269|PubMed:24960071, ECO:0000269|PubMed:25093298, ECO:0000269|PubMed:25692065, ECO:0000269|PubMed:25724124}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı 13, 612244; IBD13 (ABCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 13, 612244; IBD13 (ABCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB1	ABCB1, PGY1, MDR1, IBD13, CLCs	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 13 (IBD13) [MIM:612244]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı 14, 612245; IBD14 (IRF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 14, 612245; IBD14 (IRF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF5	IRF5, IBD14, SLEB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis (RA) [MIM:180300]: An inflammatory disease with autoimmune features and a complex genetic component. It primarily affects the joints and is characterized by inflammatory changes in the synovial membranes and articular structures, widespread fibrinoid degeneration of the collagen fibers in mesenchymal tissues, and by atrophy and rarefaction of bony structures. {ECO:0000269|PubMed:17599733}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı, 17 Karşı Koruma, 612261 (IL23R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 17, protection against, 612261 (IL23R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL23R	IL23R, IBD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 17 (IBD17) [MIM:612261]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:17068223, ECO:0000269|PubMed:17804789}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı 25, Erken Başlangıç, Otozomal Resesif, 612567, Otozomal Resesif (Otozomal Resesif Erken Başlangıçlı İnflamatuar Bağırsak Hastalığı) (IL10RB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 25, early onset, autosomal recessive, 612567, Autosomal recessive (Autosomal recessive early-onset inflammatory bowel disease) (IL10RB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10RB	CRFB4, IBD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 25 (IBD25) [MIM:612567]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:18690111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Barsak Hastalığı 28 Erken Başlangıçlı, Otozomal Resesif, 613148, Otozomal Resesif (Otozomal Resesif Erken Başlangıçlı İnflamatuar Bağırsak Hastalığı) (IL10RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory bowel disease 28, early onset, autosomal recessive, 613148, Autosomal recessive (Autosomal recessive early-onset inflammatory bowel disease) (IL10RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10RA	IL10RA, IL10R, IBD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İnflamatuvar Deri ve Barsak Hastalığı, Neonatal, 1, 614328, Otozomal Resesif; NISBD1 (Neonatal İnflamatuar Deri ve Bağırsak Hastalığı) (ADAM17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory skin and bowel disease, neonatal, 1, 614328, Autosomal recessive; NISBD1 (Neonatal inflammatory skin and bowel disease) (ADAM17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAM17	ADAM17, TACE, NISBD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory skin and bowel disease, neonatal, 1 (NISBD1) [MIM:614328]: A disorder characterized by inflammatory features with neonatal onset, involving the skin, hair, and gut. The skin lesions involve perioral and perianal erythema, psoriasiform erythroderma, with flares of erythema, scaling, and widespread pustules. Gastrointestinal symptoms include malabsorptive diarrhea that is exacerbated by intercurrent gastrointestinal infections. The hair is short or broken, and the eyelashes and eyebrows are wiry and disorganized. {ECO:0000269|PubMed:22010916}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnflamatuvar Deri ve Barsak Hastalığı, Neonatal, 2, 616069, Otozomal Resesif; NISBD2 (Neonatal İnflamatuar Deri ve Bağırsak Hastalığı) (EGFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Inflammatory skin and bowel disease, neonatal, 2, 616069, Autosomal recessive; NISBD2 (Neonatal inflammatory skin and bowel disease) (EGFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGFR	EGFR, NISBD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lung cancer (LNCR) [MIM:211980]: A common malignancy affecting tissues of the lung. The most common form of lung cancer is non-small cell lung cancer (NSCLC) that can be divided into 3 major histologic subtypes: squamous cell carcinoma, adenocarcinoma, and large cell lung cancer. NSCLC is often diagnosed at an advanced stage and has a poor prognosis. {ECO:0000269|PubMed:15118125, ECO:0000269|PubMed:16533793, ECO:0000269|PubMed:16672372}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Influenza, Ağır, Yatkınlık, 614680 (IFITM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Influenza, severe, susceptibility to, 614680 (IFITM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFITM3	IFITM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ağrı Duyarsızlığı, Konjenital, 243000, Otozomal Resesif (Kanalopati İlişkili Konjenital Ağrı Duyarsızlığı) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insensitivity to pain, congenital, 243000, Autosomal recessive (Channelopathy-associated congenital insensitivity to pain) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağrı Duyarsızlığı, Konjenital, 243000, Otozomal Resesif (Kanalopati İlişkili Konjenital Ağrı Duyarsızlığı) (Prenatal) (SCN9A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Insensitivity to pain, congenital, 243000, Autosomal recessive (Channelopathy-associated congenital insensitivity to pain) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (IKİSİ BIRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağrı Duyarsızlığı, Konjenital, Anhidrosis İle, 256800, Otozomal Resesif; CIPA (Aklesel Duysal ve Otonomik Nöropati Tip 4) (NTRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insensitivity to pain, congenital, with anhidrosis, 256800, Autosomal recessive; CIPA (Hereditary sensory and autonomic neuropathy type 4) (NTRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTRK1	NTRK1, TRKA, MTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital insensitivity to pain with anhidrosis (CIPA) [MIM:256800]: Characterized by a congenital insensitivity to pain, anhidrosis (absence of sweating), absence of reaction to noxious stimuli, self-mutilating behavior, and mental retardation. This rare autosomal recessive disorder is also known as congenital sensory neuropathy with anhidrosis or hereditary sensory and autonomic neuropathy type IV or familial dysautonomia type II. {ECO:0000269|PubMed:10090906, ECO:0000269|PubMed:10233776, ECO:0000269|PubMed:10330344, ECO:0000269|PubMed:10567924, ECO:0000269|PubMed:10861667, ECO:0000269|PubMed:10982191, ECO:0000269|PubMed:11310631, ECO:0000269|PubMed:22302274, ECO:0000269|PubMed:8696348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ağrı Duyarsızlığı, Konjenital, Anhidrosis İle, 256800, Otozomal Resesif, CIPA (Allesel Duysal ve Otonomik Nöropati Tip 4) (Prenatal) (NTRK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Insensitivity to pain, congenital, with anhidrosis, 256800, Autosomal recessive; CIPA (Hereditary sensory and autonomic neuropathy type 4) (NTRK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NTRK1	NTRK1, TRKA, MTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital insensitivity to pain with anhidrosis (CIPA) [MIM:256800]: Characterized by a congenital insensitivity to pain, anhidrosis (absence of sweating), absence of reaction to noxious stimuli, self-mutilating behavior, and mental retardation. This rare autosomal recessive disorder is also known as congenital sensory neuropathy with anhidrosis or hereditary sensory and autonomic neuropathy type IV or familial dysautonomia type II. {ECO:0000269|PubMed:10090906, ECO:0000269|PubMed:10233776, ECO:0000269|PubMed:10330344, ECO:0000269|PubMed:10567924, ECO:0000269|PubMed:10861667, ECO:0000269|PubMed:10982191, ECO:0000269|PubMed:11310631, ECO:0000269|PubMed:22302274, ECO:0000269|PubMed:8696348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Uykusuzluk, Fatal Familyal, 600072, Otozomal Dominant (Fatal Allesel Insomnia) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insomnia, fatal familial, 600072, Autosomal dominant (Fatal familial insomnia) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
İnsülin Direnci, Şiddetli, Dijenik, 125853, Otozomal Resesif (PPP1R3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insulin resistance, severe, digenic, 125853, Autosomal dominant (PPP1R3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP1R3A	PPP1R3A, PPP1R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:7926294}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
İnsülin Direnci, Şiddetli, Dijenik, 604367, Otozomal Dominant (PPARG İle İlişkili Allesel Parsiyel Lipodistrofi) (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insulin resistance, severe, digenic, 604367, Autosomal dominant (PPARG-related familial partial lipodystrophy) (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glioma 1 (GLM1) [MIM:137800]: Gliomas are benign or malignant central nervous system neoplasms derived from glial cells. They comprise astrocytomas and glioblastoma multiforme that are derived from astrocytes, oligodendrogliomas derived from oligodendrocytes and ependymomas derived from ependymocytes. {ECO:0000269	PubMed:10851250}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Polymorphic PPARG alleles have been found to be significantly over-represented among a cohort of American patients with sporadic glioblastoma multiforme suggesting a possible contribution to disease susceptibility.;	EDTA Blood Sample
İnsülin Direnci, Yatkınlık, 125853, Otozomal Dominant (PTPN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insulin resistance, susceptibility to, 125853, Autosomal dominant (PTPN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN1	PTPN1, PTP1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İnsülin-Benzeri Büyüme Faktörü I, Direnç, 270450, Otozomal Resesif, Otozomal Dominant (Büyüme Gecikmesi, İnsülin Benzeri Büyüme Faktörü Tip 1 (IGF1) Direncine Bağlı) (IGF1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Insulin-like growth factor I, resistance to, 270450, Autosomal recessive, Autosomal dominant (Growth delay due to insulin-like growth factor I resistance) (IGF1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGF1R	IGF1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Insulin-like growth factor 1 resistance (IGF1RES) [MIM:270450]: A disorder characterized by intrauterine growth retardation, poor postnatal growth and increased plasma IGF1 levels. {ECO:0000269|PubMed:14657428, ECO:0000269|PubMed:15923254}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Entellektüel Gelişim Bozukluğu, Kardiyak Aritmi, 617173, Otozomal Resesif; IDDCA (GNB5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intellectual developmental disorder with cardiac arrhythmia, 617173, Autosomal recessive; IDDCA (GNB5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB5	GNB5, GB5, IDDCA, LADCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Entellektüel Gelişim Bozukluğu, Kardiyak Aritmi, 617173, Otozomal Resesif; IDDCA (Prenatal) (GNB5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Intellectual developmental disorder with cardiac arrhythmia, 617173, Autosomal recessive; IDDCA (GNB5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNB5	GNB5, GB5, IDDCA, LADCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Entellektüel Gelişim Bozukluğu, Dismorfik Yüz ve Pitozis, 617333, Otozomal Dominant; IDDDFP (BRPF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333, Autosomal dominant; IDDDFP (BRPF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRPF1	BRPF1, BR140, IDDDFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Entellektüel Gelişim Bozukluğu, Dismorfik Yüz ve Pitozis, 617333, Otozomal Dominant; IDDDFP (Prenatal) (BRPF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Intellectual developmental disorder with dysmorphic facies and ptosis, 617333, Autosomal dominant; IDDDFP (BRPF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRPF1	BRPF1, BR140, IDDDFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnterferon Alfa, Eksikliği (IFNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Interferon, alpha, deficiency (IFNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNA1	IFNA1, IFNA@	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İnterferon Alfa, Eksikliği (Prenatal) (IFNA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Interferon, alpha, deficiency (IFNA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFNA1	IFNA1, IFNA@	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnterlökin-1 Reseptör Antagonist Yetersizliği, 612852, Otozomal Resesif (Steril Multifokal Osteomiyelit-Periostit ve Püstülozis) (IL1RN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Interleukin 1 receptor antagonist deficiency, 612852, Autosomal recessive (Sterile multifocal osteomyelitis with periostitis and pustulosis) (IL1RN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL1RN	IL1RN, MVCD4, DIRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Interleukin 1 receptor antagonist deficiency (DIRA) [MIM:612852]: A rare autoinflammatory disease of skin and bone resulting in sterile multifocal osteomyelitis, periostitis, and pustulosis from birth. The term autoinflammatory disease describes a group of disorders characterized by attacks of seemingly unprovoked inflammation without significant levels of autoantibodies and autoreactive T-cells. {ECO:0000269	PubMed:19494218}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnterlökin-1 Reseptör Antagonist Yetersizliği, 612852, Otozomal Resesif (Steril Multifokal Osteomiyelit-Periostit ve Püstülozis) (Prenatal) (IL1RN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Interleukin 1 receptor antagonist deficiency, 612852, Autosomal recessive (Sterile multifocal osteomyelitis with periostitis and pustulosis) (IL1RN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL1RN	IL1RN, MVCD4, DIRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Interleukin 1 receptor antagonist deficiency (DIRA) [MIM:612852]: A rare autoinflammatory disease of skin and bone resulting in sterile multifocal osteomyelitis, periostitis, and pustulosis from birth. The term autoinflammatory disease describes a group of disorders characterized by attacks of seemingly unprovoked inflammation without significant levels of autoantibodies and autoreactive T-cells. {ECO:0000269	PubMed:19494218}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnterstisyel Akciğer ve Karaciğer Hastalığı, 615486, Otozomal Resesif; ILLD (Ağır Erken Başlangıçlı Pulmoner Alveoler Proteinozis, MARS Eksikliğine Bağlı) (MARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Interstitial lung and liver disease, 615486, Autosomal recessive; ILLD (Severe early-onset pulmonary alveolar proteinosis due to MARS dorficiency) (MARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARS	MARS, MTRNS, METRS, ILLD, CMT2U	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Interstitial lung and liver disease (ILLD) [MIM:615486]: An autosomal recessive, life-threatening disorder characterized by respiratory insufficiency and progressive liver disease with onset in infancy or early childhood. Clinical features include failure to thrive, hypotonia, intermittent lactic acidosis, aminoaciduria, hypothyroidism, interstitial lung disease, pulmonary alveolar proteinosis, anemia, and liver canalicular cholestasis, steatosis, and iron deposition. {ECO:0000269	PubMed:24103465}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İnterstisyel Akciğer Hastalığı, Nefrotik Sendrom, ve Epidermolitiz Büllöza, Konjenital, 614748, Otozomal Resesif; ILNEB (Konjenital Nefrotik Sendrom-İnterstisyel Akciğer Hastalığı-Epidermolitis Bulosa Sendromu) (ITGA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748, Autosomal recessive; ILNEB (Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome) (ITGA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA3	ITGA3, CD49C, GAPB3, ILNEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital (ILNEB) [MIM:614748]: A multiorgan disorder characterized by congenital nephrotic syndrome, interstitial lung disease, and epidermolysis bullosa. The respiratory and renal features predominate, and lung involvement accounts for the lethal course of the disease. {ECO:0000269	PubMed:22512483}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnterstisyel Akciğer Hastalığı, Nefrotik Sendrom, ve Epidermolitiz Büllöza, Konjenital, 614748, Otozomal Resesif; ILNEB (Konjenital Nefrotik Sendrom-İnterstisyel Akciğer Hastalığı-Epidermolitis Bulosa Sendromu) (Prenatal) (ITGA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital, 614748, Autosomal recessive; ILNEB (Congenital nephrotic syndrome-interstitial lung disease-epidermolysis bullosa syndrome) (ITGA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGA3	ITGA3, CD49C, GAPB3, ILNEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Interstitial lung disease, nephrotic syndrome, and epidermolysis bullosa, congenital (ILNEB) [MIM:614748]: A multiorgan disorder characterized by congenital nephrotic syndrome, interstitial lung disease, and epidermolysis bullosa. The respiratory and renal features predominate, and lung involvement accounts for the lethal course of the disease. {ECO:0000269	PubMed:22512483}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnterstisyel Nefrit, Karyomegalik, 614817, Otozomal Resesif; KMIN (Karyomegalik İnterstisyel Nefrit) (FAN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Interstitial nephritis, karyomegalic, 614817, Autosomal recessive; KMIN (Karyomegalic interstitial nephritis) (FAN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAN1	FAN1, MTMR15, KIAA1018, KMIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Interstitial nephritis, karyomegalic (KMIN) [MIM:614817]: A rare kidney disease characterized by chronic tubulointerstitial nephritis associated with massively enlarged tubular epithelial cell nuclei. The clinical picture is associated with recurrent upper respiratory tract infections in addition to chronic kidney disease beginning in the third decade of life. {ECO:0000269	PubMed:22772369}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Intervertebral Disk Hastalığı, Yatkınlık, 603932; IDD (COL9A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intervertebral disc disease, susceptibility to, 603932; IDD (COL9A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A3	COL9A3, EDM3, IDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 3 (EDM3) [MIM:600969]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269	PubMed:10090888}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İntestinal Psödoobstrüksiyon, Nöronal, 300048, X'e Bağlı Resesif (Kronik Intestinal Psödoobstrüksiyon) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intestinal pseudoobstruction, neuronal, 300048, X-linked recessive (Chronic intestinal pseudoobstruction) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İntestinal Psödoobstrüksiyon, Nöronal, 300048, X'e Bağlı Resesif (Kronik Intestinal Psödoobstrüksiyon) (MLPA)	Intestinal pseudoobstruction, neuronal, 300048, X-linked recessive (Chronic intestinal pseudoobstruction) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Beyin Serebrovasküler Malformasyonlar Intrakranial Kanama, Yatkınlık, 108010, Otozomal Dominant (Serebral Arteriovendz Malformasyon) (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intracranial hemorrhage in brain cerebrovascular malformations, susceptibility to, 108010, Autosomal dominant (Cerebral arteriovenous malformation) (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis systemic juvenile (RASJ) [MIM:604302]: An inflammatory articular disorder with systemic-onset beginning before the age of 16. It represents a subgroup of juvenile arthritis associated with severe extraarticular features and occasionally fatal complications. During active phases of the disorder, patients display a typical daily spiking fever, an evanescent macular rash, lymphadenopathy, hepatosplenomegaly, serositis, myalgia and arthritis. {ECO:0000269	PubMed:9769329}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
İntrinsik Faktör Eksikliği, 261000, Otozomal Resesif; IFD (Konjenital İntrinsek Faktör Eksikliği) (GIF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Intrinsic factor deficiency, 261000, Autosomal recessive; IFD (Congenital intrinsic factor deficiency) (GIF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GIF	GIF, IF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
inv(16) (p13;q22) (CBFB/ MYH11)/ t(16;16) (p13;q22) (CBFB/ MYH11) FISH	inv (16) (p13;q22) (CBFB / MYH11)/ t (16;16) (p13;q22) (CBFB / MYH11) (FISH)	16p13-16q22	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal		Heparinized blood sample / Heparinized bone marrow
inv(16) (p13;q22) (CBFB/ MYH11)/ t(16;16) (p13;q22) (CBFB/ MYH11) (REAL - TIME PCR)	inv (16) (p13;q22) (CBFB / MYH11)/ t (16;16) (p13;q22) (CBFB / MYH11) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
İnvasif Pnömokok Hastalığı, Tekrarlayan İzole, 1, 610799; IPD1 (IRAK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Invasive pneumococcal disease, recurrent isolated, 1, 610799; IPD1 (IRAK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRAK4	IRAK4, REN64, IPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: IRAK4 deficiency (IRAK4D) [MIM:607676]: Causes extracellular pyogenic bacterial and fungal infections in otherwise healthy children. {ECO:0000269|PubMed:12637671, ECO:0000269|PubMed:12925671, ECO:0000269|PubMed:17878374, ECO:0000269|PubMed:19663824, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnvasif Pnömokok Hastalığı, Tekrarlayan İzole, 1, 610799; IPD1 (Prenatal) (IRAK4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Invasive pneumococcal disease, recurrent isolated, 1, 610799; IPD1 (IRAK4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRAK4	IRAK4, REN64, IPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: IRAK4 deficiency (IRAK4D) [MIM:607676]: Causes extracellular pyogenic bacterial and fungal infections in otherwise healthy children. {ECO:0000269|PubMed:12637671, ECO:0000269|PubMed:12925671, ECO:0000269|PubMed:17878374, ECO:0000269|PubMed:19663824, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnvasif Pnömokok Hastalığı, Tekrarlayan İzole, 2, 300640; IPD2 (X'e Bağlı Mikobakteriyel Hastalıklara Mendeleyen Yatkınlık, IKBKG Eksikliğine Bağlı) (IKBKG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Invasive pneumococcal disease, recurrent isolated, 2, 300640; IPD2 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAD) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ On Ice
İnvasif Pnömokok Hastalığı, Tekrarlayan İzole, 2, 300640; IPD2 (X'e Bağlı Mikobakteriyel Hastalıklara Mendeleyen Yatkınlık, IKBKG Eksikliğine Bağlı) (Prenatal) (IKBKG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Invasive pneumococcal disease, recurrent isolated, 2, 300640; IPD2 (X-linked mendelian susceptibility to mycobacterial diseases due to IKBKG deficiency) (IKBKG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IKBKG	IKBKG, NEMO, FIP3, IP, IPD2, AMCBX1, IMD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, anhidrotic, with immunodeficiency X-linked (EDAD) [MIM:300291]: A form of ectoderma dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by absence of sweat glands, sparse scalp hair, rare conical teeth and immunological abnormalities resulting in severe infectious diseases. {ECO:0000269|PubMed:11047757, ECO:0000269|PubMed:11224521, ECO:0000269|PubMed:11242109, ECO:0000269|PubMed:12045264, ECO:0000269|PubMed:15100680}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
IRAK4 Eksikliği, 607676 (İmmun Yetmezlik, Interlökin-1 Reseptörü İle Bağlantılı Kinaz-4 Eksikliğine Bağlı (IRAK4 Eksikliği)) (IRAK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	IRAK4 deficiency, 607676 (Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency) (IRAK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRAK4	IRAK4, REN64, IPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: IRAK4 deficiency (IRAK4D) [MIM:607676]: Causes extracellular pyogenic bacterial and fungal infections in otherwise healthy children. {ECO:0000269|PubMed:12637671, ECO:0000269|PubMed:12925671, ECO:0000269|PubMed:17878374, ECO:0000269|PubMed:19663824, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
IRAK4 Eksikliği, 607676 (İmmun Yetmezlik, Interlökin-1 Reseptörü İle Bağlantılı Kinaz-4 Eksikliğine Bağlı (IRAK4 Eksikliği)) (Prenatal) (IRAK4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	IRAK4 deficiency, 607676 (Immunodeficiency due to interleukin-1 receptor-associated kinase-4 deficiency) (IRAK4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRAK4	IRAK4, REN64, IPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: IRAK4 deficiency (IRAK4D) [MIM:607676]: Causes extracellular pyogenic bacterial and fungal infections in otherwise healthy children. {ECO:0000269|PubMed:12637671, ECO:0000269|PubMed:12925671, ECO:0000269|PubMed:17878374, ECO:0000269|PubMed:19663824, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Demir Dirençli Demir Eksikliği Anemisi, 206200, Otozomal Resesif; IRIDA (IRIDA Sendromu) (TMPRSS6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Iron-refractory iron deficiency anemia, 206200, Autosomal recessive; IRIDA (IRIDA syndrome) (TMPRSS6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMPRSS6	TMPRSS6, IRIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iron-refractory iron deficiency anemia (IRIDA) [MIM:206200]: Key features include congenital hypochromic microcytic anemia, very low mean corpuscular erythrocyte volume, low transferrin saturation, abnormal iron absorption characterized by no hematologic improvement following treatment with oral iron, and abnormal iron utilization characterized by a sluggish, incomplete response to parenteral iron. {ECO:0000269|PubMed:18408718, ECO:0000269|PubMed:18603562, ECO:0000269|PubMed:19357398, ECO:0000269|PubMed:19592582, ECO:0000269|PubMed:19708871, ECO:0000269|PubMed:19747362, ECO:0000269|PubMed:20232450, ECO:0000269|PubMed:20704562, ECO:0000269|PubMed:21618415, ECO:0000269|PubMed:22581667, ECO:0000269|PubMed:25156943, ECO:0000269|PubMed:25588876}. Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations leading to abrogation of TMPRSS6 activity are associated with IRIDA due to elevated levels of hepcidin, a negative regulator of plasma iron pool (PubMed:20232450). {ECO:0000269|PubMed:20232450	EDTA Blood Sample
İskemik İnme, Yatkınlık, 601367, Multifaktöryel (Erken Başlangıçlı Otozomal Dominant Alzheimer Hastalığı) (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ischemic stroke, susceptibility to, 601367, Multifactorial (Early-onset autosomal dominant Alzheimer disease) (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Variation in NOS3 seem to be associated with susceptibility to coronary spasm. {ECO:0000269|PubMed:11740345, ECO:0000269|PubMed:9737779}.;	EDTA Blood Sample
İskokoksospodopatellar Sendrom, 147891, Otozomal Dominant; ICPPS (Koksopodopatellar Sendromi) (TBX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ischiocoxopodopatellar syndrome, 147891, Autosomal dominant; ICPPS (Coxopodopatellar syndrome) (TBX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX4	TBX4, ICPPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Small patella syndrome (SPS) [MIM:147891]: Autosomal dominant skeletal dysplasia characterized by patellar aplasia or hypoplasia and by anomalies of the pelvis and feet, including disrupted ossification of the ischia and inferior pubic rami. {ECO:0000269	PubMed:15106123}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İskokoksospodopatellar Sendrom, 147891, Otozomal Dominant; ICPPS (Koksopodopatellar Sendromi) (Prenatal) (TBX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ischiocoxopodopatellar syndrome, 147891, Autosomal dominant; ICPPS (Coxopodopatellar syndrome) (TBX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX4	TBX4, ICPPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Small patella syndrome (SPS) [MIM:147891]: Autosomal dominant skeletal dysplasia characterized by patellar aplasia or hypoplasia and by anomalies of the pelvis and feet, including disrupted ossification of the ischia and inferior pubic rami. {ECO:0000269	PubMed:15106123}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İzobutiril-CoA Dehidrojenaz Yetmezliği, 611283 (İzobutiril-CoA Dehidrogenaz Eksikliği) (ACAD8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Isobutyryl-CoA dehydrogenase deficiency, 611283 (Isobutyryl-CoA dehydrogenase deficiency) (ACAD8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAD8	ACAD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İzobutiril-CoA Dehidrojenaz Yetmezliği, 611283 (İzobutiril-CoA Dehidrogenaz Eksikliği) (Prenatal) (ACAD8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Isobutyryl-CoA dehydrogenase deficiency, 611283 (Isobutyryl-CoA dehydrogenase deficiency) (ACAD8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACAD8	ACAD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İzole Büyüme Hormonu Eksikliği, GHRF eksikliğine bağlı (GHRH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Isolated growth hormone deficiency due to defect in GHRF (GHRH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHRH	GHRH, GHRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
İzole Büyüme Hormonu Eksikliği, Tip 3; Igh63 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (BTK geni) (Tüm Gen Dizi Analizi) (Postnatal)	ISOLATED GROWTH HORMONE DEFICIENCY, TYPE III; IGHD3 (Non-acquired isolated growth hormone deficiency) (BTK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BTK	BTK, AGMX1, IMD1, XLA, AT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. {ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678660, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633429, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9260283	EDTA Blood Sample

İzole Büyüme Hormonu Eksikliği, Tip 3; IghD3 (Non-Aktüz İzole Büyüme Hormonu Eksikliği) (MLPA)	ISOLATED GROWTH HORMONE DEFICIENCY, TYPE III; IGHD3 (Non-acquired isolated growth hormone deficiency) (MLPA)	BTK	BTK, AGMX1, IMD1, XLA, AT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: X-linked agammaglobulinemia (XLA) [MIM:300755]: Humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin. (ECO:0000269|PubMed:10220140, ECO:0000269|PubMed:10612838, ECO:0000269|PubMed:10678860, ECO:0000269|PubMed:7627183, ECO:0000269|PubMed:7633420, ECO:0000269|PubMed:7633425, ECO:0000269|PubMed:7711734, ECO:0000269|PubMed:7809124, ECO:0000269|PubMed:7849006, ECO:0000269|PubMed:7849697, ECO:0000269|PubMed:7849721, ECO:0000269|PubMed:7880320, ECO:0000269|PubMed:7897635, ECO:0000269|PubMed:8013627, ECO:0000269|PubMed:8162018, ECO:0000269|PubMed:8162056, ECO:0000269|PubMed:8634718, ECO:0000269|PubMed:8695804, ECO:0000269|PubMed:8723128, ECO:0000269|PubMed:8834236, ECO:0000269|PubMed:9260159, ECO:0000269|PubMed:9280283).	EDTA Blood Sample
İzovalerik Asidemi, 243500, Otozomal Resesif (İzovalerik Asidemi) (IVD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Isovaleric acidemia, 243500, Autosomal recessive (isovaleric acidemia) (IVD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IVD	IVD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Isovaleric acidemia (IVA) [MIM:243500]: A metabolic disorder characterized by retarded psychomotor development, a peculiar odor resembling sweaty feet, an aversion to dietary protein, and pernicious vomiting, leading to acidosis and coma. The acute neonatal form leads to massive metabolic acidosis from the first days of life and rapid death. (ECO:0000269|PubMed:2063866, ECO:0000269|PubMed:2204070, ECO:0000269|PubMed:2235054S, ECO:0000269|PubMed:23587913, ECO:0000269|PubMed:9665741). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İzovalerik Asidemi, 243500, Otozomal Resesif (İzovalerik Asidemi) (Prenatal) (IVD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Isovaleric acidemia, 243500, Autosomal recessive (isovaleric acidemia) (IVD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IVD	IVD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Isovaleric acidemia (IVA) [MIM:243500]: A metabolic disorder characterized by retarded psychomotor development, a peculiar odor resembling sweaty feet, an aversion to dietary protein, and pernicious vomiting, leading to acidosis and coma. The acute neonatal form leads to massive metabolic acidosis from the first days of life and rapid death. (ECO:0000269|PubMed:2063866, ECO:0000269|PubMed:2204070, ECO:0000269|PubMed:2235054S, ECO:0000269|PubMed:23587913, ECO:0000269|PubMed:9665741). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
IVIC Sendromu, 147750, Otozomal Dominant (IVIC Sendromu) (SALL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	IVIC syndrome, 147750, Autosomal dominant (IVIC syndrome) (SALL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SALL4	SALL4, HSAL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculotoradial syndrome (OORS) [MIM:147750]: Autosomal dominant condition characterized by upper limbs anomalies (radial ray defects, carpal bones fusion), extraocular motor disturbances, congenital bilateral non-progressive mixed hearing loss. Other less consistent malformations include heart involvement, mild thrombocytopenia and leukocytosis (before age 50), shoulder girdle hypoplasia, imperforate anus, kidney malrotation or rectovaginal fistula. The IVIC syndrome is an allelic disorder of Duane-radial ray syndrome (DRRS) with a similar phenotype. (ECO:0000269	PubMed:17256792). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
IVIC Sendromu, 147750, Otozomal Dominant (IVIC Sendromu) (Prenatal) (SALL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	IVIC syndrome, 147750, Autosomal dominant (IVIC syndrome) (SALL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SALL4	SALL4, HSAL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Oculotoradial syndrome (OORS) [MIM:147750]: Autosomal dominant condition characterized by upper limbs anomalies (radial ray defects, carpal bones fusion), extraocular motor disturbances, congenital bilateral non-progressive mixed hearing loss. Other less consistent malformations include heart involvement, mild thrombocytopenia and leukocytosis (before age 50), shoulder girdle hypoplasia, imperforate anus, kidney malrotation or rectovaginal fistula. The IVIC syndrome is an allelic disorder of Duane-radial ray syndrome (DRRS) with a similar phenotype. (ECO:0000269	PubMed:17256792). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant (Jackson-Weiss Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jackson-Weiss syndrome, 123150, autosomal dominant (Jackson-Weiss syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. (ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant (Jackson-Weiss Sendromu) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Jackson-Weiss syndrome, 123150, autosomal dominant (Jackson-Weiss syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. (ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant; JWS (Jackson-Weiss Sendromu) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jackson-Weiss syndrome, 123150, Autosomal dominant; JWS (Jackson-Weiss syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7674169). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant; JWS (Jackson-Weiss Sendromu) (MLPA)	Jackson-Weiss syndrome, 123150, Autosomal dominant; JWS (Jackson-Weiss syndrome) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7674169). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant; JWS (Jackson-Weiss Sendromu) (Prenatal) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Jackson-Weiss syndrome, 123150, Autosomal dominant; JWS (Jackson-Weiss syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7674169). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jackson-Weiss Sendromu, 123150, Otozomal Dominant; JWS (Jackson-Weiss Sendromu) (Prenatal) (MLPA)	Jackson-Weiss syndrome, 123150, Autosomal dominant; JWS (Jackson-Weiss syndrome) (Prenatal) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7674169). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
JAK2 Mutasyonu (Dizi Analizi) V617F Mutasyonu ve (Ekzon 12-14) (JAK2 geni) (Dizi Analizi) (Postnatal)	JAK2 mutation V617F Mutation and (Exon 12-14) (Sequence analysis) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
JAK2 V617F Mutasyonu ve (Ekzon 12-14) (Dizi Analizi) (JAK2 geni) (Dizi Analizi) (Postnatal)	JAK2 V617F Mutation and (Exon 12-14) (Sequence analysis) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow
Jalili Sendromu, 217080, Otozomal Resesif (Jalili Sendromu) (CNNM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jalili syndrome, 217080, Autosomal recessive (Jalili syndrome) (CNNM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNNM4	CNNM4, ACDP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Jalili Sendromu, 217080, Otozomal Resesif (Jalili Sendromu) (Prenatal) (CNNM4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Jalili syndrome, 217080, Autosomal recessive (Jalili syndrome) (CNNM4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNNM4	CNNM4, ACDP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jawad Sendromu, 251255, Otozomal Resesif; JWDS (Jawad Sendromu) (RBBP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jawad syndrome, 251255, Autosomal recessive; JWDS (Jawad syndrome) (RBBP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBBP8	RBBP8, RIM, SCKL2, JWDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Jawad syndrome (JWDS) [MIM:251255]: A syndrome characterized by congenital microcephaly, moderately severe mental retardation, and symmetrical digital anomalies. Digital malformations of variable degree include hallux valgus, syndactyly of toes 4 and 5, short fifth fingers, single flexion crease of fifth fingers, polydactyly and syndactyly. (ECO:0000269	PubMed:21998596). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Jawad Sendromu, 251255, Otozomal Resesif; JWDS (Jawad Sendromu) (Prenatal) (RBBP8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Jawad syndrome, 251255, Autosomal recessive; JWDS (Jawad syndrome) (RBBP8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBBP8	RBBP8, RIM, SCKL2, JWDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Jawad syndrome (JWDS) [MIM:251255]: A syndrome characterized by congenital microcephaly, moderately severe mental retardation, and symmetrical digital anomalies. Digital malformations of variable degree include hallux valgus, syndactyly of toes 4 and 5, short fifth fingers, single flexion crease of fifth fingers, polydactyly and syndactyly. {ECO:0000269	PubMed:21998596}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jervell Lange-Nielsen Sendromu 2, 612347, Otozomal Resesif; JLNS2 (Alesel Uzun QT Sendromu) (KCNE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jervell and Lange-Nielsen syndrome 2, 612347, Autosomal recessive; JLNS2 (Familial long QT syndrome) (KCNE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNE1	KCNE1, JLNS, LQT5, JLNS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Jervell and Lange-Nielsen syndrome 2 (JLNS2) [MIM:612347]: An autosomal recessive disorder characterized by congenital deafness, prolongation of the QT interval, syncopal attacks due to ventricular arrhythmias, and a high risk of sudden death. {ECO:0000269|PubMed:10409698, ECO:0000269|PubMed:9328483, ECO:0000269|PubMed:9354783}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jervell Lange-Nielsen Sendromu, 220400, Otozomal Resesif; JLNS1 (Alesel Uzun QT Sendromu) (KCNQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Jervell and Lange-Nielsen syndrome, 220400, Autosomal recessive; JLNS1 (Familial long QT syndrome) (KCNQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB3, SQT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:18711366, ECO:0000269|PubMed:18711367, ECO:0000269|PubMed:24390345}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Johanson-Blizzard Sendromu, 243800, Otozomal Resesif; JBS (Johanson-Blizzard Syndrome) (UBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Johanson-Blizzard syndrome, 243800, Autosomal recessive; JBS (Johanson-Blizzard syndrome) (UBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBR1	UBR1, JBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Johanson-Blizzard syndrome (JBS) [MIM:243800]: This disorder includes congenital exocrine pancreatic insufficiency, multiple malformations such as nasal wing aplasia, and frequent mental retardation. Pancreas of individuals with JBS do not express UBR1 and show intrauterine-onset destructive pancreatitis. {ECO:0000269	PubMed:16311597}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Johanson-Blizzard Sendromu, 243800, Otozomal Resesif; JBS (Johanson-Blizzard Syndrome) (Prenatal) (UBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Johanson-Blizzard syndrome, 243800, Autosomal recessive; JBS (Johanson-Blizzard syndrome) (UBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBR1	UBR1, JBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Johanson-Blizzard syndrome (JBS) [MIM:243800]: This disorder includes congenital exocrine pancreatic insufficiency, multiple malformations such as nasal wing aplasia, and frequent mental retardation. Pancreas of individuals with JBS do not express UBR1 and show intrauterine-onset destructive pancreatitis. {ECO:0000269	PubMed:16311597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 1, 213300, Otozomal Resesif; JBTS1 (Joubert Sendromu) (INPP5E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 1, 213300, Autosomal recessive; JBTS1 (Joubert syndrome) (INPP5E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INPP5E	INPP5E, MORMS, JBTS1, CORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, truncal obesity, retinal dystrophy, and microperis (MORMS) [MIM:610156]: An autosomal recessive disorder characterized by moderate mental retardation, truncal obesity, congenital non-progressive retinal dystrophy, and microperis in males. The phenotype is similar to Bardet-Biedl syndrome and Cohen syndrome Distinguishing features are the age of onset, the non-progressive nature of the visual impairment, lack of dysmorphic facies, skin or gingival infection, microcephaly, mottled retina, polydactyly, and testicular anomalies. {ECO:0000269	PubMed:19682215}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Joubert Sendromu 1, 213300, Otozomal Resesif; JBTS1 (Joubert Sendromu) (Prenatal) (INPP5E geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 1, 213300, Autosomal recessive; JBTS1 (Joubert syndrome) (INPP5E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INPP5E	INPP5E, MORMS, JBTS1, CORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, truncal obesity, retinal dystrophy, and microperis (MORMS) [MIM:610156]: An autosomal recessive disorder characterized by moderate mental retardation, truncal obesity, congenital non-progressive retinal dystrophy, and microperis in males. The phenotype is similar to Bardet-Biedl syndrome and Cohen syndrome Distinguishing features are the age of onset, the non-progressive nature of the visual impairment, lack of dysmorphic facies, skin or gingival infection, microcephaly, mottled retina, polydactyly, and testicular anomalies. {ECO:0000269	PubMed:19682215}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 10, 300804, X'e Bağlı Resesif; JBTS10 (Joubert Sendromu-Orofasiyodijital Defekti) (OFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 10, 300804, X-linked recessive; JBTS10 (Joubert syndrome with orofaciodigital defect) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. {ECO:0000269	PubMed:16783569}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 10, 300804, X'e Bağlı Resesif; JBTS10 (Joubert Sendromu-Orofasiyodijital Defekti) (Prenatal) (OFD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 10, 300804, X-linked recessive; JBTS10 (Joubert syndrome with orofaciodigital defect) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. {ECO:0000269	PubMed:16783569}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 12, 200990, Otozomal Resesif (Akrokallosal Sendrom) (KIF7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 12, 200990, Autosomal recessive (Acrocallosal syndrome) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:21552264}. Note=The gene represented in this entry may be involved in disease pathogenesis..	EDTA Blood Sample
Joubert Sendromu 12, 200990, Otozomal Resesif (Akrokallosal Sendrom) (Prenatal) (KIF7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 12, 200990, Autosomal recessive (Acrocallosal syndrome) (KIF7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF7	KIF7, HLS2, ACLS, JBTS12, AGBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:21552264}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 13, 614173, Otozomal Resesif; JBTS13 (Joubert Sendromu) (TCTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 13, 614173, Autosomal recessive; JBTS13 (Joubert syndrome) (TCTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCTN1	TECT1, JBTS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 13 (JBTS13) [MIM:614173]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:21725307}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Joubert Sendromu 13, 614173, Otozomal Resesif; JBTS13 (Joubert Sendromu) (Prenatal) (TCTN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 13, 614173, Autosomal recessive; JBTS13 (Joubert syndrome) (TCTN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCTN1	TECT1, JBTS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 13 (JBTS13) [MIM:614173]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:21725307}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 14, 614424, Otozomal Resesif; JBTS14 (Joubert Sendromu-Okulorenal Defekti) (TMEM237 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 14, 614424, Autosomal recessive; JBTS14 (Joubert syndrome with oculorenal defect) (TMEM237 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM237	TMEM237, ALS2CR4, JBTS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 14 (JBTS14) [MIM:614424]: An autosomal recessive disorder characterized by severe mental retardation, hypotonia, breathing abnormalities in infancy, and dysmorphic facial features. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include renal disease, abnormal eye movements, and postaxial polydactyly. {ECO:0000269	PubMed:22152675}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Joubert Sendromu 14, 614424, Otozomal Resesif; JBTS14 (Joubert Sendromu-Okulorenal Defekti) (Prenatal) (TMEM237 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 14, 614424, Autosomal recessive; JBTS14 (Joubert syndrome with oculorenal defect) (TMEM237 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM237	TMEM237, ALS2CR4, JBTS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 14 (JBTS14) [MIM:614424]: An autosomal recessive disorder characterized by severe mental retardation, hypotonia, breathing abnormalities in infancy, and dysmorphic facial features. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include renal disease, abnormal eye movements, and postaxial polydactyly. {ECO:0000269	PubMed:22152675}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 15, 614464, Otozomal Resesif; JBTS15 (Joubert Sendromu) (CEP41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 15, 614464, Autosomal recessive; JBTS15 (Joubert syndrome) (CEP41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP41	CEP41, TSGA14, JBTS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 15 (JBTS15) [MIM:614464]: An autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis and polydactyly. {ECO:0000269	PubMed:22246503}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Joubert Sendromu 15, 614464, Otozomal Resesif, JBTS15 (Joubert Sendromu) (Prenatal) (CEP41 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 15, 614464, Autosomal recessive; JBTS15 (Joubert syndrome) (CEP41 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP41	CEP41, TSGA14, JBTS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 15 (JBTS15) [MIM:614464]: An autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis and polydactyly. {ECO:0000269	PubMed:22246503}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 16, 614465, Otozomal Resesif, JBTS16 (Joubert Sendromu-Okulorenal Defekt) (TMEM138 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 16, 614465, Autosomal recessive; JBTS16 (Joubert syndrome with oculorenal defect) (TMEM138 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM138	TMEM138, JBTS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 16 (JBTS16) [MIM:614465]: An autosomal recessive disorder characterized by oculomotor apraxia, variable coloboma, and rare kidney involvement. Neuroradiologically, it is characterized by an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and polydactyly. {ECO:0000269	PubMed:22282472}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 16, 614465, Otozomal Resesif, JBTS16 (Joubert Sendromu-Okulorenal Defekti) (Prenatal) (TMEM138 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 16, 614465, Autosomal recessive; JBTS16 (Joubert syndrome with oculorenal defect) (TMEM138 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM138	TMEM138, JBTS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 16 (JBTS16) [MIM:614465]: An autosomal recessive disorder characterized by oculomotor apraxia, variable coloboma, and rare kidney involvement. Neuroradiologically, it is characterized by an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and polydactyly. {ECO:0000269	PubMed:22282472}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 17, 614615, Otozomal Resesif, JBTS17 (Joubert Sendromu) (C5orf42 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 17, 614615, Autosomal recessive; JBTS17 (Joubert syndrome) (C5orf42 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C5orf42	C5orf42, JBTS17, OFD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 6 (OFD6) [MIM:277170]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD6 is characterized by metacarpal abnormalities with central polydactyly, cerebellar abnormalities including the molar tooth sign, tongue hamartomas, additional frenula, and upper lip notch. {ECO:0000269	PubMed:24178751}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 17, 614615, Otozomal Resesif, JBTS17 (Joubert Sendromu) (Prenatal) (C5orf42 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 17, 614615, Autosomal recessive; JBTS17 (Joubert syndrome) (C5orf42 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C5orf42	C5orf42, JBTS17, OFD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 6 (OFD6) [MIM:277170]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD6 is characterized by metacarpal abnormalities with central polydactyly, cerebellar abnormalities including the molar tooth sign, tongue hamartomas, additional frenula, and upper lip notch. {ECO:0000269	PubMed:24178751}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 18, 614815, Otozomal Resesif, JBTS18 (Joubert Sendromu-Orofasiyodijital Defekt) (TCTN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 18, 614815, Autosomal recessive; JBTS18 (Joubert syndrome with orofaciodigital defect) (TCTN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCTN3	TCTN3, TECT3, C10orf61, OFD4, JBTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 4 (OFD4) [MIM:258860]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD4 patients have tongue nodules, multiple frenulae, broad flat nose, hypertelorism, and short rib polydactyly with tibial dysplasia (Majewski syndrome). The presence of severe tibial aplasia differentiates OFD4 from OFD1. Additional features of cystic dysplastic kidneys and brain malformation, including occipital encephalocele, are observed in severely affected patients. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 18, 614815, Otozomal Resesif, JBTS18 (Joubert Sendromu-Orofasiyodijital Defekt) (Prenatal) (TCTN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 18, 614815, Autosomal recessive; JBTS18 (Joubert syndrome with orofaciodigital defect) (TCTN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCTN3	TCTN3, TECT3, C10orf61, OFD4, JBTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 4 (OFD4) [MIM:258860]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD4 patients have tongue nodules, multiple frenulae, broad flat nose, hypertelorism, and short rib polydactyly with tibial dysplasia (Majewski syndrome). The presence of severe tibial aplasia differentiates OFD4 from OFD1. Additional features of cystic dysplastic kidneys and brain malformation, including occipital encephalocele, are observed in severely affected patients. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 19, 614844, Otozomal Resesif, Otozomal Dominant; NPHP14 (Joubert Sendromu-Okulorenal Defekt) (ZNF423 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 19, 614844, Autosomal recessive, Autosomal dominant; NPHP14 (Joubert syndrome with oculorenal defect) (ZNF423 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF423	ZNF423, ZFP423, OAZ, KIAA0760, NPHP14, JBTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 14 (NPHP14) [MIM:614844]: An autosomal recessive disorder manifesting as infantile-onset kidney disease, cerebellar vermis hypoplasia, and situs inversus. Nephronophthisis is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. {ECO:0000269	PubMed:23863007}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 19, 614844, Otozomal Resesif, Otozomal Dominant; NPHP14 (Joubert Sendromu-Okulorenal Defekti) (Prenatal) (ZNF423 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 19, 614844, Autosomal recessive, Autosomal dominant; NPHP14 (Joubert syndrome with oculorenal defect) (ZNF423 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNF423	ZNF423, ZFP423, OAZ, KIAA0760, NPHP14, JBTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 14 (NPHP14) [MIM:614844]: An autosomal recessive disorder manifesting as infantile-onset kidney disease, cerebellar vermis hypoplasia, and situs inversus. Nephronophthisis is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. {ECO:0000269	PubMed:23863007}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 2, 608091, Otozomal Resesif, JBTS2 (Joubert Sendromu-Okulorenal Defekt) (TMEM216 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 2, 608091, Autosomal recessive; JBTS2 (Joubert syndrome with oculorenal defect) (TMEM216 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM216	TMEM216, JBTS2, CORS2, MKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 2 (MKS2) [MIM:603194]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:20512146}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 2, 608091, Otozomal Resesif, JBTS2 (Joubert Sendromu-Okulorenal Defekti) (Prenatal) (TMEM216 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 2, 608091, Autosomal recessive; JBTS2 (Joubert syndrome with oculorenal defect) (TMEM216 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM216	TMEM216, JBTS2, CORS2, MKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 2 (MKS2) [MIM:603194]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:20512146}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 20, 614970, Otozomal Resesif, JBTS20 (Joubert Sendromu) (TMEM231 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 20, 614970, Autosomal recessive; JBTS20 (Joubert syndrome) (TMEM231 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM231	TMEM231, JBTS20, MKS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 20 (JBTS20) [MIM:614970]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:23012439}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 20, 614970, Otozomal Resesif, JBTS20 (Joubert Sendromu) (Prenatal) (TMEM231 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 20, 614970, Autosomal recessive; JBTS20 (Joubert syndrome) (TMEM231 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM231	TMEM231, JBTS20, MKS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 20 (JBTS20) [MIM:614970]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:23012439}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 21, 615636, Otozomal Resesif, JBTS21 (Joubert Sendromu) (CSPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 21, 615636, Autosomal recessive; JBTS21 (Joubert syndrome) (CSPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSPP1	CSPP1, CSPP, JBTS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 21 (JBTS21) [MIM:615636]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269|PubMed:24360803, ECO:0000269|PubMed:24360807, ECO:0000269|PubMed:24360808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 21, 615636, Otozomal Resesif, JBTS21 (Joubert Sendromu) (Prenatal) (CSPP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 21, 615636, Autosomal recessive; JBTS21 (Joubert syndrome) (CSPP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSPP1	CSPP1, CSPP, JBTS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 21 (JBTS21) [MIM:615636]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269|PubMed:24360803, ECO:0000269|PubMed:24360807, ECO:0000269|PubMed:24360808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 22 615665, Otozomal Resesif, JBTS22 (Joubert Sendromu-Orofasiyodijital Defekt) (PDE6D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 22, 615665, Autosomal recessive; JBTS22 (Joubert syndrome with orofaciodigital defect) (PDE6D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6D	PDE6D, JBTS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Joubert Sendromu 22 615665, Otozomal Resesif, JBTS22 (Joubert Sendromu-Orofasiyodijital Defekt) (Prenatal) (PDE6D gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 22, 615665, Autosomal recessive; JBTS22 (Joubert syndrome with orofaciodigital defect) (PDE6D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDE6D	PDE6D, JBTS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 23, 616490, Otozomal Resesif, JBTS23 (Joubert Sendromu) (KIAA0586 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 23, 616490, Autosomal recessive; JBTS23 (Joubert syndrome) (KIAA0586 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA0586	KIAA0586, TALPID3, JBTS23, SRTD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 23 (JBTS23) [MIM:616490]: A mild form of Joubert syndrome, a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269	PubMed:26096313}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Joubert Sendromu 23, 616490, Otozomal Resesif, JBTS23 (Joubert Sendromu) (Prenatal) (KIAA0586 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 23, 616490, Autosomal recessive; JBTS23 (Joubert syndrome) (KIAA0586 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIAA0586	KIAA0586, TALPID3, JBTS23, SRTD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 23 (JBTS23) [MIM:616490]: A mild form of Joubert syndrome, a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269	PubMed:26096313}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 24, 616654, Otozomal Resesif, JBTS24 (Joubert Sendromu) (TCTN2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 24, 616654, Autosomal recessive; JBTS24 (Joubert syndrome) (TCTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCTN2	TCTN2, TECT2, MKS8, JBTS24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 8 (MKS8) [MIM:613885]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21462283}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 24, 616654, Otozomal Resesif, JBTS24 (Joubert Sendromu) (Prenatal) (TCTN2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 24, 616654, Autosomal recessive; JBTS24 (Joubert syndrome) (TCTN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCTN2	TCTN2, TECT2, MKS8, JBTS24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 8 (MKS8) [MIM:613885]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21462283}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 25, 616781, Otozomal Resesif, JBTS25 (Joubert Sendromu) (CEP104 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 25, 616781, Autosomal recessive; JBTS25 (Joubert syndrome) (CEP104 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP104	CEP104, GLYBP, KIAA0562, JBTS25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Joubert Sendromu 25, 616781, Otozomal Resesif, JBTS25 (Joubert Sendromu) (Prenatal) (CEP104 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 25, 616781, Autosomal recessive; JBTS25 (Joubert syndrome) (CEP104 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP104	CEP104, GLYBP, KIAA0562, JBTS25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 26, 616784, Otozomal Resesif, JBTS26 (Joubert Sendromu) (KIAA0556 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 26, 616784, Autosomal recessive; JBTS26 (Joubert syndrome) (KIAA0556 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA0556	KATNIP, KIAA0556	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Joubert Sendromu 26, 616784, Otozomal Resesif, JBTS26 (Joubert Sendromu) (Prenatal) (KIAA0556 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 26, 616784, Autosomal recessive; JBTS26 (Joubert syndrome) (KIAA0556 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIAA0556	KATNIP, KIAA0556	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 27 617120, Otozomal Resesif, JBTS27 (Joubert Sendromu) (B9D1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 27, 617120, Autosomal recessive; JBTS27 (Joubert syndrome) (B9D1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B9D1	B9D1, MKSR1, MKS9, JBTS27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 9 (MKS9) [MIM:614209]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21493627}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Joubert Sendromu 27 617120, Otozomal Resesif, JBTS27 (Joubert Sendromu) (Prenatal) (B9D1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 27, 617120, Autosomal recessive; JBTS27 (Joubert syndrome) (B9D1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B9D1	B9D1, MKSR1, MKS9, JBTS27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 9 (MKS9) [MIM:614209]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21493627}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 28, 617121, Otozomal Resesif, JBTS28 (Joubert Sendromu) (MKS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 28, 617121, Autosomal recessive; JBTS28 (Joubert syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 1 (MKS1) [MIM:249000]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269|PubMed:16415886, ECO:0000269|PubMed:19466712}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 28, 617121, Otozomal Resesif, JBTS28 (Joubert Sendromu) (Prenatal) (MKS1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 28, 617121, Autosomal recessive; JBTS28 (Joubert syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 1 (MKS1) [MIM:249000]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269|PubMed:16415886, ECO:0000269|PubMed:19466712}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 4, 609583, Otozomal Resesif, JBTS4 (Joubert Sendromu-Renal Defekt) (NPHP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 4, 609583, Autosomal recessive; JBTS4 (Joubert syndrome with renal defect) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 4, 609583, Otozomal Resesif, JBTS4 (Joubert Sendromu-Renal Defekt) (MLPA)	Joubert syndrome 4, 609583, Autosomal recessive; JBTS4 (Joubert syndrome with renal defect) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 4, 609583, Otozomal Resesif, JBTS4 (Joubert Sendromu-Renal Defekt) (Prenatal) (NPHP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 4, 609583, Autosomal recessive; JBTS4 (Joubert syndrome with renal defect) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 4, 609583, Otozomal Resesif, JBTS4 (Joubert Sendromu-Renal Defekt) (Prenatal) (MLPA)	Joubert syndrome 4, 609583, Autosomal recessive; JBTS4 (Joubert syndrome with renal defect) (Prenatal) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 5, 610188, Otozomal Resesif, JBTS5 (Joubert Sendromu-Okulorenal Defekt) (CEP290 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 5, 610188, Autosomal recessive; JBTS5 (Joubert syndrome with oculorenal defect) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Joubert Sendromu 5, 610188, Otozomal Resesif; JBTS5 (Joubert Sendromu-Okulorenal Defekt) (Prenatal) (CEP290 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 5, 610188, Autosomal recessive; JBTS5 (Joubert syndrome with oculorenal defect) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP290	CEP290, KIAA0373, SHL1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 6, 610688, Otozomal Resesif (Joubert Sendromu) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 6, 610688, Autosomal recessive; JBTS6 (Joubert syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:18327255}. Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	EDTA Blood Sample
Joubert Sendromu 6, 610688, Otozomal Resesif (Joubert Sendromu) (Prenatal) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 6, 610688, Autosomal recessive; JBTS6 (Joubert syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:18327255}. Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 7, 611560, Otozomal Resesif; JBTS7 (Joubert Sendromu-Renal Defekt) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 7, 611560, Autosomal recessive; JBTS7 (Joubert syndrome with renal defect) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269	PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 7, 611560, Otozomal Resesif; JBTS7 (Joubert Sendromu-Renal Defekt) (Prenatal) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 7, 611560, Autosomal recessive; JBTS7 (Joubert syndrome with renal defect) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269	PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 8, 612291, Otozomal Resesif; JBTS8 (Joubert Sendromu) (ARL13B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 8, 612291, Autosomal recessive; JBTS8 (Joubert syndrome) (ARL13B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL13B	ARL13B, ARL2L1, JBTS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 8 (JBTS8) [MIM:612291]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:18674751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Joubert Sendromu 8, 612291, Otozomal Resesif; JBTS8 (Joubert Sendromu) (Prenatal) (ARL13B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 8, 612291, Autosomal recessive; JBTS8 (Joubert syndrome) (ARL13B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARL13B	ARL13B, ARL2L1, JBTS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 8 (JBTS8) [MIM:612291]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:18674751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu 9, 612285, Otozomal Resesif; JBTS9 (Joubert Sendromu-Okulorenal Defekt) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome 9, 612285, Autosomal recessive; JBTS9 (Joubert syndrome with oculorenal defect) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269	PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Joubert Sendromu 9, 612285, Otozomal Resesif; JBTS9 (Joubert Sendromu-Okulorenal Defekt) (Prenatal) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome 9, 612285, Autosomal recessive; JBTS9 (Joubert syndrome with oculorenal defect) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. {ECO:0000269	PubMed:19574260}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Joubert Sendromu-3, 608629, Otozomal Resesif (Joubert Sendromu-Oküler Defekt) (AH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Joubert syndrome-3, 608629, Autosomal recessive (Joubert syndrome with ocular defect) (AH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AH1	AH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 3 (JBTS3) [MIM:608629]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 3 shows minimal extra cerebral nervous system involvement and appears not to be associated with renal dysfunction. {ECO:0000269|PubMed:15322546, ECO:0000269|PubMed:15467982, ECO:0000269|PubMed:16453322, ECO:0000269|PubMed:22425360, ECO:0000269|PubMed:23532844}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Joubert Sendromu-3, 608629, Otozomal Resesif (Joubert Sendromu-Oküler Defekt) (Prenatal) (AH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Joubert syndrome-3, 608629, Autosomal recessive (Joubert syndrome with ocular defect) (AH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AH1	AH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 3 (JBTS3) [MIM:608629]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 3 shows minimal extra cerebral nervous system involvement and appears not to be associated with renal dysfunction. {ECO:0000269|PubMed:15322546, ECO:0000269|PubMed:15467982, ECO:0000269|PubMed:16453322, ECO:0000269|PubMed:22425360, ECO:0000269|PubMed:23532844}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Jüvenil Miyelomonositik Lösemi, 607785, Otozomal Dominant, Somatik Mutasyon (Jüvenil Miyelomonositik Lösemi) (CBL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Juvenile myelomonocytic leukemia, 607785, Autosomal dominant, Somatic mutation (Juvenile myelomonocytic leukemia) (CBL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBL	CBL, CBL2, NSLL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL) [MIM:613563]: A syndrome characterized by a phenotype reminiscent of Noonan syndrome. Clinical features are highly variable, including facial dysmorphism, short neck, developmental delay, hyperextensible joints and thorax abnormalities with widely spaced nipples. The facial features consist of triangular face with hypertelorism, large low-set ears, ptosis, and flat nasal bridge. Some patients manifest cardiac defects. Some have an increased risk for certain malignancies, particularly juvenile myelomonocytic leukemia. {ECO:0000269	PubMed:20619386}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Jüvenil polipozis sendromu (JPS) (BMPR1A 10q22, SMAD4 18q21, PTEN 10q23) (MLPA)	Juvenile polyposis syndrome (JPS) (BMPR1A 10q22, SMAD4 18q21, PTEN 10q23) (MLPA)	BMPR1A 10q22, SMAD4 18q21, PTEN 10q23	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Jüvenil Polipozis Sendromu, İnfantil Form, 174900, Otozomal Dominant; JPS (Jüvenil Polipozis Sendromu) (BMPR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Juvenile polyposis syndrome, infantile form, 174900, Autosomal dominant; JPS (Juvenile polyposis syndrome) (BMPR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1A	BMPR1A, ACVRLK3, ALK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jüvenil Polipozis Sendromu, İnfantil Form, 174900, Otozomal Dominant; JPS (Jüvenil Polipozis Sendromu) (MLPA)	Juvenile polyposis syndrome, infantile form, 174900, Autosomal dominant; JPS (Juvenile polyposis syndrome) (MLPA)	BMPR1A	BMPR1A, ACVRLK3, ALK3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Jüvenil Polipozis / Kalıtsal Hemorajik Telanjiyektazi Sendromu, 175050, Otozomal Dominant; JPHT (Jüvenil Polipozis Sendromu) (SMAD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050, Autosomal dominant; JPHT (Juvenile polyposis syndrome) (SMAD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Jüvenil Polipozis / Kalıtsal Hemorajik Telanjiyektazi Sendromu, 175050, Otozomal Dominant; JPHT (Jüvenil Polipozis Sendromu) (MLPA)	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050, Autosomal dominant; JPHT (Juvenile polyposis syndrome) (MLPA)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kabuki Sendromu 1, 147920, Otozomal Dominant; KABUK1 (Kabuki Sendromu) (KMT2D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kabuki syndrome 1, 147920, Autosomal dominant; KABUK1 (Kabuki syndrome) (KMT2D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KMT2D	KMT2D, MLL2, ALR, KABUK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kabuki syndrome 1 (KABUK1) [MIM:147920]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269|PubMed:20711175, ECO:0000269|PubMed:21280141, ECO:0000269|PubMed:21607748, ECO:0000269|PubMed:21658225, ECO:0000269|PubMed:21671394, ECO:0000269|PubMed:22126750, ECO:0000269|PubMed:23320472, ECO:0000269|PubMed:23913813, ECO:0000269|PubMed:24739679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kabuki Sendromu 1, 147920, Otozomal Dominant; KABUK1 (Kabuki Sendromu) (Prenatal) (KMT2D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kabuki syndrome 1, 147920, Autosomal dominant; KABUK1 (Kabuki syndrome) (KMT2D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KMT2D	KMT2D, MLL2, ALR, KABUK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kabuki syndrome 1 (KABUK1) [MIM:147920]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269|PubMed:20711175, ECO:0000269|PubMed:21280141, ECO:0000269|PubMed:21607748, ECO:0000269|PubMed:21658225, ECO:0000269|PubMed:21671394, ECO:0000269|PubMed:22126750, ECO:0000269|PubMed:23320472, ECO:0000269|PubMed:23913813, ECO:0000269|PubMed:24739679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kabuki Sendromu 2, 300867, X'e Bağlı Dominant; KABUK2 (Kabuki Sendromu) (KDM6A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kabuki syndrome 2, 300867, X-linked dominant; KABUK2 (Kabuki syndrome) (KDM6A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDM6A	KDM6A, UTX, KABUK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kabuki syndrome 2 (KABUK2) [MIM:300867]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269	PubMed:22197486}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kabuki Sendromu 2, 300867, X'e Bağlı Dominant; KABUK2 (Kabuki Sendromu) (MLPA)	Kabuki syndrome 2, 300867, X-linked dominant; KABUK2 (Kabuki syndrome) (MLPA)	KDM6A	KDM6A, UTX, KABUK2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Kabuki syndrome 2 (KABUK2) [MIM:300867]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269	PubMed:22197486}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kabuki Sendromu 2, 300867, X'e Bağlı Dominant; KABUK2 (Kabuki Sendromu) (Prenatal) (KDM6A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kabuki syndrome 2, 300867, X-linked dominant; KABUK2 (Kabuki syndrome) (KDM6A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KDM6A	KDM6A, UTX, KABUK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kabuki syndrome 2 (KABUK2) [MIM:300867]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269	PubMed:22197486}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kabuki Sendromu 2, 300867, X'e Bağlı Dominant; KABUK2 (Kabuki Sendromu) (Prenatal) (MLPA)	Kabuki syndrome 2, 300867, X-linked dominant; KABUK2 (Kabuki syndrome) (Prenatal) (MLPA)	KDM6A	KDM6A, UTX, KABUK2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kabuki syndrome 2 (KABUK2) [MIM:300867]: A congenital mental retardation syndrome with additional features, including postnatal dwarfism, a peculiar facies characterized by long palpebral fissures with eversion of the lateral third of the lower eyelids, a broad and depressed nasal tip, large prominent earlobes, a cleft or high-arched palate, scoliosis, short fifth finger, persistence of fingerpads, radiographic abnormalities of the vertebrae, hands, and hip joints, and recurrent otitis media in infancy. {ECO:0000269	PubMed:22197486}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kahrizi Sendromu, 612713, Otozomal Resesif; KHR2 (Kahrizi Sendromu) (SRDSA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kahrizi syndrome, 612713, Autosomal recessive; KHR2 (Kahrizi syndrome) (SRDSA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRDSA3	SRDSA3, SRDSA2L, CDG1Q, KRIZI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kahrizi syndrome (KHRZ) [MIM:612713]: An autosomal recessive neurodevelopmental disorder characterized by mental retardation, cataracts, coloboma, kyphosis, and coarse facial features. {ECO:0000269	PubMed:20700148}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kahrizi Sendromu, 612713, Otozomal Resesif; KHR2 (Kahrizi Sendromu) (Prenatal) (SRDSA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kahrizi syndrome, 612713, Autosomal recessive; KHR2 (Kahrizi syndrome) (SRDSA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRDSA3	SRDSA3, SRDSA2L, CDG1Q, KRIZI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kahrizi syndrome (KHRZ) [MIM:612713]: An autosomal recessive neurodevelopmental disorder characterized by mental retardation, cataracts, coloboma, kyphosis, and coarse facial features. {ECO:0000269	PubMed:20700148}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kallmann sendromu paneli (FGFR1 8p11.2, GNRHR 8p21, KISSR1 19p13.3 , GNRH1, NELF 9q34.3) (MLPA)	Kallmann syndrome (FGFR1 8p11.2, GNRHR 8p21, KISSR1 19p13.3 , GNRH1, NELF 9q34.3) (MLPA)	FGFR1 8p11.2, GNRHR 8p21, KISSR1 19p13.3 , GNRH1, NELF 9q34.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kallmann Sendromu (KAL1) (Xp22.31 Mikrodelesyonu) (FISH)	Kallmann syndrome (KAL1) (Xp22.31 microdeletion) (FISH)	Xp22.31	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
KANSL1 kopya sayısı (17q21.31) (MLPA)	KANSL1 copy number (17q21.31) (MLPA)	17q21.31	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Karzaki Hastalığı, 609242, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (NAGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Karzaki disease, 609242, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schindler disease (SCHND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Karzaki Hastalığı, 609242, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (Prenatal) (NAGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Karzaki disease, 609242, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schindler disease (SCHND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kaposi Sarkomu, Yatkınlık, 148000, Otozomal Dominant (Castleman Hastalığı) (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kaposi sarcoma, susceptibility to, 148000, Autosomal dominant (Castleman disease) (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis systemic juvenile (RASJ) [MIM:604302]: An inflammatory articular disorder with systemic-onset beginning before the age of 16. It represents a subgroup of juvenile arthritis associated with severe extraarticular features and occasionally fatal complications. During active phases of the disorder, patients display a typical daily spiking fever, an evanescent macular rash, lymphadenopathy, hepatosplenomegaly, serositis, myalgia and arthritis. {ECO:0000269	PubMed:9769329}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kappa Hafif Zincir Eksikliği, 614102, Otozomal Resesif (Tekrarlayan Enfeksiyonlar-Nadir İmmünglobulin İzotip Eksikliğine Bağlı) (IGKC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kappa light chain deficiency, 614102, Autosomal recessive (Recurrent infections associated with rare immunoglobulin isotypes deficiency) (IGKC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGKC	IGKC, IGKCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunoglobulin kappa light chain deficiency (IGKCD) [MIM:614102]: A disease characterized by the complete absence of immunoglobulin kappa chains. {ECO:0000269	PubMed:3931219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kappa Hafif Zincir Eksikliği, 614102, Otozomal Resesif (Tekrarlayan Enfeksiyonlar-Nadir İmmünglobülin İzotip Eksikliğine Bağlı) (IGKC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kappa light chain deficiency, 614102, Autosomal recessive (Recurrent infections associated with rare immunoglobulin isotypes deficiency) (IGKC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IGKC	IGKC, IGKCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunoglobulin kappa light chain deficiency (IGKCD) [MIM:614102]: A disease characterized by the complete absence of immunoglobulin kappa chains. (ECO:0000269	PubMed:33911219). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kaufman Oküloserebrofasiyal Sendrom, 244450, Otozomal Resesif; KOS (Oküloserebrofasiyel Sendrom, Kaufman Tipi) (UBE3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kaufman oculocerebrofacial syndrome, 244450, Autosomal recessive; KOS (Oculocerebrofacial syndrome, Kaufman type) (UBE3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBE3B	UBE3B, BPIDS, KOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kaufman Oküloserebrofasiyal Sendrom, 244450, Otozomal Resesif; KOS (Oküloserebrofasiyel Sendrom, Kaufman Tipi) (Prenatal) (UBE3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kaufman oculocerebrofacial syndrome, 244450, Autosomal recessive; KOS (Oculocerebrofacial syndrome, Kaufman type) (UBE3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBE3B	UBE3B, BPIDS, KOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kawasaki Hastalığı, Yatkınlık, 611775 (Kawasaki Hastalığı (ITPKC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kawasaki disease, susceptibility to, 611775 (Kawasaki disease) (ITPKC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPKC	ITPKC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
KBG Sendromu, 148050, Otozomal Dominant; KBGS (KBG Sendromu) (ANKRD11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	KBG syndrome, 148050, Autosomal dominant; KBGS (KBG syndrome) (ANKRD11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKRD11	ANKRD11, ANCO1, KBGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: KBG syndrome (KBGS) [MIM:148050]: A syndrome characterized by macrodontia of the upper central incisors, distinctive craniofacial findings, short stature, skeletal anomalies, and neurologic involvement that includes global developmental delay, seizures, and intellectual disability. (ECO:0000269	PubMed:21782149). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
KBG Sendromu, 148050, Otozomal Dominant; KBGS (KBG Sendromu) (Prenatal) (ANKRD11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	KBG syndrome, 148050, Autosomal dominant; KBGS (KBG syndrome) (ANKRD11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANKRD11	ANKRD11, ANCO1, KBGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: KBG syndrome (KBGS) [MIM:148050]: A syndrome characterized by macrodontia of the upper central incisors, distinctive craniofacial findings, short stature, skeletal anomalies, and neurologic involvement that includes global developmental delay, seizures, and intellectual disability. (ECO:0000269	PubMed:21782149). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kenny-Caffey Sendromu, Tip 1, 244460, Otozomal Resesif; KCS1 (Kenny-Caffey Sendromu) (TBCE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kenny-Caffey syndrome, type 1, 244460, Autosomal recessive; KCS1 (Kenny-Caffey syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoparathyroidism-retardation-dysmorphism syndrome (HRD) [MIM:241410]: Autosomal recessive disorder reported almost exclusively in Middle Eastern populations. (ECO:0000269	PubMed:12389028). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kenny-Caffey Sendromu, Tip 1, 244460, Otozomal Resesif; KCS1 (Kenny-Caffey Sendromu) (Prenatal) (TBCE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kenny-Caffey syndrome, type 1, 244460, Autosomal recessive; KCS1 (Kenny-Caffey syndrome) (TBCE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBCE	TBCE, KCS, KCS1, HRD, PEAMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoparathyroidism-retardation-dysmorphism syndrome (HRD) [MIM:241410]: Autosomal recessive disorder reported almost exclusively in Middle Eastern populations. (ECO:0000269	PubMed:12389028). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kenny-Caffey Sendromu, Tip 2, 127000, Otozomal Dominant; KCS2 (Kenny-Caffey Sendromu) (FAM111A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kenny-Caffey syndrome, type 2, 127000, Autosomal dominant; KCS2 (Kenny-Caffey syndrome) (FAM111A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM111A	FAM111A, KIAA1895, KCS2, GCLEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kenny-Caffey syndrome 2 (KCS2) [MIM:127000]: A disorder characterized by impaired skeletal development with small and dense bones, short stature, and primary hypoparathyroidism with hypocalcemia. Clinical features include cortical thickening and medullary stenosis of the tubular bones, delayed closure of fontanels, defective dentition, small eyes with hypermetropia, and frontal bossing with a triangular face. (ECO:0000269|PubMed:23684011, ECO:0000269|PubMed:23996431, ECO:0000269|PubMed:24635597). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kenny-Caffey Sendromu, Tip 2, 127000, Otozomal Dominant; KCS2 (Kenny-Caffey Sendromu) (Prenatal) (FAM111A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kenny-Caffey syndrome, type 2, 127000, Autosomal dominant; KCS2 (Kenny-Caffey syndrome) (FAM111A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAM111A	FAM111A, KIAA1895, KCS2, GCLEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kenny-Caffey syndrome 2 (KCS2) [MIM:127000]: A disorder characterized by impaired skeletal development with small and dense bones, short stature, and primary hypoparathyroidism with hypocalcemia. Clinical features include cortical thickening and medullary stenosis of the tubular bones, delayed closure of fontanels, defective dentition, small eyes with hypermetropia, and frontal bossing with a triangular face. (ECO:0000269|PubMed:23684011, ECO:0000269|PubMed:23996431, ECO:0000269|PubMed:24635597). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Keppen-Lubinsky Sendromu, 614098, Otozomal Dominant; KPLBS (Keppen-Lubinsky Sendromu) (KCNJ6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keppen-Lubinsky syndrome, 614098, Autosomal dominant; KPLBS (Keppen-Lubinsky syndrome) (KCNJ6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ6	KCNJ6, GIRK2, KCNJ7, KPLBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Keppen-Lubinsky Sendromu, 614098, Otozomal Dominant; KPLBS (Keppen-Lubinsky Sendromu) (Prenatal) (KCNJ6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Keppen-Lubinsky syndrome, 614098, Autosomal dominant; KPLBS (Keppen-Lubinsky syndrome) (KCNJ6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNJ6	KCNJ6, GIRK2, KCNJ7, KPLBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Keratit-İktiyoz-Sağırlık Sendromu, 148210, Otozomal Dominant (KID Sendromu) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratitis-ichthyosis-deafness syndrome, 148210, Autosomal dominant (KID syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vohwinkel syndrome (VS) [MIM:124500]: VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on fingers and toes and congenital deafness. (ECO:0000269|PubMed:10369869, ECO:0000269|PubMed:15954104, ECO:0000269|PubMed:18688874). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratit, 148190, Otozomal Dominant (Otozomal Dominant Keratit) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratitis, 148190, Autosomal dominant (Autosomal dominant keratitis) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. (ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratit, 148190, Otozomal Dominant (Otozomal Dominant Keratit) (MLPA)	Keratitis, 148190, Autosomal dominant (Autosomal dominant keratitis) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. (ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratokonus 1, 148300, Otozomal Dominant; KTCN1 (VSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratoconus 1, 148300, Autosomal dominant; KTCN1 (VSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VSX1	VSX1, RINX, KTCN1, CAASD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratoconus 1 (KTCN1) [MIM:148300]: Frequent corneal dystrophy with an incidence that varies from 50 to 230 per 100'000. The cornea assumes a conical shape as a result of a progressive non-inflammatory thinning of the corneal stroma. Keratoconus is most often an isolated sporadic condition with cases of autosomal dominant and autosomal recessive transmission. (ECO:0000269|PubMed:11978762, ECO:0000269|PubMed:15623752, ECO:0000269|PubMed:19954409). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratoderma, Palmoplantar, Noktasal Tip 1A, 148600, Otozomal Dominant; PPKP1A (Punktat Palmoplantar Keratoderma Tip 1) (AAGAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratoderma, palmoplantar, punctate type IA, 148600, Autosomal dominant; PPKP1A (Punctate palmoplantar keratoderma type 1) (AAGAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AAGAB	AAGAB, p34, PPKP1A, PPKP1, KPPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Keratoderma, Palmoplantar, Sağırlık, 148350, Otozomal Dominant (Palmoplantar Keratoderma Sağırlık Sendromu) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratoderma, palmoplantar, with deafness, 148350, Autosomal dominant (Palmoplantar keratoderma-deafness syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vohwinkel syndrome (VS) [MIM:124500]: VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on fingers and toes and congenital deafness. (ECO:0000269|PubMed:10369869, ECO:0000269|PubMed:15954104, ECO:0000269|PubMed:18688874). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratosis follicularis spinulosa decalvans, X'e Bağlı, 308800 (Keratozis follikularis spinulosa dekalvans) (MBTPS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis follicularis spinulosa decalvans, X-linked, 308800 (Keratozis follicularis spinulosa decalvans) (MBTPS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBTPS2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Olmsted syndrome, X-linked (OLMSX) [MIM:309918]: A rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma and periorificial keratotic plaques with severe itching at all lesions. Diffuse alopecia, constriction of digits, and onychodystrophy have also been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes. (ECO:0000269	PubMed:22931912). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratosis Follikularis Spinulosa Decalvans, X'e Bağlı, 308800, X'e Bağlı Resesif, KFSDX (Keratozis Follikularis Spinulosa Dekalvans) (MBTPS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis follicularis spinulosa decalvans, X-linked, 308800, X-linked recessive; KFSDX (Keratosis follicularis spinulosa decalvans) (MBTPS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBTPS2	MBTPS2, S2P, IFAP, KFSDX, OLMSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Olmsted syndrome, X-linked (OLMSX) [MIM:309918]: A rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma and periorificial keratotic plaques with severe itching at all lesions. Diffuse alopecia, constriction of digits, and onychodystrophy have also been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes. (ECO:0000269	PubMed:22931912). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Keratozis Linearis, İktiyozis Konjenita ve Sklerozan Keratoderma, 601952, Otozomal Resesif; KLICK (Keratozis Linearis-İktiyozis Konjenita-Sklerozan Keratoderma Sendromu) (POMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, 601952, Autosomal recessive; KLICK (Keratosis linearis-ichthyosis congenita-sclerosing keratoderma syndrome) (POMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMP	POMP, UMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratosis linearis with ichthyosis congenita and sclerosing keratoderma (KLICK) [MIM:601952]: A keratinizing disorder characterized by ichthyosis, palmoplantar keratoderma with constricting bands around fingers, flexural deformities of fingers and keratotic papules in a linear distribution on the flexural side of large joints. Histological examination of the skin of affected individuals shows hyperkeratosis and hyperplasia of the spinous, granular and horny epidermal layer. {ECO:0000269	PubMed:20226437}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Keratosis Palmoplantaris Striata I, AD, 148700, Otozomal Dominant (Fokal Palmoplantar Keratoderma - Eklem Keratozları) (DSG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis palmoplantaris striata I, AD, 148700, Autosomal dominant (Focal palmoplantar keratoderma with joint keratoses) (DSG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG1	DSG1, PPKS1, SPPK1, EPKHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Palmoplantar keratoderma 1, striate, focal, or diffuse (PPKS1) [MIM:148700]: A dermatological disorder characterized by thickening of the skin on the palms and soles, and longitudinal hyperkeratotic lesions on the palms, running the length of each finger. {ECO:0000269	PubMed:10333028}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratosis Striata 2 612908 Palmoplantaris; PPKS2 (Keratozis Palmoplantaris Striata) (DSP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis palmoplantaris striata II, 612908; PPKS2 (Keratosis palmoplantaris striata) (DSP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated, with woolly hair and keratoderma (DCWHK) [MIM:605676]: An autosomal recessive cardiocutaneous syndrome characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. {ECO:0000269	PubMed:11063735}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratosis Striata 3 607654 Palmoplantaris; PPKS3 (Keratozis Palmoplantaris Striata) (KRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis palmoplantaris striata III, 607654; PPKS3 (Keratosis palmoplantaris striata) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9858486}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Keratosis Pilaris Atrofikans, 604093, Otozomal Resesif; KPA (Keratozis Follikülaris Spinulosa Dekalvans) (LRP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis pilaris atrophicans, 604093, Autosomal recessive; KPA (Keratosis follicularis spinulosa decalvans) (LRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP1	LRP1, A2MR, KPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Keratoz, Seboreik, Somatik, 182000 (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keratosis, seborrheic, somatic, 182000 (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Keutel Sendromu, 245150, Otozomal Resesif; KTLS (Keutel Sendromu) (MGP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Keutel syndrome, 245150, Autosomal recessive; KTLS (Keutel syndrome) (MGP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MGP	MGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keutel syndrome (KTLS) [MIM:245150]: An autosomal recessive disorder characterized by abnormal cartilage calcification, peripheral pulmonary stenosis neural hearing loss and midfacial hypoplasia. {ECO:0000269|PubMed:9916809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Keutel Sendromu, 245150, Otozomal Resesif; KTLS (Keutel Sendromu) (Prenatal) (MGP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Keutel syndrome, 245150, Autosomal recessive; KTLS (Keutel syndrome) (MGP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MGP	MGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Keutel syndrome (KTLS) [MIM:245150]: An autosomal recessive disorder characterized by abnormal cartilage calcification, peripheral pulmonary stenosis neural hearing loss and midfacial hypoplasia. {ECO:0000269|PubMed:9916809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kindler Sendromu, 173650, Otozomal Resesif; KINDLRS (Kalıtsal Atrokeratotik Poikiloderma, Kindler-Weary Tipi) (FERMT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kindler syndrome, 173650, Autosomal recessive; KINDLRS (Hereditary acrokeratotic poikiloderma of Kindler-Weary) (FERMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FERMT1	KIND1, URP1, C20orf42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Kindler syndrome (KINDS) [MIM:173650]: Autosomal recessive skin disorder characterized by skin blistering, photosensitivity, progressive poikiloderma, and extensive skin atrophy. Additional clinical features include gingival erosions, ocular, esophageal, gastrointestinal and urogenital involvement, and an increased risk of mucocutaneous malignancy. {ECO:0000269|PubMed:12698616, ECO:0000269|PubMed:12789646, ECO:0000269|PubMed:21936020}. Note=The disease is caused by mutations affecting the gene represented in this entry. Although most FERMT1 mutations are predicted to lead to premature termination of translation, and to loss of FERMT1 function, significant clinical variability is observed among patients. There is an association of FERMT1 missense and in-frame deletion mutations with milder disease phenotypes, and later onset of complications (PubMed:21936020). {ECO:0000269|PubMed:21936020}.;	EDTA Blood Sample
King-Denborough Sendromu, 145600, Otozomal Dominant (King-Denborough Sendromu) (RYR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	King-Denborough syndrome, 145600, Autosomal dominant (King-Denborough syndrome) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23588838, ECO:0000269|PubMed:24561095,	EDTA Blood Sample
King-Denborough Sendromu, 145600, Otozomal Dominant (King-Denborough Sendromu) (Prenatal) (RYR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	King-Denborough syndrome, 145600, Autosomal dominant (King-Denborough syndrome) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23588838, ECO:0000269|PubMed:24561095,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kleefstra Sendromu, 610253, Otozomal Dominant (Kleefstra Sendromu) (2200)	Kleefstra syndrome, 610253, Autosomal dominant (Kleefstra syndrome) (440)	EHMT1	EHMT1, EUHMTASE1, DEL9q34	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	DISEASE: Kleefstra syndrome (KLEST3) [MIM:610253]: A syndrome characterized by severe mental retardation, hypotonia, brachybrachycephaly, and facial dysmorphisms. Additionally, congenital heart defects, urogenital defects, epilepsy and behavioral problems are frequently observed. {ECO:0000269|PubMed:16826528, ECO:0000269|PubMed:19264732}. Note=The disease is caused by mutations affecting the gene represented in this entry (PubMed:16826528). The syndrome can be either caused by intragenic EHMT1 mutations leading to haploinsufficiency of the EHMT1 gene or by a submicroscopic 9q34.3 deletion. Although it is not known if and to what extent other genes in the 9q34.3 region contribute to the syndrome observed in deletion cases, EHMT1 seems to be the major determinant of the core disease phenotype (PubMed:19264732). {ECO:0000269|PubMed:16826528, ECO:0000269|PubMed:19264732}.;	EDTA Blood Sample and Heparinized blood sample

Kleefstra Sendromu, 610253, Otozomal Dominant (Kleefstra Sendromu) (Prenatal)	Kleefstra syndrome, 610253, Autosomal dominant (Kleefstra syndrome) (Prenatal)	EHMT1	EHMT1, EUHMTASE1, DEL9q34	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Kleefstra syndrome (KLESTS) [MIM:610253]: A syndrome characterized by severe mental retardation, hypotonia, brachy(micro)cephaly, and facial dysmorphisms. Additionally, congenital heart defects, urogenital defects, epilepsy and behavioral problems are frequently observed. {ECO:0000269|PubMed:16826528, ECO:0000269|PubMed:19264732}. Note=The disease is caused by mutations affecting the gene represented in this entry (PubMed:16826528). The syndrome can be either caused by intragenic EHMT1 mutations leading to haploinsufficiency of the EHMT1 gene or by a submicroscopic 9q34.3 deletion. Although it is not known if and to what extent other genes in the 9q34.3 region contribute to the syndrome observed in deletion cases. EHMT1 seems to be the major determinant of the core disease phenotype (PubMed:19264732). {ECO:0000269|PubMed:16826528, ECO:0000269|PubMed:19264732};.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Klippel-Feil Sendromu 1, Otozomal Dominant, 118100, Otozomal Dominant; KFS1 (özle Klippel-Feil Sendromu) (GDF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Klippel-Feil syndrome 1, autosomal dominant, 118100, Autosomal dominant; KFS1 (isolated Klippel-Feil syndrome) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. {ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Klippel-Feil Sendromu 1, Otozomal Dominant, 118100, Otozomal Dominant; KFS1 (özle Klippel-Feil Sendromu) (Prenatal) (GDF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Klippel-Feil syndrome 1, autosomal dominant, 118100, Autosomal dominant; KFS1 (isolated Klippel-Feil syndrome) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. {ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Klippel-Feil Sendromu 2, 214300, Otozomal Resesif; KFS2 (özle Klippel-Feil Sendromu) (MEOX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Klippel-Feil syndrome 2, 214300, Autosomal recessive; KFS2 (isolated Klippel-Feil syndrome) (MEOX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEOX1	MEOX1, MOX1, KFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 2, autosomal recessive (KFS2) [MIM:214300]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. {ECO:0000269|PubMed:23290072, ECO:0000269|PubMed:24073994}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Klippel-Feil Sendromu 2, 214300, Otozomal Resesif; KFS2 (özle Klippel-Feil Sendromu) (Prenatal) (MEOX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Klippel-Feil syndrome 2, 214300, Autosomal recessive; KFS2 (isolated Klippel-Feil syndrome) (MEOX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MEOX1	MEOX1, MOX1, KFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Klippel-Feil syndrome 2, autosomal recessive (KFS2) [MIM:214300]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. {ECO:0000269|PubMed:23290072, ECO:0000269|PubMed:24073994}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Klippel-Feil Sendromu 3, Otozomal Dominant, 613702; KFS3 (özle Klippel-Feil Sendromu) (GDF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Klippel-Feil syndrome 3, autosomal dominant, 613702; KFS3 (isolated Klippel-Feil syndrome) (GDF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF3	GDF3, KFS3, MCOPCB6, MCOP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 3, autosomal dominant (KFS3) [MIM:613702]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. {ECO:0000269	PubMed:19864492}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Klippel-Feil Sendromu 3, Otozomal Dominant, 613702; KFS3 (özle Klippel-Feil Sendromu) (Prenatal) (GDF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Klippel-Feil syndrome 3, autosomal dominant, 613702; KFS3 (isolated Klippel-Feil syndrome) (GDF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF3	GDF3, KFS3, MCOPCB6, MCOP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Klippel-Feil syndrome 3, autosomal dominant (KFS3) [MIM:613702]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. {ECO:0000269	PubMed:19864492}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Klippel-Feil Sendromu 4, Miyopati ve Dismorfizm Otozomal Resesif, 616549, Otozomal Resesif; KFS4 (Klippel-Feil Anomalisi-Miyopati-Fasiyal Dismorfizmi Sendromu) (MYO18B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549, Autosomal recessive; KFS4 (Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome) (MYO18B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO18B	MYO18B, KFS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Klippel-Feil Sendromu 4, Miyopati ve Dismorfizm Otozomal Resesif, 616549, Otozomal Resesif; KFS4 (Klippel-Feil Anomalisi-Miyopati-Fasiyal Dismorfizmi Sendromu) (Prenatal) (MYO18B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Klippel-Feil syndrome 4, autosomal recessive, with myopathy and facial dysmorphism, 616549, Autosomal recessive; KFS4 (Klippel-Feil anomaly-myopathy-facial dysmorphism syndrome) (MYO18B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYO18B	MYO18B, KFS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kniest Displazi, 156550, Otozomal Dominant (Kniest Displazi) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kniest dysplasia, 156550, Autosomal dominant (Kniest dysplasia) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kniest Displazi, 156550, Otozomal Dominant (Kniest Displazi) (MLPA)	Kniest dysplasia, 156550, Autosomal dominant (Kniest dysplasia) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kniest Displazi, 156550, Otozomal Dominant (Kniest Displazi) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kniest dysplasia, 156550, Autosomal dominant (Kniest dysplasia) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kniest Displazi, 156550, Otozomal Dominant (Kniest Displazi) (Prenatal) (MLPA)	Kniest dysplasia, 156550, Autosomal dominant (Kniest dysplasia) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Knobloch Sendromu, Tip 1, 267750, Otozomal Resesif; KNO1 (Knobloch Sendromu) (COL18A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Knobloch syndrome, type 1, 267750, Autosomal recessive; KNO1 (Knobloch syndrome) (COL18A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL18A1	COL18A1, KNO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Knobloch syndrome 1 (KNO1) [MIM:267750]: A developmental disorder primarily characterized by typical eye abnormalities, including high myopia, cataracts, dislocated lens, vitreoretinal degeneration, and retinal detachment, with occipital skull defects, which can range from occipital encephalocele to occult cutis aplasia. {ECO:0000269|PubMed:10942434, ECO:0000269|PubMed:23667181}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Knobloch Sendromu, Tip 1, 267750, Otozomal Resesif; KNO1 (Knobloch Sendromu) (Prenatal) (COL18A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Knobloch syndrome, type 1, 267750, Autosomal recessive; KNO1 (Knobloch syndrome) (COL18A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL18A1	COL18A1, KNO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Knobloch syndrome 1 (KNO1) [MIM:267750]: A developmental disorder primarily characterized by typical eye abnormalities, including high myopia, cataracts, dislocated lens, vitreoretinal degeneration, and retinal detachment, with occipital skull defects, which can range from occipital encephalocele to occult cutis aplasia. {ECO:0000269|PubMed:10942434, ECO:0000269|PubMed:23667181}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Parmak Ucu Yastıkçıkları, Lökonişi ve Sensorinöral Sağırlık (Parmak Yastıkları-Lökonişi-Sensorinöral İşitme Kaybı-Palmoplantar Hiperkeratoz Sendromu (Bart-Pumphrey Sendromu)) (GJB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	KNUCKLE PADS, LEUKONYCHIA, AND SENSORINEURAL DEAFNESS (Knuckle pads-leukonychia-sensorineural deafness-palmoplantar hyperkeratosis syndrome) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vohwinkel syndrome (VS) [MIM:124500]: VS is an autosomal dominant disease characterized by hyperkeratosis, constriction on fingers and toes and congenital deafness. {ECO:0000269|PubMed:10369869, ECO:0000269|PubMed:15954104, ECO:0000269|PubMed:18688874}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kohlschutter-Tonz Sendromu, 226750, Otozomal Resesif; KTZS (Amelo-Serebro-Hipohidrotik Sendrom (Kohlschutter-Tonz Sendromu)) (ROGDI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kohlschutter-Tonz syndrome, 226750, Autosomal recessive; KTZS (Amelo-cerebro-hypohidrotic syndrome) (ROGDI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROGDI	ROGDI, KTZS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kohlschutter-Tonz Sendromu, 226750, Otozomal Resesif; KTZS (Amelo-Serebro-Hipohidrotik Sendrom (Kohlschutter-Tonz Sendromu)) (Prenatal) (ROGDI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Kohlschutter-Tonz syndrome, 226750, Autosomal recessive; KTZS (Amelo-cerebro-hypohidrotic syndrome) (ROGDI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ROGDI	ROGDI, KTZS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Koolen-De Vries Sendromu, 610443, Otozomal Dominant; KDVS (17q21.31 Mikrodelesyon Sendromu) (2200)	Koolen-De Vries syndrome, 610443, Autosomal dominant; KDVS (17q21.31 microdeletion syndrome) (4400)	KANSL1	KANSL1, KIAA1267, MSL1V1, KDVS	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal		EDTA Blood Sample and Heparinized blood sample
Koolen-De Vries Sendromu, 610443, Otozomal Dominant; KDVS (17q21.31 Mikrodelesyon Sendromu) (Prenatal)	Koolen-De Vries syndrome, 610443, Autosomal dominant; KDVS (17q21.31 microdeletion syndrome) (Prenatal)	KANSL1	KANSL1, KIAA1267, MSL1V1, KDVS	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kosaki Aşırı Büyüme Sendromu, 616592, Otozomal Dominant; KOGS (Iskelet Aşırı Büyümesi-Kraniofasyal Dismorfizm-Hiperelastik Cilt-Beyaz Cevher Lezyonları Sendromu) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kosaki overgrowth syndrome, 616592, Autosomal dominant; KOGS (Skeletal overgrowth-craniofacial dysmorphism-hyperelastic skin-white matter lesions syndrome) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloproliferative disorder chronic with eosinophilia (MPD) [MIM:131440]: A hematologic disorder characterized by malignant eosinophils proliferation. Note=The gene represented in this entry may be involved in disease pathogenesis. Chromosomal aberrations involving PDGFRB have been found in many instances of chronic myeloproliferative disorder with eosinophilia. Translocation t(5;12) with ETV6 on chromosome 12 creating an PDGFRB-ETV6 fusion protein (PubMed:12181402). Translocation t(5;15)(q33;q22) with TPS3BP1 creating a PDGFRB-TPS3BP1 fusion protein (PubMed:15492236). Translocation t(1;5)(p23;q33) that forms a POE4DP-PDGFRB fusion protein (PubMed:12907457). Translocation t(5;6)(q33-34;q23) with CEP85L that fuses the 5'-end of CEP85L (isoform 4) to the 3'-end of PDGFRB (PubMed:21938754). {ECO:0000269|PubMed:12181402, ECO:0000269|PubMed:12907457, ECO:0000269|PubMed:15492236, ECO:0000269|PubMed:21938754}.	EDTA Blood Sample
Kowarski Sendromu, 262650, Otozomal Resesif (Büyüme Hormonu Kalıtsal Anomalisine Bağlı Boy Kısalığı) (GH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kowarski syndrome, 262650, Autosomal recessive (Short stature due to growth hormone qualitative anomaly) (GH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GH1	GH1, GHN, IGHD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Growth hormone deficiency, isolated, 2 (IGHD2) [MIM:173100]: An autosomal dominant deficiency of growth hormone leading to short stature. Clinical severity is variable. Patients have a positive response and immunologic tolerance to growth hormone therapy. {ECO:0000269|PubMed:11502836, ECO:0000269|PubMed:9152628}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kowarski Sendromu, 262650, Otozomal Resesif (Büyüme Hormonu Kalıtsal Anomalisine Bağlı Boy Kısalığı) (MLPA)	Kowarski syndrome, 262650, Autosomal recessive (Short stature due to growth hormone qualitative anomaly) (MLPA)	GH1	GH1, GHN, IGHD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Growth hormone deficiency, isolated, 2 (IGHD2) [MIM:173100]: An autosomal dominant deficiency of growth hormone leading to short stature. Clinical severity is variable. Patients have a positive response and immunologic tolerance to growth hormone therapy. {ECO:0000269|PubMed:11502836, ECO:0000269|PubMed:9152628}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Krabbe Hastalığı, 245200, Otozomal Resesif (Krabbe Hastalığı) (GALC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Krabbe disease, 245200, Autosomal recessive (Krabbe disease) (GALC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALC	GALC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, globoid cell (GLD) [MIM:245200]: An autosomal recessive disorder characterized by insufficient catabolism of several galactolipids that are important for normal myelin production. Four clinical forms are recognized. The infantile form accounts for 90% of cases. It manifests before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. Cases with later onset present with unexplained blindness, weakness and sensorimotor peripheral neuropathy, mental deterioration and death. {ECO:0000269|PubMed:10234611, ECO:0000269|PubMed:10477434, ECO:0000269|PubMed:17579360, ECO:0000269|PubMed:20886637, ECO:0000269|PubMed:23462331, ECO:0000269|PubMed:8595408, ECO:0000269|PubMed:8786069, ECO:0000269|PubMed:8940268, ECO:0000269|PubMed:9272171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Krabbe Hastalığı, 245200, Otozomal Resesif (Krabbe Hastalığı) (MLPA)	Krabbe disease, 245200, Autosomal recessive (Krabbe disease) (MLPA)	GALC	GALC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukodystrophy, globoid cell (GLD) [MIM:245200]: An autosomal recessive disorder characterized by insufficient catabolism of several galactolipids that are important for normal myelin production. Four clinical forms are recognized. The infantile form accounts for 90% of cases. It manifests before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. Cases with later onset present with unexplained blindness, weakness and sensorimotor peripheral neuropathy, mental deterioration and death. {ECO:0000269|PubMed:10234611, ECO:0000269|PubMed:10477434, ECO:0000269|PubMed:17579360, ECO:0000269|PubMed:20886637, ECO:0000269|PubMed:23462331, ECO:0000269|PubMed:8595408, ECO:0000269|PubMed:8786069, ECO:0000269|PubMed:8940268, ECO:0000269|PubMed:9272171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Krabbe Hastalığı, 245200, Otozomal Resesif (Krabbe Hastalığı) (Prenatal) (GALC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Krabbe disease, 245200, Autosomal recessive (Krabbe disease) (GALC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GALC	GALC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, globoid cell (GLD) [MIM:245200]: An autosomal recessive disorder characterized by insufficient catabolism of several galactolipids that are important for normal myelin production. Four clinical forms are recognized. The infantile form accounts for 90% of cases. It manifests before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. Cases with later onset present with unexplained blindness, weakness and sensorimotor peripheral neuropathy, mental deterioration and death. {ECO:0000269|PubMed:10234611, ECO:0000269|PubMed:10477434, ECO:0000269|PubMed:17579360, ECO:0000269|PubMed:20886637, ECO:0000269|PubMed:23462331, ECO:0000269|PubMed:8595408, ECO:0000269|PubMed:8786069, ECO:0000269|PubMed:8940268, ECO:0000269|PubMed:9272171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Krabbe Hastalığı, 245200, Otozomal Resesif (Krabbe Hastalığı) (Prenatal) (MLPA)	Krabbe disease, 245200, Autosomal recessive (Krabbe disease) (Prenatal) (MLPA)	GALC	GALC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, globoid cell (GLD) [MIM:245200]: An autosomal recessive disorder characterized by insufficient catabolism of several galactolipids that are important for normal myelin production. Four clinical forms are recognized. The infantile form accounts for 90% of cases. It manifests before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. Cases with later onset present with unexplained blindness, weakness and sensorimotor peripheral neuropathy, mental deterioration and death. {ECO:0000269|PubMed:10234611, ECO:0000269|PubMed:10477434, ECO:0000269|PubMed:17579360, ECO:0000269|PubMed:20886637, ECO:0000269|PubMed:23462331, ECO:0000269|PubMed:8595408, ECO:0000269|PubMed:8786069, ECO:0000269|PubMed:8940268, ECO:0000269|PubMed:9272171}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Krabbe Hastalığı, Atipik, Saposin A Eksikliğine Bağlı, 611722, Otozomal Resesif (Krabbe Hastalığı) (PSAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	KRABBE DISEASE, ATYPICAL, DUE TO SAPOSIN A DEFICIENCY, 611722, Autosomal recessive (Krabbe disease) (PSAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy metachromatic due to saposin-B deficiency (MLD-SAPB) [MIM:249900]: An atypical form of metachromatic leukodystrophy. It is characterized by tissue accumulation of cerebroside-3-sulfate, demyelination, periventricular white matter abnormalities, peripheral neuropathy. Additional neurological features include dysarthria, ataxic gait, psychomotor regression, seizures, cognitive decline and spastic quadriparesis. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Krabbe Hastalığı, Atipik, Saposin A Eksikliğine Bağlı, 611722, Otozomal Resesif (Krabbe Hastalığı) (Prenatal) (PSAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	KRABBE DISEASE, ATYPICAL, DUE TO SAPOSIN A DEFICIENCY, 611722, Autosomal recessive (Krabbe disease) (PSAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy metachromatic due to saposin-B deficiency (MLD-SAPB) [MIM:249900]: An atypical form of metachromatic leukodystrophy. It is characterized by tissue accumulation of cerebroside-3-sulfate, demyelination, periventricular white matter abnormalities, peripheral neuropathy. Additional neurological features include dysarthria, ataxic gait, psychomotor regression, seizures, cognitive decline and spastic quadriparesis. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
KRAS Kodon 12, 13, 61, 146 Mutasyonu (Dizi Analizi) (KRAS geni) (Dizi Analizi) (Postnatal)	KRAS codon 12,13,61,146 mutations (Sequence analysis) (KRAS gene) (Dizi Analizi) (Postnatal)	KRAS	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269	PubMed:8955068). Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
NRAS Kodon 12, 13, 61, 146 Mutasyonu (Dizi Analizi) (NRAS geni) (Dizi Analizi) (Postnatal)	NRAS codon 12,13,61,146 mutations (Sequence analysis) (NRAS gene) (Dizi Analizi) (Postnatal)	NRAS	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Kufor-Rakeb Sendromu, 606693, Otozomal Resesif (Parkinsonizm, ATP13A2 Eksikliğine Bağlı) (ATP13A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kufor-Rakeb syndrome, 606693, Autosomal recessive (Parkinsonism due to ATP13A2 deficiency) (ATP13A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP13A2	ATP13A2, PARK9, KRPPD, SPG78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 12 (CLN12) [MIM:606693]: A form of neuronal ceroid lipofuscinosis characterized by rapidly progressive visual loss due to retinal dystrophy, seizures, cerebellar ataxia, and cerebellar atrophy. Cognitive decline may also occur. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material. (ECO:0000269	PubMed:22388936). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kuru, Yatkınlık, 245300 (Kuru) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Kuru, susceptibility to, 245300 (Kuru) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L-2-Hidroksiglutarik Asidüri, 236792, Otozomal Resesif; L2HGA (L-2-Hidroksi Glutarik Asidüri) (L2HGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	L-2-hydroxyglutaric aciduria, 236792, Autosomal recessive; L2HGA (L-2-hydroxyglutaric aciduria) (L2HGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L2HGDH	L2HGDH, C14orf160, L2HGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L-2-Hidroksiglutarik Asidüri, 236792, Otozomal Resesif; L2HGA (L-2-Hidroksi Glutarik Asidüri) (MLPA)	L-2-hydroxyglutaric aciduria, 236792, Autosomal recessive; L2HGA (L-2-hydroxyglutaric aciduria) (MLPA)	L2HGDH	L2HGDH, C14orf160, L2HGA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
L-2-Hidroksiglutarik Asidüri, 236792, Otozomal Resesif; L2HGA (L-2-Hidroksi Glutarik Asidüri) (Prenatal) (L2HGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	L-2-hydroxyglutaric aciduria, 236792, Autosomal recessive; L2HGA (L-2-hydroxyglutaric aciduria) (L2HGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L2HGDH	L2HGDH, C14orf160, L2HGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
L-2-Hidroksiglutarik Asidüri, 236792, Otozomal Resesif; L2HGA (L-2-Hidroksi Glutarik Asidüri) (Prenatal) (MLPA)	L-2-hydroxyglutaric aciduria, 236792, Autosomal recessive; L2HGA (L-2-hydroxyglutaric aciduria) (Prenatal) (MLPA)	L2HGDH	L2HGDH, C14orf160, L2HGA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
L-Ferritin Eksikliği, Dominant ve Resesif, 615604, Otozomal Resesif, Otozomal Dominant; LFTD (L-Ferritin Eksikliği) (FTL geni) (Tüm Gen Dizi Analizi) (Postnatal)	L-ferritin deficiency, dominant and recessive, 615604, Autosomal recessive, Autosomal dominant; LFTD (L-ferritin deficiency) (FTL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTL	FTL, NBIA3, LFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. (ECO:0000269	PubMed:19176363). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
L-Treonin Dehidrogenaz, Psödojen; TDH (TDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	L-THREONINE DEHYDROGENASE, PSEUDOGENE; TDH (TDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TDH	TDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L-Treonin Dehidrogenaz, Psödojen; TDH (Prenatal) (TDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	L-THREONINE DEHYDROGENASE, PSEUDOGENE; TDH (TDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TDH	TDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lakrimal Kanal Kusur, 149700, Otozomal Resesif; LCDD (Alleset Konjenital Nazolakrimal Kanal Tıkanıklığı) (IGSF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lacrimal duct defect, 149700, Autosomal recessive; LCDD (Familial congenital nasolacrimal duct obstruction) (IGSF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGSF3	IGSF3, LCDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lakrimoaurikülodentodigital sendrom (LADD sendromu) (FGF10 5p13, FGFR2 10q26) (Postnatal)	Lacrimoauriculodentodigital syndrome (LADD syndrome) (FGF10 5p13, FGFR2 10q26) (MLPA)	FGF10 5p13, FGFR2 10q26	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Lakrimoaurikülodentodigital sendrom (LADD sendromu) (FGF10 5p13, FGFR2 10q26) (MLPA) (Prenatal)	Lacrimoauriculodentodigital syndrome (LADD syndrome) (FGF10 5p13, FGFR2 10q26) (MLPA) (Prenatal)	FGF10 5p13, FGFR2 10q26	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laktaz Eksikliği, Konjenital, 223000, Otozomal Resesif (Konjenital Laktaz Eksikliği) (LCT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lactase deficiency, congenital, 223000, Autosomal recessive (Congenital lactase deficiency) (LCT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LCT	LCT, LAC, LPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital lactase deficiency (COLACD) [MIM:223000]: Autosomal recessive, rare and severe gastrointestinal disorder. It is characterized by watery diarrhea in infants fed with breast milk or other lactose-containing formulas. An almost total lack of LCT activity is found in jejunal biopsy material of patients with congenital lactase deficiency. Opposite to congenital lactase deficiency, also known as lactose intolerance, is the most common enzyme deficiency worldwide. It is caused by developmental down- regulation of lactase activity during childhood or early adulthood. The decline of lactase activity is a normal physiological phenomenon; however, the majority of Northern Europeans have the ability to maintain lactase activity and digest lactose throughout life (lactase persistence). The down-regulation of lactase activity operates at the transcriptional level and it is associated with a noncoding variation in the MCM6 gene, located in the upstream vicinity of LCT. (ECO:0000269	PubMed:16400612). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Laktaz Eksikliği, Konjenital, 223000, Otozomal Resesif (Konjenital Laktaz Eksikliği) (Prenatal) (LCT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lactase deficiency, congenital, 223000, Autosomal recessive (Congenital lactase deficiency) (LCT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LCT	LCT, LAC, LPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital lactase deficiency (COLACD) [MIM:223000]: Autosomal recessive, rare and severe gastrointestinal disorder. It is characterized by watery diarrhea in infants fed with breast milk or other lactose-containing formulas. An almost total lack of LCT activity is found in jejunal biopsy material of patients with congenital lactase deficiency. Opposite to congenital lactase deficiency, also known as lactose intolerance, is the most common enzyme deficiency worldwide. It is caused by developmental down- regulation of lactase activity during childhood or early adulthood. The decline of lactase activity is a normal physiological phenomenon; however, the majority of Northern Europeans have the ability to maintain lactase activity and digest lactose throughout life (lactase persistence). The down-regulation of lactase activity operates at the transcriptional level and it is associated with a noncoding variation in the MCM6 gene, located in the upstream vicinity of LCT. (ECO:0000269	PubMed:16400612). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laktaz Persistansı / Nonpersistans, 223100, Otozomal Dominant (MCM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lactase persistence/nonpersistence, 223100, Autosomal dominant (MCM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCM6	MCM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Laktikasidemi, PDX1 Eksikliğine Bağlı, 245349, Otozomal Resesif; PDHKD (Piruvat Dehidrogenaz Eksikliği) (PDHX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lacticacidemia due to PDX1 deficiency, 245349, Autosomal recessive; PDHKD (Pyruvate dehydrogenase deficiency) (PDHX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDHK	PDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase E3-binding protein deficiency (PDHXD) [MIM:245349]: A metabolic disorder characterized by decreased activity of the pyruvate dehydrogenase complex without observable reduction in the activities of enzymes E1, E2, or E3. Clinical features include hypotonia and psychomotor retardation. (ECO:0000269	PubMed:9399911). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Laktikasidemi, PDX1 Eksikliğine Bağlı, 245349, Otozomal Resesif; PDHKD (Piruvat Dehidrogenaz Eksikliği) (Prenatal) (PDHX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lacticacidemia due to PDX1 deficiency, 245349, Autosomal recessive; PDHKD (Pyruvate dehydrogenase deficiency) (PDHX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDHK	PDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase E3-binding protein deficiency (PDHXD) [MIM:245349]: A metabolic disorder characterized by decreased activity of the pyruvate dehydrogenase complex without observable reduction in the activities of enzymes E1, E2, or E3. Clinical features include hypotonia and psychomotor retardation. (ECO:0000269	PubMed:9399911). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodigital Sendromu) (FGF10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LADD syndrome, 149730, Autosomal dominant (Lacrimoauriculodentodigital syndrome) (FGF10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF10	FGF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]: An autosomal dominant ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Lacrimo-auriculo-dento-digital syndrome is characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed. (ECO:0000269|PubMed:16501574, ECO:0000269|PubMed:16630169). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodigital Sendromu) (MLPA)	LADD syndrome, 149730, Autosomal dominant (Lacrimoauriculodentodigital syndrome) (MLPA)	FGF10	FGF10	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]: An autosomal dominant ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Lacrimo-auriculo-dento-digital syndrome is characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed. (ECO:0000269|PubMed:16501574, ECO:0000269|PubMed:16630169). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodijital Sendrom) (Prenatal) (FGF10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LADD syndrome, 149730, Autosomal dominant (Lacrimoauriculodentodigital syndrome) (FGF10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGF10	FGF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]: An autosomal dominant ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Lacrimo-auriculo-dento-digital syndrome is characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed. {ECO:0000269|PubMed:16501574, ECO:0000269|PubMed:16630169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodijital Sendrom) (Prenatal) (MLPA)	LADD syndrome, 149730, Autosomal dominant (Lacrimoauriculodentodigital syndrome) (Prenatal) (MLPA)	FGF10	FGF10	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lacrimo-auriculo-dento-digital syndrome (LADDS) [MIM:149730]: An autosomal dominant ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Lacrimo-auriculo-dento-digital syndrome is characterized by aplastic/hypoplastic lacrimal and salivary glands and ducts, cup-shaped ears, hearing loss, hypodontia and enamel hypoplasia, and distal limb segments anomalies. In addition to these cardinal features, facial dysmorphism, malformations of the kidney and respiratory system and abnormal genitalia have been reported. Craniosynostosis and severe syndactyly are not observed. {ECO:0000269|PubMed:16501574, ECO:0000269|PubMed:16630169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant; LADD (Lakrimoaurikülodentodijital Sendromi) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LADD syndrome, 149730, Autosomal dominant; LADD (Lacrimoauriculodentodigital syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant; LADD (Lakrimoaurikülodentodijital Sendromi) (MLPA)	LADD syndrome, 149730, Autosomal dominant; LADD (Lacrimoauriculodentodigital syndrome) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant; LADD (Lakrimoaurikülodentodijital Sendromi) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LADD syndrome, 149730, Autosomal dominant; LADD (Lacrimoauriculodentodigital syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant; LADD (Lakrimoaurikülodentodijital Sendromi) (Prenatal) (MLPA)	LADD syndrome, 149730, Autosomal dominant; LADD (Lacrimoauriculodentodigital syndrome) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodijital Sendromi) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LADD syndrome,149730, autosomal dominant (Lacrimoauriculodentodigital syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicononal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7698257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LADD Sendromu, 149730, Otozomal Dominant (Lakrimoaurikülodentodijital Sendromi) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LADD syndrome,149730, autosomal dominant (Lacrimoauriculodentodigital syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicononal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7698257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laing Distal Miyopati, 160500, Otozomal Dominant; MPD1 (Laing Erken Başlangıçlı Distal Miyopati) (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Laing distal myopathy, 160500, Autosomal dominant; MPD1 (Laing early-onset distal myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Laing Distal Miyopati, 160500, Otozomal Dominant; MPD1 (Laing Erken Başlangıçlı Distal Miyopati) (Prenatal) (MYH7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Laing distal myopathy, 160500, Autosomal dominant; MPD1 (Laing early-onset distal myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lamb-Shaffer Sendromu, 616803, Otozomal Dominant (Gelişme ve Konuşma Gecikmesi, SOX5 Eksikliğine Bağlı) (SOX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lamb-Shaffer syndrome, 616803, Autosomal dominant (Developmental and speech delay due to SOX5 deficiency) (SOX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX5	SOX5, LAMSHF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Lamb-Shaffer Sendromu, 616803, Otozomal Dominant (Gelişme ve Konuşma Gecikmesi, SOX5 Eksikliğine Bağlı) (Prenatal) (SOX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lamb-Shaffer syndrome, 616803, Autosomal dominant (Developmental and speech delay due to SOX5 deficiency) (SOX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX5	SOX5, LAMSHF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Langer Mesomelik Displazi, 249700, Otozomal Resesif; LMD (Langer Mezomelik Displazi) (SHOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Langer mesomelic dysplasia, 249700, Autosomal recessive; LMD (Langer mesomelic dysplasia) (SHOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHOX	SHOX, GCFX, SS, PHOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. {ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Langer Mesomelik Displazi, 249700, Otozomal Resesif; LMD (Langer Mezomelik Displazi) (MLPA)	Langer mesomelic dysplasia, 249700, Autosomal recessive; LMD (Langer mesomelic dysplasia) (MLPA)	SHOX	SHOX, GCFX, SS, PHOG	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. {ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Langer Mesomelik Displazi, 249700, Otozomal Resesif; LMD (Langer Mezomelik Displazi) (Prenatal) (SHOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Langer mesomelic dysplasia, 249700, Autosomal recessive; LMD (Langer mesomelic dysplasia) (SHOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHOX	SHOX, GCFX, SS, PHOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. {ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Langer Mesomelik Displazi, 249700, Otozomal Resesif; LMD (Langer Mezomelik Displazi) (Prenatal) (MLPA)	Langer mesomelic dysplasia, 249700, Autosomal recessive; LMD (Langer mesomelic dysplasia) (Prenatal) (MLPA)	SHOX	SHOX, GCFX, SS, PHOG	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. {ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Dil Gecikmesi ve DEHB / Kognitif Bozukluk, x Kardiyak Artmi, 617182, Otozomal Resesif; LADCI (GNB5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182, Autosomal recessive; LADCI (GNB5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB5	GNB5, GB5, IDDCA, LADCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Dil Gecikmesi ve DEHB / Kognitif Bozukluk, x Kardiyak Artmi, 617182, Otozomal Resesif; LADCI (Prenatal) (GNB5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Language delay and ADHD/cognitive impairment with or without cardiac arrhythmia, 617182, Autosomal recessive; LADCI (GNB5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNB5	GNB5, GB5, IDDCA, LADCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Laron Tipi Kısa Boy, 262500, Otozomal Resesif (Laron Sendromu) (GHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Laron dwarfism, 262500, Autosomal recessive (Laron syndrome) (GHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHR	GHR, GHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laron syndrome (LARS) [MIM:262500]: A severe form of growth hormone insensitivity characterized by growth impairment, short stature, dysfunctional growth hormone receptor, and failure to generate insulin-like growth factor I in response to growth hormone. {ECO:0000269|PubMed:10870033, ECO:0000269|PubMed:14678285, ECO:0000269|PubMed:2779634, ECO:0000269|PubMed:8137822, ECO:0000269|PubMed:8421103, ECO:0000269|PubMed:8504296, ECO:0000269|PubMed:9024232, ECO:0000269|PubMed:9661642, ECO:0000269|PubMed:9851797}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Laron Tipi Kısa Boy, 262500, Otozomal Resesif (Laron Sendromu) (Prenatal) (GHR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Laron dwarfism, 262500, Autosomal recessive (Laron syndrome) (GHR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GHR	GHR, GHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laron syndrome (LARS) [MIM:262500]: A severe form of growth hormone insensitivity characterized by growth impairment, short stature, dysfunctional growth hormone receptor, and failure to generate insulin-like growth factor I in response to growth hormone. {ECO:0000269|PubMed:10870033, ECO:0000269|PubMed:14678285, ECO:0000269|PubMed:2779634, ECO:0000269|PubMed:8137822, ECO:0000269|PubMed:8421103, ECO:0000269|PubMed:8504296, ECO:0000269|PubMed:9024232, ECO:0000269|PubMed:9661642, ECO:0000269|PubMed:9851797}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Larsen Sendromu, 150250, Otozomal Dominant; LRS (Larsen Sendromu, Otozomal Dominant) (FLNB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Larsen syndrome, 150250, Autosomal dominant; LRS (Autosomal dominant Larsen syndrome) (FLNB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269|PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Larsen Sendromu, 150250, Otozomal Dominant; LRS (Larsen Sendromu, Otozomal Dominant) (Prenatal) (FLNB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Larsen syndrome, 150250, Autosomal dominant; LRS (Autosomal dominant Larsen syndrome) (FLNB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269|PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laringoonikolutanöz Sendrom, 245660, Otozomal Resesif; LOCS (LOC Sendromu) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Laryngonychocutaneous syndrome, 245660, Autosomal recessive; LOCS (LOC syndrome) (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. {ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Laringoonikolutanöz Sendrom, 245660, Otozomal Resesif; LOCS (LOC Sendromu) (MLPA)	Laryngonychocutaneous syndrome, 245660, Autosomal recessive; LOCS (LOC syndrome) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. {ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Laringoonikolutanöz Sendrom, 245660, Otozomal Resesif; LOCS (LOC Sendromu) (Prenatal) (LAMA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Laryngonychocutaneous syndrome, 245660, Autosomal recessive; LOCS (LOC syndrome) (LAMA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA3	LAMA3, LOCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. {ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laringoonikolutanöz Sendrom, 245660, Otozomal Resesif; LOCS (LOC Sendromu) (MLPA)	Laryngonychocutaneous syndrome, 245660, Autosomal recessive; LOCS (LOC syndrome) (Prenatal) (MLPA)	LAMA3	LAMA3, LOCS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epidermolysis bullosa, junctional, Herlitz type (H-JEB) [MIM:226700]: An infantile and lethal form of junctional epidermolysis bullosa, a group of blistering skin diseases characterized by tissue separation which occurs within the dermo- epidermal basement In the Herlitz type, death occurs usually within the first six months of life. Occasionally, children survive to teens. It is marked by bullous lesions at birth and extensive denudation of skin and mucous membranes that may be hemorrhagic. {ECO:0000269|PubMed:7633458, ECO:0000269|PubMed:8530087}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lateral Meningosel Sendromu, 130720, Otozomal Dominant; LMNS (Lateral Meningosel Sendromu) (NOTCH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lateral meningocele syndrome, 130720, Autosomal dominant; LMNS (Lateral meningocele syndrome) (NOTCH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH3	NOTCH3, CADASIL1, CASIL, IMF2, LMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, autosomal dominant (CADASIL) [MIM:125310]: A cerebrovascular disease characterized by multiple subcortical infarcts, pseudobulbar palsy, dementia, and the presence of granular deposits in small cerebral arteries producing ischemic stroke. {ECO:0000269|PubMed:10227618, ECO:0000269|PubMed:10371548, ECO:0000269|PubMed:10802807, ECO:0000269|PubMed:10854111, ECO:0000269|PubMed:11058919, ECO:0000269|PubMed:11102981, ECO:0000269|PubMed:11559313, ECO:0000269|PubMed:11755616, ECO:0000269|PubMed:11810186, ECO:0000269|PubMed:12136071, ECO:0000269|PubMed:12146805, ECO:0000269|PubMed:12589106, ECO:0000269|PubMed:12810003, ECO:0000269|PubMed:15229130, ECO:0000269|PubMed:15300988, ECO:0000269|PubMed:15364702, ECO:0000269|PubMed:15378071, ECO:0000269|PubMed:15818833, ECO:0000269|PubMed:16009764, ECO:0000269|PubMed:24000151, ECO:0000269|PubMed:9388399}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lateral Meningosel Sendromu, 130720, Otozomal Dominant; LMNS (Lateral Meningosel Sendromu) (Prenatal) (NOTCH3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lateral meningocele syndrome, 130720, Autosomal dominant; LMNS (Lateral meningocele syndrome) (NOTCH3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOTCH3	NOTCH3, CADASIL1, CASIL, IMF2, LMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, autosomal dominant (CADASIL) [MIM:125310]: A cerebrovascular disease characterized by multiple subcortical infarcts, pseudobulbar palsy, dementia, and the presence of granular deposits in small cerebral arteries producing ischemic stroke. {ECO:0000269|PubMed:10227618, ECO:0000269|PubMed:10371548, ECO:0000269|PubMed:10802807, ECO:0000269|PubMed:10854111, ECO:0000269|PubMed:11058919, ECO:0000269|PubMed:11102981, ECO:0000269|PubMed:11559313, ECO:0000269|PubMed:11755616, ECO:0000269|PubMed:11810186, ECO:0000269|PubMed:12136071, ECO:0000269|PubMed:12146805, ECO:0000269|PubMed:12589106, ECO:0000269|PubMed:12810003, ECO:0000269|PubMed:15229130, ECO:0000269|PubMed:15300988, ECO:0000269|PubMed:15364702, ECO:0000269|PubMed:15378071, ECO:0000269|PubMed:15818833, ECO:0000269|PubMed:16009764, ECO:0000269|PubMed:24000151, ECO:0000269|PubMed:9388399}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Latosterolosis, 607330, Otozomal Resesif (Latosterolozis) (SCSD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lathosterolosis, 607330, Autosomal recessive (Lathosterolosis) (SCSD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCSD	SCSDL, ERG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lathosterolosis (LATHST) [MIM:607330]: Autosomal recessive disorder characterized by a complex phenotype, including multiple congenital anomalies, mental retardation, and liver disease. {ECO:0000269|PubMed:12189593, ECO:0000269|PubMed:12812989}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Latosterolosis, 607330, Otozomal Resesif (Latosterolozis) (Prenatal) (SCSD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lathosterolosis, 607330, Autosomal recessive (Lathosterolosis) (SCSD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCSD	SCSDL, ERG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lathosterolosis (LATHST) [MIM:607330]: Autosomal recessive disorder characterized by a complex phenotype, including multiple congenital anomalies, mental retardation, and liver disease. {ECO:0000269|PubMed:12189593, ECO:0000269|PubMed:12812989}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Laurence-Moon Sendromu, 245800, Otozomal Resesif; LNMS (Laurence-Moon Sendromu) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Laurence-Moon syndrome, 245800, Autosomal recessive; LNMS (Laurence-Moon syndrome) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA6	PNPLA6, NTE, SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. {ECO:0000269|PubMed:24355708, ECO:0000269|PubMed:25033069}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Laurence-Moon Sendromu, 245800, Otozomal Resesif; LNMS (Laurence-Moon Sendromu) (Prenatal) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Laurence-Moon syndrome, 245800, Autosomal recessive; LNMS (Laurence-Moon syndrome) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPLA6	PNPLA6, NTE, SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. {ECO:0000269|PubMed:24355708, ECO:0000269|PubMed:25033069}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Laurin-Sandrow Sendromu, 135750, Otozomal Dominant; LSS (Laurin-Sandrow Sendromu) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Laurin-Sandrow syndrome, 135750, Autosomal dominant; LSS (Laurin-Sandrow syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159},.	EDTA Blood Sample
Laurin-Sandrow Sendromu, 135750, , Otozomal Dominant; LSS (Laurin-Sandrow Sendromu) (Prenatal) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Laurin-Sandrow syndrome, 135750, Autosomal dominant; LSS (Laurin-Sandrow syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159},.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LCHAD Eksikliği, 609016, Otozomal Resesif (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (HADHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	LCHAD deficiency, 609016, Autosomal recessive (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269	PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LCHAD Eksikliği, 609016, Otozomal Resesif (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (Prenatal) (HADHA geni) (Tüm Gen Dizi Analizi) (Prenatal)	LCHAD deficiency, 609016, Autosomal recessive (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269	PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LDL Kolesterol Seviyesi QTL2'nin, 143890, Otozomal Dominant (Homozigot Allesel Hiperkolesterolemi) (LDLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	LDL cholesterol level QTL2, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (LDLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDLR	LDLR, FHC, FH, LDLCQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hypercholesterolemia (FH) [MIM:143890]: Common autosomal semi-dominant disease that affects about 1 in 500 individuals. The receptor defect impairs the catabolism of LDL, and the resultant elevation in plasma LDL-cholesterol promotes deposition of cholesterol in the skin (xanthelasma), tendons (xanthomas), and coronary arteries (atherosclerosis). {ECO:0000269|PubMed:10090484, ECO:0000269|PubMed:10206683, ECO:0000269|PubMed:10422803, ECO:0000269|PubMed:10447263, ECO:0000269|PubMed:10532689, ECO:0000269|PubMed:10660340, ECO:0000269|PubMed:10882754, ECO:0000269|PubMed:10978268, ECO:0000269|PubMed:10980548, ECO:0000269|PubMed:11298688, ECO:0000269|PubMed:11462246, ECO:0000269|PubMed:1446662, ECO:0000269|PubMed:1464748, ECO:0000269|PubMed:17142622, ECO:0000269|PubMed:17347910, ECO:0000269|PubMed:1867200, ECO:0000269|PubMed:1931802S, ECO:0000269|PubMed:19319977, ECO:0000269|PubMed:22160468, ECO:0000269|PubMed:2318961, ECO:0000269|PubMed:24529145, ECO:0000269|PubMed:2569482, ECO:0000269|PubMed:2726768, ECO:0000269|PubMed:3263645, ECO:0000269|PubMed:3955657, ECO:0000269|PubMed:7502709	EDTA Blood Sample
LDL Kolesterol Seviyesi QTL2'nin, 143890, Otozomal Dominant (Homozigot Allesel Hiperkolesterolemi) (MLPA)	LDL cholesterol level QTL2, 143890, Autosomal dominant (Homozygous familial hypercholesterolemia) (MLPA)	LDLR	LDLR, FHC, FH, LDLCQ2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hypercholesterolemia (FH) [MIM:143890]: Common autosomal semi-dominant disease that affects about 1 in 500 individuals. The receptor defect impairs the catabolism of LDL, and the resultant elevation in plasma LDL-cholesterol promotes deposition of cholesterol in the skin (xanthelasma), tendons (xanthomas), and coronary arteries (atherosclerosis). {ECO:0000269|PubMed:10090484, ECO:0000269|PubMed:10206683, ECO:0000269|PubMed:10422803, ECO:0000269|PubMed:10447263, ECO:0000269|PubMed:10532689, ECO:0000269|PubMed:10660340, ECO:0000269|PubMed:10882754, ECO:0000269|PubMed:10978268, ECO:0000269|PubMed:10980548, ECO:0000269|PubMed:11298688, ECO:0000269|PubMed:11462246, ECO:0000269|PubMed:1446662, ECO:0000269|PubMed:1464748, ECO:0000269|PubMed:17142622, ECO:0000269|PubMed:17347910, ECO:0000269|PubMed:1867200, ECO:0000269|PubMed:1931802S, ECO:0000269|PubMed:19319977, ECO:0000269|PubMed:22160468, ECO:0000269|PubMed:2318961, ECO:0000269|PubMed:24529145, ECO:0000269|PubMed:2569482, ECO:0000269|PubMed:2726768, ECO:0000269|PubMed:3263645, ECO:0000269|PubMed:3955657, ECO:0000269|PubMed:7502709	EDTA Blood Sample
Kurşun Zehirlenmesi, Yatkınlık, 612740, Otozomal Resesif (ALAD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lead poisoning, susceptibility to, 612740, Autosomal recessive (ALAD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALAD	ALAD, ALADH, PBGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Kurşun Zehirlenmesi, Yatkınlık, 612740, Otozomal Resesif (MLPA)	Lead poisoning, susceptibility to, 612740, Autosomal recessive (MLPA)	ALAD	ALAD, ALADH, PBGS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Zayıflık, Kalıtsal (AGRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leanness, Inherited (AGRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGRP	AGRP, ART, AGRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Leber konjenital amarozu (LCA) paneli (Miks 1) (AIPL1 17p13, CRB1 1q31, CRX 19q13, RPE65 1q31) (MLPA)	Leber congenital amaurosis (LCA) panel (Mix 1) (AIPL1 17p13, CRB1 1q31, CRX 19q13, RPE65 1q31) (MLPA)	AIPL1 17p13, CRB1 1q31, CRX 19q13, RPE65 1q31	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Leber'in Konjenital Amarozu 1, 204000, Otozomal Resesif; LCA1 (Leber Konjenital Amarozis) (GUCY2D gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 1, 204000, Autosomal recessive; LCA1 (Leber congenital amaurosis) (GUCY2D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCY2D	GUCY2D, GUC2D, LCA1, CORD6, RCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 6 (CORD6) [MIM:601777]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269|PubMed:12525567, ECO:0000269|PubMed:15111605, ECO:0000269|PubMed:18332321, ECO:0000269|PubMed:18487367, ECO:0000269|PubMed:20517349, ECO:0000269|PubMed:21552474, ECO:0000269|PubMed:22194653, ECO:0000269|PubMed:23734073, ECO:0000269|PubMed:24480840, ECO:0000269|PubMed:9618177, ECO:0000269|PubMed:9683616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 10, 611755; LCA10 (Leber Konjenital Amarozis) (CEP290 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 10, 611755; LCA10 (Leber congenital amaurosis) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 11, 613837; LCA11 (Leber Konjenital Amarozis) (IMPDH1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 11, 613837; LCA11 (Leber congenital amaurosis) (IMPDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPDH1	IMPDH1, RP10, LCA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 10 (RP10) [MIM:180105]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:11875049, ECO:0000269|PubMed:11875050, ECO:0000269|PubMed:16384941}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 12, 610612, Otozomal Resesif; LCA12 (Leber Konjenital Amarozis) (RD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 12, 610612, Autosomal recessive; LCA12 (Leber congenital amaurosis) (RD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RD3	RD3, LCA12, C1orf36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Leber'in Konjenital Amarozu 13, 612712, Otozomal Resesif; LCA13 (Leber Konjenital Amarozis) (RDH12 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 13, 612712, Autosomal recessive; LCA13 (Leber congenital amaurosis) (RDH12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RDH12	RDH12, LCA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 53 (RP53) [MIM:612712]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:19140180, ECO:0000269|PubMed:19956407}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 14, 613341, Otozomal Resesif; LCA14 (Leber Konjenital Amarozis) (LRAT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 14, 613341, Autosomal recessive; LCA14 (Leber congenital amaurosis) (LRAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRAT	LRAT, LCA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 14 (LCA14) [MIM:613341]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:11381255, ECO:0000269|PubMed:17011878, ECO:0000269|PubMed:18055821}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 15, 613843, Otozomal Resesif; LCA15 (Leber Konjenital Amarozis) (TULP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 15, 613843, Autosomal recessive; LCA15 (Leber congenital amaurosis) (TULP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TULP1	TULP1, RP14, LCA15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 15 (LCA15) [MIM:613843]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:17962469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 16, 614186, Otozomal Resesif; LCA16 (Leber Konjenital Amarozis) (KCNJ13 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 16, 614186, Autosomal recessive; LCA16 (Leber congenital amaurosis) (KCNJ13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ13	KCNJ13, SVD, LCA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Snowflake vitreoretinal degeneration (SVD) [MIM:193230]: Developmental and progressive hereditary eye disorder that affects multiple tissues within the eye. Diagnostic features of SVD include fibrillar degeneration of the vitreous humor, early-onset cataract, minute crystalline deposits in the neurosensory retina, and retinal detachment. {ECO:0000269	PubMed:18179896}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 17, 615360, Otozomal Resesif; LCA17 (Leber Konjenital Amarozis) (GDF6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 17, 615360, Autosomal recessive; LCA17 (Leber congenital amaurosis) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. {ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19120173}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 18, 608133, Otozomal Resesif, Otozomal Dominant (Retinitis Pigmentoza) (PRPH2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 18, 608133, Autosomal recessive, Autosomal dominant (Retinitis pigmentosa) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 2, 204100, Otozomal Resesif; LCA2 (Leber Konjenital Amarozis) (RPE65 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 2, 204100, Autosomal recessive; LCA2 (Leber congenital amaurosis) (RPE65 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPE65	RPE65, RP20, LCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 2 (LCA2) [MIM:204100]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:10090910, ECO:0000269|PubMed:10766140, ECO:0000269|PubMed:11462243, ECO:0000269|PubMed:14611946, ECO:0000269|PubMed:14962443, ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:16205573, ECO:0000269|PubMed:17297704, ECO:0000269|PubMed:17724218, ECO:0000269|PubMed:18682808, ECO:0000269|PubMed:9326941, ECO:0000269|PubMed:9801879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 2, 204100, Otozomal Resesif; LCA2 (Leber Konjenital Amarozis) (MLPA)	Leber congenital amaurosis 2, 204100, Autosomal recessive; LCA2 (Leber congenital amaurosis) (MLPA)	RPE65	RPE65, RP20, LCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leber congenital amaurosis 2 (LCA2) [MIM:204100]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:10090910, ECO:0000269|PubMed:10766140, ECO:0000269|PubMed:11462243, ECO:0000269|PubMed:14611946, ECO:0000269|PubMed:14962443, ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:16205573, ECO:0000269|PubMed:17297704, ECO:0000269|PubMed:17724218, ECO:0000269|PubMed:18682808, ECO:0000269|PubMed:9326941, ECO:0000269|PubMed:9801879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 3, 604232; LCA3 (Leber Konjenital Amarozis) (SPATA7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 3, 604232; LCA3 (Leber congenital amaurosis) (SPATA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPATA7	SPATA7, HSD3, LCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa autosomal recessive (ARRP) [MIM:268000]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:19268277}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 4, 604393, Otozomal Resesif; LCA4 (Leber Konjenital Amarozis) (AIPL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 4, 604393, Autosomal recessive; LCA4 (Leber konjenital amarozis) (AIPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIPL1	AIPL1, LCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leber'in Konjenital Amarozu 4, 604393, Otozomal Resesif; LCA4 (Leber Konjenital Amarozis) (MLPA)	Leber congenital amaurosis 4, 604393, Autosomal recessive; LCA4 (Leber konjenital amarozis) (MLPA)	AIPL1	AIPL1, LCA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Leber'in Konjenital Amarozu 5, 604537; LCA5 (Leber Konjenital Amaurozis) (LCA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 5, 604537; LCA5 (Leber congenital amaurosis) (LCA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LCA5	LCA5, C6orf152	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 5 (LCA5) [MIM:604537]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:17546029, ECO:0000269|PubMed:18000884, ECO:0000269|PubMed:18334959}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 6, 613826; LCA6 (Leber Konjenital Amaurozis) (RPGRIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 6, 613826; LCA6 (Leber congenital amaurosis) (RPGRIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGRIP1	RPGRIP1, LCA6, CORD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 13 (CORD13) [MIM:608194]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269|PubMed:10958648, ECO:0000269|PubMed:12920076}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 7, 613829; LCA7 (Leber Konjenital Amaurozis) (CRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 7, 613829; LCA7 (Leber congenital amaurosis) (CRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRX	CRX, CORD2, CRD, LCA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 2 (CORD2) [MIM:120970]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269|PubMed:9390563, ECO:0000269|PubMed:9427255, ECO:0000269|PubMed:9792858}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 7, 613829; LCA7 (Leber Konjenital Amaurozis) (MLPA)	Leber congenital amaurosis 7, 613829; LCA7 (Leber congenital amaurosis) (MLPA)	CRX	CRX, CORD2, CRD, LCA7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cone-rod dystrophy 2 (CORD2) [MIM:120970]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269|PubMed:9390563, ECO:0000269|PubMed:9427255, ECO:0000269|PubMed:9792858}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leber'in Konjenital Amarozu 8, 613835; LCA8 (Leber Konjenital Amaurozis) (CRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 8, 613835; LCA8 (Leber congenital amaurosis) (CRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRB1	CRB1, RP12, LCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269|PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 8, 613835; LCA8 (Leber Konjenital Amaurozis) (MLPA)	Leber congenital amaurosis 8, 613835; LCA8 (Leber congenital amaurosis) (MLPA)	CRB1	CRB1, RP12, LCA8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269|PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leber'in Konjenital Amarozu 9, 608553, Otozomal Resesif; LCA9 (Leber Konjenital Amaurozis) (NMNAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leber congenital amaurosis 9, 608553, Autosomal recessive; LCA9 (Leber congenital amaurosis) (NMNAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NMNAT1	NMNAT1, NMNAT, PNAT1, LCA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lesitin: Kolesterol Açiltransferaz Eksikliği - Norum Hastalığı, 245900, Otozomal Resesif (LCAT Eksikliği) (LCAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY - Norum disease, 245900, Autosomal recessive (LCAT deficiency) (LCAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LCAT	LCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fish-eye disease (FED) [MIM:136120]: A disorder of lipoprotein metabolism due to partial lecithin-cholesterol acyltransferase deficiency that affects only alpha-LCAT activity. FED is characterized by low plasma HDL and corneal opacities due to accumulation of cholesterol deposits in the cornea ('fish-eye'). {ECO:0000269|PubMed:1516702, ECO:0000269|PubMed:1571050, ECO:0000269|PubMed:15994445, ECO:0000269|PubMed:1737840, ECO:0000269|PubMed:21901787, ECO:0000269|PubMed:8620346, ECO:0000269|PubMed:9261271}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lesitin: Kolesterol Açiltransferaz Eksikliği - Norum Hastalığı, 245900, Otozomal Resesif (LCAT Eksikliği) (Prenatal) (LCAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	LECITHIN:CHOLESTEROL ACYLTRANSFERASE DEFICIENCY - Norum disease, 245900, Autosomal recessive (LCAT deficiency) (LCAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LCAT	LCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fish-eye disease (FED) [MIM:136120]: A disorder of lipoprotein metabolism due to partial lecithin-cholesterol acyltransferase deficiency that affects only alpha-LCAT activity. FED is characterized by low plasma HDL and corneal opacities due to accumulation of cholesterol deposits in the cornea ('fish-eye'). {ECO:0000269|PubMed:1516702, ECO:0000269|PubMed:1571050, ECO:0000269|PubMed:15994445, ECO:0000269|PubMed:1737840, ECO:0000269|PubMed:21901787, ECO:0000269|PubMed:8620346, ECO:0000269|PubMed:9261271}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sol Ventriküler Noncompaction 1, ± Konjenital Kalp Kusurları, 604169, Otozomal Dominant; LVNC1 (Sol Ventriküler Noncompaction) (DTNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 1, with or without congenital heart defects, 604169, Autosomal dominant; LVNC1 (Left ventricular noncompaction) (DTNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DTNA	DTNA, D18S892E, DRP3, LVNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 1 (LVNC1) [MIM:604169]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies such as ventricular septal defects, pulmonic stenosis and atrial septal defects. The right ventricle may also be affected. {ECO:0000269|PubMed:11238270}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sol Ventriküler Noncompaction 10, 615396, Otozomal Dominant; LVNC10 (Sol Ventriküler Noncompaction) (MYBPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 10, 615396, Autosomal dominant; LVNC10 (Left ventricular noncompaction) (MYBPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYBPC3	MYBPC3, CMH4, CMD1MM, LVNC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 4 (CMH4) [MIM:115197]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:11499718, ECO:0000269|PubMed:11499719, ECO:0000269|PubMed:11815426, ECO:0000269|PubMed:12379228, ECO:0000269|PubMed:12628722, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:12818575, ECO:0000269|PubMed:12951062, ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:14563344, ECO:0000269|PubMed:15114369, ECO:0000269|PubMed:15519027, ECO:0000269|PubMed:15563892, ECO:0000269|PubMed:16004897, ECO:0000269|PubMed:16199542, ECO:0000269|PubMed:18403758, ECO:0000269|PubMed:18929575, ECO:0000269|PubMed:18957093, ECO:0000269|PubMed:23840593, ECO:0000269|PubMed:7744002, ECO:0000269|PubMed:9048664, ECO:0000269|PubMed:9541104, ECO:0000269|PubMed:9541115, ECO:0000269|PubMed:9562578}. Note=The disease is caused	EDTA Blood Sample
Sol Ventriküler Noncompaction 3, 601493, Otozomal Dominant (Sol Ventriküler Noncompaction) (LDB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 3, 601493, Autosomal dominant (Left ventricular noncompaction) (LDB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDB3	LDB3, ZASP, CYPHER, KIAA01613, MFM4, CMD1C, CMH24, LVNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 3 (LVNC3) [MIM:601493]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sol Ventriküler Noncompaction 4, 613424, Otozomal Dominant (Ailesel İzole Dilate Kardiyomiyopati) (ACTC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 4, 613424, Autosomal dominant (Familial isolated dilated cardiomyopathy) (ACTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTC1	ACTC1, CMD1R, CMH11, ASD5, LVNC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 11 (CMH11) [MIM:612098]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:10330430, ECO:0000269|PubMed:10966831, ECO:0000269|PubMed:14729850, ECO:0000269|PubMed:18403758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sol Ventriküler Noncompaction 5, 613426, Otozomal Dominant (Sol Ventriküler Noncompaction) (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 5, 613426, Autosomal dominant (Left ventricular noncompaction) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Sol Ventrikiler Noncompaction 6, 601494, Otozomal Dominant (Sol Ventrikiler Noncompaction) (TNNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 6, 601494, Autosomal dominant (Left ventricular noncompaction) (TNNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT2	TNNT2, CMH2, CMD1D, RCM3, LVNC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 2 (CMH2) [MIM:115195]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:10525521, ECO:0000269|PubMed:11034944, ECO:0000269|PubMed:12707239, ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:15563892, ECO:0000269|PubMed:16199542, ECO:0000269|PubMed:21846512, ECO:0000269|PubMed:7898523, ECO:0000269|PubMed:8205619, ECO:0000269|PubMed:8981109, ECO:0000269|PubMed:9060892, ECO:0000269|PubMed:9140840, ECO:0000269|PubMed:9482583, ECO:0000269|Ref.19}. Note=The disease is caused by mutations affecting the gene conversented in this entry.	EDTA Blood Sample
Sol Ventrikiler Noncompaction 7, 615092, Otozomal Dominant; LVNC7 (Sol Ventrikiler Noncompaction) (MIB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 7, 615092, Autosomal dominant; LVNC7 (Left ventricular noncompaction) (MIB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIB1	MIB1, MIB, DIP1, KIAA1323, LVNC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 7 (LVNC7) [MIM:615092]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. {ECO:0000269|PubMed:23314057}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sol Ventrikiler Noncompaction 8, 615373, Otozomal Dominant; LVNC8 (Sol Ventrikiler Noncompaction) (PRDM16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 8, 615373, Autosomal dominant; LVNC8 (Left ventricular noncompaction) (PRDM16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM16	PRDM16, MEL1, LVNC8, CMD1LL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1LL (CMD1LL) [MIM:615373]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:23768516}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sol Ventrikiler Noncompaction 9, 611878, Otozomal Dominant (Sol Ventrikiler Noncompaction) (TPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left ventricular noncompaction 9, 611878, Autosomal dominant (Left ventricular noncompaction) (TPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM1	TPM1, CMH3, CMD1Y , LVNC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 3 (CMH3) [MIM:115196]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:12974739, ECO:0000269|PubMed:7898523, ECO:0000269|PubMed:8205619, ECO:0000269|PubMed:8523464, ECO:0000269|PubMed:9822100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağ-Sol Eksen Malformasyonlar (LEFTY2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Left-right axis malformations (LEFTY2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEFTY2	EBAF, TGFB4, LEFTY2, LEFTA, LEFTYA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left-right axis malformations (LRAM) [MIM:601877]: The defect includes left pulmonary isomerism, with cardiac anomalies characterized by complete atrioventricular canal defect and hypoplastic left ventricle, and interrupted inferior vena cava. {ECO:0000269|PubMed:10053005}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sağ-Sol Eksen Malformasyonlar (Prenatal) (LEFTY2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Left-right axis malformations (LEFTY2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LEFTY2	EBAF, TGFB4, LEFTY2, LEFTA, LEFTYA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Left-right axis malformations (LRAM) [MIM:601877]: The defect includes left pulmonary isomerism, with cardiac anomalies characterized by complete atrioventricular canal defect and hypoplastic left ventricle, and interrupted inferior vena cava. {ECO:0000269|PubMed:10053005}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Legg-Calve-Perthes Hastalığı, 150600, Otozomal Dominant; LCPD (Legg-Calvé-Perthes Hastalığı) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Legg-Calve-Perthes disease, 150600, Autosomal dominant; LCPD (Legg-Calvé-Perthes disease) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chroric uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Legg-Calve-Perthes Hastalığı, 150600, Otozomal Dominant; LCPD (Legg-Calvé-Perthes Hastalığı) (MLPA)	Legg-Calve-Perthes disease, 150600, Autosomal dominant; LCPD (Legg-Calvé-Perthes disease) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chroric uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lejyoner Hastalığı, Yatkınlık, 608556 (TLR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Legionaire disease, susceptibility to, 608556 (TLR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR5	TLR5, TIL3, SLEB1, MELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 1 (SLEB1) [MIM:601744]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Legius Sendromu, 611431, Otozomal Dominant (Legius Sendromu) (SPRED1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Legius syndrome, 611431, Autosomal dominant (Legius syndrome) (SPRED1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPRED1	SPRED1, NFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurofibromatosis 1-like syndrome (NFLS) [MIM:611431]: A disorder characterized mainly by cafe au lait macules without neurofibromas or other tumor manifestations of neurofibromatosis type 1, axillary freckling, and macrocephaly. Additional clinical manifestations include Noonan-like facial dysmorphism, lipomas, learning disabilities and attention deficit-hyperactivity. {ECO:0000269|PubMed:17704776, ECO:0000269|PubMed:19443465, ECO:0000269|PubMed:20108422, ECO:0000269|PubMed:21089071}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, Sitokrom C Oksidaz Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (COX15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to cytochrome c oxidase deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (COX15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX15	COX15, CEMCOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:15235026, ECO:0000269|PubMed:15863660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, Sitokrom C Oksidaz Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (COX15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to cytochrome c oxidase deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (COX15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX15	COX15, CEMCOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:15235026, ECO:0000269|PubMed:15863660}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu Mitokondriyal Kompleks 1 Eksikliği, 256000, Otozomal Resesif Mitokondriyal Nedeniyle (Leigh Sendromu) (NDUFA12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex 1 deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA12	NDUFA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:21617257}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Leigh Sendromu Mitokondriyal Kompleks 1 Eksikliği, 256000, Otozomal Resesif Mitokondriyal Nedeniyle (Leigh Sendromu) (Prenatal) (NDUFA12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA12	NDUFA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269	PubMed:21617257}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Izole Kompleks 1 Eksikliği) (NDUFS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Isolated complex I deficiency) (NDUFS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS3	NDUFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Izole Kompleks 1 Eksikliği) (Prenatal) (NDUFS3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Isolated complex I deficiency) (NDUFS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS3	NDUFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFS8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS8	NDUFS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269	PubMed:9837812}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFAF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFAF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF6	NDUFAF6, C8orf38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:18614015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFA9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA9	NDUFA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA2	NDUFA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (FOXRED1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (FOXRED1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXRED1	FOXRED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:20818383, ECO:0000269|PubMed:20858599}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFS8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS8	NDUFS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269	PubMed:9837812}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFAF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFAF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF6	NDUFAF6, C8orf38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:18614015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFA9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA9	NDUFA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA2	NDUFA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal Kompleks 1 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (FOXRED1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial complex I deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (FOXRED1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXRED1	FOXRED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:20818383, ECO:0000269|PubMed:20858599}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Mitokondriyal COX4 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (COX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome due to mitochondrial COX4 deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (COX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX10	COX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. {ECO:0000269|PubMed:10767350, ECO:0000269|PubMed:12928484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leigh Sendromu, Mitokondriyal COX4 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (COX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome due to mitochondrial COX4 deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (COX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX10	COX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. {ECO:0000269|PubMed:10767350, ECO:0000269|PubMed:12928484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFS7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS7	NDUFS7, PSST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:10330338}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFS4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS4	NDUFS4, AQDQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:9463323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF2	NDUFAF2, NDUFA12L, MMTN, B17.2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:18200211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (NDUFA10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA10	NDUFA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFS7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS7	NDUFS7, PSST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:10330338}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFS4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFS4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS4	NDUFS4, AQDQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:9463323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFAF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFAF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF2	NDUFAF2, NDUFA12L, MMTN, B17.2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:16200211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (NDUFA10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (NDUFA10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA10	NDUFA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal; LS (Leigh Sendromu) (BCS1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial; LS (Leigh syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 1 (MC3DN1) [MIM:124000]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:11528392, ECO:0000269|PubMed:12910490, ECO:0000269|PubMed:17314340, ECO:0000269|PubMed:17403714, ECO:0000269|PubMed:19162478, ECO:0000269|PubMed:22991165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leigh Sendromu, 256000, Otozomal Resesif, Mitokondriyal; LS (Leigh Sendromu) (Prenatal) (BCS1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, 256000, Autosomal recessive, Mitochondrial; LS (Leigh syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 1 (MC3DN1) [MIM:124000]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:11528392, ECO:0000269|PubMed:12910490, ECO:0000269|PubMed:17314340, ECO:0000269|PubMed:17403714, ECO:0000269|PubMed:19162478, ECO:0000269|PubMed:22991165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, COX 4 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (SURF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, due to COX IV deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (SURF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SURF1	SURF1, CMT4K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:10647889, ECO:0000269|PubMed:10746561, ECO:0000269|PubMed:14564068, ECO:0000269|PubMed:21937992, ECO:0000269|PubMed:22410471, ECO:0000269|PubMed:22488715, ECO:0000269|PubMed:9843204}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leigh Sendromu, COX 4 Eksikliğine Bağlı, 256000, Otozomal Resesif, Mitokondriyal (Leigh Sendromu) (Prenatal) (SURF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, due to COX IV deficiency, 256000, Autosomal recessive, Mitochondrial (Leigh syndrome) (SURF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SURF1	SURF1, CMT4K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. {ECO:0000269|PubMed:10647889, ECO:0000269|PubMed:10746561, ECO:0000269|PubMed:14564068, ECO:0000269|PubMed:21937992, ECO:0000269|PubMed:22410471, ECO:0000269|PubMed:22488715, ECO:0000269|PubMed:9843204}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leigh Sendromu, Fransız-Kanada Tipi, 220111, Otozomal Resesif; LSFC (Konjenital Laktik Asidoz, Saguenay-Lac-Saint-Jean Tipi) (LRPPRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leigh syndrome, French-Canadian type, 220111, Autosomal recessive; LSFC (Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type) (LRPPRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRPPRC	LRPPRC, LRP130, LSFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leigh Sendromu, Fransız-Kanada Tipi, 220111, Otozomal Resesif; LSFC (Konjenital Laktik Asidoz, Saguenay-Lac-Saint-Jean Tipi) (Prenatal) (LRPPRC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leigh syndrome, French-Canadian type, 220111, Autosomal recessive; LSFC (Congenital lactic acidosis, Saguenay-Lac-Saint-Jean type) (LRPPRC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRPPRC	LRPPRC, LRP130, LSFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leiomyoma, Uterin, Somatik, 150699 (HMGA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leiomyoma, uterine, somatic, 150699 (HMGA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGA2	HMGA2, HMGIC, BABL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving HMGA2 is associated with a subclass of benign mesenchymal tumors known as lipomas. Translocation t(3;12)(q27-q28;q13-q15) with LPP is shown in lipomas. HMGA2 is also fused with a number of other genes in lipomas. {ECO:0000269	PubMed:8824803}.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Leiomiyomatozis ve Renal Hücre Kanseri, 150800, Otozomal Dominant; HLRCC (Kalıtsal Leyomiyomatozis ve Renal Hücreli Kanseri) (FH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leiomyomatosis and renal cell cancer, 150800, Autosomal dominant; HLRCC (Hereditary leiomyomatosis and renal cell cancer) (FH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FH	FH, HLRCC, MCUL1, FMRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary leiomyomatosis and renal cell cancer (HLRCC) [MIM:150800]: A disorder characterized by predisposition to cutaneous and uterine leiomyomas, and papillary type 2 renal cancer which occurs in about 20% of patients. {ECO:0000269	PubMed:11865300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenz-Majewski Hiperostotik Cücelik, 151050, Otozomal Dominant; LMHD (Lenz-Majewski Hiperostotik Dwarfizm) (PTDSS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lenz-Majewski hyperostotic dwarfism, 151050, Autosomal dominant; LMHD (Lenz-Majewski hyperostotic dwarfism) (PTDSS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTDSS1	PTDSS1, PSS1, KIAA0024, LMHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lenz-Majewski hyperostotic dwarfism (LMHD) [MIM:151050]: A syndrome of intellectual disability and multiple congenital anomalies that features generalized craniotubular hyperostosis. LMHD is characterized by the combination of sclerosing bone dysplasia, intellectual disability and distinct craniofacial, dental, cutaneous and distal limb anomalies. The progressive generalized hyperostosis associated with this syndrome affects the cranium, the vertebrae and the diaphyses of tubular bones, leading to severe growth restriction. {ECO:0000269	PubMed:24241535}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenz-Majewski Hiperostotik Cücelik, 151050, Otozomal Dominant; LMHD (Lenz-Majewski Hiperostotik Dwarfizm) (Prenatal) (PTDSS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lenz-Majewski hyperostotic dwarfism, 151050, Autosomal dominant; LMHD (Lenz-Majewski hyperostotic dwarfism) (PTDSS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTDSS1	PTDSS1, PSS1, KIAA0024, LMHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lenz-Majewski hyperostotic dwarfism (LMHD) [MIM:151050]: A syndrome of intellectual disability and multiple congenital anomalies that features generalized craniotubular hyperostosis. LMHD is characterized by the combination of sclerosing bone dysplasia, intellectual disability and distinct craniofacial, dental, cutaneous and distal limb anomalies. The progressive generalized hyperostosis associated with this syndrome affects the cranium, the vertebrae and the diaphyses of tubular bones, leading to severe growth restriction. {ECO:0000269	PubMed:24241535}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LEOPARD Sendromu 1, 151100, Otozomal Dominant; LPRD1 (Noonan Sendromu-Multipl Lentijinli) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LEOPARD syndrome 1, 151100, Autosomal dominant; LPRD1 (Noonan syndrome with multiple lentigines) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. {ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

LEOPARD Sendromu 1, 151100, Otozomal Dominant; LPRD1 (Noonan Sendromu-Multipl Lentijinli) (Prenatal) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LEOPARD syndrome 1, 151100, Autosomal dominant; LPRD1 (Noonan syndrome with multiple lentigines) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. (ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LEOPARD Sendromu 2, 611554; LPRD2 (Noonan Sendromu- Multipl Lentijinli) (RAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LEOPARD syndrome 2, 611554; LPRD2 (Noonan syndrome with multiple lentigines) (RAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAF1	RAF1, CRAF, NS5, CMD1NN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome 5 (NS5) [MIM:611553]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyeslant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. (ECO:0000269|PubMed:17603482, ECO:0000269|PubMed:17603483, ECO:0000269|PubMed:20683980). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LEOPARD Sendromu 2, 611554; LPRD2 (Noonan Sendromu- Multipl Lentijinli) (Prenatal) (RAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LEOPARD syndrome 2, 611554; LPRD2 (Noonan syndrome with multiple lentigines) (RAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAF1	RAF1, CRAF, NS5, CMD1NN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Noonan syndrome 5 (NS5) [MIM:611553]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyeslant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. (ECO:0000269|PubMed:17603482, ECO:0000269|PubMed:17603483, ECO:0000269|PubMed:20683980). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
LEOPARD Sendromu 3, 613707, Otozomal Dominant; LPRD3 (Noonan Sendromu-Multipl Lentijinli) (BRAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	LEOPARD syndrome 3, 613707, Autosomal dominant; LPRD3 (Noonan syndrome with multiple lentigines) (BRAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. (ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
LEOPARD Sendromu 3, 613707, Otozomal Dominant; LPRD3 (Noonan Sendromu-Multipl Lentijinli) (Prenatal) (BRAF geni) (Tüm Gen Dizi Analizi) (Prenatal)	LEOPARD syndrome 3, 613707, Autosomal dominant; LPRD3 (Noonan syndrome with multiple lentigines) (BRAF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. (ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leprechaunism, 246200, Otozomal Resesif (Leprechaunizm) (Donohue Sendromu) (INSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprechaunism, 246200, Autosomal recessive (Leprechaunism) (DONOHUE SYNDROME) (INSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INSR	INSR, HHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269|PubMed:1470163, ECO:0000269|PubMed:1607076, ECO:0000269|PubMed:7657032). Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Leprechaunism, 246200, Otozomal Resesif (Leprechaunizm) (Donohue Sendromu) (Prenatal) (INSR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leprechaunism, 246200, Autosomal recessive (Leprechaunism) (DONOHUE SYNDROME) (INSR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INSR	INSR, HHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269|PubMed:1470163, ECO:0000269|PubMed:1607076, ECO:0000269|PubMed:7657032). Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lepra, Karşı Koruma, 613223 (Lepra) (TLR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprosy, protection against, 613223 (Leprosy) (TLR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR1	TLR1, TIL, LPRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lepra Yatkınlık, 246300, Otozomal Dominant; LPRS3 (Lepra) (TLR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprosy, susceptibility to, 246300, Autosomal dominant; LPRS3 (Leprosy) (TLR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR2	TLR2, TIL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Lepra, Yatkınlık, 4, 610988; LPRS4 (Lepra) (LTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprosy, susceptibility to, 4, 610988; LPRS4 (Leprosy) (LTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTA	LTA, TNFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet; arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). (ECO:0000269	PubMed:12746914). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Lepra Yatkınlık, 5, 613223; LPRS5 (Lepra) (TLR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprosy, susceptibility to, 5, 613223; LPRS5 (Leprosy) (TLR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR1	TLR1, TIL, LPRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lepra, Yatkınlık, 607572; LPRS2 (Lepra) (PRKN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leprosy, susceptibility to, 607572; LPRS2 (Leprosy) (PRKN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKN	PRKN, PARK2, PDJ, LPRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Leptin Eksikliği Veya Disfonksiyonu; Lepd (Obezite, Konjenital Leptin Eksikliğine Bağlı) (LEP geni) (Tüm Gen Dizi Analizi) (Postnatal)	LEPTIN DEFICIENCY OR DYSFUNCTION; LEPD (Obesity due to congenital leptin deficiency) (LEP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEP	LEP, OB, LEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leptin deficiency (LEPD) [MIM:614962]: A rare disease characterized by low levels of serum leptin, severe hyperphagia and intractable obesity from an early age. (ECO:0000269	PubMed:9500540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leptin Eksikliği Veya Disfonksiyonu; Lepd (Obezite, Konjenital Leptin Eksikliğine Bağlı) (MLPA)	LEPTIN DEFICIENCY OR DYSFUNCTION; LEPD (Obesity due to congenital leptin deficiency) (MLPA)	LEP	LEP, OB, LEPD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leptin deficiency (LEPD) [MIM:614962]: A rare disease characterized by low levels of serum leptin, severe hyperphagia and intractable obesity from an early age. (ECO:0000269	PubMed:9500540). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leri Pleonosteoz Kromozom 8q22.1 Duplikasyon Sendromu, 151200, Otozomal Dominant (Leri Pleonosteozis) (2200)	Leri pleonosteosis chromosome 8q22.1 duplication syndrome, 151200, Autosomal dominant (Leri pleonosteosis) (440)	.	DUP8q22.1, C8DUPq22.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Leri-Weill Diskondrosteozis, 127300, Otozomal Dominant; LWD (Leri-Weill Diskondrosteoz) (SHOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leri-Weill dyschondrosteosis, 127300, Autosomal dominant; LWD (Leri-Weill dyschondrosteosis) (SHOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHOX	SHOX, GCFX, SS, PHOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. (ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Leri-Weill Diskondrosteozis, 127300, Otozomal Dominant; LWD (Leri-Weill Diskondrosteoz) (MLPA)	Leri-Weill dyschondrosteosis, 127300, Autosomal dominant; LWD (Léri-Weill dyschondrosteosis) (MLPA)	SHOX	SHOX, GCFX, SS, PHOG	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. (ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Lesch-Nyhan Sendromu, 300322, X'e Bağlı Resesif; LNS (Lesch-Nyhan Sendromu) (HPRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lesch-Nyhan syndrome, 300322, X-linked recessive; LNS (Lesch-Nyhan syndrome) (HPRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPRT1	HPRT1, HPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lesch-Nyhan syndrome (LNS) [MIM:300322]: Characterized by complete lack of enzymatic activity that results in hyperuricemia, choreoathetosis, mental retardation, and compulsive self- mutilation. {ECO:0000269|PubMed:15571223, ECO:0000269|PubMed:17027311, ECO:0000269|PubMed:2071157, ECO:0000269|PubMed:2246854, ECO:0000269|PubMed:2347587, ECO:0000269|PubMed:2358296, ECO:0000269|PubMed:24904672, ECO:0000269|PubMed:2910902, ECO:0000269|PubMed:3265398, ECO:0000269|PubMed:3384338, ECO:0000269|PubMed:6853716, ECO:0000269|PubMed:7627191, ECO:0000269|PubMed:9452051}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lesch-Nyhan Sendromu, 300322, X'e Bağlı Resesif; LNS (Lesch-Nyhan Sendromu) (MLPA)	Lesch-Nyhan syndrome, 300322, X-linked recessive; LNS (Lesch-Nyhan syndrome) (MLPA)	HPRT1	HPRT1, HPRT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lesch-Nyhan syndrome (LNS) [MIM:300322]: Characterized by complete lack of enzymatic activity that results in hyperuricemia, choreoathetosis, mental retardation, and compulsive self- mutilation. {ECO:0000269|PubMed:15571223, ECO:0000269|PubMed:17027311, ECO:0000269|PubMed:2071157, ECO:0000269|PubMed:2246854, ECO:0000269|PubMed:2347587, ECO:0000269|PubMed:2358296, ECO:0000269|PubMed:24904672, ECO:0000269|PubMed:2910902, ECO:0000269|PubMed:3265398, ECO:0000269|PubMed:3384338, ECO:0000269|PubMed:6853716, ECO:0000269|PubMed:7627191, ECO:0000269|PubMed:9452051}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lesch-Nyhan Sendromu, 300322, X'e Bağlı Resesif; LNS (Lesch-Nyhan Sendromu) (Prenatal) (HPRT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lesch-Nyhan syndrome, 300322, X-linked recessive; LNS (Lesch-Nyhan syndrome) (HPRT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPRT1	HPRT1, HPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lesch-Nyhan syndrome (LNS) [MIM:300322]: Characterized by complete lack of enzymatic activity that results in hyperuricemia, choreoathetosis, mental retardation, and compulsive self- mutilation. {ECO:0000269|PubMed:15571223, ECO:0000269|PubMed:17027311, ECO:0000269|PubMed:2071157, ECO:0000269|PubMed:2246854, ECO:0000269|PubMed:2347587, ECO:0000269|PubMed:2358296, ECO:0000269|PubMed:24904672, ECO:0000269|PubMed:2910902, ECO:0000269|PubMed:3265398, ECO:0000269|PubMed:3384338, ECO:0000269|PubMed:6853716, ECO:0000269|PubMed:7627191, ECO:0000269|PubMed:9452051}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lesch-Nyhan Sendromu, 300322, X'e Bağlı Resesif; LNS (Lesch-Nyhan Sendromu) (Prenatal) (MLPA)	Lesch-Nyhan syndrome, 300322, X-linked recessive; LNS (Lesch-Nyhan syndrome) (Prenatal) (MLPA)	HPRT1	HPRT1, HPRT	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lesch-Nyhan syndrome (LNS) [MIM:300322]: Characterized by complete lack of enzymatic activity that results in hyperuricemia, choreoathetosis, mental retardation, and compulsive self- mutilation. {ECO:0000269|PubMed:15571223, ECO:0000269|PubMed:17027311, ECO:0000269|PubMed:2071157, ECO:0000269|PubMed:2246854, ECO:0000269|PubMed:2347587, ECO:0000269|PubMed:2358296, ECO:0000269|PubMed:24904672, ECO:0000269|PubMed:2910902, ECO:0000269|PubMed:3265398, ECO:0000269|PubMed:3384338, ECO:0000269|PubMed:6853716, ECO:0000269|PubMed:7627191, ECO:0000269|PubMed:9452051}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 2, 607598, Otozomal Resesif; LCCS2 (Letal Konjenital Kontraktür Sendromu Tip 2) (ERBB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contractural syndrome 2, 607598, Autosomal recessive; LCCS2 (Lethal congenital contracture syndrome type 2) (ERBB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERBB3	ERBB3, LCCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal congenital contracture syndrome 2 (LCCS2) [MIM:607598]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS2 patients manifest craniofacial/ocular findings, lack of hydrops, multiple pterygia, and fractures, as well as a normal duration of pregnancy and a unique feature of a markedly distended urinary bladder (neurogenic bladder defect). The phenotype suggests a spinal cord neuropathic etiology. {ECO:0000269	PubMed:17701904}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 2, 607598, Otozomal Resesif; LCCS2 (Letal Konjenital Kontraktür Sendromu Tip 2) (Prenatal) (ERBB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contractural syndrome 2, 607598, Autosomal recessive; LCCS2 (Lethal congenital contracture syndrome type 2) (ERBB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERBB3	ERBB3, LCCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal congenital contracture syndrome 2 (LCCS2) [MIM:607598]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS2 patients manifest craniofacial/ocular findings, lack of hydrops, multiple pterygia, and fractures, as well as a normal duration of pregnancy and a unique feature of a markedly distended urinary bladder (neurogenic bladder defect). The phenotype suggests a spinal cord neuropathic etiology. {ECO:0000269	PubMed:17701904}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 3, 611369, Otozomal Resesif; LCCS3 (Letal Konjenital Kontraktürü Sendromu Tip 3) (PIP5K1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contractural syndrome 3, 611369, Autosomal recessive; LCCS3 (Lethal congenital contracture syndrome type 3) (PIP5K1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIP5K1C	PIP5K1C, LCCS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal congenital contracture syndrome 3 (LCCS3) [MIM:611369]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS3 patients present at birth with severe multiple joint contractures and severe muscle wasting and atrophy, mainly in the legs. Death occurs minutes to hours after birth due to respiratory insufficiency. The phenotype can be distinguished from that of LCCS1 by the absence of hydrops, fractures and multiple pterygia, and from LCCS2 by the absence of neurogenic bladder defect. {ECO:0000269	PubMed:17701898}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 3, 611369, Otozomal Resesif; LCCS3 (Letal Konjenital Kontraktürü Sendromu Tip 3) (Prenatal) (PIP5K1C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contractural syndrome 3, 611369, Autosomal recessive; LCCS3 (Lethal congenital contracture syndrome type 3) (PIP5K1C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIP5K1C	PIP5K1C, LCCS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal congenital contracture syndrome 3 (LCCS3) [MIM:611369]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS3 patients present at birth with severe multiple joint contractures and severe muscle wasting and atrophy, mainly in the legs. Death occurs minutes to hours after birth due to respiratory insufficiency. The phenotype can be distinguished from that of LCCS1 by the absence of hydrops, fractures and multiple pterygia, and from LCCS2 by the absence of neurogenic bladder defect. {ECO:0000269	PubMed:17701898}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 1, 253310, Otozomal Resesif; LCCS1 (Letal Konjenital Kontraktür Sendromu Tip 1) (GLE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 1, 253310, Autosomal recessive; LCCS1 (Lethal congenital contracture syndrome type 1) (GLE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLE1	GLE1, GLE1L, LCCS, LCCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal congenital contracture syndrome 1 (LCCS1) [MIM:253310]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS1 patients manifest early fetal hydrops and akinesia, micrognathia, pulmonary hypoplasia, pterygia, and multiple joint contractures. It leads to prenatal death. {ECO:0000269	PubMed:18204449}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 1, 253310, Otozomal Resesif; LCCS1 (Letal Konjenital Kontraktür Sendromu Tip 1) (Prenatal) (GLE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 1, 253310, Autosomal recessive; LCCS1 (Lethal congenital contracture syndrome type 1) (GLE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLE1	GLE1, GLE1L, LCCS, LCCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal congenital contracture syndrome 1 (LCCS1) [MIM:253310]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy, and congenital non-progressive joint contractures (arthrogryposis). The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS1 patients manifest early fetal hydrops and akinesia, micrognathia, pulmonary hypoplasia, pterygia, and multiple joint contractures. It leads to prenatal death. {ECO:0000269	PubMed:18204449}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 10, 617022, Otozomal Resesif; LCCS10 (NEK9 ile İlişkili Letal İskelet Displazisi) (NEK9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 10, 617022, Autosomal recessive; LCCS10 (NEK9-related lethal skeletal dysplasia) (NEK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK9	NEK9, NERCC1, LCCS10, APUG, NC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Letal Konjenital Kontraktür Sendromu 10, 617022, Otozomal Resesif; LCCS10 (NEK9 İle İlişkili Letal İskelet Displazisi) (Prenatal) (NEK9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 10, 617022, Autosomal recessive; LCCS10 (NEK9-related lethal skeletal dysplasia) (NEK9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEK9	NEK9, NERCC1, LCCS10, APUG, NC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 11, 617194, Otozomal Resesif; LCCS11 (GLDN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 11, 617194, Autosomal recessive; LCCS11 (GLDN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLDN	GLDN, CRGL2, LCCS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 11, 617194, Otozomal Resesif; LCCS11 (Prenatal) (GLDN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 11, 617194, Autosomal recessive; LCCS11 (GLDN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLDN	GLDN, CRGL2, LCCS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 4, 614915, Otozomal Resesif; LCCS4 (Letal Konjenital Kontraktürü Sendromu Tip 3) (MYBPC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 4, 614915, Autosomal recessive; LCCS4 (Lethal congenital contracture syndrome type 3) (MYBPC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYBPC1	MYBPC1, LCCS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 1B (DA1B) [MIM:614335]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269	PubMed:20045868}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 4, 614915, Otozomal Resesif; LCCS4 (Letal Konjenital Kontraktürü Sendromu Tip 3) (MYBPC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 4, 614915, Autosomal recessive; LCCS4 (Lethal congenital contracture syndrome type 3) (MYBPC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYBPC1	MYBPC1, LCCS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 1B (DA1B) [MIM:614335]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269	PubMed:20045868}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 5, 615368, Otozomal Resesif; LCCS5 (Fetal Akinezi-Beyin ve Retinal Kanama Sendromu) (DNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 5, 615368, Autosomal recessive; LCCS5 (Fetal akinesia-cerebral and retinal hemorrhage syndrome) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, LCCS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, centronuclear, 1 (CNM1) [MIM:160150]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:16227997, ECO:0000269|PubMed:17825552, ECO:0000269|PubMed:17932957, ECO:0000269|PubMed:19120038, ECO:0000269|PubMed:19623537, ECO:0000269|PubMed:19932619, ECO:0000269|PubMed:19932620, ECO:0000269|PubMed:20227276, ECO:0000269|PubMed:22396310}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 5, 615368, Otozomal Resesif; LCCS5 (Fetal Akinezi-Beyin ve Retinal Kanama Sendromu) (Prenatal) (DNM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 5, 615368, Autosomal recessive; LCCS5 (Fetal akinesia-cerebral and retinal hemorrhage syndrome) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, LCCS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, centronuclear, 1 (CNM1) [MIM:160150]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:16227997, ECO:0000269|PubMed:17825552, ECO:0000269|PubMed:17932957, ECO:0000269|PubMed:19120038, ECO:0000269|PubMed:19623537, ECO:0000269|PubMed:19932619, ECO:0000269|PubMed:19932620, ECO:0000269|PubMed:20227276, ECO:0000269|PubMed:22396310}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktürpi Sendromu 6, 616248, Otozomal Resesif; LCCS6 (ZBTB42 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 6, 616248, Autosomal recessive; LCCS6 (ZBTB42 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB42	ZBTB42, ZNF925, LCCS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Letal Konjenital Kontraktürpi Sendromu 6, 616248, Otozomal Resesif; LCCS6 (Prenatal) (ZBTB42 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 6, 616248, Autosomal recessive; LCCS6 (ZBTB42 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZBTB42	ZBTB42, ZNF925, LCCS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 7, 616286, Otozomal Resesif; LCCS7 (Hipomiyelinasyon Nöropatisi-Artrogrifozis Sendromu) (CNTNAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 7, 616286, Autosomal recessive; LCCS7 (Hypomyelination neuropathy-arthrogryposis syndrome) (CNTNAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTNAP1	CNTNAP1, CASPR, P190	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 7, 616286, Otozomal Resesif; LCCS7 (Hipomiyelinasyon Nöropatisi-Artrogrifozis Sendromu) (Prenatal) (CNTNAP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 7, 616286, Autosomal recessive; LCCS7 (Hypomyelination neuropathy-arthrogryposis syndrome) (CNTNAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNTNAP1	CNTNAP1, CASPR, P190	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 8, 616287, Otozomal Resesif; LCCS8 (Hipomiyelinasyon Nöropatisi-Artrogrifozis Sendromu) (ADCY6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 8, 616287, Autosomal recessive; LCCS8 (Hypomyelination neuropathy-arthrogryposis syndrome) (ADCY6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADCY6	ADCY6, LCCS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal congenital contracture syndrome 8 (LCCS8) [MIM:616287]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy and congenital non-progressive joint contractures. The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS8 is an anogilal form of arthrogryposis multiplex congenita, characterized by congenital distal joint contractures, reduced fetal movements, and severe motor paralysis leading to death early in the neonatal period. {ECO:0000269	PubMed:24319099}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 8, 616287, Otozomal Resesif; LCCS8 (Hipomiyelinasyon Nöropatisi-Artrogrifozis Sendromu) (Prenatal) (ADCY6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 8, 616287, Autosomal recessive; LCCS8 (Hypomyelination neuropathy-arthrogryposis syndrome) (ADCY6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADCY6	ADCY6, LCCS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal congenital contracture syndrome 8 (LCCS8) [MIM:616287]: A form of lethal congenital contracture syndrome, an autosomal recessive disorder characterized by degeneration of anterior horn neurons, extreme skeletal muscle atrophy and congenital non-progressive joint contractures. The contractures can involve the upper or lower limbs and/or the vertebral column, leading to various degrees of flexion or extension limitations evident at birth. LCCS8 is an anogilal form of arthrogryposis multiplex congenita, characterized by congenital distal joint contractures, reduced fetal movements, and severe motor paralysis leading to death early in the neonatal period. {ECO:0000269	PubMed:24319099}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 9, 616503, Otozomal Resesif; LCCS9 (ADGRG6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lethal congenital contracture syndrome 9, 616503, Autosomal recessive; LCCS9 (ADGRG6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRG6	ADGRG6, PR126, VIGR, LCCS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Letal Konjenital Kontraktür Sendromu 9, 616503, Otozomal Resesif; LCCS9 (Prenatal) (ADGRG6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lethal congenital contracture syndrome 9, 616503, Autosomal recessive; LCCS9 (ADGRG6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADGRG6	ADGRG6, PR126, VIGR, LCCS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lösemi, Akut Lenfoblastik, 613065 (Acute Lymphoblastic Leukemia) (NBN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute lymphoblastic, 613065 (Nijmegen breakage syndrome) (NBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBN	NBN, NBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:14694699}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Lenfoblastik, Somatik, 613065 (Akut Lenfoblastik Lösemi) (GNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute lymphoblastic, somatic, 613065 (Acute lymphoblastic leukemia) (GNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB1	GNB1, MRD42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, Akut Lenfoblastik, Somatik, 613065 (Akut Lenfoblastik Lösemi) (FLT3 geni) (Ekzon14 - 20) (Postnatal)	Leukemia, acute lymphoblastic, somatic, 613065 (Acute lymphoblastic leukemia) (FLT3 gene) (Sequence Analysis) (Postnatal)	FLT3	FLT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:11090077, ECO:0000269|PubMed:11293608, ECO:0000269|PubMed:11442493, ECO:0000269|PubMed:14504097, ECO:0000269|PubMed:16266983, ECO:0000269|PubMed:18305215, ECO:0000269|PubMed:8946935, ECO:0000269|PubMed:9737679}. Note=The gene represented in this entry may be involved in disease pathogenesis. Somatic mutations that lead to constitutive activation of FLT3 are frequent in AML patients. These mutations fall into two classes, the most common being in-frame internal tandem duplications of variable length in the juxtamembrane region that disrupt the normal regulation of the kinase activity. Likewise, point mutations in the activation loop of the kinase domain can result in a constitutively activated kinase.;	EDTA blood sample / EDTA bone marrow

Lösemi, Akut Lenfoblastik, Yatkınlık, 3, 615545; ALL3 (Prekürsör B-Hücreli Akut Lenfoblastik Lösemi) (PAX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute lymphoblastic, susceptibility to, 3, 615545; ALL3 (Precursor B-cell acute lymphoblastic leukemia) (PAX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX5	PAX5, BSAP, ALL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute lymphoblastic, 3 (ALL3) [MIM:613065]: A subtype of acute leukemia, a cancer of the white blood cells. Acute lymphoblastic anemia is a malignant disease of bone marrow and the most common malignancy diagnosed in children. The malignant cells are lymphoid precursor cells (lymphoblasts) that are arrested in an early stage of development. The lymphoblasts replace the normal marrow elements, resulting in a marked decrease in the production of normal blood cells. Consequently, anemia, thrombocytopenia, and neutropenia occur to varying degrees. The lymphoblasts also proliferate in organs other than the marrow, particularly the liver, spleen, and lymphnodes. (ECO:0000269	PubMed:24013638). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Lösemi, Akut Myeloid (NPM1 geni) (Tüm Gen - Dizi Analizi) (NPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid (NPM1 gen) (All coding exons - Sequence analysis) (NPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPM1	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NPM1 is found in a form of non-Hodgkin lymphoma. Translocation t(2;5)(p23;q35) with ALK. The resulting chimeric NPM1-ALK protein homodimerize and the kinase becomes constitutively activated. (ECO:0000269|PubMed:8122112, ECO:0000269|PubMed:8633027).	EDTA Blood Sample/ EDTA Bone Marrow
Lösemi, Akut Myeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (TERT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (TERT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (SH3GL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (SH3GL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH3GL1	SH3GL1, EEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=In some cases of acute leukemia, a translocation results in the formation of a KMT2A/MLL1-EEN fusion gene. (ECO:0000269	PubMed:9122235).;	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (RUNX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (RUNX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUNX1	RUNX1, CBFA2, AML1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial platelet disorder with associated myeloid malignancy (FPDMM) [MIM:601399]: Autosomal dominant disease characterized by qualitative and quantitative platelet defects, and propensity to develop acute myelogenous leukemia. (ECO:0000269	PubMed:10508512). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (NSD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (NSD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSD3	WHSC1L1, NSD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (NSD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (NSD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSD1	NSD1, ARA267, STO, SOTOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. (ECO:0000269	PubMed:14997421). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (MLLT10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (MLLT10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLLT10	AF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MLLT10 is associated with diffuse histiocytic lymphomas. Translocation t(10;11)(p13;q14) with PICALM.;	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (MLF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (MLF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLF1	MLF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MLF1 is a cause of myelodysplastic syndrome (MDS). Translocation t(3;5)(q25.1;q34) with NPM1/NPM. (ECO:0000269	PubMed:8670004).	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (LPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (LPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPP	LPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving LPP is associated with pulmonary chondroid hamartomas. Translocation t(3;12)(q27- q28;q14-q15) with HMGA2 is detected in pulmonary chondroid hamartomas.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269	PubMed:8955068). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (KIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (KIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIT	KIT, PBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Piebald trait (PBT) [MIM:172800]: Autosomal dominant genetic developmental abnormality of pigmentation characterized by congenital patches of white skin and hair that lack melanocytes. (ECO:0000269|PubMed:11074500, ECO:0000269|PubMed:1370874, ECO:0000269|PubMed:1376329, ECO:0000269|PubMed:1717985, ECO:0000269|PubMed:7687257, ECO:0000269|PubMed:8680409, ECO:0000269|PubMed:9450866, ECO:0000269|PubMed:9699740). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (CHIC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (CHIC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHIC2	CHIC2, BTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving CHIC2 is found in a form of acute myeloid leukemia (AML). Translocation t(4;12)(q12;p13) with ETV6. (ECO:0000269	PubMed:10477709).;	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Akut Miyeloid Lösemi) (MLPA)	Leukemia, acute myeloid, 601626, Autosomal dominant (Acute myeloid leukemia) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269	PubMed:8955068). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, 601626, Otozomal Dominant (Sınıflandırılmamış Akut Miyeloid Lösemi) (CEBPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, 601626, Autosomal dominant (Unclassified acute myeloid leukemia) (CEBPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEBPA	CEBPA, CEBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269|PubMed:11242107, ECO:0000269|PubMed:12661007, ECO:0000269|PubMed:15575056). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lösemi, Akut Miyeloid, Hayatta Kalmada Azalma, Somatik, 601626 (Sınıflandırılmamış Akut Miyeloid Lösemi) (FLT3 geni) (Ekzon14 - 20) (Postnatal)	Leukemia, acute myeloid, reduced survival in, somatic, 601626 (Unclassified acute myeloid leukemia) (FLT3 gene) (Sequence Analysis) (Postnatal)	FLT3	FLT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269|PubMed:11090077, ECO:0000269|PubMed:11290608, ECO:0000269|PubMed:11442493, ECO:0000269|PubMed:14504097, ECO:0000269|PubMed:16266983, ECO:0000269|PubMed:18305215, ECO:0000269|PubMed:8946935, ECO:0000269|PubMed:9737679). Note=The gene represented in this entry may be involved in disease pathogenesis. Somatic mutations that lead to constitutive activation of FLT3 are frequent in AML patients. These mutations fall into two classes, the most common being in-frame internal tandem duplications of variable length in the juxtamembrane region that disrupt the normal regulation of the kinase activity. Likewise, point mutations in the activation loop of the kinase domain can result in a constitutively activated kinase.;	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Akut Miyeloid Lösemi-Tekrarlayan Genetik Bozukluk) V617F Mutasyonu ve (Ekzon 12-14) (JAK2 geni) (Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Acute myeloid leukemia with recurrent genetic anomaly) V617F Mutation and (Exon 12-14) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	JAK2, THCYT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Myelofibrosis (MYELOF) [MIM:254450]: A disorder characterized by replacement of the bone marrow by fibrous tissue, occurring in association with a myeloproliferative disorder. Clinical manifestations may include anemia, pallor, splenomegaly, hypermetabolic state, petechiae, ecchymosis, bleeding, lymphadenopathy, hepatomegaly, portal hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Akut Miyeloid Lösemi) (PICALM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Acute myeloid leukemia) (PICALM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PICALM	PICALM, CALM, CLTH, LAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Akut Miyeloid Lösemi) (NPM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Acute myeloid leukemia) (NPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPM1	NPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NPM1 is found in a form of non-Hodgkin lymphoma. Translocation t(2;5)(p23;q35) with ALK. The resulting chimeric NPM1-ALK protein homodimerize and the kinase becomes constitutively activated. (ECO:0000269|PubMed:8122112, ECO:0000269|PubMed:8633027).	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Akut Miyeloid Lösemi) (ETV6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Acute myeloid leukemia) (ETV6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETV6	ETV6, TEL, THC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269	PubMed:15806161). Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Sınıflandırılmamış Akut Miyeloid Lösemi) (NUP214 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Unclassified acute myeloid leukemia) (NUP214 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP214	NUP214, D9S46E, CAN, CAIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NUP214 is found in a subset of acute myeloid leukemia (AML); also known as acute non-lymphocytic leukemia. Translocation t(6;9)(p23;q34) with DEK. It results in the formation of a DEK-CAN fusion gene. (ECO:0000269	PubMed:1549122).	EDTA blood sample / EDTA bone marrow

Lösemi, Akut Miyeloid, Somatik, 601626 (Sınıflandırılmamış Akut Miyeloid Lösemi) (Ekzon14 - 20) (FLT3 geni) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Unclassified acute myeloid leukemia) (FLT3 gene) (Sequence Analysis) (Postnatal)	FLT3	FLT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:11090077, ECO:0000269|PubMed:11290608, ECO:0000269|PubMed:11442493, ECO:0000269|PubMed:14504097, ECO:0000269|PubMed:16266983, ECO:0000269|PubMed:18305215, ECO:0000269|PubMed:8949930, ECO:0000269|PubMed:9737679}. Note=The gene represented in this entry may be involved in disease pathogenesis. Somatic mutations that lead to constitutive activation of FLT3 are frequent in AML patients. These mutations fall into two classes, the most common being in-frame internal tandem duplications of variable length in the juxtamembrane region that disrupt the normal regulation of the kinase activity. Likewise, point mutations in the activation loop of the kinase domain can result in a constitutively activated kinase.;	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Somatik, 601626 (Sınıflandırılmamış Akut Miyeloid Lösemi) (CEBPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, somatic, 601626 (Unclassified acute myeloid leukemia) (CEBPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEBPA	CEBPA, CEBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:11242107, ECO:0000269|PubMed:12681007, ECO:0000269|PubMed:15575056}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Miyeloid, Yatkinlik, 601626, Otozomal Dominant (Sınıflandırılmamış Akut Miyeloid Lösemi) (GATA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, susceptibility to, 601626, Autosomal dominant (Unclassified acute myeloid leukemia) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Miyeloid, Tedavi İle İlişkili (SEPT9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, therapy-related (SEPT9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEPT9	SEPT9, MSF, MSF1, NAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, Akut Miyeloid, Eozinofili (ABL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute myeloid, with eosinophilia (ABL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABL2	ABL2, ABLL, ARG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, Akut Miyeloid, Anti (Akut Miyeloid Lösemi) (GATA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LEUKEMIA, ACUTE MYELOID; AML (Acute myeloid leukemia) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Non-Lenfositik, 125264 (DEK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute nonlymphocytic, 125264 (DEK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DEK	DEK, D6S231E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving DEK is found in a subset of acute myeloid leukemia (AML); also known as acute non- lymphocytic leukemia (PubMed:1549122). Translocation t(6;9)(p23;q34) with NUP214/CAN (PubMed:1549122). It results in the formation of a DEK-NUP214 fusion gene (PubMed:1549122). {ECO:0000269	PubMed:1549122}.	EDTA Blood Sample
Lösemi, Akut Pre-B-Hücresi, 176310 (PBX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute pre-B-cell, 176310 (PBX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PBX1	PBX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, Akut Promiyelositik, 612376; APL (Akut Promiyelositik Lösemi) (RARA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute promyelocytic, 612376; APL (Acute promyelocytic leukemia) (RARA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RARA	RARA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Chromosomal aberrations involving RARA are commonly found in acute promyelocytic leukemia. Translocation t(11;17)(q32;q21) with ZBTB16/PLZF; translocation t(15;17)(q21;q21) with PML; translocation t(5;17)(q32;q11) with NPM. The PML-RARA oncoprotein requires both the PML ring structure and coiled-coil domain for both interaction with UBE2I, nuclear microspecle location and sumoylation. In addition, the coiled- coil domain functions in blocking RA-mediated transactivation and cell differentiation. {ECO:0000269|PubMed:12691149, ECO:0000269|PubMed:8302850, ECO:0000269|PubMed:8562653}.	EDTA Blood Sample
Lösemi, Akut Promiyelositik, PLZF / RARA Tipi (ZBTB16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute promyelocytic, PLZF/RARA type (ZBTB16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB16	ZBTB16, ZNF145, PLZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Skeletal defects, genital hypoplasia, and mental retardation (SGYMR) [MIM:612447]: A disorder characterized by mental retardation, craniofacial dysmorphism, microcephaly and short stature. Additional features include absence of the thumbs, hypoplasia of the radii and ulnae, additional vertebrae and ribs, retarded bone age and genital hypoplasia. {ECO:0000269	PubMed:18611983}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Akut Promiyelositik, PML / RARA Tipi (PML geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute promyelocytic, PML/RARA type (PML gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PML	PML, MYL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving PML may be a cause of acute promyelocytic leukemia (APL). Translocation t(15;17)(q21;q21) with RARA. The PML breakpoints (type A and type B) lie on either side of an alternatively spliced exon. {ECO:0000269|PubMed:1652369, ECO:0000269|PubMed:17205701}.	EDTA Blood Sample
Lösemi, Akut Promiyelositik, Somatik, 102578 (STAT5B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute promyelocytic, somatic, 102578 (STAT5B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT5B	STAT5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Lösemi, Akut Promiyelositik, Somatik, 612376 (Akut Promiyelositik Lösemi) (NUMA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute promyelocytic, somatic, 612376 (Acute promyelocytic leukemia) (NUMA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUMA1	NUMA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Lösemi, Akut T Hücresi, 180385 (LMO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, acute T-cell, 180385 (LMO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMO2	LMO2, RBTNL1, RHOM2, TTG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving LMO2 may be a cause of a form of T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(11;14)(p13;q11) with TCRD.;	EDTA Blood Sample
Lösemi, Jüvenil Miyelomonositik, 607785, Otozomal Dominant, Somatik Mutasyon, JMML (Jüvenil Miyelomonositik Lösemi) (NF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, juvenile myelomonocytic, 607785, Autosomal dominant, Somatic mutation; JMML (Juvenile myelomonocytic leukemia) (NF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Lösemi, Jüvenil Miyelomonositik, 607785, Otozomal Dominant, Somatik Mutasyon; JMML (Jüvenil Miyelomonositik Lösemi) (MLPA)	Leukemia, juvenile myelomonocytic, 607785, Autosomal dominant, Somatic mutation; JMML (Juvenile myelomonocytic leukemia) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		20	60	.	.	30	Postnatal		EDTA blood sample / EDTA bone marrow
Lösemi, Jüvenil Miyelomonositik, Somatik, 607785 (Jüvenil Miyelomonositik Lösemi) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, juvenile myelomonocytic, somatic, 607785 (Juvenile myelomonocytic leukemia) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Lösemi, Jüvenil Miyelomonositik, Somatik, 607785 (Jüvenil Miyelomonositik Lösemi) (ARHGAP26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, juvenile myelomonocytic, somatic, 607785 (Juvenile myelomonocytic leukemia) (ARHGAP26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGAP26	ARHGAP26, GRAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, juvenile myelomonocytic (JMML) [MIM:607785]: An aggressive pediatric myelodysplastic syndrome/myeloproliferative disorder characterized by malignant transformation in the hematopoietic stem cell compartment with proliferation of differentiated progeny. Patients have splenomegaly, enlarged lymph nodes, rashes, and hemorrhages. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal translocation t(5:11)(q31;q23) with KMT2A/MLL1 has been found in leukemic cells from JMML patients, also carrying inactivating mutations on the second allele (PubMed:10908648). {ECO:0000269	PubMed:10908648}.;	EDTA blood sample / EDTA bone marrow
Lösemi, Megakaryoblastik, ± Down Sendromu, 190685 (Trombositopeni-Konjenital Diseritropoietik Anemi) (GATA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, megakaryoblastic, with or without Down syndrome, somatic, 190685 (Thrombocytopenia with congenital dyserythropoietic anemia) (GATA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA1	GATA1, GF1, ERYF1, NFE1, XLTDA, XLTT, XLANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked dyserythropoietic anemia and thrombocytopenia (XDAT) [MIM:300367]: Disorder characterized by erythrocytes with abnormal size and shape, and paucity of platelets in peripheral blood. The bone marrow contains abundant and abnormally small megakaryocytes. {ECO:0000269|PubMed:10700180, ECO:0000269|PubMed:11418466, ECO:0000269|PubMed:11675338, ECO:0000269|PubMed:11809723}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Miyeloid / Lenfoid Veya Karışık-Hücre Dizisi, 159555, Otozomal Dominant (KMT2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, myeloid/lymphoid or mixed-lineage, 159555, Autosomal dominant (KMT2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KMT2A	KMT2A, MLL, HRX, HTRX1, WDSTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Wiedemann-Steiner syndrome (WDSTS) [MIM:605130]: A syndrome characterized by hairy elbows (hypertrichosis cubiti), intellectual disability, a distinctive facial appearance, and short stature. Facial characteristics include long eyelashes, thick or arched eyebrows with a lateral flare, and downslanting and vertically narrow palpebral fissures. {ECO:0000269	PubMed:22795537}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lösemi, Philadelphia Kromozom Pozitif, İmatinib Dirençli (ABL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, Philadelphia chromosome-positive, resistant to imatinib (ABL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABL1	ABL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, chronic myeloid (CML) [MIM:608232]: A clonal myeloproliferative disorder of a pluripotent stem cell with a specific cytogenetic abnormality, the Philadelphia chromosome (Ph), involving myeloid, erythroid, megakaryocytic, B-lymphoid, and sometimes T-lymphoid cells, but not marrow fibroblasts. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Lösemi, Kemoterapi Sonrası, Yatkinlik (NQO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, post-chemotherapy, susceptibility to (NQO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NQO1	NQO1, DIA4, NMOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, T Hücresi Akut Lenfoblastik, 186921 (LMO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, T-cell acute lymphoblastic, 186921 (LMO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMO1	LMO1, RBTN1, RHOM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lösemi, T Hücresi Akut Lenfoblastik, Somatik, 613065 (Akut Lenfoblastik Lösemi) (NUP214 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, T-cell acute lymphoblastic, somatic, 613065 (Acute lymphoblastic leukemia) (NUP214 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP214	NUP214, D9S46E, CAN, CAIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NUP214 is found in a subset of acute myeloid leukemia (AML); also known as acute non-lymphocytic leukemia. Translocation t(6;9)(p23;q34) with DEK. It results in the formation of a DEK-CAN fusion gene. {ECO:0000269	PubMed:15491223}.	EDTA blood sample / EDTA bone marrow
Lösemi, T-Hücresi Akut Lenfoblastoid, 151440 (LYL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, T-cell acute lymphoblastoid, 151440 (LYL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LYL1	LYL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving LYL1 may be a cause of a form of T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(7;19)(q35;q13.3) with TCRB.;	EDTA Blood Sample
Lösemi, T-Hücreli Akut Lenfositik, Somatik, 613065 (Akut Lenfoblastik Lösemi) (TAL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, T-cell acute lymphocytic, somatic, 613065 (Acute lymphoblastic leukemia) (TAL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAL2	TAL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving TAL2 may be a cause of some T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(7;9)(q34;q32) with TCRB. {ECO:0000269	PubMed:17630561}.	EDTA blood sample / EDTA bone marrow

Lösemi, T-Hücreli Akut Lenfositik, Somatik, 613065 (Akut Lenfoblastik Lösemi) (TAL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia, T-cell acute lymphocytic, somatic, 613065 (Acute lymphoblastic leukemia) (TAL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAL1	TAL1, TCL5, SCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving TAL1 may be a cause of some T-cell acute lymphoblastic leukemias (T-ALL). Translocation t(1;14)(p32;q11) with T-cell receptor alpha chain (TCRA) genes. (ECO:0000269	PubMed:2303035).	EDTA blood sample / EDTA bone marrow
Lösemi / Lenfoma, B Hücreli, 2 (BCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia/lymphoma, B-cell, 2 (BCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL2	BCL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving BCL2 has been found in chronic lymphatic leukemia. Translocation t(14;18)(q32;q21) with immunoglobulin gene regions. BCL2 mutations found in non-Hodgkin lymphomas carrying the chromosomal translocation could be attributed to the Ig somatic hypermutation mechanism resulting in nucleotide transitions. (ECO:0000269	PubMed:2875799, ECO:0000269	PubMed:3365001)	EDTA Blood Sample
Lösemi / Lenfoma, B-Hücresi, 3, 109560 (BCL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia/lymphoma, B-cell, 3, 109560 (BCL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL3	BCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving BCL3 may be a cause of B-cell chronic lymphocytic leukemia (B-CLL). Translocation t(14;19)(q32;q13.1) with immunoglobulin gene regions. (ECO:0000269	PubMed:2180580, ECO:0000269	PubMed:7896265).	EDTA Blood Sample
Lösemi / Lenfoma, T-Hücreli, 186960 (TCL1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia/lymphoma, T-cell, 186960 (TCL1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCL1A	TCL1A, TCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Lösemi / Lenfoma, T-Hücreli, 603769 (TCL1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukemia/lymphoma, T-cell, 603769 (TCL1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCL1B	TCL1B, TML1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökosit Adezyon Eksikliği, 116920, Otozomal Resesif; LAD (Lökosit Adezyon Eksikliği) (ITGB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukocyte adhesion deficiency, 116920, Autosomal recessive; LAD (Leukocyte adhesion deficiency) (ITGB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB2	ITGB2, CD18, LCAMB, LAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukocyte adhesion deficiency 1 (LAD1) [MIM:116920]: LAD1 patients have recurrent bacterial infections and their leukocytes are deficient in a wide range of adhesion-dependent functions. (ECO:0000269|PubMed:1346613, ECO:0000269|PubMed:1347532, ECO:0000269|PubMed:1352501, ECO:0000269|PubMed:1590804, ECO:0000269|PubMed:1694220, ECO:0000269|PubMed:1968911, ECO:0000269|PubMed:20529581, ECO:0000269|PubMed:20549317, ECO:0000269|PubMed:7509236, ECO:0000269|PubMed:7686755, ECO:0000269|PubMed:9884339). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökosit Adezyon Eksikliği, 116920, Otozomal Resesif; LAD (Lökosit Adezyon Eksikliği) (Prenatal) (ITGB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukocyte adhesion deficiency, 116920, Autosomal recessive; LAD (Leukocyte adhesion deficiency) (ITGB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGB2	ITGB2, CD18, LCAMB, LAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukocyte adhesion deficiency 1 (LAD1) [MIM:116920]: LAD1 patients have recurrent bacterial infections and their leukocytes are deficient in a wide range of adhesion-dependent functions. (ECO:0000269|PubMed:1346613, ECO:0000269|PubMed:1347532, ECO:0000269|PubMed:1352501, ECO:0000269|PubMed:1590804, ECO:0000269|PubMed:1694220, ECO:0000269|PubMed:1968911, ECO:0000269|PubMed:20529581, ECO:0000269|PubMed:20549317, ECO:0000269|PubMed:7509236, ECO:0000269|PubMed:7686755, ECO:0000269|PubMed:9884339). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökosit Adezyon Eksikliği, Tip 3, 612840, Otozomal Resesif; LAD3 (Lökosit Adezyon Eksikliği) (FERMT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukocyte adhesion deficiency, type III, 612840, Autosomal recessive; LAD3 (Leukocyte adhesion deficiency) (FERMT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FERMT3	KIND3, URP2, MIG2B, FERMT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukocyte adhesion deficiency 3 (LAD3) [MIM:612840]: A disorder characterized by recurrent bacterial infections without pus formation, leukocytosis and major bleeding disorders. (ECO:0000269|PubMed:18779414, ECO:0000269|PubMed:19064721, ECO:0000269|PubMed:19234460, ECO:0000269|PubMed:19234463, ECO:0000269|PubMed:19617577, ECO:0000269|PubMed:26359933). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökosit Adezyon Eksikliği, Tip 3, 612840, Otozomal Resesif; LAD3 (Lökosit Adezyon Eksikliği) (Prenatal) (FERMT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukocyte adhesion deficiency, type III, 612840, Autosomal recessive; LAD3 (Leukocyte adhesion deficiency) (FERMT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FERMT3	KIND3, URP2, MIG2B, FERMT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukocyte adhesion deficiency 3 (LAD3) [MIM:612840]: A disorder characterized by recurrent bacterial infections without pus formation, leukocytosis and major bleeding disorders. (ECO:0000269|PubMed:18779414, ECO:0000269|PubMed:19064721, ECO:0000269|PubMed:19234460, ECO:0000269|PubMed:19234463, ECO:0000269|PubMed:19617577, ECO:0000269|PubMed:26359933). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Kazanılmış Mikrosefali, ± Distoni 616763, Otozomal Resesif (PLEKHG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy and acquired microcephaly with or without dystonia, 616763, Autosomal recessive (PLEKHG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEKHG2	PLEKHG2, CLG, LDAMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökodistrofi, Kazanılmış Mikrosefali, ± Distoni 616763, Otozomal Resesif (Prenatal) (PLEKHG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy and acquired microcephaly with or without dystonia, 616763, Autosomal recessive (PLEKHG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEKHG2	PLEKHG2, CLG, LDAMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi paneli (LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12) (MLPA) (Prenatal)	Leukodystrophy panel (LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12) (MLPA) (Prenatal)	LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Lökodistrofi paneli (LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12) (MLPA) (Prenatal)	Leukodystrophy panel (LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12) (MLPA) (Prenatal)	LMNB1 5q23.2, PLP1 Xq22.2, NOTCH3 19p13.12	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Yetişkin Başlangıçlı, Otozomal Dominant, 169500, Otozomal Dominant; ADLD (Yetişkin Başlangıçlı Otozomal Dominant Lökodistrofi) (LMNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, adult-onset, autosomal dominant, 169500, Autosomal dominant; ADLD (Adult-onset autosomal dominant leukodystrophy) (LMNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNB1	LMNB1, ADLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, demyelinating, autosomal dominant, adult- onset (ADLD) [MIM:169500]: A slowly progressive and fatal demyelinating leukodystrophy, presenting in the fourth or fifth decade of life. Clinically characterized by early autonomic abnormalities, pyramidal and cerebellar dysfunction, and symmetric demyelination of the CNS. It differs from multiple sclerosis and other demyelinating disorders in that neuropathology shows preservation of oligodendroglia in the presence of subtotal demyelination and lack of astrogliosis. [ECO:0000269	PubMed:16951681]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lökodistrofi, Yetişkin Başlangıçlı, Otozomal Dominant, 169500, Otozomal Dominant; ADLD (Yetişkin Başlangıçlı Otozomal Dominant Lökodistrofi) (MLPA)	Leukodystrophy, adult-onset, autosomal dominant, 169500, Autosomal dominant; ADLD (Adult-onset autosomal dominant leukodystrophy) (MLPA)	LMNB1	LMNB1, ADLD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukodystrophy, demyelinating, autosomal dominant, adult- onset (ADLD) [MIM:169500]: A slowly progressive and fatal demyelinating leukodystrophy, presenting in the fourth or fifth decade of life. Clinically characterized by early autonomic abnormalities, pyramidal and cerebellar dysfunction, and symmetric demyelination of the CNS. It differs from multiple sclerosis and other demyelinating disorders in that neuropathology shows preservation of oligodendroglia in the presence of subtotal demyelination and lack of astrogliosis. [ECO:0000269	PubMed:16951681]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lökodistrofi, Yetişkin Başlangıçlı, Otozomal Dominant, 169500, Otozomal Dominant; ADLD (Yetişkin Başlangıçlı Otozomal Dominant Lökodistrofi) (Prenatal) (LMNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, adult-onset, autosomal dominant, 169500, Autosomal dominant; ADLD (Adult-onset autosomal dominant leukodystrophy) (LMNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMNB1	LMNB1, ADLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, demyelinating, autosomal dominant, adult- onset (ADLD) [MIM:169500]: A slowly progressive and fatal demyelinating leukodystrophy, presenting in the fourth or fifth decade of life. Clinically characterized by early autonomic abnormalities, pyramidal and cerebellar dysfunction, and symmetric demyelination of the CNS. It differs from multiple sclerosis and other demyelinating disorders in that neuropathology shows preservation of oligodendroglia in the presence of subtotal demyelination and lack of astrogliosis. [ECO:0000269	PubMed:16951681]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Yetişkin Başlangıçlı, Otozomal Dominant, 169500, Otozomal Dominant; ADLD (Yetişkin Başlangıçlı Otozomal Dominant Lökodistrofi) (Prenatal) (MLPA)	Leukodystrophy, adult-onset, autosomal dominant, 169500, Autosomal dominant; ADLD (Adult-onset autosomal dominant leukodystrophy) (Prenatal) (MLPA)	LMNB1	LMNB1, ADLD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, demyelinating, autosomal dominant, adult- onset (ADLD) [MIM:169500]: A slowly progressive and fatal demyelinating leukodystrophy, presenting in the fourth or fifth decade of life. Clinically characterized by early autonomic abnormalities, pyramidal and cerebellar dysfunction, and symmetric demyelination of the CNS. It differs from multiple sclerosis and other demyelinating disorders in that neuropathology shows preservation of oligodendroglia in the presence of subtotal demyelination and lack of astrogliosis. [ECO:0000269	PubMed:16951681]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 10, 616420, Otozomal Resesif; HLD10 (PYCR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 10, 616420, Autosomal recessive; HLD10 (PYCR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYCR2	PYCR2, HLD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 10, 616420, Otozomal Resesif; HLD10 (Prenatal) (PYCR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 10, 616420, Autosomal recessive; HLD10 (PYCR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYCR2	PYCR2, HLD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 11, 616494, Otozomal Resesif; HLD11 (Hipomiyelinizasyon-Hipogonadotropik-Hipogonadizm-Hipodonti Sendromu) (POLR1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 11, 616494, Autosomal recessive; HLD11 (Hypomyelination-hypogonadotropic-hypogonadism-hypodontia syndrome) (POLR1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR1C	POLR1C, RPA39, RPA40, RPAC1, RPA5, TCS3, HLD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Treacher Collins syndrome 3 (TCS3) [MIM:248390]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. [ECO:0000269	PubMed:21131976]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 11, 616494, Otozomal Resesif; HLD11 (Hipomiyelinizasyon-Hipogonadotropik-Hipogonadizm-Hipodonti Sendromu) (Prenatal) (POLR1C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 11, 616494, Autosomal recessive; HLD11 (Hypomyelination-hypogonadotropic-hypogonadism-hypodontia syndrome) (POLR1C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR1C	POLR1C, RPA39, RPA40, RPAC1, RPA5, TCS3, HLD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Treacher Collins syndrome 3 (TCS3) [MIM:248390]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. [ECO:0000269	PubMed:21131976]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 12, 616683, Otozomal Resesif; HLD12 (VPS11 İlişkili Otozomal Resesif Hipomiyelinizan Lökodistrofi) (VPS11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 12, 616683, Autosomal recessive; HLD12 (VPS11-related autosomal recessive hypomyelinating leukodystrophy) (VPS11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS11	VPS11, HLD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Lökodistrofi, Hipomiyelinizan, 12, 616683, Otozomal Resesif; HLD12 (VPS11 İlişkili Otozomal Resesif Hipomiyelinizan Lökodistrofi) (Prenatal) (VPS11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 12, 616683, Autosomal recessive; HLD12 (VPS11-related autosomal recessive hypomyelinating leukodystrophy) (VPS11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS11	VPS11, HLD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 13, 616881, Otozomal Resesif; HLD13 (HIKESHI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 13, 616881, Autosomal recessive; HLD13 (HIKESHI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HIKESHI	C11orf73, HIKESHI, HLD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 13, 616881, Otozomal Resesif; HLD13 (Prenatal) (HIKESHI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 13, 616881, Autosomal recessive; HLD13 (HIKESHI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HIKESHI	C11orf73, HIKESHI, HLD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 2, 608804, Otozomal Resesif; HLD2 (Pelizaeus-Merzbacher Benzeri Hastalık, GJC2 Mutasyonuna Bağlı) (GJC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 2, 608804, Autosomal recessive; HLD2 (Pelizaeus-Merzbacher-like disease due to GJC2 mutation) (GJC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJC2	GJC2, GJA12, CX47, PMLDAR, HLD2, SPG44, LMPH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 44, autosomal recessive (SPG44) [MIM:613206]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:19056803}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 2, 608804, Otozomal Resesif; HLD2 (Pelizaeus-Merzbacher Benzeri Hastalık, GJC2 Mutasyonuna Bağlı) (Prenatal) (GJC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 2, 608804, Autosomal recessive; HLD2 (Pelizaeus-Merzbacher-like disease due to GJC2 mutation) (GJC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJC2	GJC2, GJA12, CX47, PMLDAR, HLD2, SPG44, LMPH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 44, autosomal recessive (SPG44) [MIM:613206]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:19056803}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 3, 260600, Otozomal Resesif; HLD3 (Pelizaeus-Merzbacher Benzeri Hastalık, AIMP1 Mutasyonuna Bağlı) (AIMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 3, 260600, Autosomal recessive; HLD3 (Pelizaeus-Merzbacher-like disease due to AIMP1 mutation) (AIMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIMP1	AIMP1, SCYE1, EMAP2, EMAPII, HLD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 3 (HLD3) [MIM:260600]: A severe autosomal recessive hypomyelinating leukodystrophy characterized by early infantile onset of global developmental delay, lack of development, lack of speech acquisition, and peripheral spasticity associated with decreased myelination in the central nervous system. {ECO:0000269	PubMed:21092922}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 3, 260600, Otozomal Resesif; HLD3 (Pelizaeus-Merzbacher Benzeri Hastalık, AIMP1 Mutasyonuna Bağlı) (Prenatal) (AIMP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 3, 260600, Autosomal recessive; HLD3 (Pelizaeus-Merzbacher-like disease due to AIMP1 mutation) (AIMP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AIMP1	AIMP1, SCYE1, EMAP2, EMAPII, HLD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 3 (HLD3) [MIM:260600]: A severe autosomal recessive hypomyelinating leukodystrophy characterized by early infantile onset of global developmental delay, lack of development, lack of speech acquisition, and peripheral spasticity associated with decreased myelination in the central nervous system. {ECO:0000269	PubMed:21092922}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 4, 612233, Otozomal Resesif; HLD4 (Pelizaeus-Merzbacher Benzeri Hastalık HSPD1 Mutasyonuna Bağlı) (HSPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 4, 612233, Autosomal recessive; HLD4 (Pelizaeus-Merzbacher-like disease due to HSPD1 mutation) (HSPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPD1	HSPD1, SPG13, HSP60, HLD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 13, autosomal dominant (SPG13) [MIM:605280]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:11898127}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 4, 612233, Otozomal Resesif; HLD4 (Pelizaeus-Merzbacher Benzeri Hastalık HSPD1 Mutasyonuna Bağlı) (Prenatal) (HSPD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 4, 612233, Autosomal recessive; HLD4 (Pelizaeus-Merzbacher-like disease due to HSPD1 mutation) (HSPD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPD1	HSPD1, SPG13, HSP60, HLD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 13, autosomal dominant (SPG13) [MIM:605280]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:11898127}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 5, 610532, Otozomal Resesif; HLD5 (Hipomyelinizasyon- Konjenital Katarakt Sendromu) (FAM126A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 5, 610532, Autosomal recessive; HLD5 (Hypomyelination-congenital cataract syndrome) (FAM126A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM126A	FAM126A, DRCTNNB1A, HLD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 5 (HLD5) [MIM:610532]: A hypomyelinating leukodystrophy associated with congenital cataract. It is clinically characterized by congenital cataract, progressive neurologic impairment, and diffuse myelin deficiency. Affected individuals experience progressive pyramidal and cerebellar dysfunction, muscle weakness and wasting prevailingly in the lower limbs. Mental deficiency ranges from mild to moderate. HLD5 shows clinical variability, but features of hypomyelination combined with increased perivitricular white matter water content are consistently observed. {ECO:0000269|PubMed:16951682, ECO:0000269|PubMed:21911699, ECO:0000269|PubMed:23998934}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 5, 610532, Otozomal Resesif; HLD5 (Hipomyelinizasyon- Konjenital Katarakt Sendromu) (Prenatal) (FAM126A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 5, 610532, Autosomal recessive; HLD5 (Hypomyelination-congenital cataract syndrome) (FAM126A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAM126A	FAM126A, DRCTNNB1A, HLD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 5 (HLD5) [MIM:610532]: A hypomyelinating leukodystrophy associated with congenital cataract. It is clinically characterized by congenital cataract, progressive neurologic impairment, and diffuse myelin deficiency. Affected individuals experience progressive pyramidal and cerebellar dysfunction, muscle weakness and wasting prevailingly in the lower limbs. Mental deficiency ranges from mild to moderate. HLD5 shows clinical variability, but features of hypomyelination combined with increased perivitricular white matter water content are consistently observed. {ECO:0000269|PubMed:16951682, ECO:0000269|PubMed:21911699, ECO:0000269|PubMed:23998934}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 6, 612438, Otozomal Dominant; HLD6 (Hipomyelinizasyon-Bazal Gangliyon ve Serebellum Atrofisi) (TUBB4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 6, 612438, Autosomal dominant; HLD6 (Hypomyelination with atrophy of basal ganglia and cerebellum) (TUBB4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB4A	TUBB4A, DYT4, HLD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 4, torsion, autosomal dominant (DYT4) [MIM:128101]: A form of torsion dystonia, a disease defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. 'Torsion' refers to the twisting nature of body movements, often affecting the trunk. DYT4 is characterized by onset in the second to third decade of progressive laryngeal dysphonia followed by the involvement of other muscles, such as the neck or limbs. Some patients develop an ataxic gait. {ECO:0000269	PubMed:23424103}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 6, 612438, Otozomal Dominant; HLD6 (Hipomyelinizasyon-Bazal Gangliyon ve Serebellum Atrofisi) (Prenatal) (TUBB4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 6, 612438, Autosomal dominant; HLD6 (Hypomyelination with atrophy of basal ganglia and cerebellum) (TUBB4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB4A	TUBB4A, DYT4, HLD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dystonia 4, torsion, autosomal dominant (DYT4) [MIM:128101]: A form of torsion dystonia, a disease defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. 'Torsion' refers to the twisting nature of body movements, often affecting the trunk. DYT4 is characterized by onset in the second to third decade of progressive laryngeal dysphonia followed by the involvement of other muscles, such as the neck or limbs. Some patients develop an ataxic gait. {ECO:0000269	PubMed:23424103}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 7, ± Oligodontia ve / Veya Hipogonadotropik Hipogonadizm, 607694, Otozomal Resesif; HLD7 (Lökoensefalopati-Ataksi-Hipodonti-Hipomiyelinizasyon Sendromu) (POLR3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694, Autosomal recessive; HLD7 (Leukoencephalopathy-ataxia-hypodontia-hypomyelination syndrome) (POLR3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR3A	POLR3A, RPC1, RPC155, ADDH, HLD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 7, ± Oligodontia ve / Veya Hipogonadotropik Hipogonadizm, 607694, Otozomal Resesif; HLD7 (Lökoensefalopati-Ataksi-Hipodonti-Hipomiyelinizasyon Sendromu) (Prenatal) (POLR3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694, Autosomal recessive; HLD7 (Leukoencephalopathy-ataxia-hypodontia-hypomyelination syndrome) (POLR3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR3A	POLR3A, RPC1, RPC155, ADDH, HLD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 8, ± Oligodontia ve / Veya Hipogonadotropik Hipogonadizm, 614381, Otozomal Resesif; HLD8 (Lökoensefalopati-Ataksi-Hipodonti-Hipomiyelinizasyon Sendromu) (POLR3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381, Autosomal recessive; HLD8 (Leukoencephalopathy-ataxia-hypodontia-hypomyelination syndrome) (POLR3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR3B	POLR3B, RPC2, C128, HLD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism (HLD8) [MIM:614381]: An autosomal recessive neurodegenerative disorder characterized by early childhood onset of cerebellar ataxia and mild intellectual disabilities associated with diffuse hypomyelination apparent on brain MRI. Variable features include oligodontia and/or hypogonadotropic hypogonadism. {ECO:0000269|PubMed:22036171, ECO:0000269|PubMed:22036172, ECO:0000269|PubMed:23355746}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 8, ± Oligodontia ve / Veya Hipogonadotropik Hipogonadizm, 614381, Otozomal Resesif; HLD8 (Lökoensefalopati-Ataksi-Hipodonti-Hipomiyelinizasyon Sendromu) (Prenatal) (POLR3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381, Autosomal recessive; HLD8 (Leukoencephalopathy-ataxia-hypodontia-hypomyelination syndrome) (POLR3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR3B	POLR3B, RPC2, C128, HLD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism (HLD8) [MIM:614381]: An autosomal recessive neurodegenerative disorder characterized by early childhood onset of cerebellar ataxia and mild intellectual disabilities associated with diffuse hypomyelination apparent on brain MRI. Variable features include oligodontia and/or hypogonadotropic hypogonadism. {ECO:0000269|PubMed:22036171, ECO:0000269|PubMed:22036172, ECO:0000269|PubMed:23355746}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lökodistrofi, Hipomiyelinizan, 9, 616140, Otozomal Resesif; HLD9 (RARS İle İlişkili Otozomal Resesif Hipomiyelinizan Lökodistrofi) (RARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukodystrophy, hypomyelinating, 9, 616140, Autosomal recessive; HLD9 (RARS-related autosomal recessive hypomyelinating leukodystrophy) (RARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RARS	RARS, HLD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 9 (HLD9) [MIM:616140]: An autosomal recessive neurodegenerative disorder characterized by delayed psychomotor development, severe spasticity, nystagmus, and ataxia associated with diffuse hypomyelination apparent on brain MRI. {ECO:0000269	PubMed:24777941}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökodistrofi, Hipomiyelinizan, 9, 616140, Otozomal Resesif; HLD9 (RARS İle İlişkili Otozomal Resesif Hipomiyelinizan Lökodistrofi) (Prenatal) (RARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukodystrophy, hypomyelinating, 9, 616140, Autosomal recessive; HLD9 (RARS-related autosomal recessive hypomyelinating leukodystrophy) (RARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RARS	RARS, HLD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 9 (HLD9) [MIM:616140]: An autosomal recessive neurodegenerative disorder characterized by delayed psychomotor development, severe spasticity, nystagmus, and ataxia associated with diffuse hypomyelination apparent on brain MRI. {ECO:0000269	PubMed:24777941}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökoensefalopati-Ataksi, 615651, Otozomal Resesif; LKPAT (Lökoensefalopati-Hafif Serebellar Ataksi ve Beyaz Cevher Ödemi) (CLCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with ataxia, 615651, Autosomal recessive; LKPAT (Leukoencephalopathy with mild cerebellar ataxia and white matter edema) (CLCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN2	CLCN2, EGMA, ECA2, EGI11, EJM8, LKPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, idiopathic generalized 11 (EIG11) [MIM:607628]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269	PubMed:19191339}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Lökoensefalopati-Ataksi, 615651, Otozomal Resesif; LKPAT (Lökoensefalopati-Hafif Serebellar Ataksi ve Beyaz Cevher Ödemi) (Prenatal) (CLCN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy with ataxia, 615651, Autosomal recessive; LKPAT (Leukoencephalopathy with mild cerebellar ataxia and white matter edema) (CLCN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN2	CLCN2, EGMA, ECA2, EGI11, EJM8, LKPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Epilepsy, idiopathic generalized 11 (EIG11) [MIM:607628]: A disorder characterized by recurring generalized seizures in the absence of detectable brain lesions and/or metabolic abnormalities. Generalized seizures arise diffusely and simultaneously from both hemispheres of the brain. {ECO:0000269	PubMed:19191339}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökoensefalopati, Beyin Sapı ve Omurilik Tutulumu ve Laktat Yüksekliği, 611105, Otozomal Resesif; LBSL (Lökoensefalopati-Beyin Sapı ve Omurilik Tutulumu-Yüksek Laktat Sendromu) (DARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105, Autosomal recessive; LBSL (Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome) (DARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DARS2	DARS2, ASPRS. LBSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL) [MIM:611105]: Autosomal recessive disease and is defined on the basis of a highly characteristic constellation of abnormalities observed by magnetic resonance imaging and spectroscopy. Affected individuals develop slowly progressive cerebellar ataxia, spasticity, and dorsal column dysfunction, sometimes with a mild cognitive deficit or decline. {ECO:0000269	PubMed:17384640}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökoensefalopati, Beyin Sapı ve Omurilik Tutulumu ve Laktat Yüksekliği, 611105, Otozomal Resesif; LBSL (Lökoensefalopati-Beyin Sapı ve Omurilik Tutulumu-Yüksek Laktat Sendromu) (Prenatal) (DARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation, 611105, Autosomal recessive; LBSL (Leukoencephalopathy with brain stem and spinal cord involvement-high lactate syndrome) (DARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DARS2	DARS2, ASPRS. LBSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL) [MIM:611105]: Autosomal recessive disease and is defined on the basis of a highly characteristic constellation of abnormalities observed by magnetic resonance imaging and spectroscopy. Affected individuals develop slowly progressive cerebellar ataxia, spasticity, and dorsal column dysfunction, sometimes with a mild cognitive deficit or decline. {ECO:0000269	PubMed:17384640}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Distoni ve Motor Nöropati İle Birlikte Olan Lökoensefalopati, 613724, Otozomal Resesif (Lökoensefalopati-Distoni-Motor Nöropati Sendromu) (SCP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with dystonia and motor neuropathy, 613724, Autosomal recessive (Leukoencephalopathy-dystonia-motor neuropathy syndrome) (SCP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCP2	SCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukoencephalopathy, with dystonia and motor neuropathy (LDMN) [MIM:613724]: A syndrome characterized by leukoencephalopathy, dystonic head tremor, spasmodic torticollis and reduced tendon reflexes in lower extremities. Additional features include hyposmia, pathologic saccadic eye movements, a slight hypoacusis, accumulation of branched-chain pristanic acid in plasma, and the presence of abnormal bile alcohol glucuronides in urine. {ECO:0000269	PubMed:16885654}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddeli, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B5	EIF2B5, LVWM, CACH, CLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12325082, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:19158808, ECO:0000269|PubMed:21484434}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddeli, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B2	EIF2B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:21484434, ECO:0000269|PubMed:22285377, ECO:0000269|PubMed:22729508}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddeli, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (Prenatal) (EIF2B5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B5	EIF2B5, LVWM, CACH, CLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12325082, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:19158808, ECO:0000269|PubMed:21484434}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddeli, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (Prenatal) (EIF2B2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B2	EIF2B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:21484434, ECO:0000269|PubMed:22285377, ECO:0000269|PubMed:22729508}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökoensefalopati, Beyin Kalsifikasyon ve Kistleri, 614561, Otozomal Resesif; LCC (SNORD118 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy, brain calcifications, and cysts, 614561, Autosomal recessive; LCC (SNORD118 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNORD118	SNORD118, LCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökoensefalopati, Beyin Kalsifikasyon ve Kistleri, 614561, Otozomal Resesif; LCC (Prenatal) (SNORD118 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy, brain calcifications, and cysts, 614561, Autosomal recessive; LCC (SNORD118 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNORD118	SNORD118, LCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökoensefalopati, Kistik, Megalensefali, 612951, Otozomal Resesif (Kistik Lökoensefalopati- Megalensefali Olmadan) (RNASET2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy, cystic, without megalencephaly, 612951, Autosomal recessive (Cystic leukoencephalopathy without megalencephaly) (RNASET2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASET2	RNASET2, RNASE6PL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökoensefalopati, Kistik, Megalensefali, 612951, Otozomal Resesif (Kistik Lökoensefalopati- Megalensefali Olmadan) (Prenatal) (RNASET2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy, cystic, without megalencephaly, 612951, Autosomal recessive (Cystic leukoencephalopathy without megalencephaly) (RNASET2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNASET2	RNASET2, RNASE6PL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lökoensefalopati, Yaygın Kaitsal, Sferoidler İle, 221820, Otozomal Dominant (Kaitsal Yaygın Lökoensefalopatisi-Aksonal Sferoidler ve Pigmentli Glia) (CSF1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820, Autosomal dominant (Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia) (CSF1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSF1R	CSF1R, FMS, HDLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukoencephalopathy, diffuse hereditary, with spheroids (HDLS) [MIM:221820]: An autosomal dominant adult-onset rapidly progressive neurodegenerative disorder characterized by variable behavioral, cognitive, and motor changes. Patients often die of dementia within 6 years of onset. Brain imaging shows patchy abnormalities in the cerebral white matter, predominantly affecting the frontal and parietal lobes. {ECO:0000269|PubMed:22197934, ECO:0000269|PubMed:24532199}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lökoensefalopati, Yaygın Kaitsal, Sferoidler İle, 221820, Otozomal Dominant (Kaitsal Yaygın Lökoensefalopatisi-Aksonal Sferoidler ve Pigmentli Glia) (Prenatal) (CSF1R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy, diffuse hereditary, with spheroids, 221820, Autosomal dominant (Hereditary diffuse leukoencephalopathy with axonal spheroids and pigmented glia) (CSF1R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSF1R	CSF1R, FMS, HDLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukoencephalopathy, diffuse hereditary, with spheroids (HDLS) [MIM:221820]: An autosomal dominant adult-onset rapidly progressive neurodegenerative disorder characterized by variable behavioral, cognitive, and motor changes. Patients often die of dementia within 6 years of onset. Brain imaging shows patchy abnormalities in the cerebral white matter, predominantly affecting the frontal and parietal lobes. {ECO:0000269|PubMed:22197934, ECO:0000269|PubMed:24532199}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökoensefalopati, Progresif, Over Yetmezliği, 615889, Otozomal Resesif; LKENP (CACH Sendromu) (AARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy, progressive, with ovarian failure, 615889, Autosomal recessive; LKENP (CACH syndrome) (AARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AARS2	AARS2, KIAA1270, MTALARS, COXPD8, LKENP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 8 (COXPD8) [MIM:614096]: A mitochondrial disease characterized by a lethal infantile hypertrophic cardiomyopathy, generalized muscle dysfunction and some neurologic involvement. The liver is not affected. (ECO:0000269|PubMed:21549344). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lökoensefalopati, Progresif, Over Yetmezliği, 615889, Otozomal Resesif; LKENP (CACH Sendromu) (Prenatal) (AARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephalopathy, progressive, with ovarian failure, 615889, Autosomal recessive; LKENP (CACH syndrome) (AARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AARS2	AARS2, KIAA1270, MTALARS, COXPD8, LKENP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined oxidative phosphorylation deficiency 8 (COXPD8) [MIM:614096]: A mitochondrial disease characterized by a lethal infantile hypertrophic cardiomyopathy, generalized muscle dysfunction and some neurologic involvement. The liver is not affected. (ECO:0000269|PubMed:21549344). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddel, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephaly with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B4	EIF2B4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11835386, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddel, 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (Prenatal) (EIF2B4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukoencephaly with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B4	EIF2B4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. {ECO:0000269|PubMed:11835386, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lökotrien C4 Sentaz Eksikliği, 614037, Otozomal Resesif (Hipotoni-Gelişme Geriliği-Mikrosefali Sendromu (Lökotrien C4 Sentaz Eksikliği)) (LTC4S geni) (Tüm Gen Dizi Analizi) (Prognatal)	Leukotriene C4 synthase deficiency, 614037, Autosomal recessive (Hypotonia-failure to thrive-microcephaly syndrome) (LTC4S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTC4S	LTC4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lökotrien C4 Sentaz Eksikliği, 614037, Otozomal Resesif (Hipotoni-Gelişme Geriliği-Mikrosefali Sendromu (Lökotrien C4 Sentaz Eksikliği)) (Prenatal) (LTC4S geni) (Tüm Gen Dizi Analizi) (Prenatal)	Leukotriene C4 synthase deficiency, 614037, Autosomal recessive (Hypotonia-failure to thrive-microcephaly syndrome) (LTC4S gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LTC4S	LTC4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lewy Cismi Demansı, Yatkınlık, 127750, Otozomal Dominant (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lewy body dementia, susceptibility to, 127750, Autosomal dominant (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Parkinson disease (PARK) [MIM:168600]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features. {ECO:0000269|PubMed:12847165, ECO:0000269|PubMed:16148263, ECO:0000269|PubMed:19286695}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Lewy Cismi Demansı, Yatkınlık, 127750, Otozomal Dominant (MLPA)	Lewy body dementia, susceptibility to, 127750, Autosomal dominant (MLPA)	GBA	GBA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Parkinson disease (PARK) [MIM:168600]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features. {ECO:0000269|PubMed:12847165, ECO:0000269|PubMed:16148263, ECO:0000269|PubMed:19286695}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Lewy Cismi Demansı, Yatkınlık, 127750, Otozomal Dominant (Prenatal) (GBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lewy body dementia, susceptibility to, 127750, Autosomal dominant (GBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Parkinson disease (PARK) [MIM:168600]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features. {ECO:0000269|PubMed:12847165, ECO:0000269|PubMed:16148263, ECO:0000269|PubMed:19286695}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lewy Cismi Demansı, Yatkınlık, 127750, Otozomal Dominant (Prenatal) (MLPA)	Lewy body dementia, susceptibility to, 127750, Autosomal dominant (Prenatal) (MLPA)	GBA	GBA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Parkinson disease (PARK) [MIM:168600]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features. {ECO:0000269|PubMed:12847165, ECO:0000269|PubMed:16148263, ECO:0000269|PubMed:19286695}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Leydig Hücre Adenomu, Somatik, Erken Puberte, 176410 (LHCGR geni) (Tüm Gen Dizi Analizi) (Prognatal)	Leydig cell adenoma, somatic, with precocious puberty, 176410 (LHCGR gene) (Sequence Analysis-All Coding Exons) (Prognatal)	LHCGR	LHCGR, LHR, LCGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Leydig Hücre Hipoplazi-Hipergonadotropik Hipogonadizm, 238320, Otozomal Resesif (Leydig Hücre Hipoplazisi) (LHCGR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320, Autosomal recessive (Leydig cell hypoplasia) (LHCGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHCGR	LHCGR, LHR, LCGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Luteinizing hormone resistance (LHR) [MIM:238320]: An autosomal recessive disorder characterized by unresponsiveness to luteinizing hormone, defective sexual development in males, and defective follicular development and ovulation, amenorrhea and infertility in females. Two forms of the disorder have been defined in males. Type 1 is a severe form characterized by complete 46,XY male pseudohermaphroditism, low testosterone and high luteinizing hormone levels, total lack of responsiveness to luteinizing and chorionic gonadotropin hormones, lack of breast development, and absent development of secondary male sex characteristics. Type 2, a milder form, displays a broader range of phenotypic expression ranging from micropenis to severe hypospadias. {ECO:0000269|PubMed:12050206, ECO:0000269|PubMed:15377531, ECO:0000269|PubMed:15472221, ECO:0000269|PubMed:19551906, ECO:0000269|PubMed:7719343, ECO:0000269|PubMed:8559204, ECO:0000269|PubMed:9215288, ECO:0000269|PubMed:9514160, ECO:0000269|PubMed:9626144, ECO:0000269|PubMed:9626653}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lhermitte-Duclos Sendromu, 158350, Otozomal Dominant (Lhermitte-Duclos Disease) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lhermitte-Duclos syndrome, 158350, Autosomal dominant (Lhermitte-Duclos disease) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lhermitte-Duclos Sendromu, 158350, Otozomal Dominant (Lhermitte-Duclos Disease) (MLPA)	Lhermitte-Duclos syndrome, 158350, Autosomal dominant (Lhermitte-Duclos disease) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lhermitte-Duclos Sendromu, 158350, Otozomal Dominant (Lhermitte-Duclos Disease) (Prenatal) (PTEN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lhermitte-Duclos syndrome, 158350, Autosomal dominant (Lhermitte-Duclos disease) (PTEN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lhermitte-Duclos Sendromu, 158350, Otozomal Dominant (Lhermitte-Duclos Disease) (Prenatal) (MLPA)	Lhermitte-Duclos syndrome, 158350, Autosomal dominant (Lhermitte-Duclos disease) (Prenatal) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Li-Fraumeni Sendromu, 151623, Otozomal Dominant; LFS1 (Li-Fraumeni Sendromu) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Li-Fraumeni syndrome, 151623, Autosomal dominant; LFS1 (Li-Fraumeni syndrome) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Li-Fraumeni Sendromu, 151623, Otozomal Dominant; LFS1 (Li-Fraumeni Sendromu) (MLPA)	Li-Fraumeni syndrome, 151623, Autosomal dominant; LFS1 (Li-Fraumeni syndrome) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Li-Fraumeni Sendromu, 151623, Otozomal Dominant; LFS1 (Li-Fraumeni Sendromu) (MLPA)	Li-Fraumeni syndrome, 151623, Autosomal dominant; LFS1 (Li-Fraumeni syndrome) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Li-Fraumeni Sendromu, 609265; LFS2 (Li-Fraumeni Sendromu) (CHEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Li-Fraumeni syndrome, 609265; LFS2 (Li-Fraumeni syndrome) (CHEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Li-Fraumeni Sendromu, 609265; LFS2 (Li-Fraumeni Sendromu) (MLPA)	Li-Fraumeni syndrome, 609265; LFS2 (Li-Fraumeni syndrome) (MLPA)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Lichtenstein-Knorr Sendromu, 616291, Otozomal Resesif; LIKNS (Progresif Otozomal Resesif Ataksi-Sağırlık Sendromu) (SLC9A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lichtenstein-Knorr syndrome, 616291, Autosomal recessive; LIKNS (Progressive autosomal recessive ataxia-deafness syndrome) (SLC9A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC9A1	SLC9A1, NHE1, APNH, LIKNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lichtenstein-Knorr syndrome (LIKNS) [MIM:616291]: An autosomal recessive neurologic disorder characterized by progressive cerebellar ataxia and severe progressive sensorineural hearing loss. {ECO:0000269	PubMed:25205112}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Liddle Sendromu, 177200, Otozomal Dominant (Liddle Sendromu) (SCNN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Liddle syndrome, 177200, Autosomal dominant (Liddle syndrome) (SCNN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1B	SCNN1B, BESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269|PubMed:15483078, ECO:0000269|PubMed:7550319, ECO:0000269|PubMed:8524790, ECO:0000269|PubMed:8601645, ECO:0000269|PubMed:9626162, ECO:0000269|PubMed:9794716}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Liddle Sendromu, 177200, Otozomal Dominant (Liddle Sendromu) (Prenatal) (SCNN1B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Liddle syndrome, 177200, Autosomal dominant (Liddle syndrome) (SCNN1B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCNN1B	SCNN1B, BESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269|PubMed:15483078, ECO:0000269|PubMed:7550319, ECO:0000269|PubMed:8524790, ECO:0000269|PubMed:8601645, ECO:0000269|PubMed:9626162, ECO:0000269|PubMed:9794716}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Liddle Sendromu, 177200, Otozomal Dominant; LIDLS (Liddle Sendromu) (SCNN1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Liddle syndrome, 177200, Autosomal dominant; LIDLS (Liddle syndrome) (SCNN1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1G	SCNN1G, PHA1, BESC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269	PubMed:7550319}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Liddle Sendromu, 177200, Otozomal Dominant; LIDLS (Liddle Sendromu) (Prenatal) (SCNN1G geni) (Tüm Gen Dizi Analizi) (Prenatal)	Liddle syndrome, 177200, Autosomal dominant; LIDLS (Liddle syndrome) (SCNN1G gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCNN1G	SCNN1G, PHA1, BESC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269	PubMed:7550319}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Liebenberg Sendromu, 186550, Otozomal Dominant; LBNBG (Brakidaktili-Dirsek Bilek Displazisi Sendromu (Liebenberg Sendromu)) (PITX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Liebenberg syndrome, 186550, Autosomal dominant; LBNBG (Brachydactyly-elbow wrist dysplasia syndrome) (PITX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX1	PITX1, PTX1, BFT, POTX, CCF, LBNBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly (CCF) [MIM:119800]: A congenital limb deformity defined as fixation of the foot in cavus, adductus, varus, and equinus (i.e., inclined inwards, axially rotated outwards, and pointing downwards) with concomitant soft tissue abnormalities. Clubfoot may occur in isolation or as part of a syndrome. Some patients present tibial hemimelia, bilateral patellar hypoplasia, and preaxial mirror-image polydactyly. {ECO:0000269|PubMed:18950742, ECO:0000269|PubMed:22258522}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

LIG4 Sendromu, 606593 (LIG4 Sendromu) (LIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LIG4 syndrome, 606593 (LIG4 syndrome) (LIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIG4	LIG4, LIG4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269	PubMed:16357942}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
LIG4 Sendromu, 606593 (LIG4 Sendromu) (Prenatal) (LIG4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LIG4 syndrome, 606593 (LIG4 syndrome) (LIG4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIG4	LIG4, LIG4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269	PubMed:16357942}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ekstremite malformasyonları paneli (GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31) (MLPA)	Limb malformations (GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31) (MLPA)	GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ekstremite malformasyonları paneli (GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31) (MLPA) (Prenatal)	Limb malformations (GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31) (MLPA) (Prenatal)	GLI3 7p14.1, HOXD13 2q31.1, ROR2 9q22.31	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Limb-girdle tipi kas distrofisi paneli (SGCA, SGCB, SGCD, SGCG, FKRP) (MLPA)	Limb-girdle type muscular dystrophy panel (SGCA, SGCB, SGCD, SGCG, FKRP) (MLPA)	SGCA, SGCB, SGCD, SGCG, FKRP	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Ekstremite-Meme Sendromu, 603543, Otozomal Dominant; LMS (Ekstremite-Meme Sendromu) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Limb-mammary syndrome, 603543, Autosomal dominant; LMS (Limb-mammary syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ekstremite-Meme Sendromu, 603543, Otozomal Dominant; LMS (Ekstremite-Meme Sendromu) (Prenatal) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Limb-mammary syndrome, 603543, Autosomal dominant; LMS (Limb-mammary syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Limb-girdle tipi kas distrofisi, miyofibriller myopatiler (LMNA 1q22; ZMPSTE24 1p34; MYOT 5q31; CAV3 3p25) (MLPA)	Limb-type muscular dystrophy, myofibrillar myopathies (LMNA 1q22; ZMPSTE24 1p34; MYOT 5q31; CAV3 3p25) (MLPA)	LMNA 1q22; ZMPSTE24 1p34; MYOT 5q31; CAV3 3p25	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 1, 309801, X'e Bağlı Dominant; LSDMCA1 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (HCCS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Linear skin defects with multiple congenital anomalies 1, 309801, X-linked dominant; LSDMCA1 (Microphthalmia with linear skin defects syndrome) (HCCS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCCS	HCCS, MCOPS7, LSDMCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Linear skin defects with multiple congenital anomalies 1 (LSDMCA1) [MIM:309801]: A disorder characterized by dermal, ocular, neurological and cardiac abnormalities. LSDMCA1 main features are unilateral or bilateral microphthalmia, linear skin defects in affected females, and in utero lethality for males. Skin defects are limited to the face and neck, consisting of areas of aplastic skin that heal with age to form hyperpigmented areas. Additional features in female patients include agenesis of the corpus callosum, sclerocornea, chorioretinal abnormalities, infantile seizures, congenital heart defect, mental retardation, and diaphragmatic hernia. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:17033964}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 1, 309801, X'e Bağlı Dominant; LSDMCA1 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (Prenatal) (HCCS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Linear skin defects with multiple congenital anomalies 1, 309801, X-linked dominant; LSDMCA1 (Microphthalmia with linear skin defects syndrome) (HCCS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HCCS	HCCS, MCOPS7, LSDMCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Linear skin defects with multiple congenital anomalies 1 (LSDMCA1) [MIM:309801]: A disorder characterized by dermal, ocular, neurological and cardiac abnormalities. LSDMCA1 main features are unilateral or bilateral microphthalmia, linear skin defects in affected females, and in utero lethality for males. Skin defects are limited to the face and neck, consisting of areas of aplastic skin that heal with age to form hyperpigmented areas. Additional features in female patients include agenesis of the corpus callosum, sclerocornea, chorioretinal abnormalities, infantile seizures, congenital heart defect, mental retardation, and diaphragmatic hernia. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:17033964}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 2, 309887, X'e Bağlı Dominant; LSDMCA2 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (COX7B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Linear skin defects with multiple congenital anomalies 2, 309887, X-linked dominant; LSDMCA2 (Microphthalmia with linear skin defects syndrome) (COX7B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX7B	COX7B, LSDMCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Linear skin defects with multiple congenital anomalies 2 (LSDMCA2) [MIM:309887]: A distinct form of aplasia cutis congenita presenting as multiple linear skin defects on the face and neck associated with poor growth, microcephaly, and facial dysmorphism. Additional features include intellectual disability, nail dystrophy, short stature and cardiac abnormalities. {ECO:0000269	PubMed:23122588}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 2, 309887, X'e Bağlı Dominant; LSDMCA2 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (Prenatal) (COX7B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Linear skin defects with multiple congenital anomalies 2, 309887, X-linked dominant; LSDMCA2 (Microphthalmia with linear skin defects syndrome) (COX7B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX7B	COX7B, LSDMCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Linear skin defects with multiple congenital anomalies 2 (LSDMCA2) [MIM:309887]: A distinct form of aplasia cutis congenita presenting as multiple linear skin defects on the face and neck associated with poor growth, microcephaly, and facial dysmorphism. Additional features include intellectual disability, nail dystrophy, short stature and cardiac abnormalities. {ECO:0000269	PubMed:23122588}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 3, 300952, X'e Bağlı Dominant; LSDMCA3 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (NDUFB11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Linear skin defects with multiple congenital anomalies 3, 300952, X-linked dominant; LSDMCA3 (Microphthalmia with linear skin defects syndrome) (NDUFB11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFB11	NDUFB11, LSDMCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lineer Deri Defekti, Multipl Konjenital Anomalili 3, 300952, X'e Bağlı Dominant; LSDMCA3 (Mikroftalmi-Lineer Cilt Bozuklukları Sendromu) (Prenatal) (NDUFB11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Linear skin defects with multiple congenital anomalies 3, 300952, X-linked dominant; LSDMCA3 (Microphthalmia with linear skin defects syndrome) (NDUFB11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFB11	NDUFB11, LSDMCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lipaz Eksikliği, Kombine, 246650, Otozomal Resesif (LMF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipase deficiency, combined, 246650, Autosomal recessive (LMF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMF1	LMF1, TMEM112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined lipase deficiency (CLD) [MIM:246650]: Characterized by repeated episodes of pancreatitis, tuberous xanthomas and lipodystrophy and is caused by deficiency of both lipoprotein lipase (LPL) and hepatic triglyceride lipase (HTGL). {ECO:0000269	PubMed:17994020}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipid Depo Miyopatisi, Flavin Adenin Dinükleotid Sentetaz Eksikliğine Bağlı, 255100, Otozomal Resesif, LSMFLAD (FLAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipid storage myopathy due to flavin adenine dinucleotide synthetase deficiency, 255100, Autosomal recessive; LSMFLAD (FLAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLAD1	FLAD1, FADS, LSMFLAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize, Tip 1, 608594, Otozomal Resesif; CGL1 (Berardinelli-Seip Sendromu Tip 1) (Berardinelli-Seip Konjenital Lipodistrofi) (AGPAT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, congenital generalized, type 1, 608594, Autosomal recessive; CGL1 (Berardinelli-Seip Syndrome type 1) (Berardinelli-Seip congenital lipodystrophy) (AGPAT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGPAT2	AGPAT2, LPAAB, BSCL, BSCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital generalized lipodystrophy 1 (CGL1) [MIM:608594]: An autosomal recessive disorder characterized by a near complete absence of adipose tissue, extreme insulin resistance, hypertriglyceridemia, hepatic steatosis and early onset of diabetes. {ECO:0000269	PubMed:11967537}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize, Tip 1, 608594, Otozomal Resesif; CGL1 (Berardinelli-Seip Sendromu Tip 1) (Berardinelli-Seip Konjenital Lipodistrofi) (Prenatal) (AGPAT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipodystrophy, congenital generalized, type 1, 608594, Autosomal recessive; CGL1 (Berardinelli-Seip Syndrome type 1) (Berardinelli-Seip congenital lipodystrophy) (AGPAT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGPAT2	AGPAT2, LPAAB, BSCL, BSCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital generalized lipodystrophy 1 (CGL1) [MIM:608594]: An autosomal recessive disorder characterized by a near complete absence of adipose tissue, extreme insulin resistance, hypertriglyceridemia, hepatic steatosis and early onset of diabetes. {ECO:0000269	PubMed:11967537}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lipodistrofi, Konjenital Jeneralize, Tip 2, 269700, Otozomal Resesif; CGL2 (Berardinelli-Seip Konjenital Lipodistrofi) (BSCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, congenital generalized, type 2, 269700, Autosomal recessive; CGL2 (Berardinelli-Seip congenital lipodystrophy) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 17, autosomal dominant (SPG17) [MIM:270685]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG17 is characterized by prominent amyotrophy of the hand muscles, the presence of mild to severe pyramidal tract signs and spastic paraplegia. SPG17 is a motor neuron disease overlapping with distal spinal muscular atrophy type 5. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize, Tip 2, 269700, Otozomal Resesif; CGL2 (Berardinelli-Seip Konjenital Lipodistrofi) (Prenatal) (BSCL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipodystrophy, congenital generalized, type 2, 269700, Autosomal recessive; CGL2 (Berardinelli-Seip congenital lipodystrophy) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 17, autosomal dominant (SPG17) [MIM:270685]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG17 is characterized by prominent amyotrophy of the hand muscles, the presence of mild to severe pyramidal tract signs and spastic paraplegia. SPG17 is a motor neuron disease overlapping with distal spinal muscular atrophy type 5. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize Tip 3, 612526; CGL3 (Berardinelli-Seip Konjenital Lipodistrofi) (CAV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, congenital generalized, type 3, 612526; CGL3 (Berardinelli-Seip congenital lipodystrophy) (CAV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. (ECO:0000269	PubMed:18237401). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize Tip 3, 612526; CGL3 (Berardinelli-Seip Konjenital Lipodistrofi) (Prenatal) (CAV1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipodystrophy, congenital generalized, type 3, 612526; CGL3 (Berardinelli-Seip congenital lipodystrophy) (CAV1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. (ECO:0000269	PubMed:18237401). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize, Tip 4, 613327, Otozomal Resesif; CGL4 (Jeneralize Konjenital Lipodistrofi-Miyopati) (PTRF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, congenital generalized, type 4, 613327, Autosomal recessive; CGL4 (Generalized congenital lipodystrophy with myopathy) (PTRF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTRF	PTRF, CAVIN, CGL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital generalized lipodystrophy 4 (CGL4) [MIM:613327]: A disorder characterized by the association of congenital generalized lipodystrophy with muscular dystrophy and cardiac anomalies. Congenital generalized lipodystrophy is characterized by a near complete absence of adipose tissue, extreme insulin resistance, hypertriglyceridemia, hepatic steatosis and early onset of diabetes. (ECO:0000269|PubMed:19726876, ECO:0000269|PubMed:20684003). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipodistrofi, Konjenital Jeneralize, Tip 4, 613327, Otozomal Resesif; CGL4 (Jeneralize Konjenital Lipodistrofi-Miyopati) (Prenatal) (PTRF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipodystrophy, congenital generalized, type 4, 613327, Autosomal recessive; CGL4 (Generalized congenital lipodystrophy with myopathy) (PTRF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTRF	PTRF, CAVIN, CGL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital generalized lipodystrophy 4 (CGL4) [MIM:613327]: A disorder characterized by the association of congenital generalized lipodystrophy with muscular dystrophy and cardiac anomalies. Congenital generalized lipodystrophy is characterized by a near complete absence of adipose tissue, extreme insulin resistance, hypertriglyceridemia, hepatic steatosis and early onset of diabetes. (ECO:0000269|PubMed:19726876, ECO:0000269|PubMed:20684003). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 2, 151660, Otozomal Dominant; FPLD2 (Ailesel Parsiyel Lipodistrofi, Dunnigan Tipi) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, familial partial, type 2, 151660, Autosomal dominant; FPLD2 (Familial partial lipodystrophy, Dunnigan type) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. (ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 2, 151660, Otozomal Dominant; FPLD2 (Ailesel Parsiyel Lipodistrofi, Dunnigan Tipi) (MLPA)	Lipodystrophy, familial partial, type 2, 151660, Autosomal dominant; FPLD2 (Familial partial lipodystrophy, Dunnigan type) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. (ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 3, 604367, Otozomal Dominant; FPLD3 (PPARG İle İlişkili Ailesel Parsiyel Lipodistrofi) (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, familial partial, type 3, 604367, Autosomal dominant; FPLD3 (PPARG-related familial partial lipodystrophy) (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CIMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glioma 1 (GLM1) [MIM:137800]: Gliomas are benign or malignant central nervous system neoplasms derived from glial cells. They comprise astrocytomas and glioblastoma multiforme that are derived from astrocytes, oligodendrogliomas derived from oligodendrocytes and ependymomas derived from ependymocytes. (ECO:0000269	PubMed:10851250). Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Polymorphic PPARG alleles have been found to be significantly over-represented among a cohort of American patients with sporadic glioblastoma multiforme suggesting a possible contribution to disease susceptibility.;	EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 4, 613877, Otozomal Dominant; FPLD4 (Ailesel Parsiyel Lipodistrofisi, PLIN1 İle İlişkili) (PLIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, familial partial, type 4, 613877, Autosomal dominant; FPLD4 (PLIN1-related familial partial lipodystrophy) (PLIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLIN1	PLIN1, PLIN, FPLD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lipodystrophy, familial partial, 4 (FPLD4) [MIM:613877]: A form of lipodystrophy characterized by loss of subcutaneous adipose tissue primarily affecting the lower limbs, insulin- resistant diabetes mellitus, hypertriglyceridemia, and hypertension. (ECO:0000269	PubMed:21345103). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 5, 615238, Otozomal Resesif; FPLD5 (Ailesel Parsiyel Lipodistrofi, CIDEC İle İlişkili) (CIDEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, familial partial, type 5, 615238, Autosomal recessive; FPLD5 (CIDEC-related familial partial lipodystrophy) (CIDEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIDEC	CIDEC, FSP27, CIDE3, FPLD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lipodystrophy, familial partial, 5 (FPLD5) [MIM:615238]: A form of lipodystrophy characterized by loss of subcutaneous adipose tissue affecting limb, femoragluteal and subcutaneous abdominal fat, preservation of visceral, neck and axillary fat, hepatomegaly, hepatic steatosis and insulin-resistant diabetes. (ECO:0000269	PubMed:20049731). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipodistrofi, Ailesel Parsiyel, Tip 6, 615980, Otozomal Resesif; FPLD6 (Ailesel Parsiyel Lipodistrofi, LIPE İlişkili) (LIPE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, familial partial, type 6, 615980, Autosomal recessive; FPLD6 (LIPE-related familial partial lipodystrophy) (LIPE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPE	LIPE, LHS, FPLD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lipodystrophy, familial partial, 6 (FPLD6) [MIM:615980]: A form of lipodystrophy characterized by abnormal subcutaneous fat distribution. Affected individuals have increased visceral fat, impaired lipolysis, dyslipidemia, hepatic steatosis, systemic insulin resistance, and diabetes. Some patients manifest muscular dystrophy. (ECO:0000269	PubMed:24848981). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lipodistrofi, Parsiyel, Kazanılmış, Yatkınlık, 608709, Otozomal Dominant; APLD (Parsiyel Erilimiş Lipodistrofi) (LMNB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipodystrophy, partial, acquired, susceptibility to, 608709, Autosomal dominant; APLD (Partial acquired lipodystrophy) (LMNB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNB2	LMNB2, LMN2, EPM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Partial acquired lipodystrophy (APLD) [MIM:608709]: A rare childhood disease characterized by loss of subcutaneous fat from the face and trunk. Fat deposition on the pelvic girdle and lower limbs is normal or excessive. Most frequently, onset between 5 and 15 years of age. Most affected subjects are females and some show no other abnormality, but many develop glomerulonephritis, diabetes mellitus, hyperlipidemia, and complement deficiency. Mental retardation in some cases. APLD is a sporadic disorder of unknown etiology. (ECO:0000269|PubMed:16826530, ECO:0000269|PubMed:22768673). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lipoid Adrenal Hiperplazi, 201710, Otozomal Resesif; LCAH (Konjenital Adrenal Hiperplazi) (STAR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoid adrenal hyperplasia, 201710, Autosomal recessive; LCAH (Congenital adrenal hyperplasia) (STAR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAR	STAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Lipoid Adrenal Hiperplazi, 201710, Otozomal Resesif; LCAH (Konjenital Adrenal Hiperplazi) (Prenatal) (STAR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipoid adrenal hyperplasia, 201710, Autosomal recessive; LCAH (Congenital adrenal hyperplasia) (STAR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAR	STAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lipoma (LPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoma (LPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPP	LPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving LPP is associated with pulmonary chondroid hamartomas. Translocation t(3;12)(q27; q28;q14-q15) with HMGA2 is detected in pulmonary chondroid hamartomas.	EDTA Blood Sample
Lipom, Somatik (Multipl Endokrin Neoplazi Tip 1) (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoma, somatic (Multiple endocrine neoplasia type 1) (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media

Lipoprotein Glomerülopati, 611771; LPG (Lipoprotein Glomerulopatisi) (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoprotein glomerulopathy, 611771; LPG (Lipoprotein glomerulopathy) (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 2 (AD2) [MIM:104310]: A late-onset neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The APOE*4 allele is genetically associated with the common late onset familial and sporadic forms of Alzheimer disease. Risk for AD increased from 20% to 90% and mean age at onset decreased from 84 to 68 years with increasing number of APOE*4 alleles in 42 families with late onset AD. Thus APOE*4 gene dose is a major risk factor for late onset AD and, in these families, homozygosity for APOE*4 was virtually sufficient to cause AD by age 80. The mechanism by which APOE*4 participates in pathogenesis is not known.	EDTA Blood Sample
Lipoprotein Lipaz Eksikliği, 238600, Otozomal Resesif (Ailesel Lipoprotein Lipaz Eksikliği) (LPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoprotein lipase deficiency, 238600, Autosomal recessive (Familial lipoprotein lipase deficiency) (LPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPL	LPL, LIPD, HDLCQ11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lipoprotein lipase deficiency (LPL deficiency) [MIM:238600]: Recessive disorder usually manifesting in childhood. On a normal diet, patients often present with abdominal pain, hepatosplenomegaly, lipemia retinalis, eruptive xanthomata, and massive hypertriglyceridemia, sometimes complicated with acute pancreatitis. {ECO:0000269|PubMed:10660334, ECO:0000269|PubMed:10787434, ECO:0000269|PubMed:11068186, ECO:0000269|PubMed:11099402, ECO:0000269|PubMed:11134145, ECO:0000269|PubMed:11441134, ECO:0000269|PubMed:12204001, ECO:0000269|PubMed:12641539, ECO:0000269|PubMed:12966036, ECO:0000269|PubMed:1400331, ECO:0000269|PubMed:1479292, ECO:0000269|PubMed:14984478, ECO:0000269|PubMed:15185149, ECO:0000269|PubMed:1521525, ECO:0000269|PubMed:15256764, ECO:0000269|PubMed:15877202, ECO:0000269|PubMed:1598907, ECO:0000269|PubMed:1619366, ECO:0000269|PubMed:1639392, ECO:0000269|PubMed:1674945, ECO:0000269|PubMed:1702428, ECO:0000269|PubMed:1730727, ECO:0000269|PubMed:1752947, ECO:0000269|PubMed:1907278, ECO:0000269|PubMed:1969408, ECO:0000269|PubMed:1975097	EDTA Blood Sample
Lipoprotein Lipaz Eksikliği, 238600, Otozomal Resesif (Ailesel Lipoprotein Lipaz Eksikliği) (MLPA)	Lipoprotein lipase deficiency, 238600, Autosomal recessive (Familial lipoprotein lipase deficiency) (MLPA)	LPL	LPL, LIPD, HDLCQ11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lipoprotein lipase deficiency (LPL deficiency) [MIM:238600]: Recessive disorder usually manifesting in childhood. On a normal diet, patients often present with abdominal pain, hepatosplenomegaly, lipemia retinalis, eruptive xanthomata, and massive hypertriglyceridemia, sometimes complicated with acute pancreatitis. {ECO:0000269|PubMed:10660334, ECO:0000269|PubMed:10787434, ECO:0000269|PubMed:11068186, ECO:0000269|PubMed:11099402, ECO:0000269|PubMed:11134145, ECO:0000269|PubMed:11441134, ECO:0000269|PubMed:12204001, ECO:0000269|PubMed:12641539, ECO:0000269|PubMed:12966036, ECO:0000269|PubMed:1400331, ECO:0000269|PubMed:1479292, ECO:0000269|PubMed:14984478, ECO:0000269|PubMed:15185149, ECO:0000269|PubMed:1521525, ECO:0000269|PubMed:15256764, ECO:0000269|PubMed:15877202, ECO:0000269|PubMed:1598907, ECO:0000269|PubMed:1619366, ECO:0000269|PubMed:1639392, ECO:0000269|PubMed:1674945, ECO:0000269|PubMed:1702428, ECO:0000269|PubMed:1730727, ECO:0000269|PubMed:1752947, ECO:0000269|PubMed:1907278, ECO:0000269|PubMed:1969408, ECO:0000269|PubMed:1975097	EDTA Blood Sample
Lipoltransferaz 1 Eksikliği, 616299, Otozomal Resesif; LIPT1D (Lipoil Transferaz 1 Eksikliği) (LIPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lipoyltransferase 1 deficiency, 616299, Autosomal recessive; LIPT1D (Lipoyl transferase 1 deficiency) (LIPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPT1	LIPT1, LIPT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lipoltransferaz 1 Eksikliği, 616299, Otozomal Resesif; LIPT1D (Lipoil Transferaz 1 Eksikliği) (Prenatal) (LIPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lipoyltransferase 1 deficiency, 616299, Autosomal recessive; LIPT1D (Lipoyl transferase 1 deficiency) (LIPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIPT1	LIPT1, LIPT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 1, 607432, Sporadik Vakalar; LIS1 (Lizensefali, LIS1 Mutasyonuna Bağlı) (PAFAH1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 1, 607432, Isolated cases; LIS1 (Lissencephaly due to LIS1 mutation) (PAFAH1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAFAH1B1	PAFAH1B1, LIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Miller-Dieker lissencephaly syndrome (MDLS) [MIM:247200]: A contiguous gene deletion syndrome of chromosome 17p13.3, characterized by classical lissencephaly and distinct facial features. Additional congenital malformations can be part of the condition. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lizensefali 1, 607432, Sporadik Vakalar; LIS1 (Lizensefali, LIS1 Mutasyonuna Bağlı) (Prenatal) (PAFAH1B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 1, 607432, Isolated cases; LIS1 (Lissencephaly due to LIS1 mutation) (PAFAH1B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAFAH1B1	PAFAH1B1, LIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Miller-Dieker lissencephaly syndrome (MDLS) [MIM:247200]: A contiguous gene deletion syndrome of chromosome 17p13.3, characterized by classical lissencephaly and distinct facial features. Additional congenital malformations can be part of the condition. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 2 (Norman Roberts Tipi), 257320, Otozomal Resesif; LIS2 (Lizensefali Sendromu, Norman Roberts Tipi) (RELN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 2 (Norman-Roberts type), 257320, Autosomal recessive; LIS2 (Lissencephaly syndrome, Norman-Roberts type) (RELN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RELN	RELN, RL, LIS2, ETL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly 2 (LIS2) [MIM:257320]: A classic type lissencephaly associated with ataxia, mental retardation, seizures and abnormalities of the cerebellum, hippocampus and brainstem. {ECO:0000269|PubMed:10973257}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lizensefali 2 (Norman Roberts Tipi), 257320, Otozomal Resesif; LIS2 (Lizensefali Sendromu, Norman Roberts Tipi) (Prenatal) (RELN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 2 (Norman-Roberts type), 257320, Autosomal recessive; LIS2 (Lissencephaly syndrome, Norman-Roberts type) (RELN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RELN	RELN, RL, LIS2, ETL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly 2 (LIS2) [MIM:257320]: A classic type lissencephaly associated with ataxia, mental retardation, seizures and abnormalities of the cerebellum, hippocampus and brainstem. {ECO:0000269|PubMed:10973257}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 3, 611603, Otozomal Dominant; LIS3 (Lizensefali, TUBA1A Mutasyonuna Bağlı) (TUBA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 3, 611603, Autosomal dominant; LIS3 (Lissencephaly due to TUBA1A mutation) (TUBA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBA1A	TUBA1A, TUBA3, LIS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly 3 (LIS3) [MIM:611603]: A classic type lissencephaly associated with psychomotor retardation and seizures. Features include agyria or pachygyria or laminar heterotopia, severe mental retardation, motor delay, variable presence of seizures, and abnormalities of corpus callosum, hippocampus, cerebellar vermis and brainstem. {ECO:0000269|PubMed:17584854}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lizensefali 3, 611603, Otozomal Dominant; LIS3 (Lizensefali, TUBA1A Mutasyonuna Bağlı) (Prenatal) (TUBA1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 3, 611603, Autosomal dominant; LIS3 (Lissencephaly due to TUBA1A mutation) (TUBA1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBA1A	TUBA1A, TUBA3, LIS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly 3 (LIS3) [MIM:611603]: A classic type lissencephaly associated with psychomotor retardation and seizures. Features include agyria or pachygyria or laminar heterotopia, severe mental retardation, motor delay, variable presence of seizures, and abnormalities of corpus callosum, hippocampus, cerebellar vermis and brainstem. {ECO:0000269|PubMed:17584854}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 4 (Mikrosefali), 614019, Otozomal Resesif; LIS4 (Mikrolizensefali) (NDE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 4 (with microcephaly), 614019, Autosomal recessive; LIS4 (Microlissencephaly) (NDE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDE1	NDE1, NUDE, LIS4, MHAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microhydranencephaly (MHAC) [MIM:605013]: A severe neurodevelopmental disorder characterized by microcephaly, severe motor and mental retardation, spasticity, and brain malformations that include gross dilation of the ventricles with complete absence of the cerebral hemispheres or severe delay in their development. {ECO:0000269|PubMed:22526350}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lizensefali 4 (Mikrosefali), 614019, Otozomal Resesif; LIS4 (Mikrolizensefali) (Prenatal) (NDE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 4 (with microcephaly), 614019, Autosomal recessive; LIS4 (Microlissencephaly) (NDE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDE1	NDE1, NUDE, LIS4, MHAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microhydranencephaly (MHAC) [MIM:605013]: A severe neurodevelopmental disorder characterized by microcephaly, severe motor and mental retardation, spasticity, and brain malformations that include gross dilation of the ventricles with complete absence of the cerebral hemispheres or severe delay in their development. {ECO:0000269|PubMed:22526350}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 5, 615191, Otozomal Resesif; LIS5 (Parke Taşı (Cobble Stone) Lizensefali-Kas Veya Göz Tutulumu Olmayan) (LAMB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 5, 615191, Autosomal recessive; LIS5 (Cobblestone lissencephaly without muscular or ocular involvement) (LAMB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB1	LAMB1, LIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly 5 (LIS5) [MIM:615191]: An autosomal recessive brain malformation characterized by cobblestone changes in the cortex, more severe in the posterior region, and subcortical band heterotopia. Affected individuals have hydrocephalus, seizures, and severely delayed psychomotor development. {ECO:0000269|PubMed:23472759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lizensefali 5, 615191, Otozomal Resesif; LIS5 (Parke Taşı (Cobble Stone) Lizensefali-Kas Veya Göz Tutulumu Olmayan) (Prenatal) (LAMB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 5, 615191, Autosomal recessive; LIS5 (Cobblestone lissencephaly without muscular or ocular involvement) (LAMB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMB1	LAMB1, LIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly 5 (LIS5) [MIM:615191]: An autosomal recessive brain malformation characterized by cobblestone changes in the cortex, more severe in the posterior region, and subcortical band heterotopia. Affected individuals have hydrocephalus, seizures, and severely delayed psychomotor development. {ECO:0000269|PubMed:23472759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lizensefali-Mikrosefali 6, 616212, Otozomal Resesif; LIS6 (Mikrolizensefali) (KATNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 6, with microcephaly, 616212, Autosomal recessive; LIS6 (Microlissencephaly) (KATNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KATNB1	KATNB1, LIS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly 6, with microcephaly (LIS6) [MIM:616212]: A form of lissencephaly, a disorder of cortical development characterized by agyria or pachygyria and disorganization of the clear neuronal lamination of normal six-layered cortex. LIS6 features include hypoplasia of the corpus callosum, severe microcephaly and developmental delay. {ECO:0000269|PubMed:25521378, ECO:0000269|PubMed:25521379}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lizensefali-Mikrosefali 6, 616212, Otozomal Resesif; LIS6 (Mikrolizensefali) (Prenatal) (KATNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 6, with microcephaly, 616212, Autosomal recessive; LIS6 (Microlissencephaly) (KATNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KATNB1	KATNB1, LIS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly 6, with microcephaly (LIS6) [MIM:616212]: A form of lissencephaly, a disorder of cortical development characterized by agyria or pachygyria and disorganization of the clear neuronal lamination of normal six-layered cortex. LIS6 features include hypoplasia of the corpus callosum, severe microcephaly and developmental delay. {ECO:0000269|PubMed:25521378, ECO:0000269|PubMed:25521379}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lissensefali 7, Serebellar Hipoplazi İle, 616342, Otozomal Resesif; LIS7 (CDK5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 7 with cerebellar hypoplasia, 616342, Autosomal recessive; LIS7 (CDK5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDK5	CDK5, LIS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly 7, with cerebellar hypoplasia (LIS7) [MIM:616342]: A form of lissencephaly, a disorder of cortical development characterized by agyria or pachygyria and disorganization of the clear neuronal lamination of normal six-layered cortex. LIS7 patients manifest lack of psychomotor development, facial dysmorphism, arthrogryposis, and early-onset intractable seizures resulting in death in infancy. {ECO:0000269	PubMed:25560765}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lissensefali 7, Serebellar Hipoplazi İle, 616342, Otozomal Resesif; LIS7 (Prenatal) (CDK5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 7 with cerebellar hypoplasia, 616342, Autosomal recessive; LIS7 (CDK5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDK5	CDK5, LIS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly 7, with cerebellar hypoplasia (LIS7) [MIM:616342]: A form of lissencephaly, a disorder of cortical development characterized by agyria or pachygyria and disorganization of the clear neuronal lamination of normal six-layered cortex. LIS7 patients manifest lack of psychomotor development, facial dysmorphism, arthrogryposis, and early-onset intractable seizures resulting in death in infancy. {ECO:0000269	PubMed:25560765}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali 8, 617255, Otozomal Resesif; LIS8 (TMTC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly 8, 617255, Autosomal recessive; LIS8 (TMTC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMTC3	TMTC3, SMILE, LIS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lizensefali 8, 617255, Otozomal Resesif; LIS8 (Prenatal) (TMTC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly 8, 617255, Autosomal recessive; LIS8 (TMTC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMTC3	TMTC3, SMILE, LIS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali panel (LIS1, DCX, POMT1, POMGnT1, FLNA) (MLPA)	Lissencephaly panel (LIS1, DCX, POMT1, POMGnT1, FLNA) (MLPA)	LIS1, DCX, POMT1, POMGnT1, FLNA	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Lizensefali panel (LIS1, DCX, POMT1, POMGnT1, FLNA) (MLPA) (Prenatal)	Lissencephaly panel (LIS1, DCX, POMT1, POMGnT1, FLNA) (MLPA) (Prenatal)	LIS1, DCX, POMT1, POMGnT1, FLNA	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali, X'e Bağlı 2, 300215, X'e Bağlı; LISX2 (X'e Bağlı Lizensefali-Anormal Genitalya) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly, X-linked 2, 300215, X-linked; LISX2 (X-linked lissencephaly with abnormal genitalia) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lizensefali, X'e Bağlı 2, 300215, X'e Bağlı; LISX2 (X'e Bağlı Lizensefali-Anormal Genitalya) (MLPA)	Lissencephaly, X-linked 2, 300215, X-linked; LISX2 (X-linked lissencephaly with abnormal genitalia) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Lizensefali, X'e Bağlı 2, 300215, X'e Bağlı; LISX2 (X'e Bağlı Lizensefali-Anormal Genitalya) (Prenatal) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly, X-linked 2, 300215, X-linked; LISX2 (X-linked lissencephaly with abnormal genitalia) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali, X'e Bağlı 2, 300215, X'e Bağlı; LISX2 (X'e Bağlı Lizensefali-Anormal Genitalya) (Prenatal) (MLPA)	Lissencephaly, X-linked 2, 300215, X-linked; LISX2 (X-linked lissencephaly with abnormal genitalia) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali, X'e Bağlı, 300067, X'e Bağlı; LISX1 (Lizensefali Tip 1, Doublecortin Gen Mutasyonuna Bağlı) (DCX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lissencephaly, X-linked, 300067, X-linked; LISX1 (Lissencephaly type 1 due to doublecortin gene mutation) (DCX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCX	DCX, DBCN, LISX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lizensefali, X'e Bağlı, 300067, X'e Bağlı; LISX1 (Lizensefali Tip 1, Doublecortin Gen Mutasyonuna Bağlı) (MLPA)	Lissencephaly, X-linked, 300067, X-linked; LISX1 (Lissencephaly type 1 due to doublecortin gene mutation) (MLPA)	DCX	DCX, DBCN, LISX	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lizensefali, X'e Bağlı, 300067, X'e Bağlı; LISX1 (Lizensefali Tip 1, Doublecortin Gen Mutasyonuna Bağlı) (Prenatal) (DCX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lissencephaly, X-linked, 300067, X-linked; LISX1 (Lissencephaly type 1 due to doublecortin gene mutation) (DCX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCX	DCX, DBCN, LISX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizensefali, X'e Bağlı, 300067, X'e Bağlı; LISX1 (Lizensefali Tip 1, Doublecortin Gen Mutasyonuna Bağlı) (Prenatal) (MLPA)	Lissencephaly, X-linked, 300067, X-linked; LISX1 (Lissencephaly type 1 due to doublecortin gene mutation) (Prenatal) (MLPA)	DCX	DCX, DBCN, LISX	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karaciğer Yetmezliği, Geçici İnfantil, 613070, Otozomal Resesif; LFIT (Akut İnfantil Karaciğer Yetmezliği-Mtdna Kodlanmış Proteinlerin Sentez Defektine Bağlı) (TRMU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Liver failure, transient infantile, 613070, Autosomal recessive; LFIT (Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins) (TRMU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRMU	TRMU, MTO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Liver failure, infantile, transient (LFIT) [MIM:613070]: A transient disorder of hepatic function characterized by elevated liver enzymes, jaundice, vomiting, coagulopathy, hyperbilirubinemia, increased serum lactate. Patients who survive the initial acute episode can recover, show normal development and have no recurrence. {ECO:0000269	PubMed:19732863}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Karaciğer Yetmezliği, Geçici İnfantil, 613070, Otozomal Resesif, LFIT (Akut İnfantil Karaciğer Yetmezliği-Mitina Kodlanmış Proteinlerin Sentez Defektine Bağlı) (Prenatal) (TRMU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Liver failure, transient infantile, 613070, Autosomal recessive; LFIT (Acute infantile liver failure due to synthesis defect of mtDNA-encoded proteins) (TRMU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRMU	TRMU, MTO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Liver failure, infantile, transient (LFIT) [MIM:613070]: A transient disorder of hepatic function characterized by elevated liver enzymes, jaundice, vomiting, coagulopathy, hyperbilirubinemia, increased serum lactate. Patients who survive the initial acute episode can recover, show normal development and have no recurrence. {ECO:0000269	PubMed:19732863}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Loeys-Dietz Sendromu 1, 609192, Otozomal Dominant; LDS1 (Loeys-Dietz Sendromu) (TGFBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Loeys-Dietz syndrome 1, 609192, Autosomal dominant; LDS1 (Loeys-Dietz syndrome) (TGFBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBR1	TGFBR1, ALK5, AAT5, LDS1, MSSE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple self-healing squamous epithelioma (MSSE) [MIM:132800]: A disorder characterized by multiple skin tumors that undergo spontaneous regression. Tumors appear most often on sun-exposed regions, are locally invasive, and undergo spontaneous resolution over a period of months leaving pitted scars. {ECO:0000269	PubMed:21358634}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Loeys-Dietz Sendromu 1, 609192, Otozomal Dominant; LDS1 (Loeys-Dietz Sendromu) (Prenatal) (TGFBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Loeys-Dietz syndrome 1, 609192, Autosomal dominant; LDS1 (Loeys-Dietz syndrome) (TGFBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGFBR1	TGFBR1, ALK5, AAT5, LDS1, MSSE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple self-healing squamous epithelioma (MSSE) [MIM:132800]: A disorder characterized by multiple skin tumors that undergo spontaneous regression. Tumors appear most often on sun-exposed regions, are locally invasive, and undergo spontaneous resolution over a period of months leaving pitted scars. {ECO:0000269	PubMed:21358634}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Loeys-Dietz Sendromu 2, 610168, Otozomal Dominant; LDS2 (Marfan Sendromu Tip 2) (TGFBR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Loeys-Dietz syndrome 2, 610168, Autosomal dominant; LDS2 (Marfan syndrome type 2) (TGFBR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBR2	TGFBR2, HNPCC6, AAT3, MFS2, LDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10789724}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Loeys-Dietz Sendromu 2, 610168, Otozomal Dominant; LDS2 (Marfan Sendromu Tip 2) (Prenatal) (TGFBR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Loeys-Dietz syndrome 2, 610168, Autosomal dominant; LDS2 (Marfan syndrome type 2) (TGFBR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGFBR2	TGFBR2, HNPCC6, AAT3, MFS2, LDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269	PubMed:10789724}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Loeys-Dietz Sendromu 3, 613795, Otozomal Dominant; LDS3 (Anevrizma-Osteoartrit Sendromu) (SMAD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Loeys-Dietz syndrome 3, 613795, Autosomal dominant; LDS3 (Aneurysm-osteoarthritis syndrome) (SMAD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD3	SMAD3, MADH3, LDS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Loeys-Dietz Sendromu 3, 613795, Otozomal Dominant; LDS3 (Anevrizma-Osteoartrit Sendromu) (Prenatal) (SMAD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Loeys-Dietz syndrome 3, 613795, Autosomal dominant; LDS3 (Aneurysm-osteoarthritis syndrome) (SMAD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMAD3	SMAD3, MADH3, LDS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Loeys-Dietz Sendromu 4, 614816, Otozomal Dominant; LDS4 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (TGFB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Loeys-Dietz syndrome 4, 614816, Autosomal dominant; LDS4 (Familial thoracic aortic aneurysm and aortic dissection) (TGFB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFB2	TGFB2, LDS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Loeys-Dietz syndrome 4 (LDS4) [MIM:614816]: An aortic aneurysm syndrome with widespread systemic involvement. LDS4 is characterized by arterial tortuosity, aortic dissection, intracranial aneurysm and subarachnoid hemorrhage, hypertelorism, bifid uvula, pectus deformity, bicuspid aortic valve, arachnodactyly, scoliosis, foot deformities, dural ectasia, joint hyperflexibility, and thin skin with easy bruising and striae. {ECO:0000269	PubMed:22772368}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Loeys-Dietz Sendromu 4, 614816, Otozomal Dominant; LDS4 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (Prenatal) (TGFB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Loeys-Dietz syndrome 4, 614816, Autosomal dominant; LDS4 (Familial thoracic aortic aneurysm and aortic dissection) (TGFB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGFB2	TGFB2, LDS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Loeys-Dietz syndrome 4 (LDS4) [MIM:614816]: An aortic aneurysm syndrome with widespread systemic involvement. LDS4 is characterized by arterial tortuosity, aortic dissection, intracranial aneurysm and subarachnoid hemorrhage, hypertelorism, bifid uvula, pectus deformity, bicuspid aortic valve, arachnodactyly, scoliosis, foot deformities, dural ectasia, joint hyperflexibility, and thin skin with easy bruising and striae. {ECO:0000269	PubMed:22772368}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Loeys-Dietz Sendromu 5, 615582, Otozomal Dominant; LDS5 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (TGFB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Loeys-Dietz syndrome 5, 615582, Autosomal dominant; LDS5 (Familial thoracic aortic aneurysm and aortic dissection) (TGFB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFB3	TGFB3, ARVD1, RNHF, LDS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 1 (ARVD1) [MIM:107970]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. {ECO:0000269	PubMed:15639475}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Loeys-Dietz Sendromu 5, 615582, Otozomal Dominant; LDS5 (Ailesel Torakal Aort Anevrizması ve Aort Diseksiyonu) (Prenatal) (TGFB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Loeys-Dietz syndrome 5, 615582, Autosomal dominant; LDS5 (Familial thoracic aortic aneurysm and aortic dissection) (TGFB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGFB3	TGFB3, ARVD1, RNHF, LDS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arrhythmogenic right ventricular dysplasia, familial, 1 (ARVD1) [MIM:107970]: A congenital heart disease characterized by infiltration of adipose and fibrous tissue into the right ventricle and loss of myocardial cells, resulting in ventricular and supraventricular arrhythmias. {ECO:0000269	PubMed:15639475}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Uzun QT Sendromu 1, 192500, Otozomal Dominant; LQT1 (Ailesel Uzun QT Sendromu) (KCNQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 1, 192500, Autosomal dominant; LQT1 (Familial long QT syndrome) (KCNQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB3, SQT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:18711366, ECO:0000269|PubMed:18711367, ECO:0000269|PubMed:24390345}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu 1, Kazanılmış, Yatkınlık, 192500, Otozomal Dominant; LQT1 (Ailesel Uzun QT Sendromu) (KCNQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 1, acquired, susceptibility to, 192500, Autosomal dominant; LQT1 (Familial long QT syndrome) (KCNQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB3, SQT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:18711366, ECO:0000269|PubMed:18711367, ECO:0000269|PubMed:24390345}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu 12, 612955, Otozomal Dominant; LQT12 (Ailesel Uzun QT Sendromu) (SNTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 12, 612955, Autosomal dominant; LQT12 (Familial long QT syndrome) (SNTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNTA1	SNTA1, SNT1, TACIP1, LQT12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 12 (LQT12) [MIM:612955]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. {ECO:0000269|PubMed:18591664, ECO:0000269|PubMed:19684871}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Uzun QT Sendromu 13, 613485, Otozomal Dominant; LQT13 (Ailesel Uzun QT Sendromu) (KCNJ5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 13, 613485, Autosomal dominant; LQT13 (Familial long QT syndrome) (KCNJ5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ5	KCNJ5, GIRK4, KATP1, LQT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 3 (FH3) [MIM:613677]: A form of hyperaldosteronism characterized by hypertension secondary to massive adrenal mineralocorticoid production. Like patients with familial hyperaldosteronism type 1 (glucocorticoid-remediable aldosteronism), patients with FH3 present with childhood hypertension, elevated aldosteronism levels, and high levels of the hybrid steroids 18-oxocortisol and 18-hydroxycortisol. However, hypertension and aldosteronism are not reversed by administration of exogenous glucocorticoids and patients require adrenalectomy to control hypertension. {ECO:0000269|PubMed:21311022, ECO:0000269|PubMed:22203740, ECO:0000269|PubMed:22275527, ECO:0000269|PubMed:22308486, ECO:0000269|PubMed:22848660}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu 14, 616247, Otozomal Dominant; LQT14 (Ailesel Uzun QT Sendromu) (CALM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 14, 616247, Autosomal dominant; LQT14 (Familial long QT syndrome) (CALM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALM1	CALM1, PHKD, CPVT4, LQT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ventricular tachycardia, catecholaminergic polymorphic, 4 (CPVT4) [MIM:614916]: An arrhythmogenic disorder characterized by stress-induced, bidirectional ventricular tachycardia that may degenerate into cardiac arrest and cause sudden death. Patients present with recurrent syncope, seizures, or sudden death after physical activity or emotional stress. {ECO:0000269	PubMed:23040497}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu 15, 616249, Otozomal Dominant; LQT15 (Ailesel Uzun QT Sendromu) (CALM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 15, 616249, Autosomal dominant; LQT15 (Familial long QT syndrome) (CALM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALM2	CALM2, LQT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ventricular tachycardia, catecholaminergic polymorphic, 4 (CPVT4) [MIM:614916]: An arrhythmogenic disorder characterized by stress-induced, bidirectional ventricular tachycardia that may degenerate into cardiac arrest and cause sudden death. Patients present with recurrent syncope, seizures, or sudden death after physical activity or emotional stress. {ECO:0000269	PubMed:23040497}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Uzun QT Sendromu 2, 613688, Otozomal Dominant; LQT2 (Ailesel Uzun QT Sendromu) (KCNH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 2, 613688, Autosomal dominant; LQT2 (Familial long QT syndrome) (KCNH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNH2	KCNH2, LQT2, HERG, SQT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. (ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu 2, 613688, Otozomal Dominant; LQT2 (Ailesel Uzun QT Sendromu) (MLPA)	Long QT syndrome 2, 613688, Autosomal dominant; LQT2 (Familial long QT syndrome) (MLPA)	KCNH2	KCNH2, LQT2, HERG, SQT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. (ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu 2, Kazanılmış, Yatkınlık, 613688, Otozomal Dominant; LQT2 (Ailesel Uzun QT Sendromu) (KCNH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 2, acquired, susceptibility to, 613688, Autosomal dominant; LQT2 (Familial long QT syndrome) (KCNH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNH2	KCNH2, LQT2, HERG, SQT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. (ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu 2, Kazanılmış, Yatkınlık, 613688, Otozomal Dominant; LQT2 (Ailesel Uzun QT Sendromu) (MLPA)	Long QT syndrome 2, acquired, susceptibility to, 613688, Autosomal dominant; LQT2 (Familial long QT syndrome) (MLPA)	KCNH2	KCNH2, LQT2, HERG, SQT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. (ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu 4, 600919, Otozomal Dominant (Ailesel Uzun QT Sendromu) (ANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 4, 600919, Autosomal dominant (Familial long QT syndrome) (ANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANK2	ANK2, LQT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 4 (LQT4) [MIM:600919]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 4 shows many atypical features compared to classical long QT syndromes, including pronounced sinus bradycardia, polyphasic T waves and atrial fibrillation. Cardiac repolarization defects may be not as severe as in classical LQT syndromes and prolonged QT interval on EKG is not a consistent feature. (ECO:0000269|PubMed:12571597, ECO:0000269|PubMed:15178757). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu 5, 613695, Otozomal Dominant; LQT5 (Ailesel Uzun QT Sendromu) (KCNE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 5, 613695, Autosomal dominant; LQT5 (Familial long QT syndrome) (KCNE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNE1	KCNE1, JLNS, LQT5, JLNS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Jervell and Lange-Nielsen syndrome 2 (JLNS2) [MIM:612347]: An autosomal recessive disorder characterized by congenital deafness, prolongation of the QT interval, syncopal attacks due to ventricular arrhythmias, and a high risk of sudden death. (ECO:0000269|PubMed:10400998, ECO:0000269|PubMed:9328483, ECO:0000269|PubMed:9354783). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu 6, 613693, Otozomal Dominant; LQT6 (Ailesel Uzun QT Sendromu) (KCNE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 6, 613693, Autosomal dominant; LQT6 (Familial long QT syndrome) (KCNE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNE2	KCNE2, MIRP1, LQT6, ATFB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial fibrillation, familial, 4 (ATFB4) [MIM:611493]: A familial form of atrial fibrillation, a common sustained cardiac rhythm disturbance. Atrial fibrillation is characterized by disorganized atrial electrical activity and ineffective atrial contraction promoting blood stasis in the atria and reduces ventricular filling. It can result in palpitations, syncope, thromboembolic stroke, and congestive heart failure. (ECO:0000269	PubMed:15368194). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Long QT Sendromu 9, 611818; LQT9 (Ailesel Uzun QT Sendromu) (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome 9, 611818; LQT9 (Familial long QT syndrome) (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Long QT Sendromu 9, 611818; LQT9 (Ailesel Uzun QT Sendromu) (MLPA)	Long QT syndrome 9, 611818; LQT9 (Familial long QT syndrome) (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu-10, 611819, Otozomal Dominant; LQT10 (Ailesel Uzun QT Sendromu) (SCN4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome-10, 611819, Autosomal dominant; LQT10 (Familial long QT syndrome) (SCN4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4B	SCN4B, LQT10, ATFB17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 10 (LQT10) [MIM:611819]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. (ECO:0000269	PubMed:17592081). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Uzun QT Sendromu-11, 611820; LQT11 (Ailesel Uzun QT Sendromu) (AKAP9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome-11, 611820; LQT11 (Familial long QT syndrome) (AKAP9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKAP9	AKAP9, YOTIAO, AKAP450	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 11 (LQT11) [MIM:611820]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. (ECO:0000269	PubMed:18093912). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Uzun QT Sendromu-3, 603830, Otozomal Dominant; LQT3; PFHB1A (Ailesel Uzun QT Sendromu) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome-3, 603830, Autosomal dominant; LQT3; PFHB1A (Familial long QT syndrome) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial standstill 1 (ATRST1) [MIM:108770]: A rare arrhythmia characterized by the absence of electrical and mechanical activity in the atria. Electrocardiographically, it is characterized by bradycardia, the absence of P waves, and a junctional narrow complex escape rhythm. (ECO:0000269	PubMed:12522116). Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry. A mutation in SCN5A has been detected in combination with a rare GJA5 genotype in a large family with atrial standstill.	EDTA Blood Sample
Uzun QT Sendromu, Kazanılmış, Azalmış Yatkınlık, 613688, Otozomal Dominant (Ailesel Uzun QT Sendromu) (ALG10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long QT syndrome, acquired, reduced susceptibility to, 613688, Autosomal dominant (Familial long QT syndrome) (ALG10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG10	ALG10, KCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği (Uzun Zincirli 3-Hidroksiaçil-CoA Dehidrojenaz Eksikliği) (HADHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. (ECO:0000269	PubMed:9739053). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Açil Koa Dehidrojenaz, Uzun Zincirli, Eksikliği, Otozomal Resesif (LCAD Eksikliği) (ACADL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Long-chain acyl-CoA dehydrogenase deficiency, Autosomal Recessive (LCAD deficiency) (ACADL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACADL	ACADL, LCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acyl-CoA dehydrogenase very long-chain deficiency (ACADVLD) [MIM:201475]: An inborn error of mitochondrial fatty acid beta-oxidation which leads to impaired long-chain fatty acid beta-oxidation. It is clinically heterogeneous, with three major phenotypes: a severe childhood form characterized by early onset, high mortality and high incidence of cardiomyopathy; a milder childhood form with later onset, characterized by hypoketotic hypoglycemia, low mortality and rare cardiomyopathy; an adult form, with isolated skeletal muscle involvement, rhabdomyolysis and myoglobinuria, usually triggered by exercise or fasting. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Açil Koa Dehidrojenaz, Uzun Zincirli, Eksikliği, Otozomal Resesif (LCAD Eksikliği) (Prenatal) (ACADL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Long-chain acyl-CoA dehydrogenase deficiency, Autosomal Recessive (LCAD deficiency) (ACADL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACADL	ACADL, LCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Acyl-CoA dehydrogenase very long-chain deficiency (ACADVLD) [MIM:201475]: An inborn error of mitochondrial fatty acid beta-oxidation which leads to impaired long-chain fatty acid beta-oxidation. It is clinically heterogeneous, with three major phenotypes: a severe childhood form characterized by early onset, high mortality and high incidence of cardiomyopathy; a milder childhood form with later onset, characterized by hypoketotic hypoglycemia, low mortality and rare cardiomyopathy; an adult form, with isolated skeletal muscle involvement, rhabdomyolysis and myoglobinuria, usually triggered by exercise or fasting. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Düşük Yoğunluklu Lipoprotein Kolesterol Seviyesi QTL 1, 603776 (PCSK9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Low density lipoprotein cholesterol level QTL 1, 603776 (PCSK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCSK9	PCSK9, NARC1, HCHOLA3, FH3, LDLCQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypercholesterolemia, autosomal dominant, 3 (HCHOLA3) [MIM:603776]: A familial condition characterized by elevated circulating cholesterol contained in either low-density lipoproteins alone or also in very-low-density lipoproteins. (ECO:0000269|PubMed:12730697, ECO:0000269|PubMed:18799458, ECO:0000269|PubMed:24808179). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Düşük Renin Hipertansiyon, Yatkınlık (CYP11B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Low renin hypertension, susceptibility to (CYP11B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP11B2	CYP11B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperaldosteronism 1 (FH1) [MIM:103900]: A disorder characterized by hypertension, variable hyperaldosteronism, and abnormal adrenal steroid production, including 18-oxocortisol and 18-hydroxycortisol. There is significant phenotypic heterogeneity, and some individuals never develop hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry. The molecular defect causing hyperaldosteronism familial 1 is an anti-Lepore-type fusion of the CYP11B1 and CYP11B2 genes. The hybrid gene has the promoting part of CYP11B1, ACTH-sensitive, and the coding part of CYP11B2.;	EDTA Blood Sample

Lowe Sendromu, 309000, X'e Bağlı Resesif; OCRL (Lowe Okuloserebrorenal Sendromu) (OCRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lowe syndrome, 309000, X-linked recessive; OCRL (Oculocerebrorenal syndrome of Lowe) (OCRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OCRL	OCRL, LOCR, OCRL1, NPHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dent disease 2 (DD2) [MIM:300555]: A renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Characteristic abnormalities include low-molecular-weight proteinuria and other features of Fanconi syndrome, such as glycosuria, aminoaciduria, and phosphaturia, but typically do not include proximal renal tubular acidosis. Progressive renal failure is common, as are nephrocalcinosis and kidney stones. {ECO:0000269|PubMed:15627218, ECO:0000269|PubMed:17384968, ECO:0000269|PubMed:21031565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lowe Sendromu, 309000, X'e Bağlı Resesif; OCRL (Lowe Okuloserebrorenal Sendromu) (Prenatal) (OCRL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lowe syndrome, 309000, X-linked recessive; OCRL (Oculocerebrorenal syndrome of Lowe) (OCRL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OCRL	OCRL, LOCR, OCRL1, NPHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dent disease 2 (DD2) [MIM:300555]: A renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Characteristic abnormalities include low-molecular-weight proteinuria and other features of Fanconi syndrome, such as glycosuria, aminoaciduria, and phosphaturia, but typically do not include proximal renal tubular acidosis. Progressive renal failure is common, as are nephrocalcinosis and kidney stones. {ECO:0000269|PubMed:15627218, ECO:0000269|PubMed:17384968, ECO:0000269|PubMed:21031565}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lubs X'e Bağlı Mental Retardasyon Sendromu; Mrxsl (Trizomi Xq28) (2200)	LUBS X-LINKED MENTAL RETARDATION SYNDROME; MRXSL (Trisomy Xq28) (440)	Array-CGH	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Lubs X'e Bağlı Mental Retardasyon Sendromu; Mrxsl (Trizomi Xq28) (Prenatal)	LUBS X-LINKED MENTAL RETARDATION SYNDROME; MRXSL (Trisomy Xq28) (Prenatal)	Array-CGH	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Luján-Fryns Sendromu, 309520, X'e Bağlı (X'e Bağlı Mental Retardasyon- Marfanoid Habitus) (MED12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Luján-Fryns syndrome, 309520, X-linked recessive (X-linked intellectual disability with marfanoid habitus) (MED12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269|PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Luján-Fryns Sendromu, 309520, X'e Bağlı (X'e Bağlı Mental Retardasyon- Marfanoid Habitus) (Prenatal) (MED12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Luján-Fryns syndrome, 309520, X-linked recessive (X-linked intellectual disability with marfanoid habitus) (MED12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269|PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lomber Disk Dejenerasyonu, 603932 (ASPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lumbar disc degeneration, 603932 (ASPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPN	ASPN, PLAP1, OS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intervertebral disc disease (IDD) [MIM:603932]: A common musculo-skeletal disorder caused by degeneration of intervertebral disks of the lumbar spine. It results in low-back pain and unilateral leg pain. {ECO:0000269|PubMed:18304494}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to intervertebral disk disease, particularly lumbar disk degeneration, is conferred by a triplet repeat expansion polymorphism. ASPN allele having 14 aspartic acid repeats in the N-terminal region of the protein (D14), is associated with the disorder in some populations (PubMed:18304494). {ECO:0000269|PubMed:18304494}.;	EDTA Blood Sample
Lomber Disk Hastalığı, Yatkınlık, 603932 (CILP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lumbar disc disease, susceptibility to, 603932 (CILP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CILP	CILP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intervertebral disc disease (IDD) [MIM:603932]: A common musculo-skeletal disorder caused by degeneration of intervertebral disks of the lumbar spine. It results in low-back pain and unilateral leg pain. {ECO:0000269|PubMed:15864306}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:15864306}.;	EDTA Blood Sample
Brakidaktili-Sindaktili Sendromu, 610713 (Brakidaktili-Sindaktili, Zhao Tipi) (THBS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lumbar disc herniation, susceptibility to, 603932 (Brachydactyly-syndactyly, Zhao type) (THBS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THBS2	THBS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Brakidaktili-Sindaktili Sendromu, 610713 (3 (Brakidaktili-Sindaktili, Zhao Tipi)) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lumbar disc herniation, susceptibility to, 603932 (Brachydactyly-syndactyly, Zhao type) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marshall syndrome (MRSHS) [MIM:154780]: An autosomal dominant disorder characterized by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia. Clinical features include short stature; flat or retruded midface with short, depressed nose, flat nasal bridge and anteverted nares; cleft palate with or without the Pierre Robin sequence; appearance of large eyes with ocular hypertelorism; cataracts, either congenital or juvenile; esotropia; high myopia; sensorineural hearing loss; spondyloepiphyseal abnormalities; calcification of the falx cerebri; ectodermal abnormalities, including defects in sweating and dental structures. {ECO:0000269|PubMed:10486316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akciğer Kanserine Yatkınlık 2, 612052 (CHRNA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer susceptibility 2, 612052 (CHRNA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA5	CHRNA5, LNCR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Kanserine Yatkınlık 2, 612052 (CHRNA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer susceptibility 2, 612052 (CHRNA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA3	CHRNA3, LNCR2, PAOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Kanseri, 211980, Otozomal Resesif (RASSF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, 211980, Autosomal recessive (RASSF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RASSF1	RASSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Kanseri, 211980, Otozomal Resesif (PPP2R1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, 211980, Autosomal recessive (PPP2R1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP2R1B	PPP2R1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Kanseri, 211980, Otozomal Resesif (Özofageal Skuamoz Hücre Karsinması) (DLEC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, 211980, Autosomal recessive (Squamous cell carcinoma of esophagus) (DLEC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLEC1	DLEC1, DLC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:10213508}. Note=The gene represented in this entry may be involved in disease pathogenesis. DLEC1 silencing due to promoter methylation and aberrant transcription may be implicated in the development of esophageal cancer.;	EDTA Blood Sample
Akciğer Kanseri, Karşı Koruma, 211980, Otozomal Resesif (CASP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, protection against, 211980, Autosomal recessive (CASP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP8	CASP8, MCH5, ALPS2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caspase-8 deficiency (CASP8D) [MIM:607271]: Disorder resembling autoimmune lymphoproliferative syndrome (ALPS). It is characterized by lymphadenopathy, splenomegaly, and defective CD95-induced apoptosis of peripheral blood lymphocytes (PBLs). It leads to defects in activation of T-lymphocytes, B-lymphocytes, and natural killer cells leading to immunodeficiency characterized by recurrent sinopulmonary and herpes simplex virus infections and poor responses to immunization. {ECO:0000269|PubMed:12353035}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akciğer Kanseri, Karşı Koruma, Sigara İçenlerde (MPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, protection against, in smokers (MPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPO	MPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloperoxidase deficiency (MPOD) [MIM:254600]: A disorder characterized by decreased myeloperoxidase activity in neutrophils and monocytes that results in disseminated candidiasis. {ECO:0000269|PubMed:7904599, ECO:0000269|PubMed:8142659, ECO:0000269|PubMed:9354683, ECO:0000269|PubMed:9637725}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akciğer Kanseri, Direnç, 211980, Otozomal Resesif (CYP2A6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, resistance to, 211980, Autosomal recessive (CYP2A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Kanseri, Direnç, 211980, Otozomal Resesif (MLPA)	Lung cancer, resistance to, 211980, Autosomal recessive (MLPA)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Akciğer Kanseri, Somatik, 211980 (SLC22A18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, somatic, 211980 (SLC22A18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A18	SLC22A1L, BWSCR1A, IMPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Rhabdomyosarcoma, embryonal, 1 (RMSE1) [MIM:268210]: A form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines. Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Akciğer Kanseri, Somatik, 211980 (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, somatic, 211980 (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Akciğer Kanseri, Somatik, 211980 (MAP3K8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, somatic, 211980 (MAP3K8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP3K8	MAP3K8, COT, EST, TPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Akciğer Kanseri, Yatkınlık, 211980, Otozomal Resesif (FASLG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, susceptibility to, 211980, Autosomal recessive (FASLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FASLG	FASLG, TNFSF6, APT1LG1, FASL, ALPS1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 1B (ALPS1B) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269	PubMed:8787672}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akciğer Kanseri, Yatkınlık, 211980, Otozomal Resesif (ERCC6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lung cancer, susceptibility to, 211980, Autosomal recessive (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMD5, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years of life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:9443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akciğer Hastalıkları, İmmün Yetmezlik, ve Kromozom Kırılmasi Sendromu, 617241, Otozomal Resesif (NSMCE3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241, Autosomal recessive (NSMCE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSMCE3	NSMCE3, NDNL2, MAGEG1, LICS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akciğer Hastalıkları, İmmün Yetmezlik, ve Kromozom Kırılmasi Sendromu, 617241, Otozomal Resesif (Prenatal) (NSMCE3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Lung disease, immunodeficiency, and chromosome breakage syndrome, 617241, Autosomal recessive (NSMCE3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSMCE3	NSMCE3, NDNL2, MAGEG1, LICS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lupus Nefriti, Yatkınlık, 152700, Otozomal Dominant (FCGR2A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lupus nephritis, susceptibility to, 152700, Autosomal dominant (FCGR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2A	FCGR2A, IGFR2, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Luscan-Lumish Sendromu, 616831, Otozomal Dominant (SETD2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Luscan-Lumish syndrome, 616831, Autosomal dominant (SETD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETD2	SETD2, SET2, HYPB, HBP231, KIAA1732, LLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. {ECO:0000269|PubMed:20054297, ECO:0000269|PubMed:23622243, ECO:0000269|PubMed:23792563}. Note=The disease may be caused by mutations affecting the gene represented in this entry. Defects of SETD2 are associated with loss of DNA methylation at non-promoter regions (PubMed:23792563). {ECO:0000269|PubMed:23792563}.;	EDTA Blood Sample
Luscan-Lumish Sendromu, 616831, Otozomal Dominant (Prenatal) (SETD2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Luscan-Lumish syndrome, 616831, Autosomal dominant (SETD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SETD2	SETD2, SET2, HYPB, HBP231, KIAA1732, LLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. {ECO:0000269|PubMed:20054297, ECO:0000269|PubMed:23622243, ECO:0000269|PubMed:23792563}. Note=The disease may be caused by mutations affecting the gene represented in this entry. Defects of SETD2 are associated with loss of DNA methylation at non-promoter regions (PubMed:23792563). {ECO:0000269|PubMed:23792563}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Luteinize Edici Hormon Direnci, Kadın, 238320, Otozomal Resesif (Leydig Hücre Hipoplazisi) (LHCGR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Luteinizing hormone resistance, female, 238320, Autosomal recessive (Leydig cell hypoplasia) (LHCGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHCGR	LHCGR, LHR, LCGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Luteinizing hormone resistance (LHR) [MIM:238320]: An autosomal recessive disorder characterized by unresponsiveness to luteinizing hormone, defective sexual development in males, and defective follicular development and ovulation, amenorrhea and infertility in females. Two forms of the disorder have been defined in males. Type 1 is a severe form characterized by complete 46,XY male pseudohermaphroditism, low testosterone and high luteinizing hormone levels, total lack of responsiveness to luteinizing and chorionic gonadotropin hormones, lack of breast development, and absent development of secondary male sex characteristics. Type 2, a milder form, displays a broader range of phenotypic expression ranging from micropenis to severe hypospadias. {ECO:0000269|PubMed:12050206, ECO:0000269|PubMed:15372531, ECO:0000269|PubMed:15472221, ECO:0000269|PubMed:19551906, ECO:0000269|PubMed:7719343, ECO:0000269|PubMed:8559304, ECO:0000269|PubMed:9215288, ECO:0000269|PubMed:9514160, ECO:0000269|PubMed:9626144, ECO:0000269|PubMed:9626653}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfanjiyoleyomiyomatozis, 606690; LAM (Lenfanjiyoleyomiyomatozis) (TSC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphangioleiomyomatosis, 606690; LAM (Lymphangioleiomyomatosis) (TSC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSC1	TSC1, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. {ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lenfanjiyoleyomiyomatozis, 606690; LAM (Lenfanjiyoleyomiyomatozis) (MLPA)	Lymphangioleiomyomatosis, 606690; LAM (Lymphangioleiomyomatosis) (MLPA)	TSC1	TSC1, LAM	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. {ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lenfanjiyoleyomiyomatozis, Somatik, 606690 (Lenfanjiyoleyomiyomatozis) (TSC2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphangioleiomyomatosis, somatic, 606690 (Lymphangioleiomyomatosis) (TSC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSC2	TSC2, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Lymphangioleiomyomatosis (LAM) [MIM:606690]: Progressive and often fatal lung disease characterized by a diffuse proliferation of abnormal smooth muscle cells in the lungs. It affects almost exclusively young women and can occur as an isolated disorder or in association with tuberous sclerosis complex. {ECO:0000269	PubMed:10823953}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample/ Biopsy of affected site-transport media
Lenfödem-Distikiazis-Renal Hastalık ve Diyabetes Mellitus Sendromu, 153400, Otozomal Dominant (Lenfödem-Distikiasis Sendromu) (FOXC2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400, Autosomal dominant (Lymphedema-distichiasis syndrome) (FOXC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXC2	FOXC2, FKHL14, MFH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema-distichiasis (LYD) [MIM:153400]: A disorder characterized by primary limb lymphedema associated with distichiasis (double rows of eyelashes, with extra eyelashes growing from the Meibomian gland orifices). Swelling of the extremities, due to altered lymphatic flow, usually appears in late childhood or puberty. Most affected individuals have ocular findings including corneal irritation, recurrent conjunctivitis, and photophobia. Drooping of the upper eyelid (ptosis) is a variable feature of the lymphedema-distichiasis syndrome, occurring in about 30% of patients. {ECO:0000269|PubMed:11078474, ECO:0000269|PubMed:11371511, ECO:0000269|PubMed:11499682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfödem-Distikiazis-Renal Hastalık ve Diyabetes Mellitus Sendromu, 153400, Otozomal Dominant (Lenfödem-Distikiasis Sendromu) (Prenatal) (FOXC2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400, Autosomal dominant (Lymphedema-distichiasis syndrome) (FOXC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXC2	FOXC2, FKHL14, MFH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema-distichiasis (LYD) [MIM:153400]: A disorder characterized by primary limb lymphedema associated with distichiasis (double rows of eyelashes, with extra eyelashes growing from the Meibomian gland orifices). Swelling of the extremities, due to altered lymphatic flow, usually appears in late childhood or puberty. Most affected individuals have ocular findings including corneal irritation, recurrent conjunctivitis, and photophobia. Drooping of the upper eyelid (ptosis) is a variable feature of the lymphedema-distichiasis syndrome, occurring in about 30% of patients. {ECO:0000269|PubMed:11078474, ECO:0000269|PubMed:11371511, ECO:0000269|PubMed:11499682}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lenfödem-Distikiazis, 153400, Otozomal Dominant (Lenfödem-Distikiasis Sendromu) (FOXC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema-distichiasis syndrome, 153400, Autosomal dominant (Lymphedema-distichiasis syndrome) (FOXC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXC2	FOXC2, FKHL14, MFH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema-distichiasis (LYD) [MIM:153400]: A disorder characterized by primary limb lymphedema associated with distichiasis (double rows of eyelashes, with extra eyelashes growing from the Meibomian gland orifices). Swelling of the extremities, due to altered lymphatic flow, usually appears in late childhood or puberty. Most affected individuals have ocular findings including corneal irritation, recurrent conjunctivitis, and photophobia. Drooping of the upper eyelid (ptosis) is a variable feature of the lymphedema-distichiasis syndrome, occurring in about 30% of patients. {ECO:0000269|PubMed:11078474, ECO:0000269|PubMed:11371511, ECO:0000269|PubMed:11496862}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfödem-Distikiazis, 153400, Otozomal Dominant (Lenfödem-Distikiasis Sendromu) (Prenatal) (FOXC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema-distichiasis syndrome, 153400, Autosomal dominant (Lymphedema-distichiasis syndrome) (FOXC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXC2	FOXC2, FKHL14, MFH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema-distichiasis (LYD) [MIM:153400]: A disorder characterized by primary limb lymphedema associated with distichiasis (double rows of eyelashes, with extra eyelashes growing from the Meibomian gland orifices). Swelling of the extremities, due to altered lymphatic flow, usually appears in late childhood or puberty. Most affected individuals have ocular findings including corneal irritation, recurrent conjunctivitis, and photophobia. Drooping of the upper eyelid (ptosis) is a variable feature of the lymphedema-distichiasis syndrome, occurring in about 30% of patients. {ECO:0000269|PubMed:11078474, ECO:0000269|PubMed:11371511, ECO:0000269|PubMed:11496862}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfödem, Kalıtsal, IA, 153100, Otozomal Dominant; LMPH1A (Milroy Hastalığı) (FLT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema, hereditary, IA, 153100, Autosomal dominant; LMPH1A (Milroy disease) (FLT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLT4	FLT4, VEGFR3, PCL, LMPH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, hereditary, 1A (LMPH1A) [MIM:153100]: A chronic disabling condition which results in swelling of the extremities due to altered lymphatic flow. Patients with lymphedema suffer from recurrent local infections and physical impairment. {ECO:0000269|PubMed:10835628, ECO:0000269|PubMed:10856194, ECO:0000269|PubMed:16924388, ECO:0000269|PubMed:16965327, ECO:0000269|PubMed:17458866, ECO:0000269|PubMed:19289394, ECO:0000269|PubMed:26091405, ECO:0000269|PubMed:9817924}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lenfödem, Kalıtsal, IA, 153100, Otozomal Dominant; LMPH1A (Milroy Hastalığı) (Prenatal) (FLT4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema, hereditary, IA, 153100, Autosomal dominant; LMPH1A (Milroy disease) (FLT4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLT4	FLT4, VEGFR3, PCL, LMPH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema, hereditary, 1A (LMPH1A) [MIM:153100]: A chronic disabling condition which results in swelling of the extremities due to altered lymphatic flow. Patients with lymphedema suffer from recurrent local infections and physical impairment. {ECO:0000269|PubMed:10835628, ECO:0000269|PubMed:10856194, ECO:0000269|PubMed:16924388, ECO:0000269|PubMed:16965327, ECO:0000269|PubMed:17458866, ECO:0000269|PubMed:19289394, ECO:0000269|PubMed:26091405, ECO:0000269|PubMed:9817924}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfödem, Kalıtsal IC, 613480, Otozomal Dominant; LMPH1C (Milroy Hastalığı) (GJC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema, hereditary, IC, 613480, Autosomal dominant; LMPH1C (Milroy disease) (GJC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJC2	GJC2, GJA12, CX47, PMLDAR, HLD2, SPG44, LMPH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 44, autosomal recessive (SPG44) [MIM:613206]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:19056803}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lenfödem, Kalıtsal IC, 613480, Otozomal Dominant; LMPH1C (Milroy Hastalığı) (Prenatal) (GJC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema, hereditary, IC, 613480, Autosomal dominant; LMPH1C (Milroy disease) (GJC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJC2	GJC2, GJA12, CX47, PMLDAR, HLD2, SPG44, LMPH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 44, autosomal recessive (SPG44) [MIM:613206]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:19056803}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfödem, Kalıtsal ID, 615907, Otozomal Dominant; LMPH1D (Milroy Hastalığı) (VEGFC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema, hereditary, ID, 615907, Autosomal dominant; LMPH1D (Milroy disease) (VEGFC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VEGFC	VEGFC, VRP, LMPH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, hereditary, 1D (LMPH1D) [MIM:615907]: A chronic disabling condition which results in swelling of the extremities due to altered lymphatic flow. Patients with lymphedema suffer from recurrent local infections and physical impairment. {ECO:0000269|PubMed:23410910, ECO:0000269|PubMed:24744435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfödem, Kalıtsal, ID, 615907, Otozomal Dominant; LMPH1D (Milroy Hastalığı) (Prenatal) (VEGFC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema, hereditary, ID, 615907, Autosomal dominant; LMPH1D (Milroy disease) (VEGFC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VEGFC	VEGFC, VRP, LMPH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema, hereditary, 1D (LMPH1D) [MIM:615907]: A chronic disabling condition which results in swelling of the extremities due to altered lymphatic flow. Patients with lymphedema suffer from recurrent local infections and physical impairment. {ECO:0000269|PubMed:23410910, ECO:0000269|PubMed:24744435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfödem, Kalıtsal, 3, 616843, Otozomal Resesif; LMPH3 (Dehidrate Kalıtsal Stomatositoz) (PIEZO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphedema, hereditary, III, 616843, Autosomal recessive; LMPH3 (Dehydrated hereditary stomatocytosis) (PIEZO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO1	PIEZO1, FAM38A, MIB, DHS, LMPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:22529292, ECO:0000269|PubMed:23479567, ECO:0000269|PubMed:23487776, ECO:0000269|PubMed:23581886, ECO:0000269|PubMed:23695678, ECO:0000269|PubMed:23973043}. Note=The disease is caused by mutations affecting the gene represented in this entry. All disease-causing mutations characterized so far produce a gain-of- function phenotype, mutated channels exhibiting increased cation transport in erythroid cells, that could be due to slower channel inactivation rate compared to the wild-type protein.;	EDTA Blood Sample
Lenfödem, Kalıtsal, 3, 616843, Otozomal Resesif; LMPH3 (Dehidrate Kalıtsal Stomatositoz) (Prenatal) (PIEZO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphedema, hereditary, III, 616843, Autosomal recessive; LMPH3 (Dehydrated hereditary stomatocytosis) (PIEZO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO1	PIEZO1, FAM38A, MIB, DHS, LMPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema (DHS) [MIM:194380]: An autosomal dominant hemolytic anemia characterized by primary erythrocyte dehydration. DHS erythrocytes exhibit decreased total cation and potassium content that are not accompanied by a proportional net gain of sodium and water. DHS patients typically exhibit mild to moderate compensated hemolytic anemia, with an increased erythrocyte mean corpuscular hemoglobin concentration and a decreased osmotic fragility, both of which reflect cellular dehydration. Patients may also show perinatal edema and pseudohyperkalemia due to loss of potassium from red cells stored at room temperature. A minor proportion of red cells appear as stomatocytes on blood films. Complications such as splenomegaly and cholelithiasis, resulting from increased red cell trapping in the spleen and elevated bilirubin levels, respectively, may occur. The course of DHS is frequently associated with iron overload, which may lead to hepatosiderosis. {ECO:0000269|PubMed:22529292, ECO:0000269|PubMed:23479567, ECO:0000269|PubMed:23487776, ECO:0000269|PubMed:23581886, ECO:0000269|PubMed:23695678, ECO:0000269|PubMed:23973043}. Note=The disease is caused by mutations affecting the gene represented in this entry. All disease-causing mutations characterized so far produce a gain-of- function phenotype, mutated channels exhibiting increased cation transport in erythroid cells, that could be due to slower channel inactivation rate compared to the wild-type protein.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfödem, Primer-Miyelodisplazi (Sağırlık-Lenfödem-Lösemi Sendromu (Emberger Sendromu)) (GATA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LYMPHEDEMA, PRIMARY, WITH MYELODYSPLASIA (Deafness-lymphedema-leukemia syndrome) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Lenfödem, Primer-Miyelodisplazi (Sağırlık-Lenfödem-Lösemi Sendromu (Emberger Sendromu)) (Prenatal) (GATA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	LYMPHEDEMA, PRIMARY, WITH MYELODYSPLASIA (Deafness-lymphedema-leukemia syndrome) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269	PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfositik Lösemi, Akut T Hücresi (RAP1GDS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphocytic leukemia, acute T-cell (RAP1GDS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAP1GDS1	RAP1GDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Lenfoid Enhanser-Bağlama Faktörü 1, Lef1 (LEF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LYMPHOID ENHANCER-BINDING FACTOR 1, LEF1 (LEF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEF1	LEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lenfoma, B Hücreli Non-Hodgkin Somatik (ATM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, B-cell non-Hodgkin, somatic (ATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333, Note=The disease is caused	Biopsy of affected site-transport media
Lenfoma, B-Hücreli, 109565 (BCL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, B-cell, 109565 (BCL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL6	BCL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Chromosomal aberrations involving BCL6 are a cause of B-cell non-Hodgkin lymphomas (B-cell NHL), including diffuse large B-cell lymphoma and follicular lymphoma. Approximately 40% of diffuse large B-cell lymphomas and 5 to 10% of follicular lymphomas are associated with chromosomal translocations that deregulate expression of BCL6 by juxtaposing heterologous promoters to the BCL6 coding domain. Translocation t(3;14)(q27;q32). Translocation t(2;22)(q27;q11) with immunoglobulin gene regions. Translocation t(3;7)(q27;p12) with IKZF1 gene 5'non-coding region. Translocation t(3;6)(q27;p21) with Histone H4. Translocation t(3;16)(q27;p11) with IL21R. Translocation t(3;13)(q27;q14) with LCP1.	EDTA Blood Sample
Lenfoma, Foliküler, Somatik, 605027 (Non-Hodgkin Lenfoma) (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, follicular, somatic, 605027 (Non-Hodgkin lymphoma) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Lenfoma, MALT, Somatik, 137245 (MALT Lenfoma) (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, MALT, somatic, 137245 (MALT lymphoma) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Lenfoma, Mantle Hücreli, Somatik (ATM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, mantle cell, somatic (ATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333, Note=The disease is caused	Biopsy of affected site-transport media
Lenfoma, Mukoza İle İlişkili Lenfoid Tip (MALT Lenfoma) (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LYMPHOMA, MUCOSA-ASSOCIATED LYMPHOID TYPE (MALT lymphoma) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 37 (IMD37) [MIM:616098]: A form of primary combined immunodeficiency, a group of disorders characterized by severe recurrent infections, with normal numbers or an absence of T and B lymphocytes, and impaired cellular and humoral immunity. IMD37 is characterized by hypogammaglobulinemia without lymphopenia, but with profoundly reduced memory B cells and memory T cells, and increased numbers of circulating naive lymphocytes. Inheritance is autosomal recessive. {ECO:0000269|PubMed:25365219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoma, Non-Hodgkin, 605027 (Non-Hodgkin Lenfoma) (PRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, non-Hodgkin, 605027 (Non-Hodgkin lymphoma) (PRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRF1	PRF1, HPLH2, FLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. {ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoma, Non-Hodgkin, 605027 (Non-Hodgkin Lenfoma) (MLPA)	Lymphoma, non-Hodgkin, 605027 (Non-Hodgkin lymphoma) (MLPA)	PRF1	PRF1, HPLH2, FLH2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hemophagocytic lymphohistiocytosis 2 (FHL2) [MIM:603553]: A rare disorder characterized by immune dysregulation with hypercytokinemia, defective function of natural killer cell, and massive infiltration of several organs by activated lymphocytes and macrophages. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, and less frequently neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits and ataxia. {ECO:0000269|PubMed:10583959, ECO:0000269|PubMed:11179007}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoma, Non-Hodgkin, Aleksel (Non-Hodgkin Lenfoma) (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LYMPHOMA, NON-HODGKIN, FAMILIAL (Non-Hodgkin lymphoma) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 37 (IMD37) [MIM:616098]: A form of primary combined immunodeficiency, a group of disorders characterized by severe recurrent infections, with normal numbers or an absence of T and B lymphocytes, and impaired cellular and humoral immunity. IMD37 is characterized by hypogammaglobulinemia without lymphopenia, but with profoundly reduced memory B cells and memory T cells, and increased numbers of circulating naive lymphocytes. Inheritance is autosomal recessive. {ECO:0000269|PubMed:25365219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoma, Non-Hodgkin, Somatik, 605027 (Non-Hodgkin Lenfoma) (RAD54L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, non-Hodgkin, somatic, 605027 (Non-Hodgkin lymphoma) (RAD54L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD54L	RAD54L, HR54, HRAD54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Lenfoma, Non-Hodgkin, Somatik, 605027 (Non-Hodgkin Lenfoma) (RAD54B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, non-Hodgkin, somatic, 605027 (Non-Hodgkin lymphoma) (RAD54B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD54B	RAD54B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Lenfoma, Non-Hodgkin, Somatik, 605027 (Non-Hodgkin Lenfoma) (CASP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, non-Hodgkin, somatic, 605027 (Non-Hodgkin lymphoma) (CASP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP10	CASP10, MCH4, ALPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Lenfoma, Somatik (MAD1L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma, somatic (MAD1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAD1L1	MAD1L1, TXBP181	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in MAD1L1 are involved in the development and/or progression of various types of cancer. {ECO:0000269|PubMed:10597320, ECO:0000269|PubMed:11429278}.	Biopsy of affected site-transport media
Lenfoma / Lösemi, B-Hücreli, Varyant (KDSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoma/leukemia, B-cell, variant (KDSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDSR	FVT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lenfoproliferatif Sendrom 1, 613011, Otozomal Resesif; LPFS1 (Otozomal Resesif Lenfoproliferatif Hastalık) (ITK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoproliferative syndrome 1, 613011, Autosomal recessive; LPFS1 (Autosomal recessive lymphoproliferative disease) (ITK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITK	ITK, EMT, LPFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphoproliferative syndrome 1 (LPFS1) [MIM:613011]: A rare immunodeficiency characterized by extreme susceptibility to infection with Epstein-Barr virus (EBV). Inadequate immune response to EBV can have a fatal outcome. Clinical features include splenomegaly, lymphadenopathy, anemia, thrombocytopenia, pancytopenia, recurrent infections. There is an increased risk for lymphoma. {ECO:0000269|PubMed:19425169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoproliferatif Sendrom 1, 613011, Otozomal Resesif; LPFS1 (Otozomal Resesif Lenfoproliferatif Hastalık) (Prenatal) (ITK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphoproliferative syndrome 1, 613011, Autosomal recessive; LPFS1 (Autosomal recessive lymphoproliferative disease) (ITK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITK	ITK, EMT, LPFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphoproliferative syndrome 1 (LPFS1) [MIM:613011]: A rare immunodeficiency characterized by extreme susceptibility to infection with Epstein-Barr virus (EBV). Inadequate immune response to EBV can have a fatal outcome. Clinical features include splenomegaly, lymphadenopathy, anemia, thrombocytopenia, pancytopenia, recurrent infections. There is an increased risk for lymphoma. {ECO:0000269|PubMed:19425169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Lenfoproliferatif Sendrom 2, 615122, Otozomal Resesif; LPFS2 (Otozomal Resesif Lenfoproliferatif Hastalık) (CD27 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoproliferative syndrome 2, 615122, Autosomal recessive; LPFS2 (Autosomal recessive lymphoproliferative disease) (CD27 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD27	TNFRSF7, CD27, S152. LPFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphoproliferative syndrome 2 (LPFS2) [MIM:615122]: An autosomal recessive immunodeficiency disorder associated with persistent symptomatic EBV viremia, hypogammaglobulinemia, and impaired T-cell-dependent B-cell responses and T-cell dysfunction. The phenotype is highly variable, ranging from asymptomatic borderline-low hypogammaglobulinemia, to a full-blown symptomatic systemic inflammatory response with life-threatening EBV-related complications, including hemophagocytic lymphohistiocytosis, a lymphoproliferative disorder, and malignant lymphoma requiring stem cell transplantation. {ECO:0000269|PubMed:22197273, ECO:0000269|PubMed:22801960}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoproliferatif Sendrom 2, 615122, Otozomal Resesif; LPFS2 (Otozomal Resesif Lenfoproliferatif Hastalık) (Prenatal) (CD27 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphoproliferative syndrome 2, 615122, Autosomal recessive; LPFS2 (Autosomal recessive lymphoproliferative disease) (CD27 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD27	TNFRSF7, CD27, S152. LPFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphoproliferative syndrome 2 (LPFS2) [MIM:615122]: An autosomal recessive immunodeficiency disorder associated with persistent symptomatic EBV viremia, hypogammaglobulinemia, and impaired T-cell-dependent B-cell responses and T-cell dysfunction. The phenotype is highly variable, ranging from asymptomatic borderline-low hypogammaglobulinemia, to a full-blown symptomatic systemic inflammatory response with life-threatening EBV-related complications, including hemophagocytic lymphohistiocytosis, a lymphoproliferative disorder, and malignant lymphoma requiring stem cell transplantation. {ECO:0000269|PubMed:22197273, ECO:0000269|PubMed:22801960}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfoproliferatif Sendrom, X'e Bağlı 1, 308240, X'e Bağlı Resesif; XLP1 (X'e Bağlı Lenfoproliferatif Sendromj (SH2D1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoproliferative syndrome, X-linked, 1, 308240, X-linked recessive; XLP1 (X-linked lymphoproliferative disease) (SH2D1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH2D1A	SH2D1A, LYP, IMD5, XLP, XLPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Lenfoproliferatif Sendrom, X'e Bağlı 1, 308240, X'e Bağlı Resesif; XLP1 (X'e Bağlı Lenfoproliferatif Sendromj (Prenatal) (SH2D1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphoproliferative syndrome, X-linked, 1, 308240, X-linked recessive; XLP1 (X-linked lymphoproliferative disease) (SH2D1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SH2D1A	SH2D1A, LYP, IMD5, XLP, XLPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lenfoproliferatif Sendrom, 300635 2, X'e Bağlı; XLP2 (X'e Bağlı Lenfoproliferatif Sendrom) (XIAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lymphoproliferative syndrome, X-linked, 2, 300635, XLP2 (X-linked lymphoproliferative disease) (XIAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XIAP	XIAP, BIRC4, API3, XLP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphoproliferative syndrome, X-linked, 2 (XLP2) [MIM:300635]: A rare immunodeficiency characterized by extreme susceptibility to infection with Epstein-Barr virus (EBV). Symptoms include severe or fatal mononucleosis, acquired hypogammaglobulinemia, pancytopenia and malignant lymphoma. {ECO:0000269|PubMed:17080092}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lenfoproliferatif Sendrom, 300635 2, X'e Bağlı; XLP2 (X'e Bağlı Lenfoproliferatif Sendrom) (Prenatal) (XIAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lymphoproliferative syndrome, X-linked, 2, 300635, XLP2 (X-linked lymphoproliferative disease) (XIAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XIAP	XIAP, BIRC4, API3, XLP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphoproliferative syndrome, X-linked, 2 (XLP2) [MIM:300635]: A rare immunodeficiency characterized by extreme susceptibility to infection with Epstein-Barr virus (EBV). Symptoms include severe or fatal mononucleosis, acquired hypogammaglobulinemia, pancytopenia and malignant lymphoma. {ECO:0000269|PubMed:17080092}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lynch Sendromu 1 (Lynch Sendromu) (MSH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	LYNCH SYNDROME I (Lynch syndrome) (MSH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH2	MSH2, COCA1, FCC1, HNPCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11926458	EDTA Blood Sample
Lynch Sendromu 1 (Lynch Sendromu) (MLPA)	LYNCH SYNDROME I (Lynch syndrome) (MLPA)	MSH2	MSH2, COCA1, FCC1, HNPCC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11926458	EDTA Blood Sample
Lizinürik Protein İntoleransı, 222700, Otozomal Resesif; LPI (Lizinürik Protein İntoleransı) (SLC7A7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lysinuric protein intolerance, 222700, Autosomal recessive; LPI (Lysinuric protein intolerance) (SLC7A7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC7A7	SLC7A7, LPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lysinuric protein intolerance (LPI) [MIM:222700]: A metabolic disorder characterized by increased renal excretion of cationic amino acid (CAA), reduced CAA absorption from intestine, and orotic aciduria. On a normal diet, LPI patients present poor feeding, vomiting, diarrhea, episodes of hyperammonaemic coma and growth retardation. Hepatosplenomegaly, osteoporosis and a life- threatening pulmonary involvement (alveolar proteinosis) are also seen. Biochemically LPI is characterized by defective transport of dibasic amino acids at the basolateral membrane of epithelial cells in kidney and intestine. {ECO:0000269|PubMed:10080182, ECO:0000269|PubMed:10631139, ECO:0000269|PubMed:10655553, ECO:0000269|PubMed:12402335, ECO:0000269|PubMed:15776427, ECO:0000269|PubMed:17764084, ECO:0000269|PubMed:9829974}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Lizinürik Protein İntoleransı, 222700, Otozomal Resesif; LPI (Lizinürik Protein İntoleransı) (Prenatal) (SLC7A7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lysinuric protein intolerance, 222700, Autosomal recessive; LPI (Lysinuric protein intolerance) (SLC7A7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC7A7	SLC7A7, LPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lysinuric protein intolerance (LPI) [MIM:222700]: A metabolic disorder characterized by increased renal excretion of cationic amino acid (CAA), reduced CAA absorption from intestine, and orotic aciduria. On a normal diet, LPI patients present poor feeding, vomiting, diarrhea, episodes of hyperammonaemic coma and growth retardation. Hepatosplenomegaly, osteoporosis and a life- threatening pulmonary involvement (alveolar proteinosis) are also seen. Biochemically LPI is characterized by defective transport of dibasic amino acids at the basolateral membrane of epithelial cells in kidney and intestine. {ECO:0000269|PubMed:10080182, ECO:0000269|PubMed:10631139, ECO:0000269|PubMed:10655553, ECO:0000269|PubMed:12402335, ECO:0000269|PubMed:15776427, ECO:0000269|PubMed:17764084, ECO:0000269|PubMed:9829974}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizozomal Asit Fosfataz Eksikliği, 200950, Otozomal Resesif (Asit Fosfataz Eksikliği) (ACP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lysosomal acid phosphatase deficiency, 200950, Autosomal recessive (Acid phosphatase deficiency) (ACP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACP2	ACP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Lysosomal acid phosphatase has been shown to be deficient in cultured fibroblasts from patients manifesting intermittent vomiting, hypotonia, lethargy, opisthotonos, terminal bleeding and death in early infancy. {ECO:0000269|PubMed:5410815}.	EDTA Blood Sample
Lizozomal Asit Fosfataz Eksikliği, 200950, Otozomal Resesif (Asit Fosfataz Eksikliği) (Prenatal) (ACP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lysosomal acid phosphatase deficiency, 200950, Autosomal recessive (Acid phosphatase deficiency) (ACP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACP2	ACP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Lysosomal acid phosphatase has been shown to be deficient in cultured fibroblasts from patients manifesting intermittent vomiting, hypotonia, lethargy, opisthotonos, terminal bleeding and death in early infancy. {ECO:0000269|PubMed:5410815}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Lizil Hidroksilaz 3 Eksikliği, 612394, Otozomal Resesif (Bağ Dokusu Hastalığı-Lizil Hidroksilaz-3 Eksikliğine Bağlı) (PLOD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Lysyl hydroxylase 3 deficiency, 612394, Autosomal recessive (Connective tissue disorder due to lysyl hydroxylase-3 deficiency) (PLOD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLOD3	PLOD3, LH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lysyl hydroxylase 3 deficiency (LH3 deficiency) [MIM:612394]: Connective tissue disorder. The syndrome is characterized by congenital malformations severely affecting many tissues and organs and revealing features of several collagen disorders, most of them involving COL2A1 (type II collagen). The findings suggest that the failure of lysyl hydroxylation and hydroxylysyl carbohydrate addition, which affects many collagens, is the molecular basis of this syndrome. {ECO:0000269|PubMed:18834968}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Lizil Hidroksilaz 3 Eksikliği, 612394, Otozomal Resesif (Bağ Dokusu Hastalığı-Lizil Hidroksilaz-3 Eksikliğine Bağlı) (Prenatal) (PLOD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Lysyl hydroxylase 3 deficiency, 612394, Autosomal recessive (Connective tissue disorder due to lysyl hydroxylase-3 deficiency) (PLOD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLOD3	PLOD3, LH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lysyl hydroxylase 3 deficiency (LH3 deficiency) [MIM:612394]: Connective tissue disorder. The syndrome is characterized by congenital malformations severely affecting many tissues and organs and revealing features of several collagen disorders, most of them involving COL2A1 (type II collagen). The findings suggest that the failure of lysyl hydroxylation and hydroxylysyl carbohydrate addition, which affects many collagens, is the molecular basis of this syndrome. {ECO:0000269	PubMed:18834968}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Machado–Joseph Hastalığı 109150, Otozomal Dominant; MJD (Spinoserebellar Ataksi Tip 3) (ATXN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Machado-Joseph disease, 109150, Autosomal dominant; MJD (Spinocerebellar ataxia type 3) (ATXN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN3	ATXN3, MJD, SCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 3 (SCA3) [MIM:109150]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA3 belongs to the autosomal dominant cerebellar ataxias type I (ADCA-I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. The molecular defect in SCA3 is the a CAG repeat expansion in ATX3 coding region. Longer expansions result in earlier onset and more severe clinical manifestations of the disease. {ECO:0000269	PubMed:7874163}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Machado–Joseph Hastalığı 109150, Otozomal Dominant; MJD (Spinoserebellar Ataksi Tip 3) (Prenatal) (ATXN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Machado-Joseph disease, 109150, Autosomal dominant; MJD (Spinocerebellar ataxia type 3) (ATXN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATXN3	ATXN3, MJD, SCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinocerebellar ataxia 3 (SCA3) [MIM:109150]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA3 belongs to the autosomal dominant cerebellar ataxias type I (ADCA-I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. The molecular defect in SCA3 is the a CAG repeat expansion in ATX3 coding region. Longer expansions result in earlier onset and more severe clinical manifestations of the disease. {ECO:0000269	PubMed:7874163}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali, Alopesi, Cutis Laxa ve Skolyoz, 613075, Otozomal Resesif (RIN2 Sendromu) (RIN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075, Autosomal recessive (RIN2 syndrome) (RIN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIN2	RIN2, MACS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makrosefali, Alopesi, Cutis Laxa ve Skolyoz, 613075, Otozomal Resesif (RIN2 Sendromu) (Prenatal) (RIN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075, Autosomal recessive (RIN2 syndrome) (RIN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RIN2	RIN2, MACS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali, Dismorfik Yüz ve Psikomotor Retardasyon, 617011, Otozomal Resesif; MDFPMR (Megalensefali-Ağır Kifoskolyozlu-Aşırı Büyüme Sendromu) (HERC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011, Autosomal recessive; MDFPMR (Megalencephaly-severe kyphoscoliosis-overgrowth syndrome) (HERC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HERC1	HERC1, MDFPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makrosefali, Dismorfik Yüz ve Psikomotor Retardasyon, 617011, Otozomal Resesif; MDFPMR (Megalensefali-Ağır Kifoskolyozlu-Aşırı Büyüme Sendromu) (Prenatal) (HERC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Macrocephaly, dysmorphic facies, and psychomotor retardation, 617011, Autosomal recessive; MDFPMR (Megalencephaly-severe kyphoscoliosis-overgrowth syndrome) (HERC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HERC1	HERC1, MDFPMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali, Makrozomi, Yüzde Dismorfizm Sendromu, 614192; MMFD (Aşırı Büyüme-Makrosefali-Fasiyeldismorfizm Sendromu) (RNF135 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192; MMFD (Overgrowth-macrocephaly-facial dysmorphism syndrome) (RNF135 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF135	RNF135, MMFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makrosefali, Makrozomi, Yüzde Dismorfizm Sendromu, 614192; MMFD (Aşırı Büyüme-Makrosefali-Fasiyeldismorfizm Sendromu) (Prenatal) (RNF135 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Macrocephaly, macrosomia, facial dysmorphism syndrome, 614192; MMFD (Overgrowth-macrocephaly-facial dysmorphism syndrome) (RNF135 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNF135	RNF135, MMFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali / Otizm Sendromu, 605309, Otozomal Dominant (Makrosefali-Mental Retardasyon-Otizm Sendromu) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrocephaly/autism syndrome, 605309, Autosomal dominant (Macrocephaly-intellectual disability-autism syndrome) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Makrosefali / Otizm Sendromu, 605309, Otozomal Dominant (Makrosefali-Mental Retardasyon-Otizm Sendromu) (MLPA)	Macrocephaly/autism syndrome, 605309, Autosomal dominant (Macrocephaly-intellectual disability-autism syndrome) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Makrosefali / Otizm Sendromu, 605309, Otozomal Dominant (Makrosefali-Mental Retardasyon-Otizm Sendromu) (Prenatal) (PTEN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Macrocephaly/autism syndrome, 605309, Autosomal dominant (Macrocephaly-intellectual disability-autism syndrome) (PTEN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali / Otizm Sendromu, 605309, Otozomal Dominant (Makrosefali-Mental Retardasyon-Otizm Sendromu) (Prenatal) (MLPA)	Macrocephaly/autism syndrome, 605309, Autosomal dominant (Macrocephaly-intellectual disability-autism syndrome) (Prenatal) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makrosefali / Megalensefali Sendromu, Otozomal Resesif, 248000, Otozomal Resesif; MGCPH (Megalensefali) (TBC1D7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000, Autosomal recessive; MGCPH (Megalencephaly) (TBC1D7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D7	TBC1D7, PIGS1, TBC7, MGCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makrosefali / Megalensefali Sendromu, Otozomal Resesif, 248000, Otozomal Resesif; MGCPH (Megalensefali) (Prenatal) (TBC1D7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Macrocephaly/megalencephaly syndrome, autosomal recessive, 248000, Autosomal recessive; MGCPH (Megalencephaly) (TBC1D7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBC1D7	TBC1D7, PIGS1, TBC7, MGCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Makroglobulinemi, Waldenström, Somatik, 153600 (Waldenström Makroglobulinemisi) (MYD88 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macroglobulinemia, Waldenstrom, somatic, 153600 (Waldenström macroglobulinemia) (MYD88 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYD88	MYD88, MYD88D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makrotrombositopeni ve Progresif Sensorinöral İşitme Kaybı, 600208, Otozomal Dominant (MYH9 İlgili Hastalık) (MYH9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrothrombocytopenia and progressive sensorineural deafness, 600208, Autosomal dominant (MYH9-related disease) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Makrotrombositopeni, Otozomal Dominant TUBB1 İlişkili, 613112, Otozomal Dominant (Otozomal Dominant Makrotrombositopeni) (TUBB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macrothrombocytopenia, autosomal dominant, TUBB1-related, 613112, Autosomal dominant (Autosomal dominant macrothrombocytopenia) (TUBB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB1	TUBB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macrothrombocytopenia, autosomal dominant, TUBB1-related (MAD-TUBB1) [MIM:613112]: A congenital blood disorder characterized by increased platelet size and decreased number of circulating platelets. {ECO:0000269	PubMed:18849486}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküler Korneal Distrofi, 217800, Otozomal Resesif; MCD (Maküler Kornea Distrofisi) (CHST6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular corneal dystrophy, 217800, Autosomal recessive; MCD (Macular corneal dystrophy) (CHST6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHST6	CHST6, MCDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makula Dejenerasyonu, Yaş İle İlişkili, 1, 603075, Otozomal Dominant; ARMD1 (HMCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 1, 603075, Autosomal dominant; ARMD1 (HMCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMCN1	HMCN1, FBLN6, FIBL6, ARMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Makula Dejenerasyonu, Yaş İle İlişkili, 11, 611953; ARMD11 (Kalıtsal Serebral Hemoraji-Amiloidoz) (CST3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 11, 611953; ARMD11 (Hereditary cerebral hemorrhage with amyloidosis) (CST3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CST3	CST3, ARMD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyloidosis 6 (AMYL6) [MIM:105150]: A hereditary generalized amyloidosis due to cystatin C amyloid deposition. Cystatin C amyloid accumulates in the walls of arteries, arterioles, and sometimes capillaries and veins of the brain, and in various organs including lymphoid tissue, spleen, salivary glands, and seminal vesicles. Amyloid deposition in the cerebral vessels results in cerebral amyloid angiopathy, cerebral hemorrhage and premature stroke. Cystatin C levels in the cerebrospinal fluid are abnormally low. {ECO:0000269|PubMed:1352269, ECO:0000269|PubMed:2541223}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Maküla Dejenerasyonu, Yaş İle İlişkili, 12, 613784; ARMD12 (CX3CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 12, 613784; ARMD12 (CX3CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CX3CR1	CX3CR1, GPR13, V28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 12 (ARMD12) [MIM:613784]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269	PubMed:15208270}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili 13, Yatkınlık, 615439, Otozomal Dominant; ARMD13 (İmmün Yetmezlik-Faktör 1 Anomasi) (CFI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 13, susceptibility to, 615439, Autosomal dominant; ARMD13 (Immunodeficiency with factor I anomaly) (CFI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFI	CFI, FI, AHUS3, ARMD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement factor I deficiency (CFI deficiency) [MIM:610984]: Autosomal recessive condition associated with a propensity to pyogenic infections. {ECO:0000269|PubMed:12562389, ECO:0000269|PubMed:17018561, ECO:0000269|PubMed:8613545}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 14, Düşük Risk, 615489 (CFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 14, reduced risk of, 615489 (CFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFB	CFB, BF, GBG, AHUS4, ARMD14, CFBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 4 (AHUS4) [MIM:612924]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:17182750, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to the development of atypical hemolytic uremic syndrome can be conferred by mutations in various components of or regulatory factors in the complement cascade system. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 14, Düşük Risk, 615489; ARMD14 (İmmün Yetmezlik, Klasik Komponent Yolağı Kompleman Etkinliğine Bağlı) (C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 14, reduced risk of, 615489; ARMD14 (Immunodeficiency due to a classical component pathway complement deficiency) (C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C2	C2, ARMD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 2 deficiency (C2D) [MIM:217000]: A rare defect of the complement classical pathway associated with the development of autoimmune disorders, mainly systemic lupus erythematosus. Skin and joint manifestations are common and renal disease is relatively rare. Patients with complement component 2 deficiency are also reported to have recurrent invasive infections. {ECO:0000269|PubMed:8621452, ECO:0000269|PubMed:9670930}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 15, Yatkınlık, 615591; ARMD15 (C9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 15, susceptibility to, 615591; ARMD15 (C9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C9	C9, C9D, ARMD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Complement component 9 deficiency (C9D) [MIM:613825]: A rare defect of the complement classical pathway associated with susceptibility to severe recurrent infections predominantly by Neisseria gonorrhoeae or Neisseria meningitidis. Some patients may develop dermatomyositis. {ECO:0000269	PubMed:9634479}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 2, 153800, Otozomal Dominant; ARMD2 (Stargardt Hastalığı) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 2, 153800, Autosomal dominant; ARMD2 (Stargardt disease) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 2 (ARMD2) [MIM:153800]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269|PubMed:19028736, ECO:0000269|PubMed:9295268}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 3, 608895, Otozomal Dominant (Kalıtsal Sensorimotor Nöropati-Hiperelastik Deri) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 3, 608895, Autosomal dominant (Hereditary sensorimotor neuropathy with hyperelastic skin) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 1A (ARCL1A) [MIM:219100]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. Type I autosomal recessive cutis laxa is a specific, life-threatening disorder with organ involvement, lung aelectasis and emphysema, diverticula of the gastrointestinal and genitourinary systems, and vascular anomalies. Associated cranial anomalies, late closure of the fontanel, joint laxity, hip dislocation, and inguinal hernia have been observed but are uncommon. {ECO:0000269|PubMed:12189163, ECO:0000269|PubMed:16652333, ECO:0000269|PubMed:16691202, ECO:0000269|PubMed:17035250, ECO:0000269|PubMed:18185537}. Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations affecting this gene can modify the phenotype of diseases caused by ELN mutations. {ECO:0000269|PubMed:19194475}.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 4, 610698; ARMD4 (CFH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 4, 610698; ARMD4 (CFH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 4, 610698; ARMD4 (MLPA)	Macular degeneration, age-related, 4, 610698; ARMD4 (MLPA)	CFH	CFH, HF1, HUS, ARMD4, AHUS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 1 (AHUS1) [MIM:235400]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:10577907, ECO:0000269|PubMed:10762557, ECO:0000269|PubMed:11851332, ECO:0000269|PubMed:12960213, ECO:0000269|PubMed:14583443, ECO:0000269|PubMed:14978182, ECO:0000269|PubMed:20513133, ECO:0000269|PubMed:9551389}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 6, 613757; ARMD6 (Progresif Kon Distrofisi) (RAX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 6, 613757; ARMD6 (Cone rod dystrophy) (RAX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAX2	RAX2, RAXL1, QRX, CORD11, ARMD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 11 (CORD11) [MIM:610381]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269	PubMed:15228672}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 603075, Otozomal Dominant (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 603075, Autosomal dominant (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 2 (AD2) [MIM:104310]: A late-onset neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The APOE*4 allele is genetically associated with the common late onset familial and sporadic forms of Alzheimer disease. Risk for AD increased from 20% to 90% and mean age at onset decreased from 84 to 68 years with increasing number of APOE*4 alleles in 42 families with late onset AD. Thus APOE*4 gene dose is a major risk factor for late onset AD and, in these families, homozygosity for APOE*4 was virtually sufficient to cause AD by age 80. The mechanism by which APOE*4 participates in pathogenesis is not known.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 7, 610149; ARMD7 (HTRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 7, 610149; ARMD7 (HTRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA1	HTRA1, PRSS11, ARMD7, CARASIL, CADASIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy (CARASIL) [MIM:600142]: A cerebrovascular disease characterized by non-hypertensive arteriopathy of cerebral small vessels with subcortical infarcts, alopecia, and spondylosis. Small cerebral arteries show arteriosclerotic changes, fibrous intimal proliferation, and hyaline degeneration with splitting of the intima and/or the internal elastic membrane. Neurologic features include progressive dementia, gait disturbances, extrapyramidal and pyramidal signs, and demyelination of the cerebral white matter with sparing of U fibers. {ECO:0000269	PubMed:19387015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, 8, 613778; ARMD8 (ARMS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 8, 613778; ARMD8 (ARMS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARMS2	LOC387715, ARMD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Maküla Dejenerasyonu, Yaş İle İlişkili, 9, 611378; ARMD9 (C3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, 9, 611378; ARMD9 (C3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C3	C3, ARMD9, AHUS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 9 (ARMD9) [MIM:611378]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269|PubMed:17634448, ECO:0000269|PubMed:24036952}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, Neovasküler Tip, 610149 (HTRA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, neovascular type, 610149 (HTRA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA1	HTRA1, PRSS11, ARMD7, CARASIL, CADASIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy, autosomal recessive, with subcortical infarcts and leukoencephalopathy (CARASIL) [MIM:600142]: A cerebrovascular disease characterized by non-hypertensive arteriopathy of cerebral small vessels with subcortical infarcts, alopecia, and spondylosis. Small cerebral arteries show arteriosclerotic changes, fibrous intimal proliferation, and hyaline degeneration with splitting of the intima and/or the internal elastic membrane. Neurologic features include progressive dementia, gait disturbances, extrapyramidal and pyramidal signs, and demyelination of the cerebral white matter with sparing of U fibers. {ECO:0000269	PubMed:19387015}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, Düşük Risk, 603075, Otozomal Dominant (CFHR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, reduced risk of, 603075, Autosomal dominant (CFHR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFHR3	CFHR3, FHR3, HLF4, CFHL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, Düşük Risk, 603075, Otozomal Dominant (MLPA)	Macular degeneration, age-related, reduced risk of, 603075, Autosomal dominant (MLPA)	CFHR3	CFHR3, FHR3, HLF4, CFHL3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, Düşük Risk, 603075, Otozomal Dominant (MLPA)	Macular degeneration, age-related, reduced risk of, 603075, Autosomal dominant (MLPA)	CFHR1	CFHR1, FHR1, HFL1, CFHL1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Maküla Dejenerasyonu, Yaş İle İlişkili, Yatkınlık, 5, 613761 (Cockayne Sendromu) (ERCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, age-related, susceptibility to, 5, 613761 (Cockayne syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMD5, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:3443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküla Dejenerasyonu, Erken Başlangıçlı, 616118, Otozomal Dominant; EOMD (Konjenital Kontraktürel Araknodaktili) (FBN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, early-onset, 616118, Autosomal dominant; EOMD (Congenital contractural arachnodactyly) (FBN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN2	FBN2, CCA, EOMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, early-onset (EOMD) [MIM:616118]: An ocular disorder characterized by macular changes resulting in progressive loss of visual acuity. {ECO:0000269	PubMed:24899048}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, Juvenil, 248200, Otozomal Resesif (Stargardt Hastalığı) (CNGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, juvenile, 248200, Autosomal recessive (Stargardt disease) (CNGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNGB3	CNGB3, ACHM3, ACHM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achromatopsia 3 (ACHM3) [MIM:262300]: An ocular stationary disorder due to the absence of functioning cone photoreceptors in the retina. It is characterized by total colorblindness, low visual acuity, photophobia and nystagmus. Achromatopsia type 3 patients manifest severe myopia. {ECO:0000269|PubMed:10888875, ECO:0000269|PubMed:10958649, ECO:0000269|PubMed:12357335, ECO:0000269|PubMed:14757870, ECO:0000269|PubMed:15657609, ECO:0000269|PubMed:15712225}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküla Dejenerasyonu, X'e Bağlı Atrofik, 300834, X'e Bağlı Resesif (Progresif Kon Distrofisi) (RPGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular degeneration, X-linked atrophic, 300834, X-linked recessive (Cone rod dystrophy) (RPGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGR	RPGR, RP3, CRD, RP15, COD1, CORDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 3 (RP3) [MIM:300029]: A X-linked retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. In RP3, affected males have a severe phenotype, and carrier females show a wide spectrum of clinical features ranging from completely asymptomatic to severe retinitis pigmentosa. Heterozygous women can manifest a form of choroidoretinal degeneration which is distinguished from other types by the absence of visual defects in the presence of a brilliant, scintillating, golden-colored, patchy appearance most striking around the macula, called a tapetal-like retinal reflex. {ECO:0000269|PubMed:10482958, ECO:0000269|PubMed:10737996, ECO:0000269|PubMed:10932196, ECO:0000269|PubMed:10937588, ECO:0000269|PubMed:10970770, ECO:0000269|PubMed:11180598, ECO:0000269|PubMed:11992260, ECO:0000269|PubMed:12657579, ECO:0000269|PubMed:14564670, ECO:0000269|PubMed:9671101, ECO:0000269|PubMed:8817343, ECO:0000269|PubMed:9399904, ECO:0000269|PubMed:9855182}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküler Distrofi-Merkezi Kon Tutulumu, 616170, Otozomal Resesif; CCMD (MFSD8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy with central cone involvement, 616170, Autosomal recessive; CCMD (MFSD8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFSD8	MFSD8, MGC33302, CLN7, CCMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy with central cone involvement (CCMD) [MIM:616170]: An ocular disease characterized by decreased visual acuity, slight pigmentary changes and color vision abnormalities, becoming apparent in the third to sixth decade of life. Fundus anomalies are variable and include bull's eye maculopathy, severe atrophy of central fovea, relatively spared fovea with surrounding atrophic ring, central retinal pigment epithelium and/or choroid changes, pale or atrophic peripapillary area, pale optic disk, relatively spared periphery, and slightly or moderately attenuated vessels. {ECO:0000269	PubMed:25227500}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküler Distrofi, Paternli, 1, 169150, Otozomal Dominant; MDPT1 (Kelebek Şeklinde Pigment Distrofisi) (PRPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, patterned, 1, 169150, Autosomal dominant; MDPT1 (Butterfly-shaped pigment dystrophy) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküler Distrofi, Desenli, 2, 608970, Otozomal Dominant (Kelebek Şeklinde Pigment Distrofisi) (CTNNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, patterned, 2, 608970, Autosomal dominant (Butterfly-shaped pigment dystrophy) (CTNNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNA1	CTNNA1, MDPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary diffuse gastric cancer (HDGC) [MIM:137215]: A cancer predisposition syndrome with increased susceptibility to diffuse gastric cancer. Diffuse gastric cancer is a malignant disease characterized by poorly differentiated infiltrating lesions resulting in thickening of the stomach. Malignant tumors start in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. {ECO:0000269	PubMed:23208944}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	EDTA Blood Sample
Maküler Distrofi, Desenli (Paternal) 3, 617111, Otozomal Dominant; MDPT3 (Martinik Kırışık Retina Pigment Epiteliyopati) (MAPKAPK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, patterned, 3, 617111, Autosomal dominant; MDPT3 (Martinique crinkled retinal pigment epitheliopathy) (MAPKAPK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPKAPK3	MAPKAP3, 3PK, MDPT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Maküler Distrofi, Retina, 2, 608051, Otozomal Dominant; MCDR2 (Retina Maküler Distrofi Tip 2) (PROM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, retinal, 2, 608051, Autosomal dominant; MCDR2 (Retinal macular dystrophy type 2) (PROM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROM1	PROM1, PROML1, AC133, RP41, CORD12, CD133, MCDR2, STGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stargardt disease 4 (STGD4) [MIM:603786]: A common hereditary macular degeneration. It is characterized by decreased central vision, atrophy of the macula and underlying retinal pigment epithelium, and frequent presence of prominent flecks in the posterior pole of the retina. {ECO:0000269	PubMed:18654668}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküler Distrofi, Vitelliform, 2, 153700, Otozomal Dominant; VMD2 (Best Vitelliform Maküla Distrofisi) (BEST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, vitelliform, 2, 153700, Autosomal dominant; VMD2 (Best vitelliform macular dystrophy) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy, vitelliform, 2 (VMD2) [MIM:153700]: An autosomal dominant form of macular degeneration that usually begins in childhood or adolescence. VMD2 is characterized by typical 'egg yolk' macular lesions due to abnormal accumulation of lipofuscin within and beneath the retinal pigment epithelium cells. Progression of the disease leads to destruction of the retinal pigment epithelium and vision loss. {ECO:0000269|PubMed:10331951, ECO:0000269|PubMed:10394929, ECO:0000269|PubMed:10453731, ECO:0000269|PubMed:10682987, ECO:0000269|PubMed:10798642, ECO:0000269|PubMed:11241846, ECO:0000269|PubMed:11449320, ECO:0000269|PubMed:12187431, ECO:0000269|PubMed:12324875, ECO:0000269|PubMed:13129869, ECO:0000269|PubMed:14517959, ECO:0000269|PubMed:15176385, ECO:0000269|PubMed:18766995, ECO:0000269|PubMed:19357557, ECO:0000269|PubMed:9662395, ECO:0000269|PubMed:9700209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Maküler Distrofi, Vitelliform, 3, 608161, Otozomal Dominant; VMD3 (Yetişkin Başlangıçlı Foveomakülar Vitelliform Distrofi) (PRPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, vitelliform, 3, 608161, Autosomal dominant; VMD3 (Adult-onset foveomacular vitelliform dystrophy) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Maküler Distrofi, Vitelliform, 4, 616151, Otozomal Dominant, VMD4 (Yetişkin Başlangıçlı Foveomakülar Vitelliform Distrofi) (IMPG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, vitelliform, 4, 616151, Autosomal dominant; VMD4 (Adult-onset foveomacular vitelliform dystrophy) (IMPG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPG1	IMPG1, IPM150, VMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy, vitelliform, 4 (VMD4) [MIM:616151]: A form of macular dystrophy, a retinal disease in which various forms of deposits, pigmentary changes, and atrophic lesions are observed in the macula lutea. Vitelliform macular dystrophies are characterized by yellow, lipofuscin-containing deposits, usually localized at the center of the macula. VMD4 features include late- onset moderate visual impairment, small satellite drusen-like lesions in the foveal area, and preservation of retinal pigment epithelium reflectivity. {ECO:0000269	PubMed:23993198}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Maküler Distrofi, Vitelliform, 5, 616152, Otozomal Dominant, VMD5 (Yetişkin Başlangıçlı Foveomakülar Vitelliform Distrofi) (IMPG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Macular dystrophy, vitelliform, 5, 616152, Autosomal dominant; VMD5 (Adult-onset foveomacular vitelliform dystrophy) (IMPG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPG2	IMPG2, IPM200, RPS6, VMD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 56 (RP56) [MIM:613581]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:20673862}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Majeed Sendromu, 609628; MJDS (Majeed Sendromu) (LPIN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Majeed syndrome, 609628; MJDS (Majeed syndrome) (LPIN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPIN2	LPIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Majeed syndrome (MAJEEDS) [MIM:609628]: An autosomal recessive syndrome characterized by chronic recurrent multifocal osteomyelitis that is of early onset with a lifelong course, congenital dyserythropoietic anemia that presents as hypochromic, microcytic anemia during the first year of life and ranges from mild to transfusion-dependent, and transient inflammatory dermatosis, often manifesting as Sweet syndrome (neutrophilic skin infiltration). {ECO:0000269	PubMed:15994876}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Majeed Sendromu, 609628; MJDS (Majeed Sendromu) (Prenatal) (LPIN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Majeed syndrome, 609628; MJDS (Majeed syndrome) (LPIN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LPIN2	LPIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Majeed syndrome (MAJEEDS) [MIM:609628]: An autosomal recessive syndrome characterized by chronic recurrent multifocal osteomyelitis that is of early onset with a lifelong course, congenital dyserythropoietic anemia that presents as hypochromic, microcytic anemia during the first year of life and ranges from mild to transfusion-dependent, and transient inflammatory dermatosis, often manifesting as Sweet syndrome (neutrophilic skin infiltration). {ECO:0000269	PubMed:15994876}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Majör Afektif Bozukluk-7, Yatkınlık, 612371 (XBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Major affective disorder-7, susceptibility to, 612371 (XBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XBP1	XBP1, XBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Major affective disorder 7 (MAFD7) [MIM:612371]: A major psychiatric disorder that is characterized by severe mood swings, with fluctuation between two abnormal mood states (manic or major depressive episode). Mania is accompanied by symptoms of euphoria, irritability, or excitation, whereas depression is associated with low mood and decreased motivation and energy. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Majör Depresif Bozukluk ve Antidepresan Taç Tedavisine Hılandırılmış Yanıt, 608516 (FKBP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Major depressive disorder and accelerated response to antidepressant drug treatment, 608516 (FKBP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKBP5	FKBP5, FKBP51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Majör Depresif Bozukluk, Sitalopram Tedaviye Yanıt, 608516 (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Major depressive disorder, response to citalopram therapy in, 608516 (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Entellektüel, Düşük Risk, 611162 (Sıtma) (CD36 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, cerebral, reduced risk of, 611162 (Malaria) (CD36 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD36	CD36, CHDS7, BDPLT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Platelet glycoprotein IV deficiency (PG4D) [MIM:608404]: A disorder characterized by macrothrombocytopenia without notable hemostatic problems and bleeding tendency. Platelet glycoprotein IV deficiency can be divided into 2 subgroups. The type I phenotype is characterized by platelets and monocytes/macrophages exhibiting complete CD36 deficiency. The type II phenotype lacks the surface expression of CD36 in platelets, but expression in monocytes/macrophages is near normal. {ECO:0000269|PubMed:11950861, ECO:0000269|PubMed:7533763}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients also have postprandial hypertriglyceridemia, insulin resistance and hypertension increasing atherosclerotic risk. {ECO:0000269|PubMed:18753675}.	EDTA Blood Sample
Sıtma, Serebral, Yatkınlık, 611162 (Sıtma) (TNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, cerebral, susceptibility to, 611162 (Malaria) (TNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNF	TNF, TNFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet; arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). {ECO:0000269	PubMed:12746914}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sıtma, Serebral, Yatkınlık, 611162 (Sıtma) (ICAM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, cerebral, susceptibility to, 611162 (Malaria) (ICAM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ICAM1	ICAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Hafif Yatkınlık, 609148 (NCR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, mild, susceptibility to, 609148 (NCR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NCR3	NCR3, 1C7, NKP30, CD337, MALS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Karşı Koruma, 611162 (Sıtma) (TIRAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, protection against, 611162 (Malaria) (TIRAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIRAP	TIRAP, BACTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Direnç, 611162 (Sıtma) (NOS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, resistance to, 611162 (Malaria) (NOS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS2	NOS2A, NOS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Direnç, 611162 (Sıtma) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, resistance to, 611162 (Malaria) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6253091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sıtma, Direnç, 611162 (Sıtma) (GYPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, resistance to, 611162 (Malaria) (GYPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYPB	GYPB, SS, MNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Direnç, 611162 (Sıtma) (FCGR2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, resistance to, 611162 (Malaria) (FCGR2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2B	FCGR2B, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:12115230, ECO:0000269|PubMed:20385827}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sıtma, Ağır, Direnç, 611162 (Sıtma) (CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, severe, resistance to, 611162 (Malaria) (CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR1	CR1, C3BR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Ağır, Yatkınlık, 611162 (Sıtma) (FCGR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, severe, susceptibility to, 611162 (Malaria) (FCGR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2A	FCGR2A, IGFR2, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Yatkınlık (Sıtma) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MALARIA, SUSCEPTIBILITY TO (Malaria) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sıtma, Yatkınlık, 611162 (Sıtma) (CISH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, susceptibility to, 611162 (Malaria) (CISH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CISH	CISH, BACTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sıtma, Vivax, Karşı Koruma, 611162 (Sıtma) (ACKR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malaria, vivax, protection against, 611162 (Malaria) (ACKR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACKR1	DARC, FY, GPD, WBCQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Erkek Germ Hücresi Tümörü, Somatik, 273300 (Testiküler Seminomatöz Germ Hücreli Tümör) (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Male germ cell tumor, somatic, 273300 (Testicular seminomatous germ cell tumor) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Erkek İnfertilitesi (UBE2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Male infertility (UBE2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBE2B	UBE2B, RAD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Akrosin Eksikliğine Bağlı Erkek İnfertilitesi, 102480 (ACR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Male infertility due to acrosin deficiency, 102480 (ACR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACR	ACR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Malign Fibröz Histiositon, 605352 (MFHAS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malignant fibrous histiocytoma, 605352 (MFHAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFHAS1	MFHAS1, MASL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Malign Hipertermi Yatkınlık 1, 145600, Otozomal Dominant; MHS1 (Malign Hipertermi) (RYR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malignant hyperthermia susceptibility 1, 145600, Autosomal dominant; MHS1 (Malignant hyperthermia) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095.	EDTA Blood Sample
Malign Hipertermi Yatkınlık 5, 601887, Otozomal Dominant (Malign Hipertermi) (CACNA1S geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malignant hyperthermia susceptibility 5, 601887, Autosomal dominant (Malignant hyperthermia) (CACNA1S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Malign Hipertermi Yatkınlık 5, 601887, Otozomal Dominant (Malign Hipertermi) (MLPA)	Malignant hyperthermia susceptibility 5, 601887, Autosomal dominant (Malignant hyperthermia) (MLPA)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Malign Melanom, Somatik, 155600 (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malignant melanoma, somatic, 155600 (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Malonil-CoA Dekarboksilaz Eksikliği, 248360, Otozomal Resesif, ctbA (Malonik Asitüri) (MLYCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Malonyl-CoA decarboxylase deficiency, 248360, Autosomal recessive, ctbA (Malonic aciduria) (MLYCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLYCD	MLYCD, MCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Malonyl-CoA decarboxylase deficiency (MLYCD deficiency) [MIM:248360]: Autosomal recessive disease characterized by abdominal pain, chronic constipation, episodic vomiting, metabolic acidosis and malonic aciduria. {ECO:0000269|PubMed:10417274}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Malonil-CoA Dekarboksilaz Eksikliği, 248360, Otozomal Resesif, ctbA (Malonik Asitüri) (MLPA)	Malonyl-CoA decarboxylase deficiency, 248360, Autosomal recessive, ctbA (Malonic aciduria) (MLPA)	MLYCD	MLYCD, MCD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Malonyl-CoA decarboxylase deficiency (MLYCD deficiency) [MIM:248360]: Autosomal recessive disease characterized by abdominal pain, chronic constipation, episodic vomiting, metabolic acidosis and malonic aciduria. {ECO:0000269|PubMed:10417274}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Malonil-CoA Dekarboksilaz Eksikliği, 248360, Otozomal Resesif, ctbA (Malonik Asitüri) (Prenatal) (MLYCD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Malonyl-CoA decarboxylase deficiency, 248360, Autosomal recessive, ctbA (Malonic aciduria) (MLYCD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MLYCD	MLYCD, MCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Malonyl-CoA decarboxylase deficiency (MLYCD deficiency) [MIM:248360]: Autosomal recessive disease characterized by abdominal pain, chronic constipation, episodic vomiting, metabolic acidosis and malonic aciduria. {ECO:0000269|PubMed:10417274}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Malonil-CoA Dekarboksilaz Eksikliği, 248360, Otozomal Resesif, ctbA (Malonik Asitüri) (Prenatal) (MLPA)	Malonyl-CoA decarboxylase deficiency, 248360, Autosomal recessive, ctbA (Malonic aciduria) (Prenatal) (MLPA)	MLYCD	MLYCD, MCD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Malonyl-CoA decarboxylase deficiency (MLYCD deficiency) [MIM:248360]: Autosomal recessive disease characterized by abdominal pain, chronic constipation, episodic vomiting, metabolic acidosis and malonic aciduria. {ECO:0000269|PubMed:10417274}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibüler Hipoplazi, Sağırlık, Prematüre Yaşlanma ve Lipodistrofi Sendromu, 615381, Otozomal Dominant; MDPL (Mandibular Hipoplazi-Sağırlık-Prematüre Yaşlanma Sendromu) (POLD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381, Autosomal dominant; MDPL (Mandibular hypoplasia-deafness-progeroid syndrome) (POLD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLD1	POLD1, CRCS10, MDPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer 10 (CRCS10) [MIM:612591]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24501277}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Mandibüler Hipoplazi, Sağırlık, Prematüre Yaşlanma ve Lipodistrofi Sendromu, 615381, Otozomal Dominant; MDPL (Mandibular Hipoplazi-Sağırlık-Prematüre Yaşlanma Sendromu) (Prenatal) (POLD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381, Autosomal dominant; MDPL (Mandibular hypoplasia-deafness-progeroid syndrome) (POLD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLD1	POLD1, CRCS10, MDPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer 10 (CRCS10) [MIM:612591]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24501277}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibuloakral Displazi İle Tip A Lipodistrofi; Mada (Mandibuloakral Displazi) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA (Mandibuloacral dysplasia) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mandibuloakral Displazi İle Tip A Lipodistrofi; Mada (Mandibuloakral Displazi) (MLPA)	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA (Mandibuloacral dysplasia) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mandibuloakral Displazi İle Tip A Lipodistrofi; Mada (Mandibuloakral Displazi) (Prenatal) (LMNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA (Mandibuloacral dysplasia) (LMNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibuloakral Displazi İle Tip A Lipodistrofi; Mada (Mandibuloakral Displazi) (Prenatal) (MLPA)	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY; MADA (Mandibuloacral dysplasia) (Prenatal) (MLPA)	LMNA	LMNA, LMNI, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mandibuloakral Displazi, Tip B Lipodistrofi 608612, Otozomal Resesif; MADB (Mandibuloakral Displazi) (ZMPSTE24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mandibuloacral dysplasia with type B lipodystrophy, 608612, Autosomal recessive; MADB (Mandibuloacral dysplasia) (ZMPSTE24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mandibuloakral Displazi, Tip B Lipodistrofi 608612, Otozomal Resesif; MADB (Mandibuloakral Displazi) (MLPA)	Mandibuloacral dysplasia with type B lipodystrophy, 608612, Autosomal recessive; MADB (Mandibuloacral dysplasia) (MLPA)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mandibuloakral Displazi, Tip B Lipodistrofi 608612, Otozomal Resesif; MADB (Mandibuloakral Displazi) (Prenatal) (ZMPSTE24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mandibuloacral dysplasia with type B lipodystrophy, 608612, Autosomal recessive; MADB (Mandibuloacral dysplasia) (ZMPSTE24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibuloakral Displazi, Tip B Lipodistrofi 608612, Otozomal Resesif; MADB (Mandibuloakral Displazi) (Prenatal) (MLPA)	Mandibuloacral dysplasia with type B lipodystrophy, 608612, Autosomal recessive; MADB (Mandibuloacral dysplasia) (Prenatal) (MLPA)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibulofasiyal Disostoz, Akopesi 616367, Otozomal Dominant; MFDA (Mandibulofasiyal Disostoz-Akopesi) (EDNRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mandibulofacial dysostosis with akopecia, 616367, Autosomal dominant; MFDA (Mandibulofacial dysostosis with alopecia) (EDNRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDNRA	EDNRA, MFDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mandibulofasiyal Disostoz, Akopesi 616367, Otozomal Dominant; MFDA (Mandibulofasiyal Disostoz-Akopesi) (Prenatal) (EDNRA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mandibulofacial dysostosis with akopecia, 616367, Autosomal dominant; MFDA (Mandibulofacial dysostosis with alopecia) (EDNRA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDNRA	EDNRA, MFDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mandibulofasiyal Disostoz, Guion-Almeida Tipi, 610536, Otozomal Dominant; MFDGA (Mandibulofasiyal Disostoz-Mikrosefali Sendromu, Guion-Almeida Tipi) (EFTUD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mandibulofacial dysostosis, Guion-Almeida type, 610536, Autosomal dominant; MFDGA (Mandibulofacial dysostosis-microcephaly syndrome) (EFTUD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFTUD2	EFTUD2, KIAA0031, MFDGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mandibulofacial dysostosis with microcephaly (MFDM) [MIM:610536]: A rare syndrome characterized by progressive microcephaly, midface and malar hypoplasia, micrognathia, microtia, dysplastic ears, preauricular skin tags, significant developmental delay, and speech delay. Many patients have major sequelae, including choanal atresia that results in respiratory difficulties, conductive hearing loss, and cleft palate. {ECO:0000269	PubMed:22305528}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mandibulofasiyal Disostoz, Guion-Almeida Tipi, 610536, Otozomal Dominant; MFDGA (Mandibulofasiyal Disostoz-Mikrosefali Sendromu, Guion-Almeida Tipi) (Prenatal) (EFTUD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mandibulofacial dysostosis, Guion-Almeida type, 610536, Autosomal dominant; MFDGA (Mandibulofacial dysostosis-microcephaly syndrome) (EFTUD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EFTUD2	EFTUD2, KIAA0031, MFDGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mandibulofacial dysostosis with microcephaly (MFDM) [MIM:610536]: A rare syndrome characterized by progressive microcephaly, midface and malar hypoplasia, micrognathia, microtia, dysplastic ears, preauricular skin tags, significant developmental delay, and speech delay. Many patients have major sequelae, including choanal atresia that results in respiratory difficulties, conductive hearing loss, and cleft palate. {ECO:0000269	PubMed:22305528}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Manitoba Okülotrikoanal Sendrom, 248450, Otozomal Resesif; MOTA (Okülotrikoanal Sendromu) (FREM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Manitoba oculotrichoanal syndrome, 248450, Autosomal recessive; MOTA (Oculotrichoanal syndrome) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Manitoba oculotrichoanal syndrome (MOTA) [MIM:248450]: A rare condition defined by eyelid colobomas, cryptophthalmos, and anophthalmia/microphthalmia, an aberrant hairline, a bifid or broad nasal tip, and gastrointestinal anomalies such as omphalocele and anal stenosis. {ECO:0000269	PubMed:21507892}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Manitoba Okülotrikoanal Sendrom, 248450, Otozomal Resesif; MOTA (Okülotrikoanal Sendromu) (Prenatal) (FREM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Manitoba oculotrichoanal syndrome, 248450, Autosomal recessive; MOTA (Oculotrichoanal syndrome) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Manitoba oculotrichoanal syndrome (MOTA) [MIM:248450]: A rare condition defined by eyelid colobomas, cryptophthalmos, and anophthalmia/microphthalmia, an aberrant hairline, a bifid or broad nasal tip, and gastrointestinal anomalies such as omphalocele and anal stenosis. {ECO:0000269	PubMed:21507892}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mannozidoz, Alfa-, Tip 1 ve 2, 248500, Otozomal Resesif; MANSA (Alfa-Mannosidoz) (MANZB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mannosidosis, alpha-, types I and II, 248500, Autosomal recessive; MANSA (Alpha-mannosidosis) (MANZB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MANZB1	MANZB1, MANB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mannosidosis, alpha B, lysosomal (MANSA) [MIM:248500]: A lysosomal storage disease characterized by accumulation of unbranched oligosaccharide chains. This accumulation is expressed histologically as cytoplasmic vacuolation predominantly in the CNS and parenchymatous organs. Depending on the clinical findings at the age of onset, a severe infantile (type I) and a mild juvenile (type II) form of alpha-mannosidosis are recognized. There is considerable variation in the clinical expression with mental retardation, recurrent infections, impaired hearing and Hurler- like skeletal changes being the most consistent abnormalities. {ECO:0000269|PubMed:12718372, ECO:0000269|PubMed:15712269, ECO:0000269|PubMed:22161967, ECO:0000269|PubMed:9158146, ECO:0000269|PubMed:9758606, ECO:0000269|PubMed:9915946}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mannozidoz, Alfa-, Tip 1 ve 2, 248500, Otozomal Resesif; MANSA (Alfa-Mannosidoz) (Prenatal) (MANZB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mannosidosis, alpha-, types I and II, 248500, Autosomal recessive; MANSA (Alpha-mannosidosis) (MANZB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MANZB1	MANZB1, MANB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mannosidosis, alpha B, lysosomal (MANSA) [MIM:248500]: A lysosomal storage disease characterized by accumulation of unbranched oligosaccharide chains. This accumulation is expressed histologically as cytoplasmic vacuolation predominantly in the CNS and parenchymatous organs. Depending on the clinical findings at the age of onset, a severe infantile (type I) and a mild juvenile (type II) form of alpha-mannosidosis are recognized. There is considerable variation in the clinical expression with mental retardation, recurrent infections, impaired hearing and Hurler- like skeletal changes being the most consistent abnormalities. {ECO:0000269|PubMed:12718372, ECO:0000269|PubMed:15712269, ECO:0000269|PubMed:22161967, ECO:0000269|PubMed:9158146, ECO:0000269|PubMed:9758606, ECO:0000269|PubMed:9915946}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mannozidoz, Beta, 248510, Otozomal Resesif; MANSB (Beta-Mannosidozis) (MANBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mannosidosis, beta, 248510, Autosomal recessive; MANSB (Beta-mannosidosis) (MANBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MANBA	MANBA, MANB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mannosidosis, beta A, lysosomal (MANSB) [MIM:248510]: An autosomal recessive lysosomal storage disease of glycoprotein catabolism. Clinical features are heterogeneous with a wide range of symptoms and age of onset. The disease is associated with a range of neurological involvement, including various degrees of mental retardation in most of the cases, hearing loss and speech impairment, hypotonia, epilepsy and peripheral neuropathy. Affected individuals have a profound reduction in beta A mannosidase activity in plasma, fibroblasts and leukocytes. {ECO:0000269	PubMed:9384606}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mannozidoz, Beta, 248510, Otozomal Resesif; MANSB (Beta-Mannosidozis) (Prenatal) (MANBA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mannosidosis, beta, 248510, Autosomal recessive; MANSB (Beta-mannosidosis) (MANBA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MANBA	MANBA, MANB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mannosidosis, beta A, lysosomal (MANSB) [MIM:248510]: An autosomal recessive lysosomal storage disease of glycoprotein catabolism. Clinical features are heterogeneous with a wide range of symptoms and age of onset. The disease is associated with a range of neurological involvement, including various degrees of mental retardation in most of the cases, hearing loss and speech impairment, hypotonia, epilepsy and peripheral neuropathy. Affected individuals have a profound reduction in beta A mannosidase activity in plasma, fibroblasts and leukocytes. {ECO:0000269	PubMed:9384606}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Akçaağaç Şurubu Hastalığı, Hafif Varyant, 615135; MSUDMV (Akçaağaç Şurubu İdrar Hastalığı) (PPM1K geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maple syrup urine disease, mild variant, 615135; MSUDMV (Maple syrup urine disease) (PPM1K gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPM1K	PPM1K, PP2CM, PTMP, MSUDMV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maple syrup urine disease, mild variant (MSUDMV) [MIM:615135]: A mild form of maple syrup urine disease, a metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. MSUDMV is characterized by increased plasma levels of branched-chain amino acids (BCAA) apparent at birth. Treatment with a low-protein diet free of BCAA can result in normal psychomotor development and lack of metabolic episodes. {ECO:0000269|PubMed:23086801}. Note=The gene represented in this entry is involved in disease pathogenesis.;	EDTA Blood Sample
Akçaağaç Şurubu Hastalığı, Hafif Varyant, 615135; MSUDMV (Akçaağaç Şurubu İdrar Hastalığı) (Prenatal) (PPM1K geni) (Tüm Gen Dizi Analizi) (Prenatal)	Maple syrup urine disease, mild variant, 615135; MSUDMV (Maple syrup urine disease) (PPM1K gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPM1K	PPM1K, PP2CM, PTMP, MSUDMV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Maple syrup urine disease, mild variant (MSUDMV) [MIM:615135]: A mild form of maple syrup urine disease, a metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. MSUDMV is characterized by increased plasma levels of branched-chain amino acids (BCAA) apparent at birth. Treatment with a low-protein diet free of BCAA can result in normal psychomotor development and lack of metabolic episodes. {ECO:0000269|PubMed:23086801}. Note=The gene represented in this entry is involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akçaağaç Şurubu İdrar Hastalığı, Otozomal Resesif Tip 1a, 248600; MSUD (Akçaağaç Şurubu İdrar Hastalığı) (BCKDHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maple syrup urine disease, type Ia, 248600, Autosomal recessive; MSUD (Maple syrup urine disease) (BCKDHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCKDHA	BCKDHA, MSUD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maple syrup urine disease 1A (MSUD1A) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:1867199, ECO:0000269|PubMed:1885764, ECO:0000269|PubMed:2060625, ECO:0000269|PubMed:21844576, ECO:0000269|PubMed:2241958, ECO:0000269|PubMed:2703538, ECO:0000269|PubMed:7883996, ECO:0000269|PubMed:8037208, ECO:0000269|PubMed:8161368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akçaağaç Şurubu İdrar Hastalığı, Otozomal Resesif Tip 1a, 248600; MSUD (Akçaağaç Şurubu İdrar Hastalığı) (Prenatal) (BCKDHA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Maple syrup urine disease, type Ia, 248600, Autosomal recessive; MSUD (Maple syrup urine disease) (BCKDHA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCKDHA	BCKDHA, MSUD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Maple syrup urine disease 1A (MSUD1A) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:1867199, ECO:0000269|PubMed:1885764, ECO:0000269|PubMed:2060625, ECO:0000269|PubMed:21844576, ECO:0000269|PubMed:2241958, ECO:0000269|PubMed:2703538, ECO:0000269|PubMed:7883996, ECO:0000269|PubMed:8037208, ECO:0000269|PubMed:8161368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akçaağaç Şurubu İdrar Hastalığı, Tip 1b, 248600, Otozomal Resesif (Akçaağaç Şurubu İdrar Hastalığı) (BCKDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maple syrup urine disease, type Ib, 248600, Autosomal recessive (Maple syrup urine disease) (BCKDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCKDHB	BCKDHB, E1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maple syrup urine disease 1B (MSUD1B) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:11509994, ECO:0000269|PubMed:22326532, ECO:0000269|PubMed:8161368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akçaağaç Şurubu İdrar Hastalığı, Tip 1b, 248600, Otozomal Resesif (Akçaağaç Şurubu İdrar Hastalığı) (Prenatal) (BCKDHB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Maple syrup urine disease, type Ib, 248600, Autosomal recessive (Maple syrup urine disease) (BCKDHB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCKDHB	BCKDHB, E1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Maple syrup urine disease 1B (MSUD1B) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:11509994, ECO:0000269|PubMed:22326532, ECO:0000269|PubMed:8161368}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Akçaağaç Şurubu İdrar Hastalığı, Tip 2, 248600, Otozomal Resesif (Akçaağaç Şurubu İdrar Hastalığı) (DBT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maple syrup urine disease, type II, 248600, Autosomal recessive (Maple syrup urine disease) (DBT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DBT	DBT, BCATE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maple syrup urine disease 2 (MSUD2) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:1847055, ECO:0000269|PubMed:9621512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Akçaağaç Şurubu İdrar Hastalığı, Tip 2, 248600, Otozomal Resesif (Akçaağaç Şurubu İdrar Hastalığı (Prenatal) (DBT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Maple syrup urine disease, type II, 248600, Autosomal recessive (Maple syrup urine disease) (DBT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DBT	DBT, BCATE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Maple syrup urine disease 2 (MSUD2) [MIM:248600]: A metabolic disorder due to an enzyme defect in the catabolic pathway of the branched-chain amino acids leucine, isoleucine, and valine. Accumulation of these 3 amino acids and their corresponding keto acids leads to encephalopathy and progressive neurodegeneration. Clinical features include mental and physical retardation, feeding problems, and a maple syrup odor to the urine. The keto acids of the branched-chain amino acids are present in the urine. If untreated, maple syrup urine disease can lead to seizures, coma, and death. The disease is often classified by its pattern of signs and symptoms. The most common and severe form of the disease is the classic type, which becomes apparent soon after birth. Variant forms of the disorder become apparent later in infancy or childhood and are typically milder, but they still involve developmental delay and other medical problems if not treated. {ECO:0000269|PubMed:1847055, ECO:0000269|PubMed:9621512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Marden-Walker Sendromu, 248700, Otozomal Dominant; MWKS (Marden-Walker Sendromu) (PIEZO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marden-Walker syndrome, 248700, Autosomal dominant; MWKS (Marden-Walker syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 5 (DA5) [MIM:108145]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA5 features include ocular abnormalities, typically ptosis, ophthalmoplegia and/or strabismus, in addition to contractures of the skeletal muscles. Some patients have pulmonary hypertension as a result of restrictive lung disease. {ECO:0000269|PubMed:23487782}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Marden-Walker Sendromu, 248700, Otozomal Dominant; MWKS (Marden-Walker Sendromu) (PIEZO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Marden-Walker syndrome, 248700, Autosomal dominant; MWKS (Marden-Walker syndrome) (PIEZO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIEZO2	PIEZO2, FAM38B, DA5, DA3, MWKS, DAIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 5 (DA5) [MIM:108145]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA5 features include ocular abnormalities, typically ptosis, ophthalmoplegia and/or strabismus, in addition to contractures of the skeletal muscles. Some patients have pulmonary hypertension as a result of restrictive lung disease. {ECO:0000269|PubMed:23487782}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Marfan Lipodistrofi Sendromu, 616914, Otozomal Dominant; MFLS (Prematüre Yaşlanma ve Marfanoid Görünüm-Lipodistrofi Sendromu) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marfan lipodystrophy syndrome, 616914, Autosomal dominant; MFLS (Progeroid and marfanoid aspect-lipodystrophy syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marfan Lipodistrofi Sendromu, 616914, Otozomal Dominant; MFLS (Prematüre Yaşlanma ve Marfanoid Görünüm-Lipodistrofi Sendromu) (MLPA)	Marfan lipodystrophy syndrome, 616914, Autosomal dominant; MFLS (Progeroid and marfanoid aspect-lipodystrophy syndrome) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marfan sendromu (Miks 1) (FBN1 15q21.1, TGFBR2 3p22) (MLPA)/ Marfan sendromu (Miks 2) (FBN1 15q21.1) (MLPA)	Marfan syndrome (Mix 1) (FBN1 15q21.1, TGFBR2 3p22) (MLPA) / Marfan syndrome (Mix 2) (FBN1 15q21.1) (MLPA)	FBN1 15q21.1, TGFBR2 3p22	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Marfan Sendromu, 154700, Otozomal Dominant; MFS (Marfan Sendromu) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marfan syndrome, 154700, Autosomal dominant; MFS (Marfan syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marfan Sendromu, 154700, Otozomal Dominant; MFS (Marfan Sendromu) (MLPA)	Marfan syndrome, 154700, Autosomal dominant; MFS (Marfan syndrome) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marinesco-Sjögren Sendromu, 248800, Otozomal Resesif; MSS (Marinesco-Sjögren Sendromu) (SIL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marinesco-Sjogren syndrome, 248800, Autosomal recessive; MSS (Marinesco-Sjögren syndrome) (SIL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIL1	SIL1, BAP, MSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marinesco-Sjogren syndrome (MSS) [MIM:248800]: Autosomal recessive multisystem disorder which is characterized by cerebellar ataxia due to cerebellar atrophy, with Purkinje and granule cell loss and myopathy featuring marked muscle replacement with fat and connective tissue. Other cardinal features include bilateral cataracts, hypergonadotrophic hypogonadism and mild to severe mental retardation. Skeletal abnormalities, short stature, dysarthria, strabismus and nystagmus are also frequent findings. Mutational inactivation of this protein may result in ER stress- induced cell death signaling or malfunctioning chaperone machineries that mishandle client proteins which are critical for the organs targeted in MSS. {ECO:0000269|PubMed:16282977, ECO:0000269|PubMed:16282978}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marinesco-Sjögren Sendromu, 248800, Otozomal Resesif; MSS (Marinesco-Sjögren Sendromu) (Prenatal) (SIL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Marinesco-Sjogren syndrome, 248800, Autosomal recessive; MSS (Marinesco-Sjögren syndrome) (SIL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIL1	SIL1, BAP, MSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Marinesco-Sjogren syndrome (MSS) [MIM:248800]: Autosomal recessive multisystem disorder which is characterized by cerebellar ataxia due to cerebellar atrophy, with Purkinje and granule cell loss and myopathy featuring marked muscle replacement with fat and connective tissue. Other cardinal features include bilateral cataracts, hypergonadotrophic hypogonadism and mild to severe mental retardation. Skeletal abnormalities, short stature, dysarthria, strabismus and nystagmus are also frequent findings. Mutational inactivation of this protein may result in ER stress- induced cell death signaling or malfunctioning chaperone machineries that mishandle client proteins which are critical for the organs targeted in MSS. {ECO:0000269|PubMed:16282977, ECO:0000269|PubMed:16282978}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Marshall Sendromu, 154780, Otozomal Dominant; MRSHS (Marshall Sendromu) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marshall syndrome, 154780, Autosomal dominant; MRSHS (Marshall syndrome) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marshall syndrome (MRSHS) [MIM:154780]: An autosomal dominant disorder characterized by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia. Clinical features include short stature; flat or retruded midface with short, depressed nose, flat nasal bridge and anteverted nares; cleft palate with or without the Pierre Robin sequence; appearance of large eyes with ocular hypertelorism; cataracts, either congenital or juvenile; esotropia; high myopia; sensorineural hearing loss; spondyloepiphyseal abnormalities; calcification of the falx cerebri; ectodermal abnormalities, including defects in sweating and dental structures. {ECO:0000269|PubMed:10486316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Marshall Sendromu, 154780, Otozomal Dominant; MRSHS (Marshall Sendromu) (Prenatal) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Marshall syndrome, 154780, Autosomal dominant; MRSHS (Marshall syndrome) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Marshall syndrome (MRSHS) [MIM:154780]: An autosomal dominant disorder characterized by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia. Clinical features include short stature; flat or retruded midface with short, depressed nose, flat nasal bridge and anteverted nares; cleft palate with or without the Pierre Robin sequence; appearance of large eyes with ocular hypertelorism; cataracts, either congenital or juvenile; esotropia; high myopia; sensorineural hearing loss; spondyloepiphyseal abnormalities; calcification of the falx cerebri; ectodermal abnormalities, including defects in sweating and dental structures. {ECO:0000269|PubMed:10486316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Marshall-Smith Sendromu, 602535, Otozomal Dominant; MRSHSS (Marshall-Smith Sendromu) (NFIX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Marshall-Smith syndrome, 602535, Autosomal dominant; MRSHSS (Marshall-Smith syndrome) (NFIX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFIX	NFIX, NF1A, SOTOS2, MRSHSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marshall-Smith syndrome (MRSHSS) [MIM:602535]: A distinct malformation syndrome characterized by accelerated skeletal maturation, relative failure to thrive, respiratory difficulties, mental retardation, and unusual facies, including prominent forehead, shallow orbits, blue sclerae, depressed nasal bridge, and micrognathia. Additional skeletal findings include long and thin tubular bones, broad middle phalanges with relatively narrow distal phalanges, and scoliosis. {ECO:0000269	PubMed:20673863}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Marshall-Smith Sendromu, 602535, Otozomal Dominant; MRSHSS (Marshall-Smith sendromu) (Prenatal) (NFIX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Marshall-Smith syndrome, 602535, Autosomal dominant; MRSHSS (Marshall-Smith syndrome) (NFIX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFIX	NFIX, NF1A, SOTOS2, MRSHSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Marshall-Smith syndrome (MRSHSS) [MIM:602535]: A distinct malformation syndrome characterized by accelerated skeletal maturation, relative failure to thrive, respiratory difficulties, mental retardation, and unusual facies, including prominent forehead, shallow orbits, blue sclerae, depressed nasal bridge, and micrognathia. Additional skeletal findings include long and thin tubular bones, broad middle phalanges with relatively narrow distal phalanges, and scoliosis. {ECO:0000269	PubMed:20673863}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Martsolf Sendromu, 212720, Otozomal Resesif (Kataraktİ-Entelektüel Yetersizlik-Hipogonadizm Sendromu (Martsolf Sendromu)) (RAB3GAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Martsolf syndrome, 212720, Autosomal recessive (Cataract-Intellectual disability-hypogonadism syndrome) (RAB3GAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB3GAP2	RAB3GAP2, RAB3GAP150, p150, KIAA0839, WARBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Warburg micro syndrome 2 (WARBM2) [MIM:614225]: A rare syndrome characterized by microcephaly, microphthalmia, microcornia, congenital cataracts, optic atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, and hypogonadism. {ECO:0000269	PubMed:20967465}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Martsolf Sendromu, 212720, Otozomal Resesif (Kataraktİ-Entelektüel Yetersizlik-Hipogonadizm Sendromu (Martsolf Sendromu)) (Prenatal) (RAB3GAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Martsolf syndrome, 212720, Autosomal recessive (Cataract-Intellectual disability-hypogonadism syndrome) (RAB3GAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB3GAP2	RAB3GAP2, RAB3GAP150, p150, KIAA0839, WARBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Warburg micro syndrome 2 (WARBM2) [MIM:614225]: A rare syndrome characterized by microcephaly, microphthalmia, microcornia, congenital cataracts, optic atrophy, cortical dysplasia, in particular corpus callosum hypoplasia, severe mental retardation, spastic diplegia, and hypogonadism. {ECO:0000269	PubMed:20967465}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MASA Sendromu, 303350, X'e Bağlı (MASA Sendromu) (L1CAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	MASA syndrome, 303350, X-linked recessive (MASA-syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MASA Sendromu, 303350, X'e Bağlı (MASA Sendromu) (Prenatal) (L1CAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	MASA syndrome, 303350, X-linked recessive (MASA syndrome) (L1CAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	L1CAM	L1CAM, CAML1, HSAS1, MASA, SPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, aphasia, shuffling gait, and adducted thumbs syndrome (MASA) [MIM:303350]: An X-linked recessive syndrome with a highly variable clinical spectrum. Main clinical features include spasticity and hyperreflexia of lower limbs, shuffling gait, mental retardation, aphasia and adducted thumbs. The features of spasticity have been referred to as complicated spastic paraplegia type 1 (SPG1). Some patients manifest corpus callosum hypoplasia and hydrocephalus. Inter- and intrafamilial variability is very wide, such that patients with hydrocephalus, MASA, SPG1, and agenesis of corpus callosum can be present within the same family. {ECO:0000269|PubMed:10805190, ECO:0000269|PubMed:11857550, ECO:0000269|PubMed:16816908, ECO:0000269|PubMed:7920659, ECO:0000269|PubMed:7920660, ECO:0000269|PubMed:9268105, ECO:0000269|PubMed:9300653, ECO:0000269|PubMed:9452110, ECO:0000269|PubMed:9832035). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MASP2 Eksikliği, 613791, Otozomal Resesif (İmmün Yetmezlik, MASP-2 Eksikliğine Bağlı) (MASP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MASP2 deficiency, 613791, Autosomal recessive (Immunodeficiency due to MASP-2 deficiency) (MASP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MASP2	MASP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: MASP2 deficiency (MASPD) [MIM:613791]: A disorder that results in autoimmune manifestations, recurrent severe infections, and chronic inflammatory disease. {ECO:0000269|PubMed:12904520, ECO:0000269|PubMed:17252003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MASP2 Eksikliği, 613791, Otozomal Resesif (İmmün Yetmezlik, MASP-2 Eksikliğine Bağlı) (Prenatal) (MASP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MASP2 deficiency, 613791, Autosomal recessive (Immunodeficiency due to MASP-2 deficiency) (MASP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MASP2	MASP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: MASP2 deficiency (MASPD) [MIM:613791]: A disorder that results in autoimmune manifestations, recurrent severe infections, and chronic inflammatory disease. {ECO:0000269|PubMed:12904520, ECO:0000269|PubMed:17252003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MASS Sendromu, 604308 (MASS Sendromu) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MASS syndrome, 604308 (MASS syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269	PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MASS Sendromu, 604308 (MASS Sendromu) (MLPA)	MASS syndrome, 604308 (MASS syndrome) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269	PubMed:21683322}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mast Hücresi Hastalığı, 154800, Otozomal Dominant (Makülopapüler Kutanöz Mastositoz) (KIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mast cell disease, 154800, Autosomal dominant (Maculopapular cutaneous mastocytosis) (KIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIT	KIT, PBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Piebald trait (PBT) [MIM:172800]: Autosomal dominant genetic developmental abnormality of pigmentation characterized by congenital patches of white skin and hair that lack melanocytes. {ECO:0000269|PubMed:11074500, ECO:0000269|PubMed:1370874, ECO:0000269|PubMed:1378329, ECO:0000269|PubMed:1717985, ECO:0000269|PubMed:7687267, ECO:0000269|PubMed:8680409, ECO:0000269|PubMed:9450866, ECO:0000269|PubMed:9699740}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mast Sendromu, 248900, Otozomal Resesif (Otozomal Resesif Spastik Paraplejı Tip 21) (SPG21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mast syndrome, 248900, Autosomal recessive (Autosomal recessive spastic paraplegia type 21) (SPG21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG21	ACP33, MAST, SPG21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MODY Tip 6, 606394, MODY6 (MODY) (NEUROD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young 6, 606394; MODY6 (MODY) (NEUROD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEUROD1	NEUROD1, NIDDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 6 (MODY6) [MIM:606394]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269	PubMed:10540951}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MODY, Tip 10, 613370, Otozomal Dominant; MODY10 (MODY) (INS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type 10, 613370, Autosomal dominant; MODY10 (MODY) (INS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INS	INS, MODY10, IDDM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, insulin-dependent, 2 (IDDM2) [MIM:125852]: A multifactorial disorder of glucose homeostasis that is characterized by susceptibility to ketoacidosis in the absence of insulin therapy. Clinical features are polydipsia, polyphagia and polyuria which result from hyperglycemia-induced osmotic diuresis and secondary thirst. These derangements result in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:1819254O}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MODY, Tip 11, 613375, Otozomal Dominant; MODY11 (MODY) (BLK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type 11, 613375, Autosomal dominant; MODY11 (MODY) (BLK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BLK	BLK, MODY11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 11 (MODY11) [MIM:613375]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269	PubMed:19667185}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

MODY, Tip 13, 616329, Otozomal Dominant; MODY13 (MODY) (KCNJ11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type 13, 616329, Autosomal dominant; MODY13 (MODY) (KCNJ11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ11	KCNJ11, BIR, PHHI, HHF2, TNDM3, MODY13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 2 (HHF2) [MIM:601820]: Most common cause of persistent hypoglycaemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycaemia may occur. (ECO:0000269|PubMed:10204114, ECO:0000269|PubMed:12364426, ECO:0000269|PubMed:15562009, ECO:0000269|PubMed:15579781, ECO:0000269|PubMed:15807877, ECO:0000269|PubMed:15998776, ECO:0000269|PubMed:16332676, ECO:0000269|PubMed:16357843, ECO:0000269|PubMed:18596924, ECO:0000269|PubMed:19357197, ECO:0000269|PubMed:7847376, ECO:0000269|PubMed:8923010). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MODY, Maturity Onset Diyabet Of The Young, Tip 14, 616511, Otozomal Dominant; MODY14 (MODY) (APPL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type 14, 616511, Autosomal dominant; MODY14 (MODY) (APPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APPL1	APPL1, APPL, MODY14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
MODY, Tip 9, 612225, MODY9 (MODY) (PAX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type IX, 612225, MODY9 (MODY) (PAX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX4	PAX4, MODY9, KPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. (ECO:0000269	PubMed:11723072). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
MODY, Tip 7, 610508, MODY7 (MODY) (KLF11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type VII, 610508, MODY7 (MODY) (KLF11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF11	KLF11, TIEG2, FKLF1, FKLF, MODY7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 7 (MODY7) [MIM:610508]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. [ECO:0000269	PubMed:15774581]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
MODY, 8, 609812, Otozomal Dominant; MODY8 (MODY) (CEL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Maturity-onset diabetes of the young, type VIII, 609812, Autosomal dominant; MODY8 (MODY) (CEL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEL	CEL, BSSL, CELL, MODY8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
May-Hegglin Anomalisi, 155100, Otozomal Dominant; MHA (May Hegglin Trombositopeni) (MYH9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	May-Hegglin anomaly, 155100, Autosomal dominant; MHA (May-Hegglin thrombocytopenia) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. (ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mayer-Rokitansky-Küster-Hauser Sendromu (16p11.2; 17q12; 22q11.21) (MLPA)	Mayer-Rokitansky-Küster-Hauser Syndrome (16p11.2; 17q12; 22q11.21) (MLPA)	16p11.2; 17q12; 22q11.21	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Mcardle Hastalığı, 232600, Otozomal Resesif (Glikojen Depo Hastalığı, Kas Glikojen Fosforilaz Eksikliğine Bağlı, Mcardle Hastalığı) (PYGM geni) (Tüm Gen Dizi Analizi) (Postnatal)	McArdle disease, 232600, Autosomal recessive (Glycogen storage disease due to muscle glycogen phosphorylase deficiency) (PYGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYGM	PYGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 5 (GSD5) [MIM:232600]: A metabolic disorder resulting in myopathy characterized by exercise intolerance, cramps, muscle weakness and recurrent myoglobinuria. (ECO:0000269|PubMed:10382911, ECO:0000269|PubMed:10382912, ECO:0000269|PubMed:10417800, ECO:0000269|PubMed:10590419, ECO:0000269|PubMed:10681080, ECO:0000269|PubMed:10714589, ECO:0000269|PubMed:10899452, ECO:0000269|PubMed:11706962, ECO:0000269|PubMed:12031624, ECO:0000269|PubMed:7603523, ECO:0000269|PubMed:8316268, ECO:0000269|PubMed:8535454, ECO:0000269|PubMed:9506549). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mcardle Hastalığı, 232600, Otozomal Resesif (Glikojen Depo Hastalığı, Kas Glikojen Fosforilaz Eksikliğine Bağlı, Mcardle Hastalığı) (Prenatal) (PYGM geni) (Tüm Gen Dizi Analizi) (Prenatal)	McArdle disease, 232600, Autosomal recessive (Glycogen storage disease due to muscle glycogen phosphorylase deficiency) (PYGM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYGM	PYGM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 5 (GSD5) [MIM:232600]: A metabolic disorder resulting in myopathy characterized by exercise intolerance, cramps, muscle weakness and recurrent myoglobinuria. (ECO:0000269|PubMed:10382911, ECO:0000269|PubMed:10382912, ECO:0000269|PubMed:10417800, ECO:0000269|PubMed:10590419, ECO:0000269|PubMed:10681080, ECO:0000269|PubMed:10714589, ECO:0000269|PubMed:10899452, ECO:0000269|PubMed:11706962, ECO:0000269|PubMed:12031624, ECO:0000269|PubMed:7603523, ECO:0000269|PubMed:8316268, ECO:0000269|PubMed:8535454, ECO:0000269|PubMed:9506549). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mccune-Albright Sendromu, Somatik, Mozaik, 174800 (Mccune-Albright Sendromu) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	McCune-Albright syndrome, somatic, mosaic, 174800 (McCune-Albright syndrome) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Mccune-Albright Sendromu, Mas (Mccune-Albright Sendromu) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	MCCUNE-ALBRIGHT SYNDROME; MAS (McCune-Albright syndrome) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mckusick-Kaufman Sendromu, 236700, Otozomal Resesif; MKKS (Mckusick-Kaufman Sendromu) (MKKS geni) (Tüm Gen Dizi Analizi) (Postnatal)	McKusick-Kaufman syndrome, 236700, Autosomal recessive; MKKS (McKusick-Kaufman syndrome) (MKKS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKKS	MKKS, HMCS, KMS, MKS, BBS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: McKusick-Kaufman syndrome (MKKS) [MIM:236700]: Autosomal recessive developmental disorder. It is characterized by hydrometrocolpos, postaxial polydactyly and congenital heart defects. (ECO:0000269	PubMed:10802661). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mckusick-Kaufman Sendromu, 236700, Otozomal Resesif; MKKS (Mckusick-Kaufman Sendromu) (Prenatal) (MKKS geni) (Tüm Gen Dizi Analizi) (Prenatal)	McKusick-Kaufman syndrome, 236700, Autosomal recessive; MKKS (McKusick-Kaufman syndrome) (MKKS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MKKS	MKKS, HMCS, KMS, MKS, BBS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: McKusick-Kaufman syndrome (MKKS) [MIM:236700]: Autosomal recessive developmental disorder. It is characterized by hydrometrocolpos, postaxial polydactyly and congenital heart defects. (ECO:0000269	PubMed:10802661). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mcleod Sendromu, ± Kronik Granülomatoz Hastalığı, 300842, X'e Bağlı; MCLDS (Mcleod Nöroakantositoz Sendromu) (XK geni) (Tüm Gen Dizi Analizi) (Postnatal)	McLeod syndrome with or without chronic granulomatous disease, 300842, X-linked; MCLDS (McLeod neuroacanthocytosis syndrome) (XK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XK	XK, MCLDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: McLeod syndrome (MLS) [MIM:300842]: A multisystem disorder characterized by the absence of red blood cell Kx antigen, weak expression of Kell red blood cell antigens, acanthocytosis, and compensated hemolysis. Most carriers of this McLeod blood group phenotype have acanthocytosis and elevated serum creatine kinase levels and are prone to develop a severe neurologic disorder resembling Huntington disease. Additional symptoms include generalized seizures, neuromuscular symptoms leading to weakness and atrophy, and cardiomyopathy mainly manifesting with atrial fibrillation, malignant arrhythmias, and dilated cardiomyopathy. (ECO:0000269|PubMed:11761473, ECO:0000269|PubMed:11961232, ECO:0000269|PubMed:12823753). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mcleod Sendromu, ± Kronik Granülomatoz Hastalığı, 300842, X'e Bağlı; MCLDS (Mcleod Nöroakantositoz Sendromu) (Prenatal) (XK geni) (Tüm Gen Dizi Analizi) (Prenatal)	McLeod syndrome with or without chronic granulomatous disease, 300842, X-linked; MCLDS (McLeod neuroacanthocytosis syndrome) (XK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XK	XK, MCLDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: McLeod syndrome (MLS) [MIM:300842]: A multisystem disorder characterized by the absence of red blood cell Kx antigen, weak expression of Kell red blood cell antigens, acanthocytosis, and compensated hemolysis. Most carriers of this McLeod blood group phenotype have acanthocytosis and elevated serum creatine kinase levels and are prone to develop a severe neurologic disorder resembling Huntington disease. Additional symptoms include generalized seizures, neuromuscular symptoms leading to weakness and atrophy, and cardiomyopathy mainly manifesting with atrial fibrillation, malignant arrhythmias, and dilated cardiomyopathy. (ECO:0000269	PubMed:11761473, ECO:0000269	PubMed:11961232, ECO:0000269	PubMed:12823753). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meacham Sendromu, 608978 (Meacham Sendromu) (WT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meacham syndrome, 608978 (Meacham syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. (ECO:0000269	PubMed:10571943). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meacham Sendromu, 608978 (Meacham Sendromu) (MLPA)	Meacham syndrome, 608978 (Meacham syndrome) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. (ECO:0000269	PubMed:10571943). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Meacham Sendromu, 608978 (Meacham Sendromu) (Prenatal) (WT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meacham syndrome, 608978 (Meacham syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meacham Sendromu, 608978 (Meacham Sendromu) (Prenatal) (MLPA)	Meacham syndrome, 608978 (Meacham syndrome) (Prenatal) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269	PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 1, 249000, Otozomal Resesif; MKS1 (Meckel Sendromu) (MKS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 1, 249000, Autosomal recessive; MKS1 (Meckel syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 1 (MKS1) [MIM:249000]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269|PubMed:16415886, ECO:0000269|PubMed:19466712}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 1, 249000, Otozomal Resesif; MKS1 (Meckel Sendromu) (Prenatal) (MKS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 1, 249000, Autosomal recessive; MKS1 (Meckel syndrome) (MKS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MKS1	MKS1, MKS, BBS13, JBTS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 1 (MKS1) [MIM:249000]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269|PubMed:16415886, ECO:0000269|PubMed:19466712}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 10, 614175, Otozomal Resesif (Meckel Sendromu) (B9D2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 10, 614175, Autosomal recessive (Meckel syndrome) (B9D2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B9D2	B9D2, MKS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 10 (MKS10) [MIM:614175]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21763481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meckel Sendromu 10, 614175, Otozomal Resesif (Meckel Sendromu) (Prenatal) (B9D2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 10, 614175, Autosomal recessive (Meckel syndrome) (B9D2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B9D2	B9D2, MKS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 10 (MKS10) [MIM:614175]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:21763481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 11, 615397, Otozomal Resesif; MKS11 (Meckel Sendromu) (TMEM231 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 11, 615397, Autosomal recessive; MKS11 (Meckel syndrome) (TMEM231 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM231	TMEM231, JBTS20, MKS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 20 (JBTS20) [MIM:614970]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:23012439}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 11, 615397, Otozomal Resesif; MKS11 (Meckel Sendromu) (Prenatal) (TMEM231 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 11, 615397, Autosomal recessive; MKS11 (Meckel syndrome) (TMEM231 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM231	TMEM231, JBTS20, MKS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 20 (JBTS20) [MIM:614970]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. {ECO:0000269	PubMed:23012439}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 12, 616258, Otozomal Resesif; MKS12 (Letal Fetal Serebroreonogenitourinер Agenezi / Hipoplazi Sendromu) (KIF14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 12, 616258, Autosomal recessive; MKS12 (Lethal fetal cerebroreonogenitourinary agenesis/hypoplasia syndrome) (KIF14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF14	KIF14, KIAA0042, MKS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 12 (MKS12) [MIM:616258]: A form of Meckel syndrome, a disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:24128419}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meckel Sendromu 12, 616258, Otozomal Resesif; MKS12 (Letal Fetal Serebroreonogenitourinер Agenezi / Hipoplazi Sendromu) (Prenatal) (KIF14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 12, 616258, Autosomal recessive; MKS12 (Lethal fetal cerebroreonogenitourinary agenesis/hypoplasia syndrome) (KIF14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF14	KIF14, KIAA0042, MKS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 12 (MKS12) [MIM:616258]: A form of Meckel syndrome, a disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:24128419}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 2, 603194, Otozomal Resesif; MKS2 (Meckel Sendromu) (TMEM216 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 2, 603194, Autosomal recessive; MKS2 (Meckel syndrome) (TMEM216 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM216	TMEM216, JBTS2, CORS2, MKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 2 (MKS2) [MIM:603194]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:20512146}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meckel Sendromu 2, 603194, Otozomal Resesif; MKS2 (Meckel Sendromu) (Prenatal) (TMEM216 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 2, 603194, Autosomal recessive; MKS2 (Meckel syndrome) (TMEM216 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM216	TMEM216, JBTS2, CORS2, MKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 2 (MKS2) [MIM:603194]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. {ECO:0000269	PubMed:20512146}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 3, 607361, Otozomal Resesif; MKS3 (Meckel Sendromu) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 3, 607361, Autosomal recessive; MKS3 (Meckel syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:18327255}. Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	EDTA Blood Sample
Meckel Sendromu 3, 607361, Otozomal Resesif; MKS3 (Meckel Sendromu) (Prenatal) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 3, 607361, Autosomal recessive; MKS3 (Meckel syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269	PubMed:18327255}. Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 4, 611134, Otozomal Resesif; MKS4 (Meckel Sendromu) (CEP290 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 4, 611134, Autosomal recessive; MKS4 (Meckel syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 4, 611134, Otozomal Resesif; MKS4 (Meckel Sendromu) (Prenatal) (CEP290 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 4, 611134, Autosomal recessive; MKS4 (Meckel syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Meckel Sendromu 5, 611561, Otozomal Resesif; MKS5 (Meckel Sendromu) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 5, 611561, Autosomal recessive; MKS5 (Meckel syndrome) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. (ECO:0000269	PubMed:19574260). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 5, 611561, Otozomal Resesif; MKS5 (Meckel Sendromu) (Prenatal) (RPGRIP1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 5, 611561, Autosomal recessive; MKS5 (Meckel syndrome) (RPGRIP1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPGRIP1L	RPGRIP1L, KIAA1005, JBTS7, MKS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. (ECO:0000269	PubMed:19574260). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 6, 612284, Otozomal Resesif; MKS6 (Meckel Sendromu) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 6, 612284, Autosomal recessive; MKS6 (Meckel syndrome) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. (ECO:0000269	PubMed:19574260). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 6, 612284, Otozomal Resesif; MKS6 (Meckel Sendromu) (Prenatal) (CC2D2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 6, 612284, Autosomal recessive; MKS6 (Meckel syndrome) (CC2D2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CC2D2A	CC2D2A, KIAA1345, MKS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: COACH syndrome (COACHS) [MIM:216360]: A disorder characterized by mental retardation, ataxia due to cerebellar hypoplasia, and hepatic fibrosis. Patients present the molar tooth sign, a midbrain-hindbrain malformation pathognomonic for Joubert syndrome and related disorders. Other features, such as coloboma and renal cysts, may be variable. (ECO:0000269	PubMed:19574260). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 7, 267010, Otozomal Resesif; MKS7 (NPHP3 İlişkili Meckel Benzeri Sendrom) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 7, 267010, Autosomal recessive; MKS7 (NPHP3-related Meckel-like syndrome) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:18371931]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 7, 267010, Otozomal Resesif; MKS7 (NPHP3 İlişkili Meckel Benzeri Sendrom) (Prenatal) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 7, 267010, Autosomal recessive; MKS7 (NPHP3-related Meckel-like syndrome) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:18371931]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 8, 613885, Otozomal Resesif; MKS8 (Meckel Sendromu) (TCTN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 8, 613885, Autosomal recessive; MKS8 (Meckel syndrome) (TCTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCTN2	TCTN2, TECT2, MKS8, JBTS24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 8 (MKS8) [MIM:613885]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. [ECO:0000269	PubMed:21462283]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meckel Sendromu 8, 613885, Otozomal Resesif; MKS8 (Meckel Sendromu) (Prenatal) (TCTN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 8, 613885, Autosomal recessive; MKS8 (Meckel syndrome) (TCTN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCTN2	TCTN2, TECT2, MKS8, JBTS24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 8 (MKS8) [MIM:613885]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. [ECO:0000269	PubMed:21462283]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu 9, 614209, Otozomal Resesif; MKS9 (Meckel Sendromu) (B9D1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meckel syndrome 9, 614209, Autosomal recessive; MKS9 (Meckel syndrome) (B9D1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B9D1	B9D1, MKSR1, MKS9, JBTS27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 9 (MKS9) [MIM:614209]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. [ECO:0000269	PubMed:21493627]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Meckel Sendromu 9, 614209, Otozomal Resesif; MKS9 (Meckel Sendromu) (Prenatal) (B9D1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meckel syndrome 9, 614209, Autosomal recessive; MKS9 (Meckel syndrome) (B9D1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B9D1	B9D1, MKSR1, MKS9, JBTS27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meckel syndrome 9 (MKS9) [MIM:614209]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. [ECO:0000269	PubMed:21493627]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meckel Sendromu, Tip 10; Mks10 (Meckel Sendromu) (B9D2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MECKEL SYNDROME, TYPE 10; MKS10 (Meckel syndrome) (B9D2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B9D2	B9D2, MKS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meckel syndrome 10 (MKS10) [MIM:614175]: A disorder characterized by a combination of renal cysts and variably associated features including developmental anomalies of the central nervous system (typically encephalocele), hepatic ductal dysplasia and cysts, and polydactyly. [ECO:0000269	PubMed:21763481]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mekonyum İleusu, 614665, Otozomal Resesif (Neonatalin Bağırsak Tıkanıklığı, Guanilat Siklaz 2C Eksikliğine Bağlı) (GUCY2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meconium ileus, 614665, Autosomal recessive (Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency) (GUCY2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCY2C	GUCY2C, GUC2C, DIAR6, MECIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 6 (DIAR6) [MIM:614616]: A relatively mild, early-onset chronic diarrhea that may be associated with increased susceptibility to inflammatory bowel disease, small bowel obstruction, and esophagitis. [ECO:0000269	PubMed:22436048]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mekonyum İleusu, 614665, Otozomal Resesif (Neonatalin Bağırsak Tıkanıklığı, Guanilat Siklaz 2C Eksikliğine Bağlı) (Prenatal) (GUCY2C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meconium ileus, 614665, Autosomal recessive (Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency) (GUCY2C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GUCY2C	GUCY2C, GUC2C, DIAR6, MECIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 6 (DIAR6) [MIM:614616]: A relatively mild, early-onset chronic diarrhea that may be associated with increased susceptibility to inflammatory bowel disease, small bowel obstruction, and esophagitis. [ECO:0000269	PubMed:22436048]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MEDNİK Sendromu, 609313, Otozomal Resesif (Mednik Sendromu) (AP1S1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MEDNIK syndrome, 609313, Autosomal recessive (MEDNIK syndrome) (AP1S1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP1S1	AP1S1, CLAPS1, AP19, MEDNIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK) [MIM:609313]: A disorder characterized by erythematous skin lesions and hyperkeratosis, severe psychomotor retardation, peripheral neuropathy, sensorineural hearing loss, together with elevated very-long-chain fatty acids and severe congenital diarrhea. (ECO:0000269	PubMed:19057675). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MEDNİK Sendromu, 609313, Otozomal Resesif (Mednik Sendromu) (Prenatal) (AP1S1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MEDNIK syndrome, 609313, Autosomal recessive (MEDNIK syndrome) (AP1S1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP1S1	AP1S1, CLAPS1, AP19, MEDNIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, enteropathy, deafness, peripheral neuropathy, ichthyosis, and keratoderma (MEDNIK) [MIM:609313]: A disorder characterized by erythematous skin lesions and hyperkeratosis, severe psychomotor retardation, peripheral neuropathy, sensorineural hearing loss, together with elevated very-long-chain fatty acids and severe congenital diarrhea. (ECO:0000269	PubMed:19057675). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Medüller Kistik Böbrek Hastalığı 1, 174000, Otozomal Dominant; MCKD1 (Otozomal Dominant Medüller Kistik Böbrek Hastalık ± Hiperürisemi) (MUC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Medullary cystic kidney disease 1, 174000, Autosomal dominant; MCKD1 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (MUC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUC1	MUC1, PUM, MCKD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Medullary cystic kidney disease 1 (MCKD1) [MIM:174000]: A form of tubulointerstitial nephropathy characterized by formation of renal cysts at the corticomedullary junction. It is characterized by adult onset of impaired renal function and salt wasting resulting in end-stage renal failure by the sixth decade. (ECO:0000269	PubMed:23396133). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Medüller Kistik Böbrek Hastalığı 1, 174000, Otozomal Dominant; MCKD1 (Otozomal Dominant Medüller Kistik Böbrek Hastalık ± Hiperürisemi) (Prenatal) (MUC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Medullary cystic kidney disease 1, 174000, Autosomal dominant; MCKD1 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (MUC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MUC1	MUC1, PUM, MCKD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Medullary cystic kidney disease 1 (MCKD1) [MIM:174000]: A form of tubulointerstitial nephropathy characterized by formation of renal cysts at the corticomedullary junction. It is characterized by adult onset of impaired renal function and salt wasting resulting in end-stage renal failure by the sixth decade. (ECO:0000269	PubMed:23396133). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Medüller Kistik Böbrek Hastalığı 2, 603860; MCKD2 (Otozomal Dominant Medüller Kistik Böbrek Hastalık ± Hiperürisemi) (UMOD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medullary cystic kidney disease 2, 603860; MCKD2 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (UMOD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UMOD	UMOD, HNFJ1, FJHN, MCKD2, ADMCKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial juvenile hyperuricemic nephropathy 1 (HNFJ1) [MIM:162000]: A renal disease characterized by juvenile onset of hyperuricemia, polyuria, progressive renal failure, and gout. The disease is associated with interstitial pathological changes resulting in fibrosis. (ECO:0000269|PubMed:12471200, ECO:0000269|PubMed:12629136, ECO:0000269|PubMed:12900848, ECO:0000269|PubMed:14569098, ECO:0000269|PubMed:14570709, ECO:0000269|PubMed:15086896, ECO:0000269|PubMed:15575003, ECO:0000269|PubMed:15983957, ECO:0000269|PubMed:17010121, ECO:0000269|PubMed:21060763, ECO:0000269|PubMed:22776760, ECO:0000269|PubMed:23197950, ECO:0000269|PubMed:23985501). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mediller Kistik Böbrek Hastalığı 2, 603860; MCKD2 (Otozomal Dominant Mediller Kistik Böbrek Hastalık ± Hiperürisemi) (Prenatal) (UMOD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Medullary cystic kidney disease 2, 603860; MCKD2 (Autosomal dominant medullary cystic kidney disease with or without hyperuricemia) (UMOD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UMOD	UMOD, HNFJ1, FJHN, MCKD2, ADMCKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial juvenile hyperuricemic nephropathy 1 (HNF1I) [MIM:162000]: A renal disease characterized by juvenile onset of hyperuricemia, polyuria, progressive renal failure, and gout. The disease is associated with interstitial pathological changes resulting in fibrosis. {ECO:0000269|PubMed:12471200, ECO:0000269|PubMed:12629136, ECO:0000269|PubMed:12900848, ECO:0000269|PubMed:14569098, ECO:0000269|PubMed:14570709, ECO:0000269|PubMed:15086896, ECO:0000269|PubMed:15575003, ECO:0000269|PubMed:15983957, ECO:0000269|PubMed:17010121, ECO:0000269|PubMed:21060763, ECO:0000269|PubMed:22776760, ECO:0000269|PubMed:23197950, ECO:0000269|PubMed:23988501}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mediller Tiroid Karsinoması, 155240, Otozomal Dominant (Multipl Endokrin Neoplazi Tip 2) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medullary thyroid carcinoma, 155240, Autosomal dominant (Multiple endocrine neoplasia type 2) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mediller Tiroid Karsinoması, 155240, Otozomal Dominant (Multipl Endokrin Neoplazi Tip 2) (MLPA)	Medullary thyroid carcinoma, 155240, Autosomal dominant (Multiple endocrine neoplasia type 2) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mediller Tiroid Karsinoması, Ailesel, 155240, Otozomal Dominant (Ailesel Mediller Tiroid Kanseri) (NTRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medullary thyroid carcinoma, familial, 155240, Autosomal dominant; MTC (Familial medullary thyroid carcinoma) (NTRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTRK1	NTRK1, TRKA, MTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital insensitivity to pain with anhidrosis (CIPA) [MIM:256800]: Characterized by a congenital insensitivity to pain, anhidrosis (absence of sweating), absence of reaction to noxious stimuli, self-mutilating behavior, and mental retardation. This rare autosomal recessive disorder is also known as congenital sensory neuropathy with anhidrosis or hereditary sensory and autonomic neuropathy type IV or familial dysautonomia type II. {ECO:0000269|PubMed:10090906, ECO:0000269|PubMed:10233776, ECO:0000269|PubMed:10330344, ECO:0000269|PubMed:10567924, ECO:0000269|PubMed:10861667, ECO:0000269|PubMed:10982191, ECO:0000269|PubMed:11310631, ECO:0000269|PubMed:22302274, ECO:0000269|PubMed:8696348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Medulloblastoma, 155255, Otozomal Resesif, Otozomal Dominant; MDB (Medulloblastoma) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medulloblastoma, 155255, Autosomal recessive, Autosomal dominant; MDB (Medulloblastoma) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Medulloblastoma, 155255, Otozomal Resesif, Otozomal Dominant; MDB (Medulloblastoma) (MLPA)	Medulloblastoma, 155255, Autosomal recessive, Autosomal dominant; MDB (Medulloblastoma) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Medulloblastom, Desmoplastik, 155255, Otozomal Resesif, Otozomal Dominant (Medulloblastoma) (SUFU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medulloblastoma, desmoplastic, 155255, Autosomal recessive, Autosomal dominant (Medulloblastoma) (SUFU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUFU	SUFU, SUFUX1, SUFUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Medulloblastom, Somatik, 155255 (Klasik Medulloblastom) (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medulloblastoma, somatic, 155255 (Classic medulloblastoma) (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Medulloblastom, Somatik, 155255 (Medulloblastoma) (PTCH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Medulloblastoma, somatic, 155255 (Medulloblastoma) (PTCH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTCH2	PTCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Medulloblastoma (MDB) [MIM:155255]: Malignant, invasive embryonal tumor of the cerebellum with a preferential manifestation in children. {ECO:0000269|PubMed:9931336}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meesmann Korneal Distrofi, 122100, Otozomal Dominant (Meesmann Korneal Distrofisi) (KRT12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meesmann corneal dystrophy, 122100, Autosomal dominant (Meesmann corneal dystrophy) (KRT12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT12	KRT12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meesmann Korneal Distrofi, 122100, Otozomal Dominant; MECD (Meesmann Korneal Distrofisi) (KRT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meesmann corneal dystrophy, 122100, Autosomal dominant; MECD (Meesmann corneal dystrophy) (KRT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT3	KRT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meester-Loeys Sendromu, 300989, X'e Bağlı. MRLS (BGN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meester-Loeys syndrome, 300989, X-linked; MRLS (BGN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BGN	BGN, SEMDX, MRLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meester-Loeys Sendromu, 300989, X'e Bağlı. MRLS (BGN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meester-Loeys syndrome, 300989, X-linked; MRLS (BGN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BGN	BGN, SEMDX, MRLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megakaryoblastik Lösemi, Akut. (MKL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megakaryoblastic leukemia, acute (MKL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKL1	MKL1, AMKL, MAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MKL1 may be a cause of acute megakaryoblastic leukemia. Translocation t(1;22)(p13;q13) with RBM15. Although both reciprocal fusion transcripts are detected in acute megakaryoblastic leukemia (AMKL, FAB-M7), the RBM15-MKL1 chimeric protein has all the putative functional domains encoded by each gene and is the candidate oncogene.;	EDTA Blood Sample
Megakaryoblastik Lösemi, Akut, 606077 (RBM15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megakaryoblastic leukemia, acute, 606077 (RBM15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBM15	RBM15, SPEN, OTT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving RBM15 may be a cause of acute megakaryoblastic leukemia. Translocation t(1;22)(p13;q13) with MKL1. Although both reciprocal fusion transcripts are detected in acute megakaryoblastic leukemia (AMKL, FAB-M7), the RBM15-MKL1 chimeric protein has all the putative functional domains encoded by each gene and is the candidate oncogene. {ECO:0000269|PubMed:11344311, ECO:0000269|PubMed:11431911}.	EDTA Blood Sample
Megalensefalik Lökoensefalopati-Subkortikal Kistler 2A, 613925, Otozomal Resesif; MLC2A (Megalensefalik Lökoensefalopati-Subkortikal Kist) (HEPACAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925, Autosomal recessive; MLC2A (Megalencephalic leukoencephalopathy with subcortical cysts) (HEPACAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEPACAM	HEPACAM, MLC2A, MLC2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukoencephalopathy, megalencephalic, with subcortical cysts, 2A (MLC2A) [MIM:613925]: A neurodegenerative disorder characterized by infantile-onset macrocephaly and later onset of motor deterioration, with ataxia and spasticity, seizures, and cognitive decline of variable severity. The brain appears swollen on magnetic resonance imaging with white-matter abnormalities and subcortical cysts, in all stages of the disease. {ECO:0000269|PubMed:21419380}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Megalensefalik Lökoensefalopati-Subkortikal Kistler 2A, 613925, Otozomal Resesif; MLC2A (Megalensefalik Lökoensefalopati-Subkortikal Kist) (Prenatal) (HEPACAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephalic leukoencephalopathy with subcortical cysts 2A, 613925, Autosomal recessive; MLC2A (Megalencephalic leukoencephalopathy with subcortical cysts) (HEPACAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HEPACAM	HEPACAM, MLC2A, MLC2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukoencephalopathy, megalencephalic, with subcortical cysts, 2A (MLC2A) [MIM:613925]: A neurodegenerative disorder characterized by infantile-onset macrocephaly and later onset of motor deterioration, with ataxia and spasticity, seizures, and cognitive decline of variable severity. The brain appears swollen on magnetic resonance imaging with white-matter abnormalities and subcortical cysts, in all stages of the disease. {ECO:0000269|PubMed:21419380}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Megalensefalik Lökoensefalopati-Subkortikal Kistler 2B, Geri Dönüşlü, ± Mental Retardasyon, 613926, Otozomal Dominant; MLC2B (Megalensefalik Lökoensefalopati-Subkortikal Kist) (HEPACAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926, Autosomal dominant; MLC2B (Megalencephalic leukoencephalopathy with subcortical cysts) (HEPACAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEPACAM	HEPACAM, MLC2A, MLC2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukoencephalopathy, megalencephalic, with subcortical cysts, 2A (MLC2A) [MIM:613925]: A neurodegenerative disorder characterized by infantile-onset macrocephaly and later onset of motor deterioration, with ataxia and spasticity, seizures, and cognitive decline of variable severity. The brain appears swollen on magnetic resonance imaging with white-matter abnormalities and subcortical cysts, in all stages of the disease. (ECO:0000269	PubMed:21419380). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Megalensefalik Lökoensefalopati-Subkortikal Kistler 2B, Geri Dönüşlü, ± Mental Retardasyon, 613926, Otozomal Dominant; MLC2B (Megalensefalik Lökoensefalopati-Subkortikal Kist) (Prenatal) (HEPACAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephalic leukoencephalopathy with subcortical cysts 2B, remitting, with or without mental retardation, 613926, Autosomal dominant; MLC2B (Megalencephalic leukoencephalopathy with subcortical cysts) (HEPACAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HEPACAM	HEPACAM, MLC2A, MLC2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukoencephalopathy, megalencephalic, with subcortical cysts, 2A (MLC2A) [MIM:613925]: A neurodegenerative disorder characterized by infantile-onset macrocephaly and later onset of motor deterioration, with ataxia and spasticity, seizures, and cognitive decline of variable severity. The brain appears swollen on magnetic resonance imaging with white-matter abnormalities and subcortical cysts, in all stages of the disease. (ECO:0000269	PubMed:21419380). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefalik Lökoensefalopati, Subkortikal Kistler, 604004, Otozomal Resesif; MLC1 (Megalensefalik Lökoensefalopati-Subkortikal Kist) (MLC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephalic leukoencephalopathy with subcortical cysts, 604004, Autosomal recessive; MLC1 (Megalencephalic leukoencephalopathy with subcortical cysts) (MLC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLC1	MLC1, LVM, VL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Megalensefalik Lökoensefalopati, Subkortikal Kistler, 604004, Otozomal Resesif; MLC1 (Megalensefalik Lökoensefalopati-Subkortikal Kist) (MLPA)	Megalencephalic leukoencephalopathy with subcortical cysts, 604004, Autosomal recessive; MLC1 (Megalencephalic leukoencephalopathy with subcortical cysts) (MLPA)	MLC1	MLC1, LVM, VL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Megalensefalik Lökoensefalopati, Subkortikal Kistler, 604004, Otozomal Resesif; MLC1 (Megalensefalik Lökoensefalopati-Subkortikal Kist) (Prenatal) (MLC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephalic leukoencephalopathy with subcortical cysts, 604004, Autosomal recessive; MLC1 (Megalencephalic leukoencephalopathy with subcortical cysts) (MLC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MLC1	MLC1, LVM, VL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefalik Lökoensefalopati, Subkortikal Kistler, 604004, Otozomal Resesif; MLC1 (Megalensefalik Lökoensefalopati-Subkortikal Kist) (Prenatal) (MLPA)	Megalencephalic leukoencephalopathy with subcortical cysts, 604004, Autosomal recessive; MLC1 (Megalencephalic leukoencephalopathy with subcortical cysts) (Prenatal) (MLPA)	MLC1	MLC1, LVM, VL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefali-Kapiller Malformasyon-Polimikrogiri Sendromu, Somatik, 602501; MCAP (Megalensefali-Kapiller Malformasyon-Polimikrogiri Sendromu) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501; MCAP (Megalencephaly-capillary malformation-polymicrogyria syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	EDTA Blood Sample/ Biopsy of affected site-transport media
Megalensefali-Kapiller Malformasyon-Polimikrogiri Sendromu, Somatik, 602501; MCAP (Megalensefali-Kapiller Malformasyon-Polimikrogiri Sendromu) (Prenatal) (PIK3CA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501; MCAP (Megalencephaly-capillary malformation-polymicrogyria syndrome) (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269	PubMed:16353168). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 1, 603387, Otozomal Dominant; MPPH1 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (PIK3R2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387, Autosomal dominant; MPPH1 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (PIK3R2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3R2	PIK3R2, MPPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 (MPPH1) [MIM:603387]: A syndrome characterized by megalencephaly, hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269	PubMed:22729224). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 1, 603387, Otozomal Dominant; MPPH1 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (Prenatal) (PIK3R2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1, 603387, Autosomal dominant; MPPH1 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (PIK3R2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3R2	PIK3R2, MPPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 1 (MPPH1) [MIM:603387]: A syndrome characterized by megalencephaly, hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269	PubMed:22729224). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 2, 615937, Otozomal Dominant; MPPH2 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (AKT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937, Autosomal dominant; MPPH2 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (AKT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT3	AKT3, PKBG, MPPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2 (MPPH2) [MIM:615937]: A syndrome characterized by megalencephaly, hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269|PubMed:22500628, ECO:0000269|PubMed:22729223, ECO:0000269|PubMed:22729224). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 2, 615937, Otozomal Dominant; MPPH2 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (Prenatal) (AKT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937, Autosomal dominant; MPPH2 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (AKT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AKT3	AKT3, PKBG, MPPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2 (MPPH2) [MIM:615937]: A syndrome characterized by megalencephaly, hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269|PubMed:22500628, ECO:0000269|PubMed:22729223, ECO:0000269|PubMed:22729224). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 3, 615938, Otozomal Dominant; MPPH3 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (CCND2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938, Autosomal dominant; MPPH3 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (CCND2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCND2	CCND2, MPPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 (MPPH3) [MIM:615938]: A syndrome characterized by megalencephaly, ventriculomegaly that may lead to hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269	PubMed:24705253). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Megalensefali-Polimikrogiri-Polidaktili-Hidrosefali Sendromu 3, 615938, Otozomal Dominant; MPPH3 (Megalensefali-Polimikrogiri-Postaksiyel Polidaktili-Hidrosefali Sendromu) (Prenatal) (CCND2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3, 615938, Autosomal dominant; MPPH3 (Megalencephaly-polymicrogyria-postaxial polydactyly-hydrocephalus syndrome) (CCND2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCND2	CCND2, MPPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 3 (MPPH3) [MIM:615938]: A syndrome characterized by megalencephaly, ventriculomegaly that may lead to hydrocephalus, and polymicrogyria; polydactyly may also be seen. There is considerable phenotypic similarity between this disorder and the megalencephaly-capillary malformation syndrome. (ECO:0000269	PubMed:24705253). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megaloblastik Anemi, Dihidrofolat Redüktaz Eksikliğine Bağlı, 613839, Otozomal Resesif (Konstitüsyonel Megaloblastik Anemi-Ağır Nörolojik Hastalık) (DHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839, Autosomal recessive (Constitutional megaloblastic anemia with severe neurologic disease) (DHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHFR	DHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Megaloblastik Anemi, Dihidrofolat Redüktaz Eksikliğine Bağlı, 613839, Otozomal Resesif (Konstitüsyonel Megaloblastik Anemi-Ağır Nörolojik Hastalık) (Prenatal) (DHFR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megaloblastic anemia due to dihydrofolate reductase deficiency, 613839, Autosomal recessive (Constitutional megaloblastic anemia with severe neurologic disease) (DHFR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHFR	DHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megaloblastik Anemi-1, Fin Tipi, 261100, Otozomal Resesif (Grasbeck-Imerslund Hastalığı) (CUBN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megaloblastic anemia-1, Finnish type, 261100, Autosomal recessive (Gräsbeck-Imerslund disease) (CUBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CUBN	CUBN, IFCR, MGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Recessive hereditary megaloblastic anemia 1 (RH-MGA1) [MIM:261100]: Due to selective malabsorption of vitamin B12. Defects in vitamin B12 absorption lead to impaired function of thymidine synthase. As a consequence DNA synthesis is interrupted. Rapidly dividing cells involved in erythropoiesis are particularly affected. (ECO:0000269|PubMed:10080186, ECO:0000269|PubMed:10887099). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Megaloblastik Anemi-1, Fin Tipi, 261100, Otozomal Resesif (Grasbeck-Imerslund Hastalığı) (Prenatal) (CUBN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megaloblastic anemia-1, Finnish type, 261100, Autosomal recessive (Gräsbeck-Imerslund disease) (CUBN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CUBN	CUBN, IFCR, MGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Recessive hereditary megaloblastic anemia 1 (RH-MGA1) [MIM:261100]: Due to selective malabsorption of vitamin B12. Defects in vitamin B12 absorption lead to impaired function of thymidine synthase. As a consequence DNA synthesis is interrupted. Rapidly dividing cells involved in erythropoiesis are particularly affected. (ECO:0000269|PubMed:10080186, ECO:0000269|PubMed:10887099). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megaloblastik Anemi-1, Norveç Tipi, 261100, Otozomal Resesif (Grasbeck-Imerslund Hastalığı) (AMN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megaloblastic anemia-1, Norwegian type, 261100, Autosomal recessive (Gräsbeck-Imerslund disease) (AMN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMN	AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Megaloblastik Anemi-1, Norveç Tipi, 261100, Otozomal Resesif (Grasbeck-Imerslund Hastalığı) (Prenatal) (AMN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Megaloblastic anemia-1, Norwegian type, 261100, Autosomal recessive (Gräsbeck-Imerslund disease) (AMN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMN	AMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Megalokornea 1, X'e Bağlı, 309300, X'e Bağlı; MGC1 (özle Konjenital Megalokornea) (CHRDL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Megalocornea 1, X-linked, 309300, X-linked recessive; MGC1 (isolated congenital megalocornea) (CHRDL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRDL1	CHRDL1, VOPT, MGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Megalocornea 1, X-linked (MGC1) [MIM:309300]: An eye disorder in which the corneal diameter is bilaterally enlarged (greater than 13 mm) without an increase in intraocular pressure. It may also be referred to as anterior megalophthalmos, since the entire anterior segment is larger than normal. Features of megalocornea in addition to a deep anterior chamber include astigmatic refractive errors, atrophy of the iris stroma, miosis secondary to decreased function of the dilator muscle, iridodonesis, and tremulousness, subluxation, or dislocation of the lens. Whereas most affected individuals exhibit normal ocular function, complications include cataract development and glaucoma following lenticular dislocation or subluxation. (ECO:0000269	PubMed:22284829). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Meier-Gorlin Sendromu 1, 224690, Otozomal Resesif; MGORS1 (Kulak-Patella-Kısa Boy Sendromu) (ORC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 1, 224690, Autosomal recessive; MGORS1 (Ear-patella-short stature syndrome) (ORC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ORC1	ORC1, ORC1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meier-Gorlin syndrome 1 (MGORS1) [MIM:224690]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632, ECO:0000269|PubMed:21358633}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meier-Gorlin Sendromu 1, 224690, Otozomal Resesif; MGORS1 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (ORC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 1, 224690, Autosomal recessive; MGORS1 (Ear-patella-short stature syndrome) (ORC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ORC1	ORC1, ORC1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meier-Gorlin syndrome 1 (MGORS1) [MIM:224690]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632, ECO:0000269|PubMed:21358633}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 2, 613800, Otozomal Resesif; MGORS2 (Kulak-Patella-Kısa Boy Sendromu) (ORC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 2, 613800, Autosomal recessive; MGORS2 (Ear-patella-short stature syndrome) (ORC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ORC4	ORC4, ORC4L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meier-Gorlin syndrome 2 (MGORS2) [MIM:613800]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meier-Gorlin Sendromu 2, 613800, Otozomal Resesif; MGORS2 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (ORC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 2, 613800, Autosomal recessive; MGORS2 (Ear-patella-short stature syndrome) (ORC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ORC4	ORC4, ORC4L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meier-Gorlin syndrome 2 (MGORS2) [MIM:613800]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 3, 613803, Otozomal Resesif; MGORS3 (Kulak-Patella-Kısa Boy Sendromu) (ORC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 3, 613803, Autosomal recessive; MGORS3 (Ear-patella-short stature syndrome) (ORC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ORC6	ORC6, ORC6L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meier-Gorlin syndrome 3 (MGORS3) [MIM:613803]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meier-Gorlin Sendromu 3, 613803, Otozomal Resesif; MGORS3 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (ORC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 3, 613803, Autosomal recessive; MGORS3 (Ear-patella-short stature syndrome) (ORC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ORC6	ORC6, ORC6L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meier-Gorlin syndrome 3 (MGORS3) [MIM:613803]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 4, 613804, Otozomal Resesif; MGORS4 (Kulak-Patella-Kısa Boy Sendromu) (CDT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 4, 613804, Autosomal recessive; MGORS4 (Ear-patella-short stature syndrome) (CDT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDT1	CDT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meier-Gorlin syndrome 4 (MGORS4) [MIM:613804]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meier-Gorlin Sendromu 4, 613804, Otozomal Resesif; MGORS4 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (CDT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 4, 613804, Autosomal recessive; MGORS4 (Ear-patella-short stature syndrome) (CDT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDT1	CDT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meier-Gorlin syndrome 4 (MGORS4) [MIM:613804]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358631, ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 5, 613805, Otozomal Resesif; MGORS5 (Kulak-Patella-Kısa Boy Sendromu) (CDC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 5, 613805, Autosomal recessive; MGORS5 (Ear-patella-short stature syndrome) (CDC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC6	CDC6, CDC18L, MGORS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meier-Gorlin syndrome 5 (MGORS5) [MIM:613805]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meier-Gorlin Sendromu 5, 613805, Otozomal Resesif; MGORS5 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (CDC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 5, 613805, Autosomal recessive; MGORS5 (Ear-patella-short stature syndrome) (CDC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDC6	CDC6, CDC18L, MGORS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Meier-Gorlin syndrome 5 (MGORS5) [MIM:613805]: A syndrome characterized by bilateral microtia, aplasia/hypoplasia of the patellae, and severe intrauterine and postnatal growth retardation with short stature and poor weight gain. Additional clinical findings include anomalies of cranial sutures, microcephaly, apparently low-set and simple ears, microstomia, full lips, highly arched or cleft palate, micrognathia, genitourinary tract anomalies, and various skeletal anomalies. While almost all cases have primordial dwarfism with substantial prenatal and postnatal growth retardation, not all cases have microcephaly, and microtia and absent/hypoplastic patella are absent in some. Despite the presence of microcephaly, intellect is usually normal. {ECO:0000269|PubMed:21358632}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 6, 616835; MGORS6 (Kulak-Patella-Kısa Boy Sendromu) (GMNN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 6, 616835; MGORS6 (Ear-patella-short stature syndrome) (GMNN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GMNN	GMNN, MGORS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Meier-Gorlin Sendromu 6, 616835; MGORS6 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (GMNN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 6, 616835; MGORS6 (Ear-patella-short stature syndrome) (GMNN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GMNN	GMNN, MGORS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Meier-Gorlin Sendromu 7, 617063, Otozomal Resesif; MGORS7 (Kulak-Patella-Kısa Boy Sendromu) (CDC45 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meier-Gorlin syndrome 7, 617063, Autosomal recessive; MGORS7 (Ear-patella-short stature syndrome) (CDC45 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC45	CDC45L, CDC45L2, MGORS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meier-Gorlin Sendromu 7, 617063, Otozomal Resesif; MGORS7 (Kulak-Patella-Kısa Boy Sendromu) (Prenatal) (CDC45 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Meier-Gorlin syndrome 7, 617063, Autosomal recessive; MGORS7 (Ear-patella-short stature syndrome) (CDC45 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDC45	CDC45L, CDC45L2, MGORS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Melanositik Nevus Sendromu, Konjenital, Somatik, 137550 (Büyük Konjenital Melanositik Nevüs) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanocytic nevus syndrome, congenital, somatic, 137550 (Large congenital melanocytic nevus) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample/ Biopsy of affected site-transport media
Melanositik Nevus Sendromu, Konjenital, Cmns (Büyük Konjenital Melanositik Nevüs) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	MELANOCYTIC NEVUS SYNDROME, CONGENITAL, CMNS (Large congenital melanocytic nevus) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bladder cancer (BLC) [MIM:109800]: A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas that begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. {ECO:0000269|PubMed:6298635, ECO:0000269|PubMed:6844927}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom ve Sinir Sistemi Tümör Sendromu-Melanoma-Astrositoma Sendromu, 155755; Otozomal Dominant (Melanom ve Sinir Sistemi Tümör Sendromu) (CDKN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma and neural system tumor syndrome-MELANOMA-ASTROCYTOMA SYNDROME, 155755, Autosomal dominant (Melanoma and neural system tumor syndrome) (CDKN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN2A	CDKN2A, MTS1, P16, MLM, CMM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Li-Fraumeni syndrome (LFS) [MIM:151623]: Autosomal dominant familial cancer syndrome that in its classic form is defined by the existence of a proband affected by a sarcoma before 45 years with a first degree relative affected by any tumor before 45 years and another first degree relative with any tumor before 45 years or a sarcoma at any age. Other clinical definitions for LFS have been proposed (PubMed:8118819 and PubMed:8718514) and called Li-Fraumeni like syndrome (LFL). In these families affected relatives develop a diverse set of malignancies at unusually early ages. Four types of cancers account for 80% of tumors occurring in TP53 germline mutation carriers: breast cancers, soft tissue and bone sarcomas, brain tumors (astrocytomas) and adrenocortical carcinomas. Less frequent tumors include choroid plexus carcinoma or papilloma before the age of 15, rhabdomyosarcoma before the age of 5, leukemia, Wilms tumor, malignant phyllodes tumor, colorectal and gastric cancers. {ECO:0000269|PubMed:10484981}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 2, 155601, Otozomal Dominant; CMM2 (Ailesel Melanoma) (CDKN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, 2, 155601, Autosomal dominant; CMM2 (Familial melanoma) (CDKN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN2A	CDKN2A, MTS1, P16, MLM, CMM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Li-Fraumeni syndrome (LFS) [MIM:151623]: Autosomal dominant familial cancer syndrome that in its classic form is defined by the existence of a proband affected by a sarcoma before 45 years with a first degree relative affected by any tumor before 45 years and another first degree relative with any tumor before 45 years or a sarcoma at any age. Other clinical definitions for LFS have been proposed (PubMed:8118819 and PubMed:8718514) and called Li-Fraumeni like syndrome (LFL). In these families affected relatives develop a diverse set of malignancies at unusually early ages. Four types of cancers account for 80% of tumors occurring in TP53 germline mutation carriers: breast cancers, soft tissue and bone sarcomas, brain tumors (astrocytomas) and adrenocortical carcinomas. Less frequent tumors include choroid plexus carcinoma or papilloma before the age of 15, rhabdomyosarcoma before the age of 5, leukemia, Wilms tumor, malignant phyllodes tumor, colorectal and gastric cancers. {ECO:0000269|PubMed:10484981}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 3, 609048, Otozomal Dominant; CMM3 (Ailesel Melanoma) (CDK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, 3, 609048, Autosomal dominant; CMM3 (Familial melanoma) (CDK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDK4	CDK4, CMM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanoma, cutaneous malignant 3 (CMM3) [MIM:609048]: A malignant neoplasm of melanocytes, arising de novo or from a pre- existing benign nevus, which occurs most often in the skin but also may involve other sites. {ECO:0000269|PubMed:7652577, ECO:0000269|PubMed:8528263, ECO:0000269|PubMed:9311594, ECO:0000269|PubMed:9425228}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 3, 609048, Otozomal Dominant; CMM3 (Ailesel Melanoma) (MLPA)	Melanoma, cutaneous malignant, 3, 609048, Autosomal dominant; CMM3 (Familial melanoma) (MLPA)	CDK4	CDK4, CMM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Melanoma, cutaneous malignant 3 (CMM3) [MIM:609048]: A malignant neoplasm of melanocytes, arising de novo or from a pre- existing benign nevus, which occurs most often in the skin but also may involve other sites. {ECO:0000269|PubMed:7652577, ECO:0000269|PubMed:8528263, ECO:0000269|PubMed:9311594, ECO:0000269|PubMed:9425228}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 5, 613099; CMM5 (Ailesel Melanoma) (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, 5, 613099; CMM5 (Familial melanoma) (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanoma, cutaneous malignant 5 (CMM5) [MIM:613099]: A malignant neoplasm of melanocytes, arising de novo or from a pre- existing benign nevus, which occurs most often in the skin but also may involve other sites. {ECO:0000269|PubMed:17434924, ECO:0000269|PubMed:19338054}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 6, 613972; CMM6 (Ailesel Melanoma) (XRCC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, 6, 613972; CMM6 (Familial melanoma) (XRCC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XRCC3	XRCC3, CMM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12023982}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, 9, 615134; CMM9 (Ailesel Melanoma) (TERT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, 9, 615134; CMM9 (Familial melanoma) (TERT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, Yatkınlık, 10, 615848, Otozomal Dominant; CMM10 (Ailesel Melanoma) (POT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, susceptibility to, 10, 615848, Autosomal dominant; CMM10 (Familial melanoma) (POT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POT1	POT1, CMM10, GLM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Melanom, Kutanöz Malign, Yatkınlık 8, 601800 (Oküler Albinizm-Konjenital Sensorinöral Bağırlık) (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, susceptibility to, 8, 601800 (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11296837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:15146472, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1893921, ECO:0000269|PubMed:1943686, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:23504663, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Melanom, Kutanöz Malign, Yatkınlık 8, 601800 (Oküler Albinizm-Konjental Sensorinöral Sağırlık) (MLPA)	Melanoma, cutaneous malignant, susceptibility to, 8, 601800 (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Albinism, oculocutaneous, 1A (OCA1A) [MIM:203100]: An autosomal recessive disorder in which the biosynthesis of melanin pigment is absent in skin, hair, and eyes. It is characterized by complete lack of tyrosinase activity due to production of an inactive enzyme. Patients present with a life-long absence of melanin pigment after birth, and manifest increased sensitivity to ultraviolet radiation with predisposition to skin cancer. Visual anomalies include decreased acuity, nystagmus, strabismus and photophobia. {ECO:0000269|PubMed:10571953, ECO:0000269|PubMed:10671066, ECO:0000269|PubMed:10987646, ECO:0000269|PubMed:11295837, ECO:0000269|PubMed:11858948, ECO:0000269|PubMed:1487241, ECO:0000269|PubMed:1514647 2, ECO:0000269|PubMed:1642278, ECO:0000269|PubMed:1899321, ECO:0000269|PubMed:1943886, ECO:0000269|PubMed:1970634, ECO:0000269|PubMed:22981120, ECO:0000269|PubMed:2342539, ECO:0000269|PubMed:2350466 3, ECO:0000269|PubMed:24934919, ECO:0000269|PubMed:7902671, ECO:0000269|PubMed:7955413, ECO:0000269|PubMed:8128955, ECO:0000269|PubMed:8644824, ECO:0000269|PubMed:9259202}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, Yatkınlık 8, 614456; CMM8 (MITF İlişkili Melanom ve Böbrek Hücreli Karsinom Yatkınlık Sendromu) (MITF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, cutaneous malignant, susceptibility to, 8, 614456; CMM8 (MITF-related melanoma and renal-cell carcinoma predisposition syndrome) (MITF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Waardenburg syndrome 2, with ocular albinism, autosomal recessive (WS2-OA) [MIM:103470]: A disorder characterized by the association of features typical of Waardenburg syndrome type 2 with ocular albinism. Patients manifest reduced visual acuity, albinotic fundus, deafness, hypomelanosis. {ECO:0000269	PubMed:9647758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Kutanöz Malign, Yatkınlık 8, 614456; CMM8 (MITF İlişkili Melanom ve Böbrek Hücreli Karsinom Yatkınlık Sendromu) (MLPA)	Melanoma, cutaneous malignant, susceptibility to, 8, 614456; CMM8 (MITF-related melanoma and renal-cell carcinoma predisposition syndrome) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Waardenburg syndrome 2, with ocular albinism, autosomal recessive (WS2-OA) [MIM:103470]: A disorder characterized by the association of features typical of Waardenburg syndrome type 2 with ocular albinism. Patients manifest reduced visual acuity, albinotic fundus, deafness, hypomelanosis. {ECO:0000269	PubMed:9647758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanom, Malign, Somatik (STK11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, malignant, somatic (STK11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STK11	STK11, PJS, LKB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Peutz-Jeghers syndrome (PJS) [MIM:175200]: An autosomal dominant disorder characterized by melanocytic macules of the lips, multiple gastrointestinal hamartomatous polyps and an increased risk for various neoplasms, including gastrointestinal cancer. {ECO:0000269|PubMed:10408777, ECO:0000269|PubMed:12372054, ECO:0000269|PubMed:21411391, ECO:0000269|PubMed:9425897, ECO:0000269|PubMed:9428765, ECO:0000269|PubMed:9760200, ECO:0000269|PubMed:9837816}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Melanom, Malign, Somatik (BRAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melanoma, malignant, somatic (BRAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Melanozis, Nörokitan; Ncms (Nörokitan Melanositozis) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	MELANOSIS, NEUROCUTANEOUS; NCMS (Neurocutaneous melanocytosis) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melanozis, Nörokitan; Ncms (Nörokitan Melanositozis) (MLPA)	MELANOSIS, NEUROCUTANEOUS; NCMS (Neurocutaneous melanocytosis) (MLPA)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MELAS Hastalığı (A3243G, T3271C, C3093G, A3252G, C3256T, A3260G, T3291C, T3308C, A13514G) (Sık görülen Mutasyonlar) (Dizi Analizi) (Mitokondrial Genom geni) (Dizi Analizi) (Postnatal)	MELAS syndrome (Mitochondrial myopaty, encephalopathy, lactic acidosis and stroke-like episodes) (A3243G, T3271C, C3093G, A3252G, C3256T, A3260G, T3291C, T3308C, A13514G) (Frequent mutations) (Sequence analysis) (Mitokondrial Genom gene) (Dizi Analizi) (Postnatal)	Mitokondrial Genom	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meleda Hastalığı, 248300, Otozomal Resesif; MDM (Mal De Meleda) (SLURP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meleda disease, 248300, Autosomal recessive; MDM (Mal de Meleda) (SLURP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLURP1	SLURP1, MDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mal de Meleda (MDM) [MIM:248300]: A rare autosomal recessive skin disorder, characterized by diffuse transgressive palmoplantar keratoderma with keratotic lesions extending onto the dorsa of the hands and the feet (transgrediens). Patients may have hyperhidrosis. Other features include perioral erythema, lichenoid plaques on the knees and the elbows, and nail abnormalities. {ECO:0000269|PubMed:11285253, ECO:0000269|PubMed:12483299, ECO:0000269|PubMed:14756676, ECO:0000269|PubMed:17008894}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Meliodosis, Yatkınlık, 615557 (Melioidozis) (TLR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melioidosis, susceptibility to, 615557 (Melioidosis) (TLR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR5	TLR5, TIL3, SLEB1, MELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 1 (SLEB1) [MIM:601744]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Meliodosis, Yatkınlık, 615557 (Melioidozis) (Prenatal) (TLR5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Melioidosis, susceptibility to, 615557 (Melioidosis) (TLR5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TLR5	TLR5, TIL3, SLEB1, MELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Systemic lupus erythematosus 1 (SLEB1) [MIM:601744]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Melnick-Needles Sendromu, 309350, X'e Bağlı Dominant; MNS (Melnick-Needles Sendromu) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Melnick-Needles syndrome, 309350, X-linked dominant; MNS (Melnick-Needles syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melnick-Needles Sendromu, 309350, X'e Bağlı Dominant; MNS (Melnick-Needles Sendromu) (MLPA)	Melnick-Needles syndrome, 309350, X-linked dominant; MNS (Melnick-Needles syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melnick-Needles Sendromu, 309350, X'e Bağlı Dominant; MNS (Melnick-Needles Sendromu) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Melnick-Needles syndrome, 309350, X-linked dominant; MNS (Melnick-Needles syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Melnick-Needles Sendromu, 309350, X'e Bağlı Dominant; MNS (Melnick-Needles Sendromu) (Prenatal) (MLPA)	Melnick-Needles syndrome, 309350, X-linked dominant; MNS (Melnick-Needles syndrome) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Melorheostosis-Osteopoikilosis, 155950, Sporadik Vakalar (Melorheostozis) (LEMD3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Melorheostosis with osteopoikilosis, 155950, Isolated cases (Melorheostosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melorheostosis (MEL) [MIM:155950]: Rare mesenchymal dysplasia and one of the sclerosing bone disorders. It is caused by a developmental error, with a sclerotomal distribution, frequently involving one limb. It may be asymptomatic, but pain, stiffness with limitation of motion, leg-length discrepancy and limb deformity may occur. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Melorheostosis-Osteopoikilosis, 155950, Sporadik Vakalar (Melorheostozis) (Prenatal) (LEMD3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Melorheostosis with osteopoikilosis, 155950, Isolated cases (Melorheostosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Melorheostosis (MEL) [MIM:155950]: Rare mesenchymal dysplasia and one of the sclerosing bone disorders. It is caused by a developmental error, with a sclerotomal distribution, frequently involving one limb. It may be asymptomatic, but pain, stiffness with limitation of motion, leg-length discrepancy and limb deformity may occur. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hafıza Bozukluğu, Yatkınlık (BDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Memory impairment, susceptibility to (BDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. {ECO:0000269	PubMed:11840487}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MEND Sendromu, 300960, X'e Bağlı Resesif (MEND Sendromu) (EBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	MEND syndrome, 300960, X-linked recessive (MEND syndrome) (EBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EBP	EBP, CDPX2, CPXD, CPX, MEND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: MEND syndrome (MEND) [MIM:300960]: An X-linked recessive disorder associated with a defect in sterol biosynthesis. Disease manifestations and severity are highly variable. Clinical features include intellectual disability, short stature, scoliosis, digital abnormalities, cataracts, and dermatologic abnormalities. {ECO:0000269|PubMed:12503101, ECO:0000269|PubMed:20949533, ECO:0000269|PubMed:24459067, ECO:0000269|PubMed:24705572}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MEND Sendromu, 300960, X'e Bağlı Resesif (MEND Sendromu) (Prenatal) (EBP geni) (Tüm Gen Dizi Analizi) (Prenatal)	MEND syndrome, 300960, X-linked recessive (MEND syndrome) (EBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EBP	EBP, CDPX2, CPXD, CPX, MEND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: MEND syndrome (MEND) [MIM:300960]: An X-linked recessive disorder associated with a defect in sterol biosynthesis. Disease manifestations and severity are highly variable. Clinical features include intellectual disability, short stature, scoliosis, digital abnormalities, cataracts, and dermatologic abnormalities. {ECO:0000269|PubMed:12503101, ECO:0000269|PubMed:20949533, ECO:0000269|PubMed:24459067, ECO:0000269|PubMed:24705572}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Menenjyom, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, 607174, Autosomal dominant (Familial multiple meningioma) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Menenjyoma, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (MN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, 607174, Autosomal dominant (Familial multiple meningioma) (MN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MN1	MN1, MGCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MN1 may be a cause of acute myeloid leukemia (AML). Translocation t(12;22)(p13;q11) with ETV6. {ECO:0000269	PubMed:7731705}.	EDTA Blood Sample
Menenjyom, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (MLPA)	Meningioma, 607174, Autosomal dominant (Familial multiple meningioma) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Meningiom, Ailesel, Yatkınlık, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (SUFU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, familial, susceptibility to, 607174, Autosomal dominant (Familial multiple meningioma) (SUFU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUFU	SUFU, SUFUXL, SUFUH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Meningiom, Ailesel, Yatkınlık, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (SMARCE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, familial, susceptibility to, 607174, Autosomal dominant (Familial multiple meningioma) (SMARCE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCE1	SMARCE1, BAF57, CSS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Meningioma (MNGMA) [MIM:607174]: A common neoplasm of the central nervous system derived from arachnoidal cells. The majority of meningiomas are well differentiated vascular tumors which grow slowly and have a low potential to be invasive, although malignant subtypes occur. Most cases are sporadic. Familial occurrence of meningioma is rare. {ECO:0000269	PubMed:23377182}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Menenjyom, NF2 İlişkili Somatik, 607174; NF2 (NF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, NF2-related, somatic, 607174 (NF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF2	NF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Meningiom, SIS İlişkili, 607174, Otozomal Dominant (Ailesel Multipl Meningiom) (PDGFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Meningioma, SIS-related, 607174, Autosomal dominant (Familial multiple meningioma) (PDGFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFB	PDGFB, SIS, IBGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving PDGFB is found in dermatofibrosarcoma protuberans. Translocation t(17;22)(q22;q13) with PDGFB. {ECO:0000269	PubMed:12660034}.	EDTA Blood Sample
Menkes Hastalığı, 309400, X'e Bağlı (Meckel Sendromu) (ATP7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Menkes disease, 309400, X-linked recessive (Menkes disease) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Menkes Hastalığı, 309400, X'e Bağlı (Meckel Sendromu) (MLPA)	Menkes disease, 309400, X-linked recessive (Menkes disease) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Menkes Hastalığı, 309400, X'e Bağlı (Meckel Sendromu) (Prenatal) (ATP7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Menkes disease, 309400, X-linked recessive (Menkes disease) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Menkes Hastalığı, 309400, X'e Bağlı (Meckel Sendromu) (Prenatal) (MLPA)	Menkes disease, 309400, X-linked recessive (Menkes disease) (Prenatal) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon 105, 300984, X'e Bağlı (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (USP27X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation 105, 300984, X-linked recessive (X-linked non-syndromic intellectual disability) (USP27X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USP27X	USP27X, USP22L, MRX105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon 105, 300984, X'e Bağlı (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (USP27X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation 105, 300984, X-linked recessive (X-linked non-syndromic intellectual disability) (USP27X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	USP27X	USP27X, USP22L, MRX105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, Ayrıt Edici Yüz Özellikleri, x Kardiyak Defekt, 616789, Otozomal Dominant; MRFACD (Kardiyak Anomaliler-Gelişimsel Gecikme-Fasiyel Dismorfizm Sendromu) (MED13L gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation and distinctive facial features with or without cardiac defects, 616789, Autosomal dominant; MRFACD (Cardiac anomalies-developmental delay-facial dysmorphism syndrome) (MED13L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED13L	MED13L, THRAP2, PROSIT240, TRAP240L, KIAA1025, MRFACD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MED13L is found in a patient with transposition of the great arteries, dextro-looped and mental retardation. Translocation t(12;17)(q24.1;q21). {ECO:0000269	PubMed:14638541}.;	EDTA Blood Sample
Mental Retardasyon, Ayrıt Edici Yüz Özellikleri, x Kardiyak Defekt, 616789, Otozomal Dominant; MRFACD (Kardiyak Anomaliler-Gelişimsel Gecikme-Fasiyel Dismorfizm Sendromu) (Prenatal) (MED13L gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation and distinctive facial features with or without cardiac defects, 616789, Autosomal dominant; MRFACD (Cardiac anomalies-developmental delay-facial dysmorphism syndrome) (MED13L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED13L	MED13L, THRAP2, PROSIT240, TRAP240L, KIAA1025, MRFACD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving MED13L is found in a patient with transposition of the great arteries, dextro-looped and mental retardation. Translocation t(12;17)(q24.1;q21). {ECO:0000269	PubMed:14638541}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon ve Mikrosefali, Pontin ve Serebellar Hipoplazi, 300749, X'e Bağlı Dominant; MICPCH (X'e Bağlı Mental Retardasyon, Najm Tipi) (CASK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749, X-linked dominant; MICPCH (X-linked intellectual disability, Najm type) (CASK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269	PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon ve Mikrosefali, Pontin ve Serebellar Hipoplazi, 300749, X'e Bağlı Dominant; MICPCH (X'e Bağlı Mental Retardasyon, Najm Tipi) (Prenatal) (CASK gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation and microcephaly with pontine and cerebellar hypoplasia, 300749, X-linked dominant; MICPCH (X-linked intellectual disability, Najm type) (CASK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269	PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon Sendromu, X'e Bağlı, Siderius Tipi, 300263, X'e Bağlı Resesif; MRXSSD (X'e Bağlı Mental Retardasyon, Siderius Tipi) (PHF8 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation syndrome, X-linked, Siderius type, 300263, X-linked recessive; MRXSSD (X-linked intellectual disability, Siderius type) (PHF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHF8	PHF8, ZNF422, KIAA1111, MRXSSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, Siderius type (MRXSSD) [MIM:300263]: A syndrome characterized by mild to borderline mental retardation with or without cleft lip/cleft palate. {ECO:0000269|PubMed:16199551, ECO:0000269|PubMed:17661819}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon Sendromu, X'e Bağlı, Siderius Tipi, 300263, X'e Bağlı Resesif; MRXSSD (X'e Bağlı Mental Retardasyon, Siderius Tipi) (Prenatal) (PHF8 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation syndrome, X-linked, Siderius type, 300263, X-linked recessive; MRXSSD (X-linked intellectual disability, Siderius type) (PHF8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHF8	PHF8, ZNF422, KIAA1111, MRXSSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, Siderius type (MRXSSD) [MIM:300263]: A syndrome characterized by mild to borderline mental retardation with or without cleft lip/cleft palate. {ECO:0000269|PubMed:16199551, ECO:0000269|PubMed:17661819}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Konuşma Yetersizliği, ± Otistik Özellikler, 613670, Otozomal Dominant (Mental Retardasyon-Ağır Konuşma Gecikmesi-Hafif Dismorfizm Sendromu) (FOXP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation with language impairment and with or without autistic features, 613670, Autosomal dominant (Intellectual disability-severe speech delay-mild dysmorphism syndrome) (FOXP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXP1	FOXP1, QRF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation with language impairment and autistic features (MRLIAF) [MIM:613670]: A developmental disorder characterized by mild to moderate mental retardation, language impairment, and autistic features. Patients show global delay, delayed walking, severely delayed speech development, and behavioral abnormalities, including irritability, hyperactivity, aggression, and stereotypical rigid behaviors. {ECO:0000269	PubMed:20950788}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Konuşma Yetersizliği, ± Otistik Özellikler, 613670, Otozomal Dominant (Mental Retardasyon-Ağır Konuşma Gecikmesi-Hafif Dismorfizm Sendromu) (Prenatal) (FOXP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation with language impairment and with or without autistic features, 613670, Autosomal dominant (Intellectual disability-severe speech delay-mild dysmorphism syndrome) (FOXP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXP1	FOXP1, QRF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation with language impairment and autistic features (MRLIAF) [MIM:613670]: A developmental disorder characterized by mild to moderate mental retardation, language impairment, and autistic features. Patients show global delay, delayed walking, severely delayed speech development, and behavioral abnormalities, including irritability, hyperactivity, aggression, and stereotypical rigid behaviors. {ECO:0000269	PubMed:20950788}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon-Hipotonik Yüz Sendromu, X'e Bağlı, 309580, X'e Bağlı Resesif; MRXHF1 (X'e Bağlı Mental Retardasyon-Hipotonik Yüz Sendromu) (ATRX gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation-hypotonic facies syndrome, X-linked, 309580, X-linked recessive; MRXHF1 (X-linked intellectual disability-hypotonic face syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon-Hipotonik Yüz Sendromu, X'e Bağlı, 309580, X'e Bağlı Resesif; MRXHF1 (X'e Bağlı Mental Retardasyon-Hipotonik Yüz Sendromu) (MLPA)	Mental retardation-hypotonic facies syndrome, X-linked, 309580, X-linked recessive; MRXHF1 (X-linked intellectual disability-hypotonic face syndrome) (MLPA)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon-Hipotonik Yüz Sendromu, X'e Bağlı, 309580, X'e Bağlı Resesif; MRXHF1 (X'e Bağlı Mental Retardasyon-Hipotonik Yüz Sendromu) (Prenatal) (ATRX gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation-hypotonic facies syndrome, X-linked, 309580, X-linked recessive; MRXHF1 (X-linked intellectual disability-hypotonic face syndrome) (ATRX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon-Hipotonik Yüz Sendromu, X'e Bağlı, 309580, X'e Bağlı Resesif; MRXHF1 (X'e Bağlı Mental Retardasyon-Hipotonik Yüz Sendromu) (Prenatal) (MLPA)	Mental retardation-hypotonic facies syndrome, X-linked, 309580, X-linked recessive; MRXHF1 (X-linked intellectual disability-hypotonic face syndrome) (Prenatal) (MLPA)	ATRX	ATRX, XH2, XNP, SHS, SFM1, MRXHF1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Ön Maksiller Çıkıntı ve Şaşılık, 613671, Otozomal Resesif; MRAMS (SOBP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, anterior maxillary protrusion, and strabismus, 613671, Autosomal recessive; MRAMS (SOBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOBP	SOBP, JXC1, MRAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, anterior maxillary protrusion, and strabismus (MRAMS) [MIM:613671]: A syndrome characterized by severe mental retardation, strabismus and dysmorphic features such as anterior maxillary protrusion with vertical maxillary excess, open bite and prominent crowded teeth. Some patients may lack dysmorphic features and manifest temporal lobe epilepsy and psychosis. Esotropia and amblyopia are present in some individuals. {ECO:0000269	PubMed:21035105}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Ön Maksiller Çıkıntı ve Şaşılık, 613671, Otozomal Resesif; MRAMS (Prenatal) (SOBP gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, anterior maxillary protrusion, and strabismus, 613671, Autosomal recessive; MRAMS (SOBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOBP	SOBP, JXC1, MRAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, anterior maxillary protrusion, and strabismus (MRAMS) [MIM:613671]: A syndrome characterized by severe mental retardation, strabismus and dysmorphic features such as anterior maxillary protrusion with vertical maxillary excess, open bite and prominent crowded teeth. Some patients may lack dysmorphic features and manifest temporal lobe epilepsy and psychosis. Esotropia and amblyopia are present in some individuals. {ECO:0000269	PubMed:21035105}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 1, 156200, Otozomal Dominant; MRD1 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (MBD5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 1, 156200, Autosomal dominant; MRD1 (Autosomal dominant non-syndromic intellectual disability) (MBD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBD5	MBD5, KIAA1461, MRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 1 (MRD1) [MIM:156200]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:17847001}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 1, 156200, Otozomal Dominant; MRD1 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (MBD5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 1, 156200, Autosomal dominant; MRD1 (Autosomal dominant non-syndromic intellectual disability) (MBD5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MBD5	MBD5, KIAA1461, MRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 1 (MRD1) [MIM:156200]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:17847001}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 10, 614256; MRD10 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (CACNG2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 10, 614256; MRD10 (Autosomal dominant non-syndromic intellectual disability) (CACNG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNG2	CACNG2, MRD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 10 (MRD10) [MIM:614256]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21376300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 10, 614256; MRD10 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (CACNG2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 10, 614256; MRD10 (Autosomal dominant non-syndromic intellectual disability) (CACNG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CACNG2	CACNG2, MRD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 10 (MRD10) [MIM:614256]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21376300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 11, 614257; MRD11 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (EPB41L1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 11, 614257; MRD11 (Autosomal dominant non-syndromic intellectual disability) (EPB41L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPB41L1	EPB41L1, MRD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 11, 614257; MRD11 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (EPB41L1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 11, 614257; MRD11 (Autosomal dominant non-syndromic intellectual disability) (EPB41L1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPB41L1	EPB41L1, MRD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 13, 614563, Otozomal Dominant; MRD13 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (DYNC1H1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 13, 614563, Autosomal dominant; MRD13 (Autosomal dominant non-syndromic intellectual disability) (DYNC1H1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYNC1H1	DYNC1H1, DNCL, DNECL, CMT20, MRD13, SMALED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 1, autosomal dominant (SMALED1) [MIM:158600]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMALED1 is characterized by muscle weakness predominantly affecting the proximal lower extremities. {ECO:0000269|PubMed:22459677, ECO:0000269|PubMed:22847149, ECO:0000269|PubMed:25512093}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 13, 614563, Otozomal Dominant; MRD13 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (DYNC1H1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 13, 614563, Autosomal dominant; MRD13 (Autosomal dominant non-syndromic intellectual disability) (DYNC1H1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYNC1H1	DYNC1H1, DNCL, DNECL, CMT20, MRD13, SMALED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 1, autosomal dominant (SMALED1) [MIM:158600]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMALED1 is characterized by muscle weakness predominantly affecting the proximal lower extremities. {ECO:0000269|PubMed:22459677, ECO:0000269|PubMed:22847149, ECO:0000269|PubMed:25512093}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, Otozomal Dominant 18, 615074, Otozomal Dominant; MRD18 (Ağır Mental Retardasyon-Yetersiz Konuşma-Şaşılık-Negatif Yüz-Uzun Parmaklar Sendromu) (GATAD2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 18, 615074, Autosomal dominant; MRD18 (Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome) (GATAD2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATAD2B	GATAD2B, KIAA1150, p68, MRD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 18 (MRD18) [MIM:615074]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD18 patients have severe intellectual disability and dysmorphic features. {ECO:0000269|PubMed:23033978, ECO:0000269|PubMed:23644463}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 18, 615074, Otozomal Dominant; MRD18 (Ağır Mental Retardasyon-Yetersiz Konuşma-Şaşılık-Negatif Yüz-Uzun Parmaklar Sendromu) (Prenatal) (GATAD2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 18, 615074, Autosomal dominant; MRD18 (Severe intellectual disability-poor language-strabismus-grimacing face-long fingers syndrome) (GATAD2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATAD2B	GATAD2B, KIAA1150, p68, MRD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 18 (MRD18) [MIM:615074]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD18 patients have severe intellectual disability and dysmorphic features. {ECO:0000269|PubMed:23033978, ECO:0000269|PubMed:23644463}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 19, 615075, Otozomal Dominant; MRD19 (Şiddetli Mental Retardasyon-Progresif Spastik Diplejі Sendromu) (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 19, 615075, Autosomal dominant; MRD19 (Severe intellectual disability-progressive spastic diplegia syndrome) (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:9065402}. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 19, 615075, Otozomal Dominant; MRD19 (Şiddetli Mental Retardasyon-Progresif Spastik Diplejі Sendromu) (Prenatal) (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 19, 615075, Autosomal dominant; MRD19 (Severe intellectual disability-progressive spastic diplegia syndrome) (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:9065402}. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 20; Mrd20 (Sq14.3 Mikrodelesyon Sendromu) (2200)	MENTAL RETARDATION, AUTOSOMAL DOMINANT 20; MRD20 (Sq14.3 microdeletion syndrome) (440)	.	.	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal		EDTA Blood Sample and Heparinized blood sample
Mental Retardasyon, Otozomal Dominant 20; Mrd20 (Sq14.3 Mikrodelesyon Sendromu) (Prenatal) (2200)	MENTAL RETARDATION, AUTOSOMAL DOMINANT 20; MRD20 (Sq14.3 microdeletion syndrome) (Prenatal) (440)	.	.	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 21, 615502, Otozomal Dominant; MRD21 (Mental Retardasyon-Besleme Zorlukları-Gelişimsel Gecikme-Mikrosefali Sendromu) (CTCF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 21, 615502, Autosomal dominant; MRD21 (Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome) (CTCF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTCF	CTCF, MRD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 21 (MRD21) [MIM:615502]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Additional MRD21 features include short stature, microcephaly, and developmental delay. {ECO:0000269	PubMed:23746550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 21, 615502, Otozomal Dominant; MRD21 (Mental Retardasyon-Besleme Zorlukları-Gelişimsel Gecikme-Mikrosefali Sendromu) (Prenatal) (CTCF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 21, 615502, Autosomal dominant; MRD21 (Intellectual disability-feeding difficulties-developmental delay-microcephaly syndrome) (CTCF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTCF	CTCF, MRD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 21 (MRD21) [MIM:615502]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Additional MRD21 features include short stature, microcephaly, and developmental delay. {ECO:0000269	PubMed:23746550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 22, 612337, Otozomal Dominant; MRD22 (Distal Monozomi 1q (ZBTB18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 22, 612337, Autosomal dominant; MRD22 (Distal monosomy 1q (ZBTB18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB18	ZBTB18, ZNF238, RP58, MRD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 22, 612337, Otozomal Dominant; MRD22 (Distal Monozomi 1q (Prenatal) (ZBTB18 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 22, 612337, Autosomal dominant; MRD22 (Distal monosomy 1q (ZBTB18 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZBTB18	ZBTB18, ZNF238, RP58, MRD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 23 615761, Otozomal Dominant; MRD23 (Mental Retardasyon-Fasiyal Dismorfizm Sendromu, SETD5 Haploinsufficiency Bağlı) (SETD5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 23, 615761, Autosomal dominant; MRD23 (Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency) (SETD5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETD5	SETD5, KIAA1757	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 23 (MRD23) [MIM:615761]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD23 patients manifest moderate to severe intellectual disability with additional variable features of brachycephaly, a low hairline, depressed nasal bridge, prominent high nasal root, tubular nose, upslanting palpebral fissures, long and smooth philtrum, micrognathia, thin upper lip, and crowded teeth. Behavioral problems, including obsessive-compulsive disorder, hand flapping with ritualized behavior, and autism, are prominent features. {ECO:0000269	PubMed:24680889}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 23 615761, Otozomal Dominant; MRD23 (Mental Retardasyon-Fasiyal Dismorfizm Sendromu, SETD5 Haploinsufficiency Bağlı) (Prenatal) (SETD5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 23, 615761, Autosomal dominant; MRD23 (Intellectual disability-facial dysmorphism syndrome due to SETD5 haploinsufficiency) (SETD5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SETD5	SETD5, KIAA1757	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 23 (MRD23) [MIM:615761]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD23 patients manifest moderate to severe intellectual disability with additional variable features of brachycephaly, a low hairline, depressed nasal bridge, prominent high nasal root, tubular nose, upslanting palpebral fissures, long and smooth philtrum, micrognathia, thin upper lip, and crowded teeth. Behavioral problems, including obsessive-compulsive disorder, hand flapping with ritualized behavior, and autism, are prominent features. {ECO:0000269	PubMed:24680889}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 24, 615828, Otozomal Dominant; MRD24 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (DEAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 24, 615828, Autosomal dominant; MRD24 (Autosomal dominant non-syndromic intellectual disability) (DEAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DEAF1	DEAF1, SPN, ZMYND5, MRD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 24 (MRD24) [MIM:615828]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:21076407, ECO:0000269|PubMed:24726472}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 24, 615828, Otozomal Dominant; MRD24 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (DEAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 24, 615828, Autosomal dominant; MRD24 (Autosomal dominant non-syndromic intellectual disability) (DEAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DEAF1	DEAF1, SPN, ZMYND5, MRD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 24 (MRD24) [MIM:615828]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:21076407, ECO:0000269|PubMed:24726472}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 26, 615834, Otozomal Dominant; MRD26 (Otizm Spektrum Bozukluğu, AUTS2 Eksikliğine Bağlı) (AUTS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 26, 615834, Autosomal dominant; MRD26 (Autism spectrum disorder due to AUTS2 deficiency) (AUTS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AUTS2	KIAA0442, MRD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 26, 615834, Otozomal Dominant; MRD26 (Otizm Spektrum Bozukluğu, AUTS2 Eksikliğine Bağlı) (Prenatal) (AUTS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 26, 615834, Autosomal dominant; MRD26 (Autism spectrum disorder due to AUTS2 deficiency) (AUTS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AUTS2	KIAA0442, MRD26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 29 616078, Otozomal Dominant; MRD29 (Entelektüel Yetersizlik-Ekspressif Afazi-Fasiyal Dismorfizm Sendromu) (SETBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 29, 616078, Autosomal dominant; MRD29 (Intellectual disability-expressive aphasia-facial dysmorphism syndrome) (SETBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETBP1	SETBP1, KIAA0437, SEB, MRD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:23648668, ECO:0000269|PubMed:23889083}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 29 616078, Otozomal Dominant; MRD29 (Entelektüel Yetersizlik-Ekspressif Afazi-Fasiyal Dismorfizm Sendromu) (Prenatal) (SETBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 29, 616078, Autosomal dominant; MRD29 (Intellectual disability-expressive aphasia-facial dysmorphism syndrome) (SETBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SETBP1	SETBP1, KIAA0437, SEB, MRD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269|PubMed:23648668, ECO:0000269|PubMed:23889083}. Note=The gene represented in this entry is involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 3, 612580; MRD3 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (CDH15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 3, 612580; MRD3 (Autosomal dominant non-syndromic intellectual disability) (CDH15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH15	CDH15, CDH14, CDH3, MRD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 3 (MRD3) [MIM:612580]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:19012874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 3, 612580; MRD3 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (CDH15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 3, 612580; MRD3 (Autosomal dominant non-syndromic intellectual disability) (CDH15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDH15	CDH15, CDH14, CDH3, MRD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 3 (MRD3) [MIM:612580]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:19012874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 30 616083, Otozomal Dominant; MRD30 (Entelektüel Yetersizlik-Ekspressif Afazi-Fasiyal Dismorfizm Sendromu) (ZMYND11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 30, 616083, Autosomal dominant; MRD30 (Intellectual disability-expressive aphasia-facial dysmorphism syndrome) (ZMYND11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZMYND11	ZMYND11, BS69, BRAM1, MRD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 30 (MRD30) [MIM:616083]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD30 patients manifest mild intellectual disability and subtle facial dysmorphisms, including hypertelorism, ptosis, and a wide mouth. {ECO:0000269	PubMed:25217958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mental Retardasyon, Otozomal Dominant 30 616083, Otozomal Dominant; MRD30 (Entelektüel Yetersizlik-Ekspressif Afazi-Fasiyal Dismorfizm Sendromu) (Prenatal) (ZMYND11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 30, 616083, Autosomal dominant; MRD30 (Intellectual disability-expressive aphasia-facial dysmorphism syndrome) (ZMYND11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZMYND11	ZMYND11, BS69, BRAM1, MRD30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 30 (MRD30) [MIM:616083]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD30 patients manifest mild intellectual disability and subtle facial dysmorphisms, including hypertelorism, ptosis, and a wide mouth. (ECO:0000269	PubMed:25217958). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 31, 616158, Otozomal Dominant; MRD31 (PURA İle İlişkili Ağır Neonatal Hipotoni-Nöbet-Ensefalopati Sendromu, Nokta Mutasyonlarına Bağlı) (PURA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 31, 616158, Autosomal dominant; MRD31 (PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation) (PURA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PURA	PURA, PUR1, MRD31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 31 (MRD31) [MIM:616158]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD31 patients manifest neonatal hypotonia, encephalopathy with or without epilepsy, and severe developmental delay. (ECO:0000269|PubMed:25342064, ECO:0000269|PubMed:25439098). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 31, 616158, Otozomal Dominant; MRD31 (PURA İle İlişkili Ağır Neonatal Hipotoni-Nöbet-Ensefalopati Sendromu, Nokta Mutasyonlarına Bağlı) (Prenatal) (PURA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 31, 616158, Autosomal dominant; MRD31 (PURA-related severe neonatal hypotonia-seizures-encephalopathy syndrome due to a point mutation) (PURA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PURA	PURA, PUR1, MRD31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 31 (MRD31) [MIM:616158]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD31 patients manifest neonatal hypotonia, encephalopathy with or without epilepsy, and severe developmental delay. (ECO:0000269|PubMed:25342064, ECO:0000269|PubMed:25439098). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 32, 616268, Otozomal Dominant; MRD32 (Otozomal Dominant Mental Retardasyon-Kraniofasyal Anomaliler-Kalp Defekli Sendromu) (KAT6A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 32, 616268, Autosomal dominant; MRD32 (Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome) (KAT6A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KAT6A	KAT6A, MYST3, MOZ, ZNF220, MRD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 32 (MRD32) [MIM:616268]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD32 patients manifest intellectual disability, dysmorphic facial features, delayed psychomotor development, and lack of speech. (ECO:0000269|PubMed:25728775, ECO:0000269|PubMed:25728777). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 32, 616268, Otozomal Dominant; MRD32 (Otozomal Dominant Mental Retardasyon-Kraniofasyal Anomaliler-Kalp Defekli Sendromu) (Prenatal) (KAT6A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 32, 616268, Autosomal dominant; MRD32 (Autosomal dominant intellectual disability-craniofacial anomalies-cardiac defects syndrome) (KAT6A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KAT6A	KAT6A, MYST3, MOZ, ZNF220, MRD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 32 (MRD32) [MIM:616268]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD32 patients manifest intellectual disability, dysmorphic facial features, delayed psychomotor development, and lack of speech. (ECO:0000269|PubMed:25728775, ECO:0000269|PubMed:25728777). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 33, 616311, MRD33 (Otozomal Dominant Mikrosefali (DPP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 33, 616311, MRD33 (Autosomal dominant microcephaly) (DPP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPP6	DPP6, VF2, MRD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial paroxysmal ventricular fibrillation 2 (VF2) [MIM:612956]: A cardiac arrhythmia marked by fibrillary contractions of the ventricular muscle due to rapid repetitive excitation of myocardial fibers without coordinated contraction of the ventricle and by absence of atrial activity. (ECO:0000269	PubMed:19285295). Note=The disease is caused by mutations affecting the gene represented in this entry. A genetic variation 340 bases upstream from the ATG start site of the DPP6 gene is the cause of familial paroxysmal ventricular fibrillation type 2	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 33, 616311, MRD33 (Otozomal Dominant Mikrosefali (Prenatal) (DPP6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 33, 616311, MRD33 (Autosomal dominant microcephaly) (DPP6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPP6	DPP6, VF2, MRD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial paroxysmal ventricular fibrillation 2 (VF2) [MIM:612956]: A cardiac arrhythmia marked by fibrillary contractions of the ventricular muscle due to rapid repetitive excitation of myocardial fibers without coordinated contraction of the ventricle and by absence of atrial activity. (ECO:0000269	PubMed:19285295). Note=The disease is caused by mutations affecting the gene represented in this entry. A genetic variation 340 bases upstream from the ATG start site of the DPP6 gene is the cause of familial paroxysmal ventricular fibrillation type 2	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 34, 616351, Otozomal Dominant; MRD34 (COL4A3BP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 34, 616351, Autosomal dominant; MRD34 (COL4A3BP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A3BP	COL4A3BP, GPBP, CERT, MRD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 34 (MRD34) [MIM:616351]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269	PubMed:25533962). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 34, 616351, Otozomal Dominant; MRD34 (Prenatal) (COL4A3BP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 34, 616351, Autosomal dominant; MRD34 (COL4A3BP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A3BP	COL4A3BP, GPBP, CERT, MRD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 34 (MRD34) [MIM:616351]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269	PubMed:25533962). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 35, 616355, Otozomal Dominant; MRD35 (Mental Retardasyon-Makrosefali-Hipotoni-Davranışsal Anormallikler Sendromu) (PPP2R5D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 35, 616355, Autosomal dominant; MRD35 (Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome) (PPP2R5D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP2R5D	PPP2R5D, MRD35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 35 (MRD35) [MIM:616355]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269|PubMed:25533962, ECO:0000269|PubMed:26168268). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 35, 616355, Otozomal Dominant; MRD35 (Mental Retardasyon-Makrosefali-Hipotoni-Davranışsal Anormallikler Sendromu) (Prenatal) (PPP2R5D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 35, 616355, Autosomal dominant; MRD35 (Intellectual disability-macrocephaly-hypotonia-behavioral abnormalities syndrome) (PPP2R5D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPP2R5D	PPP2R5D, MRD35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 35 (MRD35) [MIM:616355]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269|PubMed:25533962, ECO:0000269|PubMed:26168268). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 36, 616362, Otozomal Dominant; MRD36 (Mikrosefali Korpus Kallozum Hipoplazisi-Mental Retardasyon-Fasiyal Dismorfizm Sendromu) (PPP2R1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 36, 616362, Autosomal dominant; MRD36 (Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome) (PPP2R1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP2R1A	PPP2R1A, MRD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 36 (MRD36) [MIM:616362]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269|PubMed:25533962, ECO:0000269|PubMed:26168268). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 36, 616362, Otozomal Dominant; MRD36 (Mikrosefali Korpus Kallozum Hipoplazisi-Mental Retardasyon-Fasiyal Dismorfizm Sendromu) (Prenatal) (PPP2R1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 36, 616362, Autosomal dominant; MRD36 (Microcephaly-corpus callosum hypoplasia-intellectual disability-facial dysmorphism syndrome) (PPP2R1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPP2R1A	PPP2R1A, MRD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 36 (MRD36) [MIM:616362]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269|PubMed:25533962, ECO:0000269|PubMed:26168268). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 38, 616393, Otozomal Dominant; MRD38 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (EEF1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 38, 616393, Autosomal dominant; MRD38 (Autosomal dominant non-syndromic intellectual disability) (EEF1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EEF1A2	EEF1A2, EIEE33, MRD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 38 (MRD38) [MIM:616393]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD38 common features are severe intellectual disability, autistic behavior, absent speech, neonatal hypotonia, epilepsy and progressive microcephaly. (ECO:0000269	PubMed:24697219). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 38, 616393, Otozomal Dominant; MRD38 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (EEF1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 38, 616393, Autosomal dominant; MRD38 (Autosomal dominant non-syndromic intellectual disability) (EEF1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EEF1A2	EEF1A2, EIEE33, MRD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 38 (MRD38) [MIM:616393]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD38 common features are severe intellectual disability, autistic behavior, absent speech, neonatal hypotonia, epilepsy and progressive microcephaly. (ECO:0000269	PubMed:24697219). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 39, 616521, Otozomal Dominant; MRD39 (MYT1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 39, 616521, Autosomal dominant; MRD39 (MYT1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYT1L	MYT1L, KIAA1106, MRD39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 39, 616521, Otozomal Dominant; MRD39 (Prenatal) (MYT1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 39, 616521, Autosomal dominant; MRD39 (MYT1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYT1L	MYT1L, KIAA1106, MRD39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 4, 612581, MRD4 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (KIRREL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 4, 612581, MRD4 (Autosomal dominant non-syndromic intellectual disability) (KIRREL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIRREL3	KIRREL3, NEPH2, KIAA1867, KIRRE, MRD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 4 (MRD4) [MIM:612581]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269	PubMed:19012874). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 4, 612581, MRD4 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (KIRREL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 4, 612581, MRD4 (Autosomal dominant non-syndromic intellectual disability) (KIRREL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIRREL3	KIRREL3, NEPH2, KIAA1867, KIRRE, MRD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 4 (MRD4) [MIM:612581]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269	PubMed:19012874). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 40, 616579, Otozomal Dominant; MRD40 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (CHAMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 40, 616579, Autosomal dominant; MRD40 (Autosomal dominant non-syndromic intellectual disability) (CHAMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHAMP1	CHAMP1, ZNF828, C13orf8, KIAA1802, MRD40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Mental Retardasyon, Otozomal Dominant 40, 616579, Otozomal Dominant; MRD40 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (CHAMP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 40, 616579, Autosomal dominant; MRD40 (Autosomal dominant non-syndromic intellectual disability) (CHAMP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHAMP1	CHAMP1, ZNF828, C13orf8, KIAA1802, MRD40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 41, 616944, Otozomal Dominant; MRD41 (TBL1XR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 41, 616944, Autosomal dominant; MRD41 (TBL1XR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBL1XR1	TBL1XR1, TBLR1, IRA1, C21, MRD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 41, 616944, Otozomal Dominant; MRD41 (Prenatal) (TBL1XR1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 41, 616944, Autosomal dominant; MRD41 (TBL1XR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBL1XR1	TBL1XR1, TBLR1, IRA1, C21, MRD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 42, 616973, Otozomal Dominant; MRD42 (GNB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 42, 616973, Autosomal dominant; MRD42 (GNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB1	GNB1, MRD42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 42, 616973, Otozomal Dominant; MRD42 (Prenatal) (GNB1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 42, 616973, Autosomal dominant; MRD42 (GNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNB1	GNB1, MRD42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 43, 616977, Otozomal Dominant; MRD43 (HIVEP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 43, 616977, Autosomal dominant; MRD43 (HIVEP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HIVEP2	HIVEP2, MRD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 43, 616977, Otozomal Dominant; MRD43 (Prenatal) (HIVEP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 43, 616977, Autosomal dominant; MRD43 (HIVEP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HIVEP2	HIVEP2, MRD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 44, 617061, Otozomal Dominant; MRD44 (Mikrognati-Tekrarlayan Enfeksiyonlar-Davranışsal Anormallikler-Hafif Mental Retardasyon Sendromu) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 44, 617061, Autosomal dominant; MRD44 (Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome) (TRIO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIO	TRIO, MRD44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 44, 617061, Otozomal Dominant; MRD44 (Mikrognati-Tekrarlayan Enfeksiyonlar-Davranışsal Anormallikler-Hafif Mental Retardasyon Sendromu) (Prenatal) (TRIO gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 44, 617061, Autosomal dominant; MRD44 (Micrognathia-recurrent infections-behavioral abnormalities-mild intellectual disability syndrome) (TRIO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIO	TRIO, MRD44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 5, 612621, Otozomal Dominant; MRD5 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (SYNGAP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 5, 612621, Autosomal dominant; MRD5 (Autosomal dominant non-syndromic intellectual disability) (SYNGAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNGAP1	SYNGAP1, MRD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 5 (MRD5) [MIM:612621]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD5 patients show global developmental delay with delayed motor development, hypotonia, moderate-to-severe mental retardation, and severe language impairment. Epilepsy and autism can be present in some patients. (ECO:0000269|PubMed:19196676, ECO:0000269|PubMed:21076407, ECO:0000269|PubMed:21237447, ECO:0000269|PubMed:23161826). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 5, 612621, Otozomal Dominant; MRD5 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (SYNGAP1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 5, 612621, Autosomal dominant; MRD5 (Autosomal dominant non-syndromic intellectual disability) (SYNGAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYNGAP1	SYNGAP1, MRD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 5 (MRD5) [MIM:612621]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD5 patients show global developmental delay with delayed motor development, hypotonia, moderate-to-severe mental retardation, and severe language impairment. Epilepsy and autism can be present in some patients. (ECO:0000269|PubMed:19196676, ECO:0000269|PubMed:21076407, ECO:0000269|PubMed:21237447, ECO:0000269|PubMed:23161826). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 6, 613970; MRD6 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (GRIN2B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 6, 613970; MRD6 (Autosomal dominant non-syndromic intellectual disability) (GRIN2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIN2B	GRIN2B, NMDAR2B, MRD6, EIEE27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 6 (MRD6) [MIM:613970]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:20890276, ECO:0000269|PubMed:23033978}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 6, 613970; MRD6 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (GRIN2B gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 6, 613970; MRD6 (Autosomal dominant non-syndromic intellectual disability) (GRIN2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIN2B	GRIN2B, NMDAR2B, MRD6, EIEE27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 6 (MRD6) [MIM:613970]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:20890276, ECO:0000269|PubMed:23033978}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 7, 614104, Otozomal Dominant; MRD7 (DYRK1A İlişkili Mental Retardasyon Sendromu) (DYRK1A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 7, 614104, Autosomal dominant; MRD7 (DYRK1A-related intellectual disability syndrome) (DYRK1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYRK1A	DYRK1A, MNBH, MNB, MRD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 7 (MRD7) [MIM:614104]: A disease characterized by primary microcephaly, severe mental retardation without speech, anxious autistic behavior, and dysmorphic features, including bitemporal narrowing, deep-set eyes, large simple ears, and a pointed nasal tip. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21294719}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 7, 614104, Otozomal Dominant; MRD7 (DYRK1A İlişkili Mental Retardasyon Sendromu) (Prenatal) (DYRK1A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 7, 614104, Autosomal dominant; MRD7 (DYRK1A-related intellectual disability syndrome) (DYRK1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYRK1A	DYRK1A, MNBH, MNB, MRD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 7 (MRD7) [MIM:614104]: A disease characterized by primary microcephaly, severe mental retardation without speech, anxious autistic behavior, and dysmorphic features, including bitemporal narrowing, deep-set eyes, large simple ears, and a pointed nasal tip. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21294719}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 8, 614254; MRD8 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (GRIN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 8, 614254; MRD8 (Autosomal dominant non-syndromic intellectual disability) (GRIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIN1	GRIN1, NMDAR1, MRD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 8 (MRD8) [MIM:614254]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21376300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 8, 614254; MRD8 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (GRIN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 8, 614254; MRD8 (Autosomal dominant non-syndromic intellectual disability) (GRIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIN1	GRIN1, NMDAR1, MRD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 8 (MRD8) [MIM:614254]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21376300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 9, 614255, Otozomal Dominant; MRD9 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (KIF1A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant 9, 614255, Autosomal dominant; MRD9 (Autosomal dominant non-syndromic intellectual disability) (KIF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1A	KIF1A, ATSV, UNC104, SPG30, HSN2C, MRD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 2C (HSN2C) [MIM:614213]: A neurodegenerative disorder characterized by onset in the first decade of progressive distal sensory loss leading to ulceration and amputation of the fingers and toes. Affected individuals also develop distal muscle weakness, primarily affecting the lower limbs. {ECO:0000269	PubMed:21820098}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant 9, 614255, Otozomal Dominant; MRD9 (Otozomal Dominant Sendromik Olmayan Mental Retardasyon) (Prenatal) (KIF1A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant 9, 614255, Autosomal dominant; MRD9 (Autosomal dominant non-syndromic intellectual disability) (KIF1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF1A	KIF1A, ATSV, UNC104, SPG30, HSN2C, MRD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuropathy, hereditary sensory, 2C (HSN2C) [MIM:614213]: A neurodegenerative disorder characterized by onset in the first decade of progressive distal sensory loss leading to ulceration and amputation of the fingers and toes. Affected individuals also develop distal muscle weakness, primarily affecting the lower limbs. {ECO:0000269	PubMed:21820098}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant, 27, 615866, Otozomal Dominant; MRD27 (Coffin-Siris Sendromu) (SOX11 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal dominant, 27, 615866, Autosomal dominant; MRD27 (Coffin-Siris syndrome) (SOX11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX11	SOX11, MRD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Dominant, 27, 615866, Otozomal Dominant; MRD27 (Coffin-Siris Sendromu) (Prenatal) (SOX11 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal dominant, 27, 615866, Autosomal dominant; MRD27 (Coffin-Siris syndrome) (SOX11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX11	SOX11, MRD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 1, 249500, Otozomal Resesif; MRT1 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (PRSS12 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 1, 249500, Autosomal recessive; MRT1 (Autosomal recessive non-syndromic intellectual disability) (PRSS12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRSS12	PRSS12, BSSP3, MRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 1, 249500, Otozomal Resesif; MRT1 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (PRSS12 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 1, 249500, Autosomal recessive; MRT1 (Autosomal recessive non-syndromic intellectual disability) (PRSS12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRSS12	PRSS12, BSSP3, MRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 12, 611090, Otozomal Resesif; MRT12 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (ST3GAL3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 12, 611090, Autosomal recessive; MRT12 (Autosomal recessive non-syndromic intellectual disability) (ST3GAL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ST3GAL3	ST3GAL3, SIAT6, ST3GALII, MRT12, EIEE15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 12 (MRT12) [MIM:611090]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:21907012}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mental Retardasyon, Otozomal Resesif 12, 611090, Otozomal Resesif; MRT12 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (ST3GAL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 12, 611090, Autosomal recessive; MRT12 (Autosomal recessive non-syndromic intellectual disability) (ST3GAL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ST3GAL3	ST3GAL3, SIAT6, ST3GALII, MRT12, EIEE15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 12 (MRT12) [MIM:611090]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269[PubMed:21907012). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 13, 613192, Otozomal Resesif; MRT13 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (TRAPPC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 13, 613192, Autosomal recessive; MRT13 (Autosomal recessive non-syndromic intellectual disability) (TRAPPC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAPPC9	TRAPPC9, NIBP, KIAA1882, MRT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 13, 613192, Otozomal Resesif; MRT13 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (TRAPPC9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 13, 613192, Autosomal recessive; MRT13 (Autosomal recessive non-syndromic intellectual disability) (TRAPPC9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAPPC9	TRAPPC9, NIBP, KIAA1882, MRT13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 14, 614020, Otozomal Resesif; MRT14 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (TECR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 14, 614020, Autosomal recessive; MRT14 (Autosomal recessive non-syndromic intellectual disability) (TECR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TECR	TECR, GPSN2, TER, SC2, MRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 14 (MRT14) [MIM:614020]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269[PubMed:21212097). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 14, 614020, Otozomal Resesif; MRT14 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (TECR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 14, 614020, Autosomal recessive; MRT14 (Autosomal recessive non-syndromic intellectual disability) (TECR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TECR	TECR, GPSN2, TER, SC2, MRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 14 (MRT14) [MIM:614020]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269[PubMed:21212097). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 15, 614202, Otozomal Resesif; MRT15 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (MAN1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 15, 614202, Autosomal recessive; MRT15 (Autosomal recessive non-syndromic intellectual disability) (MAN1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAN1B1	MAN1B1, MRT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 15, 614202, Otozomal Resesif; MRT15 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (MAN1B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 15, 614202, Autosomal recessive; MRT15 (Autosomal recessive non-syndromic intellectual disability) (MAN1B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAN1B1	MAN1B1, MRT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 18, 614249, Otozomal Resesif; MRT18 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (MED23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 18, 614249, Autosomal recessive; MRT18 (Autosomal recessive non-syndromic intellectual disability) (MED23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED23	MED23, MRT18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 18 (MRT18) [MIM:614249]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269[PubMed:21868677). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 18, 614249, Otozomal Resesif; MRT18 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (MED23 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 18, 614249, Autosomal recessive; MRT18 (Autosomal recessive non-syndromic intellectual disability) (MED23 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED23	MED23, MRT18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 18 (MRT18) [MIM:614249]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. (ECO:0000269[PubMed:21868677). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 2, 607417, Otozomal Resesif; MRT2 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (CRBN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 2, 607417, Autosomal recessive; MRT2 (Autosomal recessive non-syndromic intellectual disability) (CRBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRBN	CRBN, MRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 2A (MRT2A) [MIM:607417]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Non-syndromic mental retardation patients do not manifest other clinical signs. MRT2A patients display mild mental retardation with a standard IQ ranged from 50 to 70. IQ scores are lower in males than females. Developmental milestones are mildly delayed. There are no dysmorphic or autistic features. (ECO:0000269[PubMed:15557513). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 2, 607417, Otozomal Resesif; MRT2 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (CRBN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 2, 607417, Autosomal recessive; MRT2 (Autosomal recessive non-syndromic intellectual disability) (CRBN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRBN	CRBN, MRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 2A (MRT2A) [MIM:607417]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Non-syndromic mental retardation patients do not manifest other clinical signs. MRT2A patients display mild mental retardation with a standard IQ ranged from 50 to 70. IQ scores are lower in males than females. Developmental milestones are mildly delayed. There are no dysmorphic or autistic features. (ECO:0000269[PubMed:15557513). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 27, 614340, Otozomal Resesif; MRT27 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (LINS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 27, 614340, Autosomal recessive; MRT27 (Autosomal recessive non-syndromic intellectual disability) (LINS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LINS1	LINS1, WINS1, FLJ10583, MRT27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 27, 614340, Otozomal Resesif; MRT27 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (LINS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 27, 614340, Autosomal recessive; MRT27 (Autosomal recessive non-syndromic intellectual disability) (LINS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LINS1	LINS1, WINS1, FLJ10583, MRT27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 3, 608443, Otozomal Resesif; MRT3 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (CC2D1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 3, 608443, Autosomal recessive; MRT3 (Autosomal recessive non-syndromic intellectual disability) (CC2D1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CC2D1A	CC2D1A, MRT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 3 (MRT3) [MIM:608443]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Non-syndromic mental retardation patients do not manifest other clinical signs. (ECO:0000269[PubMed:16033914). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 3, 608443, Otozomal Resesif; MRT3 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (CC2D1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 3, 608443, Autosomal recessive; MRT3 (Autosomal recessive non-syndromic intellectual disability) (CC2D1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CC2D1A	CC2D1A, MRT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 3 (MRT3) [MIM:608443]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Non-syndromic mental retardation patients do not manifest other clinical signs. (ECO:0000269[PubMed:16033914). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 34, Varyant Lissensefali, 614499, Otozomal Resesif; MRT34 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (CRADD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499, Autosomal recessive; MRT34 (Autosomal recessive non-syndromic intellectual disability) (CRADD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRADD	CRADD, RAIDD, MRT34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 34 (MRT34) [MIM:614499]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT34 is a non-syndromic form. Affected individuals have mildly delayed development and significantly impaired cognitive function, precluding independent living and self-care. Speech is rudimentary, but articulate; autism is not present. (ECO:0000269[PubMed:22279524). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 34, Varyant Lissensefali, 614499, Otozomal Resesif; MRT34 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (CRADD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 34, with variant lissencephaly, 614499, Autosomal recessive; MRT34 (Autosomal recessive non-syndromic intellectual disability) (CRADD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRADD	CRADD, RAIDD, MRT34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 34 (MRT34) [MIM:614499]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT34 is a non-syndromic form. Affected individuals have mildly delayed development and significantly impaired cognitive function, precluding independent living and self-care. Speech is rudimentary, but articulate; autism is not present. (ECO:0000269[PubMed:22279524). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 36, 615286, Otozomal Resesif; MRT36 (Mental Retardasyon–Şaşılık Sendromu) (ADAT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 36, 615286, Autosomal recessive; MRT36 (Intellectual disability-strabismus syndrome) (ADAT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAT3	ADAT3, TAD3, MRT36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 36 (MRT36) [MIM:615286]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT36 is often associated with esotropia and failure to thrive. Other more variable features included microcephaly, hypotonia, and mild brain abnormalities on MRI, such as dilated ventricles or delayed myelination. (ECO:0000269[PubMed:23620220). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 36, 615286, Otozomal Resesif; MRT36 (Mental Retardasyon–Şaşılık Sendromu) (Prenatal) (ADAT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 36, 615286, Autosomal recessive; MRT36 (Intellectual disability-strabismus syndrome) (ADAT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADAT3	ADAT3, TAD3, MRT36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 36 (MRT36) [MIM:615286]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT36 is often associated with esotropia and failure to thrive. Other more variable features included microcephaly, hypotonia, and mild brain abnormalities on MRI, such as dilated ventricles or delayed myelination. (ECO:0000269[PubMed:23620220). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 38, 615516, Otozomal Resesif; MRT38 (Gelişme Geriliği - Otizm Spektrum Bozukluğu ve Yürüme İstabilitesi) (HERC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 38, 615516, Autosomal recessive; MRT38 (Developmental delay with autism spectrum disorder and gait instability) (HERC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HERC2	HERC2, SHEP1, MRT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 38 (MRT38) [MIM:615516]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT38 is characterized by global developmental delay affecting motor, speech, adaptive, and social development. Patients manifest autistic features, aggression, self-injury, impulsivity, and distractibility. (ECO:0000269[PubMed:23065719). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mental Retardasyon, Otozomal Resesif 38, 615516, Otozomal Resesif; MRT38 (Gelişme Geriliği - Otizm Spektrum Bozukluğu ve Yürüme İnstabilitesi) (Prenatal) (HERC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 38, 615516, Autosomal recessive; MRT38 (Developmental delay with autism spectrum disorder and gait instability) (HERC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HERC2	HERC2, SHEP1, MRT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 38 (MRT38) [MIM:615516]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT38 is characterized by global developmental delay affecting motor, speech, adaptive, and social development. Patients manifest autistic features, aggression, self-injury, impulsivity, and distractibility. {ECO:0000269	PubMed:23065719}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 39, 615541, Otozomal Resesif; MRT39 (Ağır Mental Retardasyon-Boy Kısalığı-Davranışsal Anormallikler-Fasiyel Dismorfizm Sendromu) (TTI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 39, 615541, Autosomal recessive; MRT39 (Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome) (TTI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTI2	TTI2, C8orf41, MRT39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 39 (MRT39) [MIM:615541]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT39 affected individuals show delayed psychomotor development, severe speech delay, short stature, kyphoscoliosis, and dysmorphic facial features. Behavioral abnormalities include hyperactivity, aggression, and stereotypic movements. {ECO:0000269	PubMed:23956177}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 39, 615541, Otozomal Resesif; MRT39 (Ağır Mental Retardasyon-Boy Kısalığı-Davranışsal Anormallikler-Fasiyel Dismorfizm Sendromu) (Prenatal) (TTI2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 39, 615541, Autosomal recessive; MRT39 (Severe intellectual disability-short stature-behavioral abnormalities-facial dysmorphism syndrome) (TTI2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTI2	TTI2, C8orf41, MRT39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 39 (MRT39) [MIM:615541]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT39 affected individuals show delayed psychomotor development, severe speech delay, short stature, kyphoscoliosis, and dysmorphic facial features. Behavioral abnormalities include hyperactivity, aggression, and stereotypic movements. {ECO:0000269	PubMed:23956177}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 40, 615599, Otozomal Resesif; MRT40 (Mikrosefali-İnce Korpus Kallozum-Mental Retardasyon Sendromu) (TAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 40, 615599, Autosomal recessive; MRT40 (Microcephaly-thin corpus callosum-intellectual disability syndrome) (TAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF2	TAF2, TAF2B, TAFII150, CIF150, MRT40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 40, 615599, Otozomal Resesif; MRT40 (Mikrosefali-İnce Korpus Kallozum-Mental Retardasyon Sendromu) (Prenatal) (TAF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 40, 615599, Autosomal recessive; MRT40 (Microcephaly-thin corpus callosum-intellectual disability syndrome) (TAF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAF2	TAF2, TAF2B, TAFII150, CIF150, MRT40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 41, 615637, Otozomal Resesif; MRT41 (Makrosefali-Gelişimsel Gecikme Sendromu) (KPTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 41, 615637, Autosomal recessive; MRT41 (Macrocephaly-developmental delay syndrome) (KPTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KPTN	KPTN, 2E4, MRT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 41 (MRT41) [MIM:615637]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT41 most consistent features are global developmental delay, macrocephaly with frontal bossing, high levels of anxiety, and some features suggestive of a pervasive developmental disorder. Less common features include fifth finger clinodactyly, recurrent pneumonia, and hepatosplenomegaly. {ECO:0000269	PubMed:24239382}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 41, 615637, Otozomal Resesif; MRT41 (Makrosefali-Gelişimsel Gecikme Sendromu) (Prenatal) (KPTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 41, 615637, Autosomal recessive; MRT41 (Macrocephaly-developmental delay syndrome) (KPTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KPTN	KPTN, 2E4, MRT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 41 (MRT41) [MIM:615637]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT41 most consistent features are global developmental delay, macrocephaly with frontal bossing, high levels of anxiety, and some features suggestive of a pervasive developmental disorder. Less common features include fifth finger clinodactyly, recurrent pneumonia, and hepatosplenomegaly. {ECO:0000269	PubMed:24239382}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 42, 615802, Otozomal Resesif; MRT42 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (PGAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 42, 615802, Autosomal recessive; MRT42 (Autosomal recessive non-syndromic intellectual disability) (PGAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGAP1	PGAP1, MRT42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 42 (MRT42) [MIM:615802]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:24784135}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 42, 615802, Otozomal Resesif; MRT42 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (PGAP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 42, 615802, Autosomal recessive; MRT42 (Autosomal recessive non-syndromic intellectual disability) (PGAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGAP1	PGAP1, MRT42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 42 (MRT42) [MIM:615802]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:24784135}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 43, 615817, Otozomal Resesif; MRT43 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (WASHC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 43, 615817, Autosomal recessive; MRT43 (Autosomal recessive non-syndromic intellectual disability) (WASHC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WASHC4	KIAA1033, SWIP, MRT43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 43, 615817, Otozomal Resesif; MRT43 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (WASHC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 43, 615817, Autosomal recessive; MRT43 (Autosomal recessive non-syndromic intellectual disability) (WASHC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WASHC4	KIAA1033, SWIP, MRT43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 44, 615942, Otozomal Resesif; MRT44 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (METTL23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 44, 615942, Autosomal recessive; MRT44 (Autosomal recessive non-syndromic intellectual disability) (METTL23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	METTL23	METTL23, C17orf95, MRT44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 44 (MRT44) [MIM:615942]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT44 manifestations include mild to severe cognitive impairment, delayed psychomotor development, seizures in some patients, and dysmorphic features. {ECO:0000269|PubMed:24501276, ECO:0000269|PubMed:24626631}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 44, 615942, Otozomal Resesif; MRT44 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (METTL23 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 44, 615942, Autosomal recessive; MRT44 (Autosomal recessive non-syndromic intellectual disability) (METTL23 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	METTL23	METTL23, C17orf95, MRT44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 44 (MRT44) [MIM:615942]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT44 manifestations include mild to severe cognitive impairment, delayed psychomotor development, seizures in some patients, and dysmorphic features. {ECO:0000269|PubMed:24501276, ECO:0000269|PubMed:24626631}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 45, 615979, Otozomal Resesif; MRT45 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (FBXO31 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 45, 615979, Autosomal recessive; MRT45 (Autosomal recessive non-syndromic intellectual disability) (FBXO31 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBXO31	FBXO31, FBX31, FBXO14, FBX14, MRT45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 45 (MRT45) [MIM:615979]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT45 manifestations include mild to moderate intellectual disability and dysmorphic features, including coarse facies, broad nasal bridge, fleshy nares, and thick, prominent lips. {ECO:0000269	PubMed:24623383}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 45, 615979, Otozomal Resesif; MRT45 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (FBXO31 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 45, 615979, Autosomal recessive; MRT45 (Autosomal recessive non-syndromic intellectual disability) (FBXO31 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBXO31	FBXO31, FBX31, FBXO14, FBX14, MRT45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 45 (MRT45) [MIM:615979]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT45 manifestations include mild to moderate intellectual disability and dysmorphic features, including coarse facies, broad nasal bridge, fleshy nares, and thick, prominent lips. {ECO:0000269	PubMed:24623383}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 46, 616116, Otozomal Resesif; MRT46 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (NDST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 46, 616116, Autosomal recessive; MRT46 (Autosomal recessive non-syndromic intellectual disability) (NDST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDST1	NDST1, HSST, MRT46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 46, 616116, Otozomal Resesif; MRT46 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (NDST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 46, 616116, Autosomal recessive; MRT46 (Autosomal recessive non-syndromic intellectual disability) (NDST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDST1	NDST1, HSST, MRT46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 47, 616193, Otozomal Resesif; MRT47 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (FMN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 47, 616193, Autosomal recessive; MRT47 (Autosomal recessive non-syndromic intellectual disability) (FMN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FMN2	FMN2, MRT47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 47 (MRT47) [MIM:616193]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT47 patients show delayed development, with cognition and speech more affected than motor skills. {ECO:0000269	PubMed:25480035}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 47, 616193, Otozomal Resesif; MRT47 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (FMN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 47, 616193, Autosomal recessive; MRT47 (Autosomal recessive non-syndromic intellectual disability) (FMN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FMN2	FMN2, MRT47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 47 (MRT47) [MIM:616193]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT47 patients show delayed development, with cognition and speech more affected than motor skills. {ECO:0000269	PubMed:25480035}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, Otozomal Resesif 48, 616269, Otozomal Resesif; MRT48 (Progresif Esansiyel Tremor-Konuşma Bozukluğu-Fasiyal Dismorfizm-Mental Retardasyon-Anormal Davranış Sendromu) (SLC6A17 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 48, 616269, Autosomal recessive; MRT48 (Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome) (SLC6A17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A17	SLC6A17, NTT4, MRT48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 48 (MRT48) [MIM:616269]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT48 patients show moderate to severe intellectual disability and additional features including progressive tremor, speech impairment, and sometimes behavioral problems. {ECO:0000269	PubMed:25704603}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 48, 616269, Otozomal Resesif; MRT48 (Progresif Esansiyel Tremor-Konuşma Bozukluğu-Fasiyal Dismorfizm-Mental Retardasyon-Anormal Davranış Sendromu) (Prenatal) (SLC6A17 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 48, 616269, Autosomal recessive; MRT48 (Progressive essential tremor-speech impairment-facial dysmorphism-intellectual disability-abnormal behavior syndrome) (SLC6A17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC6A17	SLC6A17, NTT4, MRT48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 48 (MRT48) [MIM:616269]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT48 patients show moderate to severe intellectual disability and additional features including progressive tremor, speech impairment, and sometimes behavioral problems. {ECO:0000269	PubMed:25704603}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 49, 616281, Otozomal Resesif; MRT49 (Postnatal Mikrosefali-Infantil Hipotoni-Spastik Diplejі-Distrtri-Mental Retardasyon Sendromu) (GPT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 49, 616281, Autosomal recessive; MRT49 (Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome) (GPT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPT2	GPT2, ALT2, MRT49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 49, 616281, Otozomal Resesif; MRT49 (Postnatal Mikrosefali-Infantil Hipotoni-Spastik Diplejі-Distrtri-Mental Retardasyon Sendromu) (Prenatal) (GPT2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 49, 616281, Autosomal recessive; MRT49 (Postnatal microcephaly-infantile hypotonia-spastic diplegia-dysarthria-intellectual disability syndrome) (GPT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPT2	GPT2, ALT2, MRT49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 5, 611091, Otozomal Resesif; MRT5 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (NSUN2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 5, 611091, Autosomal recessive; MRT5 (Autosomal recessive non-syndromic intellectual disability) (NSUN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSUN2	NSUN2, TRM4, SAKI, MISU, MRT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 5 (MRT5) [MIM:611091]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:22541559, ECO:0000269|PubMed:22541562}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 5, 611091, Otozomal Resesif; MRT5 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (NSUN2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 5, 611091, Autosomal recessive; MRT5 (Autosomal recessive non-syndromic intellectual disability) (NSUN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSUN2	NSUN2, TRM4, SAKI, MISU, MRT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 5 (MRT5) [MIM:611091]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:22541559, ECO:0000269|PubMed:22541562}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 50, 616460, Otozomal Resesif; MRT50 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (EDC3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 50, 616460, Autosomal recessive; MRT50 (Autosomal recessive non-syndromic intellectual disability) (EDC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDC3	EDC3, YJDC, MRT50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 50 (MRT50) [MIM:616460]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT50 patients show mild intellectual disability and microcephaly. {ECO:0000269	PubMed:25701870}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 50, 616460, Otozomal Resesif; MRT50 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (EDC3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 50, 616460, Autosomal recessive; MRT50 (Autosomal recessive non-syndromic intellectual disability) (EDC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDC3	EDC3, YJDC, MRT50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 50 (MRT50) [MIM:616460]: A form of mental retardation, a disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT50 patients show mild intellectual disability and microcephaly. {ECO:0000269	PubMed:25701870}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 51, 616739, Otozomal Resesif; MRT51 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (HNMT gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 51, 616739, Autosomal recessive; MRT51 (Autosomal recessive non-syndromic intellectual disability) (HNMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNMT	HNMT, MRT51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 51, 616739, Otozomal Resesif; MRT51 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (HNMT gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 51, 616739, Autosomal recessive; MRT51 (Autosomal recessive non-syndromic intellectual disability) (HNMT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HNMT	HNMT, MRT51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 53, 616917, Otozomal Resesif; MRT53 (PIGG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 53, 616917, Autosomal recessive; MRT53 (PIGG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGG	PIGG, GPI7, MRT53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 53, 616917, Otozomal Resesif; MRT53 (Prenatal) (PIGG gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 53, 616917, Autosomal recessive; MRT53 (PIGG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGG	PIGG, GPI7, MRT53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 54, 617028, Otozomal Resesif; MRT54 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (TNIK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 54, 617028, Autosomal recessive; MRT54 (Autosomal recessive non-syndromic intellectual disability) (TNIK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNIK	TNIK, KIAA0551, MRT54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 54, 617028, Otozomal Resesif; MRT54 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (TNIK gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 54, 617028, Autosomal recessive; MRT54 (Autosomal recessive non-syndromic intellectual disability) (TNIK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNIK	TNIK, KIAA0551, MRT54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 55, 617051, Otozomal Resesif; MRT55 (PUS3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 55, 617051, Autosomal recessive; MRT55 (PUS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PUS3	PUS3, MRT55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 55, 617051, Otozomal Resesif; MRT55 (Prenatal) (PUS3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 55, 617051, Autosomal recessive; MRT55 (PUS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PUS3	PUS3, MRT55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 56, 617125, Otozomal Resesif; MRT56 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (ZC3H14 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 56, 617125, Autosomal recessive; MRT56 (Autosomal recessive non-syndromic intellectual disability) (ZC3H14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZC3H14	ZC3H14, SUT2, MRT56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 56, 617125, Otozomal Resesif; MRT56 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (ZC3H14 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 56, 617125, Autosomal recessive; MRT56 (Autosomal recessive non-syndromic intellectual disability) (ZC3H14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZC3H14	ZC3H14, SUT2, MRT56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 57, 617188, Otozomal Resesif; MRT57 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (MBOAT7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 57, 617188, Autosomal recessive; MRT57 (Autosomal recessive non-syndromic intellectual disability) (MBOAT7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBOAT7	MBOAT7, BB1, LENG4, MRT57	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 57, 617188, Otozomal Resesif; MRT57 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (MBOAT7 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 57, 617188, Autosomal recessive; MRT57 (Autosomal recessive non-syndromic intellectual disability) (MBOAT7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MBOAT7	MBOAT7, BB1, LENG4, MRT57	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 58, 617270, Otozomal Resesif; MRT58 (ELP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 58, 617270, Autosomal recessive; MRT58 (ELP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELP2	ELP2, STATIP1, MRT58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 58, 617270, Otozomal Resesif; MRT58 (ELP2 gen) (Prenatal) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 58, 617270, Autosomal recessive; MRT58 (ELP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELP2	ELP2, STATIP1, MRT58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 59, 617323, Otozomal Resesif; MRT59 (IMPA1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 59, 617323, Autosomal recessive; MRT59 (IMPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPA1	IMPA1, MRT59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 59, 617323, Otozomal Resesif; MRT59 (Prenatal) (IMPA1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 59, 617323, Autosomal recessive; MRT59 (IMPA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IMPA1	IMPA1, MRT59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 7, 611093, Otozomal Resesif; MRT7 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (TUSC3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive 7, 611093, Autosomal recessive; MRT7 (Autosomal recessive non-syndromic intellectual disability) (TUSC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUSC3	TUSC3, M33, DBS1992, MRT7, MRT22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 7, 611093, Otozomal Resesif; MRT7 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (TUSC3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive 7, 611093, Autosomal recessive; MRT7 (Autosomal recessive non-syndromic intellectual disability) (TUSC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUSC3	TUSC3, M33, DBS1992, MRT7, MRT22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 37, 615493, Otozomal Resesif (Mental Retardasyon-Hipotoni-Spastisite-Uyku Bozukluğu Sendromu) (ANK3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive, 37, 615493, Autosomal recessive (Intellectual disability-hypotonia-spasticity-sleep disorder syndrome) (ANK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANK3	ANK3, MRT37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 37 (MRT37) [MIM:615493]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT37 patients manifest delayed global development with speech delay, hypotonia, spasticity, and a sleep disorder. Severe behavioral abnormalities include aggression, hyperactivity, and grinding of the teeth. Note=The disease is caused by mutations affecting the gene represented in this entry. A homozygous deletion in ANK3 predicted to result in frameshift and premature truncation, has been shown to be the cause of moderate intellectual disability, an ADHD-like phenotype and behavioral problems in a consanguineous family (PubMed:23390136). {ECO:0000269	PubMed:23390136}.;	EDTA Blood Sample

Mental Retardasyon, Otozomal Resesif 37, 615493, Otozomal Resesif (Mental Retardasyon-Hipotoni-Spastisite-Uyku Bozukluğu Sendromu) (Prenatal) (ANK3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive, 37, 615493, Autosomal recessive (Intellectual disability-hypotonia-spasticity-sleep disorder syndrome) (ANK3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANK3	ANK3, MRT37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 37 (MRT37) [MIM:615493]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRT37 patients manifest delayed global development with speech delay, hypotonia, spasticity, and a sleep disorder. Severe behavioral abnormalities include aggression, hyperactivity, and grinding of the teeth. Note=The disease is caused by mutations affecting the gene represented in this entry. A homozygous deletion in ANK3 predicted to result in frameshift and premature truncation, has been shown to be the cause of moderate intellectual disability, an ADHD-like phenotype and behavioral problems in a consanguineous family (PubMed:23390136). {ECO:0000269	PubMed:23390136}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 52, 616887, Otozomal Resesif; MRT52 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (LMAN2L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive, 52, 616887, Autosomal recessive; MRT52 (Autosomal recessive non-syndromic intellectual disability) (LMAN2L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMAN2L	LMAN2L, VIPL, MRT52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif 52, 616887, Otozomal Resesif; MRT52 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (LMAN2L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive, 52, 616887, Autosomal recessive; MRT52 (Autosomal recessive non-syndromic intellectual disability) (LMAN2L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMAN2L	LMAN2L, VIPL, MRT52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif, 6, 611092, Otozomal Resesif; MRT6 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (GRIK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, autosomal recessive, 6, 611092, Autosomal recessive; MRT6 (Autosomal recessive non-syndromic intellectual disability) (GRIK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIK2	GRIK2, GLUR6, MRT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal recessive 6 (MRT6) [MIM:611092]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic mental retardation. MRT6 patients display mild to severe mental retardation and psychomotor development delay in early childhood. Patients do not have neurologic problems, congenital malformations, or facial dysmorphism. Body height, weight, and head circumference are normal. {ECO:0000269	PubMed:17847003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Otozomal Resesif, 6, 611092, Otozomal Resesif; MRT6 (Otozomal Resesif Sendromik Olmayan Mental Retardasyon) (Prenatal) (GRIK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, autosomal recessive, 6, 611092, Autosomal recessive; MRT6 (Autosomal recessive non-syndromic intellectual disability) (GRIK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIK2	GRIK2, GLUR6, MRT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal recessive 6 (MRT6) [MIM:611092]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic mental retardation. MRT6 patients display mild to severe mental retardation and psychomotor development delay in early childhood. Patients do not have neurologic problems, congenital malformations, or facial dysmorphism. Body height, weight, and head circumference are normal. {ECO:0000269	PubMed:17847003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, FRA12A Tipi, 136630, Otozomal Dominant (DIP2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, FRA12A type, 136630, Autosomal dominant (DIP2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIP2B	DIP2B, KIAA1463	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, FRA12A Tipi, 136630, Otozomal Dominant (Prenatal) (DIP2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, FRA12A type, 136630, Autosomal dominant (DIP2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DIP2B	DIP2B, KIAA1463	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon ve Stereotipik Hareketler, Epilepsi, ve / Veya Serebral Bozukluklar, 613443, Otozomal Dominant (Sq14.3 Mikrodelesyon Sendromu) (MEF2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443, Autosomal dominant (Sq14.3 microdeletion syndrome) (MEF2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEF2C	MEF2C, CSDELq14.3, DELSq14.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. {ECO:0000269	PubMed:19592390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon ve Stereotipik Hareketler, Epilepsi, ve / Veya Serebral Bozukluklar, 613443, Otozomal Dominant (Sq14.3 Mikrodelesyon Sendromu) (MLPA)	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443, Autosomal dominant (Sq14.3 microdeletion syndrome) (MLPA)	MEF2C	MEF2C, CSDELq14.3, DELSq14.3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. {ECO:0000269	PubMed:19592390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon ve Stereotipik Hareketler, Epilepsi, ve / Veya Serebral Bozukluklar, 613443, Otozomal Dominant (Sq14.3 Mikrodelesyon Sendromu) (Prenatal) (MEF2C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443, Autosomal dominant (Sq14.3 microdeletion syndrome) (MEF2C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MEF2C	MEF2C, CSDELq14.3, DELSq14.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. {ECO:0000269	PubMed:19592390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon ve Stereotipik Hareketler, Epilepsi, ve / Veya Serebral Bozukluklar, 613443, Otozomal Dominant (Sq14.3 Mikrodelesyon Sendromu) (Prenatal) (MLPA)	Mental retardation, stereotypic movements, epilepsy, and/or cerebral malformations, 613443, Autosomal dominant (Sq14.3 microdeletion syndrome) (Prenatal) (MLPA)	MEF2C	MEF2C, CSDELq14.3, DELSq14.3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 20 (MRD20) [MIM:613443]: A disorder characterized by severe mental retardation, absent speech, hypotonia, poor eye contact and stereotypic movements. Dysmorphic features include high broad forehead with variable small chin, short nose with anteverted nares, large open mouth, upslanted palpebral fissures and prominent eyebrows. Some patients have seizures. {ECO:0000269	PubMed:19592390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, Trunkal Obezite, Retina Distrofisi ve Mikropenis, 610156, Otozomal Resesif; MORMS (MORM Sendromu) (Mental Retardasyon, Trunkal Obezite, Retina Distrofisi ve Mikropenis)) (INPP5E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156, Autosomal recessive; MORMS (MORM syndrome) (INPP5E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INPP5E	INPP5E, MORMS, JBTS1, CORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, truncal obesity, retinal dystrophy, and micropenis (MORMS) [MIM:610156]: An autosomal recessive disorder characterized by moderate mental retardation, truncal obesity, congenital non-progressive retinal dystrophy, and micropenis in males. The phenotype is similar to Bardet-Biedl syndrome and Cohen syndrome Distinguishing features are the age of onset, the non-progressive nature of the visual impairment, lack of dysmorphic facies, skin or gingival infection, microcephaly, mottled retina, polydactyly, and testicular anomalies. {ECO:0000269	PubMed:19668215}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, Trunkal Obezite, Retina Distrofisi ve Mikropenis, 610156, Otozomal Resesif; MORMS (MORM Sendromu) (Mental Retardasyon, Trunkal Obezite, Retina Distrofisi ve Mikropenis)) (Prenatal) (INPP5E geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, truncal obesity, retinal dystrophy, and micropenis, 610156, Autosomal recessive; MORMS (MORM syndrome) (INPP5E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INPP5E	INPP5E, MORMS, JBTS1, CORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, truncal obesity, retinal dystrophy, and micropenis (MORMS) [MIM:610156]: An autosomal recessive disorder characterized by moderate mental retardation, truncal obesity, congenital non-progressive retinal dystrophy, and micropenis in males. The phenotype is similar to Bardet-Biedl syndrome and Cohen syndrome Distinguishing features are the age of onset, the non-progressive nature of the visual impairment, lack of dysmorphic facies, skin or gingival infection, microcephaly, mottled retina, polydactyly, and testicular anomalies. {ECO:0000269	PubMed:19668215}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon e Nistagmus, 300422 (CASK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, with or without nystagmus, 300422 (CASK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269	PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon e Nistagmus, 300422 (Prenatal) (CASK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, with or without nystagmus, 300422 (CASK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASK	CASK, MICPCH, FGS4, CMG, MRXSNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: FG syndrome 4 (FGS4) [MIM:300422]: FG syndrome (FGS) is an X-linked disorder characterized by mental retardation, relative macrocephaly, hypotonia and constipation. {ECO:0000269	PubMed:19200522}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 1/78, 309530, X'e Bağlı Dominant; MRX1 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (IQSEC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 1/78, 309530, X-linked dominant; MRX1 (X-linked non-syndromic intellectual disability) (IQSEC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IQSEC2	IQSEC2, KIAA0522, MRX1, MRX78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 1/78, 309530, X'e Bağlı Dominant; MRX1 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (IQSEC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 1/78, 309530, X-linked dominant; MRX1 (X-linked non-syndromic intellectual disability) (IQSEC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IQSEC2	IQSEC2, KIAA0522, MRX1, MRX78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 100, 300923, X'e Bağlı Resesif; MRX100 (KIF4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 100, 300923, X-linked recessive; MRX100 (KIF4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF4A	KIF4A, KIF4, MRX100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 100, 300923, X'e Bağlı Resesif; MRX100 (KIF4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 100, 300923, X-linked recessive; MRX100 (KIF4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF4A	KIF4A, KIF4, MRX100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 101, 300928, X'e Bağlı Resesif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (MID2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 101, 300928, X-linked recessive (X-linked non-syndromic intellectual disability) (MID2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MID2	MID2, MRX101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 101 (MRX101) [MIM:300928]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX101 clinical features include global developmental delay, hyperactivity often with aggressive outbursts, and seizures in some patients. Several affected individuals have long face, prominent ears, and squint or strabismus. {ECO:0000269	PubMed:24115387}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mental Retardasyon, X'e Bağlı 101, 300928, X'e Bağlı Reseslf (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (MID2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 101, 300928, X-linked recessive (X-linked non-syndromic intellectual disability) (MID2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MID2	MID2, MRX101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 101 (MRX101) [MIM:300928]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX101 clinical features include global developmental delay, hyperactivity often with aggressive outbursts, and seizures in some patients. Several affected individuals have long face, prominent ears, and squint or strabismus. {ECO:0000269|PubMed:24115387}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 102, 300958, X'e Bağlı Reseslf, X'e Bağlı Dominant; MRX102 (X'e Bağlı Mental Retardasyon-Hipotoni-Hareket Bozukluğu Sendromu) (DDX3X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 102, 300958, X-linked recessive, X-linked dominant; MRX102 (X-linked intellectual disability-hypotonia-movement disorder syndrome) (DDX3X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDX3X	DDX3X, DDX3, DBX, MRX102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 102, 300958, X'e Bağlı Reseslf, X'e Bağlı Dominant; MRX102 (X'e Bağlı Mental Retardasyon-Hipotoni-Hareket Bozukluğu Sendromu) (Prenatal) (DDX3X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 102, 300958, X-linked recessive, X-linked dominant; MRX102 (X-linked intellectual disability-hypotonia-movement disorder syndrome) (DDX3X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDX3X	DDX3X, DDX3, DBX, MRX102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 103, 300982, X'e Bağlı Reseslf, MRX103 (KLHL15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 103, 300982, X-linked recessive; MRX103 (KLHL15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL15	KLHL15, KIAA1677	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 103, 300982, X'e Bağlı Reseslf, MRX103 (KLHL15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 103, 300982, X-linked recessive; MRX103 (KLHL15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLHL15	KLHL15, KIAA1677	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 104, 300983, X'e Bağlı Reseslf; MRX104 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (FRMPD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 104, 300983, X-linked recessive; MRX104 (X-linked non-syndromic intellectual disability) (FRMPD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRMPD4	FRMPD4, PRESO, KIAA0316, MRX104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 104, 300983, X'e Bağlı Reseslf; MRX104 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (FRMPD4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 104, 300983, X-linked recessive; MRX104 (X-linked non-syndromic intellectual disability) (FRMPD4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FRMPD4	FRMPD4, PRESO, KIAA0316, MRX104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 12/35, 300957, X'e Bağlı Reseslf (X'e Bağlı Mental Retardasyon-Boy Kısalığı-Şişmanlık Sendromu) (THOC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 12/35, 300957, X-linked recessive (X-linked intellectual disability-short stature-overweight syndrome) (THOC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THOC2	THOC2, THO2, MRX12, MRX35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 12/35, 300957, X'e Bağlı Reseslf (X'e Bağlı Mental Retardasyon-Boy Kısalığı-Şişmanlık Sendromu) (Prenatal) (THOC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 12/35, 300957, X-linked recessive (X-linked intellectual disability-short stature-overweight syndrome) (THOC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	THOC2	THOC2, THO2, MRX12, MRX35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı19, 300844, X'e Bağlı Dominant; MRX19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 19, 300844, X-linked dominant; MRX19 (X-linked non-syndromic intellectual disability) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı19, 300844, X'e Bağlı Dominant; MRX19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (MLPA)	Mental retardation, X-linked 19, 300844, X-linked dominant; MRX19 (X-linked non-syndromic intellectual disability) (MLPA)	RPS6KA3	RPS6KA3, RSK2, MRX19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı19, 300844, X'e Bağlı Dominant; MRX19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 19, 300844, X-linked dominant; MRX19 (X-linked non-syndromic intellectual disability) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı19, 300844, X'e Bağlı Dominant; MRX19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (MLPA)	Mental retardation, X-linked 19, 300844, X-linked dominant; MRX19 (X-linked non-syndromic intellectual disability) (Prenatal) (MLPA)	RPS6KA3	RPS6KA3, RSK2, MRX19	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 19; Mrx19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MENTAL RETARDATION, X-LINKED 19; MRX19 (X-linked non-syndromic intellectual disability) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 19; Mrx19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal)	MENTAL RETARDATION, X-LINKED 19; MRX19 (X-linked non-syndromic intellectual disability) (MLPA)	RPS6KA3	RPS6KA3, RSK2, MRX19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 19; Mrx19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (RPS6KA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MENTAL RETARDATION, X-LINKED 19; MRX19 (X-linked non-syndromic intellectual disability) (RPS6KA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPS6KA3	RPS6KA3, RSK2, MRX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 19; Mrx19 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (MLPA)	MENTAL RETARDATION, X-LINKED 19; MRX19 (X-linked non-syndromic intellectual disability) (Prenatal) (MLPA)	RPS6KA3	RPS6KA3, RSK2, MRX19	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 19 (MRX19) [MIM:300844]: A non-syndromic form of mild to moderate mental retardation. Mental retardation is characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. In contrast to syndromic or specific X-linked mental retardation which also present with associated physical, neurological and/or psychiatric manifestations, intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation. {ECO:0000269|PubMed:10319851, ECO:0000269|PubMed:17100996}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 21/34, 300143, X'e Bağlı Reseslf; MRX21 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (IL1RAPL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 21/34, 300143, X-linked recessive; MRX21 (X-linked non-syndromic intellectual disability) (IL1RAPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL1RAPL1	IL1RAPL1, IL1R8, MRX21, MRX34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 21/34, 300143, X'e Bağlı Reseslf; MRX21 (X`e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (IL1RAPL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 21/34, 300143, X-linked recessive; MRX21 (X-linked non-syndromic intellectual disability) (IL1RAPL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL1RAPL1	IL1RAPL1, IL1R8, MRX21, MRX34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, X'e Bağlı 29 ve Diğerleri, 300419, X'e Bağlı Reseif; MRXARX (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 29 and others, 300419, X-linked recessive; MRXARX (X-linked non-syndromic intellectual disability) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 29 ve Diğerleri, 300419, X'e Bağlı Reseif; MRXARX (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (MLPA)	Mental retardation, X-linked 29 and others, 300419, X-linked recessive; MRXARX (X-linked non-syndromic intellectual disability) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 29 ve Diğerleri, 300419, X'e Bağlı Reseif; MRXARX (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 29 and others, 300419, X-linked recessive; MRXARX (X-linked non-syndromic intellectual disability) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 29 ve Diğerleri, 300419, X'e Bağlı Reseif; MRXARX (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (MLPA)	Mental retardation, X-linked 29 and others, 300419, X-linked recessive; MRXARX (X-linked non-syndromic intellectual disability) (Prenatal) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 3 (Metilmalonik Asidemi ve Homosistinemi, Cbtx Tipi), 309541, X'e Bağlı Reseif (Metilmalonik Asidemi-Homosistinürili, Tip Cblx) (HCFC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 3 (methylmalonic acidemia and homocystinemia, cblX type), 309541, X-linked recessive (Methylmalonic acidemia with homocystinuria, type cblX) (HCFC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCFC1	HCFC1, HCF1, MRX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 3 (MRX3) [MIM:309541]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:23000143}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 3 (Metilmalonik Asidemi ve Homosistinemi, Cbtx Tipi), 309541, X'e Bağlı Reseif (Metilmalonik Asidemi-Homosistinürili, Tip Cblx) (Prenatal) (HCFC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 3 (methylmalonic acidemia and homocystinemia, cblX type), 309541, X-linked recessive (Methylmalonic acidemia with homocystinuria, type cblX) (HCFC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HCFC1	HCFC1, HCF1, MRX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 3 (MRX3) [MIM:309541]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non-syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:23000143}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 30/47, 300558, X'e Bağlı Reseif; MRX30 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (PAK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 30/47, 300558, X-linked recessive; MRX30 (X-linked non-syndromic intellectual disability) (PAK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAK3	PAK3, MRX30, MRX47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 30 (MRX30) [MIM:300558]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:10946356, ECO:0000269|PubMed:12884430, ECO:0000269|PubMed:9731525}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 30/47, 300558, X'e Bağlı Reseif; MRX30 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (PAK3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 30/47, 300558, X-linked recessive; MRX30 (X-linked non-syndromic intellectual disability) (PAK3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAK3	PAK3, MRX30, MRX47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 30 (MRX30) [MIM:300558]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:10946356, ECO:0000269|PubMed:12884430, ECO:0000269|PubMed:9731525}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 41, 300849, X'e Bağlı Dominant; MRX41 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (GDI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 41, 300849, X-linked dominant; MRX41 (X-linked non-syndromic intellectual disability) (GDI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDI1	GDI1, RABGD1A, MRX41, MRX48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 41, 300849, X'e Bağlı Dominant; MRX41 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (GDI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 41, 300849, X-linked dominant; MRX41 (X-linked non-syndromic intellectual disability) (GDI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDI1	GDI1, RABGD1A, MRX41, MRX48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 46, 300436, X'e Bağlı Reseif; MRX46 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (ARHGEF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 46, 300436, X-linked recessive; MRX46 (X-linked non-syndromic intellectual disability) (ARHGEF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGEF6	ARHGEF6, MRX46, COOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 46, 300436, X'e Bağlı Reseif; MRX46 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (ARHGEF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 46, 300436, X-linked recessive; MRX46 (X-linked non-syndromic intellectual disability) (ARHGEF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARHGEF6	ARHGEF6, MRX46, COOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 49, Mrx49 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (CLCN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MENTAL RETARDATION, X-LINKED 49; MRX49 (X-linked non-syndromic intellectual disability) (CLCN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN4	CLCN4, MRX49, MRX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 49, Mrx49 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (CLCN4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MENTAL RETARDATION, X-LINKED 49; MRX49 (X-linked non-syndromic intellectual disability) (CLCN4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN4	CLCN4, MRX49, MRX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 49/15, 300114, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (CLCN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 49/15, 300114, X-linked recessive (X-linked non-syndromic intellectual disability) (CLCN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN4	CLCN4, MRX49, MRX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 49/15, 300114, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (CLCN4 geni) (Tüm Gen Dizi Analizi (Prenatal)	Mental retardation, X-linked 49/15, 300114, X-linked recessive (X-linked non-syndromic intellectual disability) (CLCN4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN4	CLCN4, MRX49, MRX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 52, 300504, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (ATRX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 52, 300504, X-linked recessive (X-linked non-syndromic intellectual disability) (ATRX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATRX	MRXS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 52, 300504, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (MLPA)	Mental retardation, X-linked 52, 300504, X-linked recessive (X-linked non-syndromic intellectual disability) (MLPA)	ATRX	MRXS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 52, 300504, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (ATRX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 52, 300504, X-linked recessive (X-linked non-syndromic intellectual disability) (ATRX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATRX	MRXS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 52, 300504, X'e Bağlı Reseif (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (MLPA)	Mental retardation, X-linked 52, 300504, X-linked recessive (X-linked non-syndromic intellectual disability) (Prenatal) (MLPA)	ATRX	MRXS2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alpha-thalassemia myelodysplasia syndrome (ATMDS) [MIM:300448]: A disorder characterized by hypochromic, microcytic red blood cells, hemoglobin H detected in peripheral blood, and multilineage myelodysplasia. {ECO:0000269	PubMed:12858175}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 58, 300210, X'e Bağlı Reseif; MRXS8 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (TSPAN7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 58, 300210, X-linked recessive; MRXS8 (X-linked non-syndromic intellectual disability) (TSPAN7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSPAN7	TSPAN7, TM4SF2, MXS1, A15, MRXS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 58, 300210, X'e Bağlı Reseif; MRXS8 (X' e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (TSPAN7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 58, 300210, X-linked recessive; MRXS8 (X-linked non-syndromic intellectual disability) (TSPAN7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSPAN7	TSPAN7, TM4SF2, MXS1, A15, MRXS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, X'e Bağlı 61, 300978, X'e Bağlı Resesif (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (RLIM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 61, 300978, X-linked recessive (X-linked non-syndromic intellectual disability) (RLIM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RLIM	RNF12, RLIM, MRX61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 61, 300978, X'e Bağlı Resesif (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (RLIM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 61, 300978, X-linked recessive (X-linked non-syndromic intellectual disability) (RLIM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RLIM	RNF12, RLIM, MRX61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 63, 300387, X'e Bağlı Dominant; MRX63 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (ACSL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 63, 300387, X-linked dominant; MRX63 (X-linked non-syndromic intellectual disability) (ACSL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACSL4	ACSL4, FACL4, ACS4, MRX63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alport syndrome with mental retardation, midface hypoplasia and elliptocytosis (ATS-MR) [MIM:300194]: A X-linked contiguous gene deletion syndrome characterized by glomerulonephritis, sensorineural hearing loss, mental retardation, midface hypoplasia and elliptocytosis. {ECO:0000269|PubMed:9480748}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 63, 300387, X'e Bağlı Dominant; MRX63 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (ACSL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 63, 300387, X-linked dominant; MRX63 (X-linked non-syndromic intellectual disability) (ACSL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACSL4	ACSL4, FACL4, ACS4, MRX63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alport syndrome with mental retardation, midface hypoplasia and elliptocytosis (ATS-MR) [MIM:300194]: A X-linked contiguous gene deletion syndrome characterized by glomerulonephritis, sensorineural hearing loss, mental retardation, midface hypoplasia and elliptocytosis. {ECO:0000269|PubMed:9480748}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 72, 300271, X'e Bağlı Resesif; MRX72 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (RAB39B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 72, 300271, X-linked recessive; MRX72 (X-linked non-syndromic intellectual disability) (RAB39B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB39B	RAB39B, MRX72, WSMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Waisman syndrome (WSMN) [MIM:311510]: A neurologic disorder characterized by delayed psychomotor development, intellectual disability, and early-onset Parkinson disease. {ECO:0000269|PubMed:25434055}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 72, 300271, X'e Bağlı Resesif; MRX72 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (RAB39B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 72, 300271, X-linked recessive; MRX72 (X-linked non-syndromic intellectual disability) (RAB39B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB39B	RAB39B, MRX72, WSMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Waisman syndrome (WSMN) [MIM:311510]: A neurologic disorder characterized by delayed psychomotor development, intellectual disability, and early-onset Parkinson disease. {ECO:0000269|PubMed:25434055}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 9/44, 309549, X-linked recessive; MRX9 (X-linked non-syndromic intellectual-disability) (FTSJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	Mental retardation, X-linked 9/44, 309549, X-linked recessive; MRX9 (X-linked non-syndromic intellectual-disability) (FTSJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTSJ1	FTSJ1, JM23, SPB1, MRX44, MRX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 9/44, 309549, X'e Bağlı Resesif; MRX9 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (FTSJ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 9/44, 309549, X-linked recessive; MRX9 (X-linked non-syndromic intellectual disability) (FTSJ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FTSJ1	FTSJ1, JM23, SPB1, MRX44, MRX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 90, 300850, X'e Bağlı Resesif; MRX90 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (DLG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 90, 300850, X-linked recessive; MRX90 (X-linked non-syndromic intellectual disability) (DLG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLG3	DLG3, NEDLG, SAP102, MRX90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 90 (MRX90) [MIM:300850]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:15185169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 90, 300850, X'e Bağlı Resesif; MRX90 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (DLG3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 90, 300850, X-linked recessive; MRX90 (X-linked non-syndromic intellectual disability) (DLG3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLG3	DLG3, NEDLG, SAP102, MRX90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 90 (MRX90) [MIM:300850]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:15185169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 91, 300577, X'e Bağlı Dominant (ZDHHC15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 91, 300577, X-linked dominant (ZDHHC15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZDHHC15	ZDHHC15, MRX91	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 91 (MRX91) [MIM:300577]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:15915161}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 91, 300577, X'e Bağlı Dominant (Prenatal) (ZDHHC15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 91, 300577, X-linked dominant (ZDHHC15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZDHHC15	ZDHHC15, MRX91	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 91 (MRX91) [MIM:300577]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269|PubMed:15915161}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 93, 300659, X'e Bağlı Resesif (BRWD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 93, 300659, X-linked recessive (BRWD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRWD3	BRWD3, MRX93	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 93 (MRX93) [MIM:300659]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX93 is associated with macrocephaly. {ECO:0000269|PubMed:17668385}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 93, 300659, X'e Bağlı Resesif (Prenatal) (BRWD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 93, 300659, X-linked recessive (BRWD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRWD3	BRWD3, MRX93	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 93 (MRX93) [MIM:300659]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX93 is associated with macrocephaly. {ECO:0000269|PubMed:17668385}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 94, 300699, X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, GRIA3 Anomalilerine Bağlı) (GRIA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 94, 300699, X-linked recessive (X-linked intellectual disability due to GRIA3 anomalies) (GRIA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRIA3	GRIA3, GLUR3, MRX94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 94 (MRX94) [MIM:300699]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX94 patients have moderate mental retardation. Other variable features are macrocephaly, seizures, myoclonic jerks, autistic behavior, asthenic body habitus, distal muscle weakness and hyporeflexia. {ECO:0000269|PubMed:17988220}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 94, 300699, X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, GRIA3 Anomalilerine Bağlı) (Prenatal) (GRIA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 94, 300699, X-linked recessive (X-linked intellectual disability due to GRIA3 anomalies) (GRIA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRIA3	GRIA3, GLUR3, MRX94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 94 (MRX94) [MIM:300699]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. MRX94 patients have moderate mental retardation. Other variable features are macrocephaly, seizures, myoclonic jerks, autistic behavior, asthenic body habitus, distal muscle weakness and hyporeflexia. {ECO:0000269|PubMed:17988220}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 96, 300802, X'e Bağlı Dominant; MRX96 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (SYP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 96, 300802, X-linked dominant; MRX96 (X-linked non-syndromic intellectual disability) (SYP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYP	SYP, MRXSYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, SYP-related (MRXSYP) [MIM:300802]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:19377476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 96, 300802, X'e Bağlı Dominant; MRX96 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (SYP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 96, 300802, X-linked dominant; MRX96 (X-linked non-syndromic intellectual disability) (SYP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYP	SYP, MRXSYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, SYP-related (MRXSYP) [MIM:300802]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269|PubMed:19377476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 97, 300803, X'e Bağlı; MRX97 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (ZNF711 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 97, 300803, X-linked; MRX97 (X-linked non-syndromic intellectual disability) (ZNF711 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF711	ZNF711, ZNF6, CMPX1, MRX97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 97, 300803, X'e Bağlı; MRX97 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (ZNF711 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 97, 300803, X-linked; MRX97 (X-linked non-syndromic intellectual disability) (ZNF711 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNF711	ZNF711, ZNF6, CMPX1, MRX97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, X'e Bağlı 98, 300912. X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, Cantagrel Tipi) (KIAA2022 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 98, 300912, X-linked recessive (X-linked intellectual disability, Cantagrel type) (KIAA2022 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA2022	KIAA2022, MRX98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 98 (MRX98) [MIM:300912]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRX98 patients show delayed psychomotor development, absent or poor speech development, and postnatal growth retardation, often with microcephaly. Some patients show autistic behavioral features, such as stereotypic hand movements and repetitive behaviors. Additional, more variable features include spasticity, axial hypotonia, seizures, drooling, gastroesophageal reflux, and lack of sphincter control. {ECO:0000269|PubMed:15466006, ECO:0000269|PubMed:23615299}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving KIAA2022 is found in patients with severe mental retardation. Pericentric inversion inv(X)(p22.3;q13.2) with P2RY8 leading to inactivation of KIAA2022 (PubMed:15466006). {ECO:0000269|PubMed:15466006}.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 98, 300912. X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, Cantagrel Tipi) (Prenatal) (KIAA2022 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 98, 300912, X-linked recessive (X-linked intellectual disability, Cantagrel type) (KIAA2022 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIAA2022	KIAA2022, MRX98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 98 (MRX98) [MIM:300912]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRX98 patients show delayed psychomotor development, absent or poor speech development, and postnatal growth retardation, often with microcephaly. Some patients show autistic behavioral features, such as stereotypic hand movements and repetitive behaviors. Additional, more variable features include spasticity, axial hypotonia, seizures, drooling, gastroesophageal reflux, and lack of sphincter control. {ECO:0000269|PubMed:15466006, ECO:0000269|PubMed:23615299}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving KIAA2022 is found in patients with severe mental retardation. Pericentric inversion inv(X)(p22.3;q13.2) with P2RY8 leading to inactivation of KIAA2022 (PubMed:15466006). {ECO:0000269|PubMed:15466006}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 99, 300919, X'e Bağlı Resesif, MRX99 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (USP9X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 99, 300919, X-linked recessive; MRX99 (X-linked non-syndromic intellectual disability) (USP9X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USP9X	USP9X, DFFRX, MRX99, MRXS99F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 99, 300919, X'e Bağlı Resesif, MRX99 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (USP9X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 99, 300919, X-linked recessive; MRX99 (X-linked non-syndromic intellectual disability) (USP9X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	USP9X	USP9X, DFFRX, MRX99, MRXS99F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 99, Sendromik, Kadınlarda Görülen, 300968, X'e Bağlı Dominant (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (USP9X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked 99, syndromic, female-restricted, 300968, X-linked dominant (X-linked non-syndromic intellectual disability) (USP9X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USP9X	USP9X, DFFRX, MRX99, MRXS99F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı 99, Sendromik, Kadınlarda Görülen, 300968, X'e Bağlı Dominant (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal) (USP9X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked 99, syndromic, female-restricted, 300968, X-linked dominant (X-linked non-syndromic intellectual disability) (USP9X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	USP9X	USP9X, DFFRX, MRX99, MRXS99F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 10, 300220, X'e Bağlı (HSD10 Hastalığı) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic 10, 300220, X-linked recessive (HSD10 disease) (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 10 (MRXS10) [MIM:300220]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS10 patients manifest mild mental retardation, choreoathetosis and abnormal behavior. {ECO:0000269|PubMed:17236142}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 10, 300220, X'e Bağlı (HSD10 Hastalığı) (Prenatal) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic 10, 300220, X-linked recessive (HSD10 disease) (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 10 (MRXS10) [MIM:300220]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS10 patients manifest mild mental retardation, choreoathetosis and abnormal behavior. {ECO:0000269|PubMed:17236142}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 16, 305400, X'e Bağlı Resesif (Aarskog-Scott Sendromu) (FGD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic 16, 305400, X-linked recessive (Aarskog-Scott syndrome) (FGD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGD1	FGD1, FGDY, AAS, MRXS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aarskog-Scott syndrome (AAS) [MIM:305400]: A rare multisystemic disorder characterized by disproportionately short stature, and by facial, skeletal and urogenital anomalies. Some patients manifest mental retardation, attention deficit disorder and hyperactivity. {ECO:0000269|PubMed:10930571, ECO:0000269|PubMed:11093277, ECO:0000269|PubMed:14560308}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 16, 305400, X'e Bağlı Resesif (Aarskog-Scott Sendromu) (Prenatal) (FGD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic 16, 305400, X-linked recessive (Aarskog-Scott syndrome) (FGD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGD1	FGD1, FGDY, AAS, MRXS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aarskog-Scott syndrome (AAS) [MIM:305400]: A rare multisystemic disorder characterized by disproportionately short stature, and by facial, skeletal and urogenital anomalies. Some patients manifest mental retardation, attention deficit disorder and hyperactivity. {ECO:0000269|PubMed:10930571, ECO:0000269|PubMed:11093277, ECO:0000269|PubMed:14560308}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 5, 304340, X'e Bağlı Resesif (X'e Bağlı Ensefalokist Yetersizlikli-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı) Nöbet Sendromu)(Fried Sendromu) (X'e Bağlı Ensefalokist Yetersizlik-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı-Nöbet Sendromu) (AP1S2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic 5, 304340, X-linked recessive (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (Fried syndrome) (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (AP1S2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP1S2	AP1S2, MRX59, MRXSF, MRXS21, MRXSS, PGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik 5, 304340, X'e Bağlı Resesif (X'e Bağlı Ensefalokist Yetersizlikli-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı) Nöbet Sendromu)(Fried Sendromu) (X'e Bağlı Ensefalokist Yetersizlik-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı-Nöbet Sendromu) (Prenatal) (AP1S2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic 5, 304340, X-linked recessive (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (Fried syndrome) (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (AP1S2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP1S2	AP1S2, MRX59, MRXSF, MRXS21, MRXSS, PGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Christianson Tipi, 300243, X'e Bağlı Dominant; MRXSCH (Christianson Sendromu) (SLC9A6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic, Christianson type, 300243, X-linked dominant; MRXSCH (Christianson syndrome) (SLC9A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC9A6	SLC9A6, NHE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Christianson Tipi, 300243, X'e Bağlı Dominant; MRXSCH (Christianson Sendromu) (Prenatal) (SLC9A6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic, Christianson type, 300243, X-linked dominant; MRXSCH (Christianson syndrome) (SLC9A6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC9A6	SLC9A6, NHE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Lubs Tipi, 300260, X'e Bağlı Resesif (Trizomi Xq28) (MECP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic, Lubs type, 300260, X-linked recessive (Trisomy Xq28) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Lubs Tipi, 300260, X'e Bağlı Resesif (Trizomi Xq28) (MLPA)	Mental retardation, X-linked syndromic, Lubs type, 300260, X-linked recessive (Trisomy Xq28) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Lubs Tipi, 300260, X'e Bağlı Resesif (Trizomi Xq28) (Prenatal) (MECP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic, Lubs type, 300260, X-linked recessive (Trisomy Xq28) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Lubs Tipi, 300260, X'e Bağlı Resesif (Trizomi Xq28) (Prenatal) (MLPA)	Mental retardation, X-linked syndromic, Lubs type, 300260, X-linked recessive (Trisomy Xq28) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Nascimento-Tipi, 300860, X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, Nascimento Tipi) (UBE2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic, Nascimento-type, 300860, X-linked recessive (X-linked intellectual disability, Nascimento type) (UBE2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBE2A	UBE2A, RAD6A, MRXSN, MRXS30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, Nascimento-type (MRXSN) [MIM:300860]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXSN features include dysmorphic facies, hirsutism, skin and nails abnormalities, obesity, speech anomalies and seizures. {ECO:0000269|PubMed:16990393, ECO:0000269|PubMed:20412111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mental Retardasyon, X'e Bağlı Sendromik, Nascimento-Tipi, 300860, X'e Bağlı Resesif (X'e Bağlı Mental Retardasyon, Nascimento Tipi) (Prenatal) (UBE2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic, Nascimento-type, 300860, X-linked recessive (X-linked intellectual disability, Nascimento type) (UBE2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBE2A	UBE2A, RAD6A, MRXSN, MRXS30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, Nascimento-type (MRXSN) [MIM:300860]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXSN features include dysmorphic facies, hirsutism, skin and nails abnormalities, obesity, speech anomalies and seizures. {ECO:0000269|PubMed:16909393, ECO:0000269|PubMed:20412111}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Raymond Tipi, 300799 (X'e Bağlı Mental Retardasyon, Raymond Tipi) (ZDHHC9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic, Raymond type, 300799 (X-linked intellectual disability, Raymond type) (ZDHHC9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZDHHC9	ZDHHC9, DHHC9, MRXSZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, ZDHHC9-related (MRXSZ) [MIM:300799]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Some MRXSZ patients have marfanoid habitus as an additional feature. {ECO:0000269|PubMed:17436253, ECO:0000269|PubMed:19377476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Raymond Tipi, 300799 (X'e Bağlı Mental Retardasyon, Raymond Tipi) (Prenatal) (ZDHHC9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic, Raymond type, 300799 (X-linked intellectual disability, Raymond type) (ZDHHC9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZDHHC9	ZDHHC9, DHHC9, MRXSZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, ZDHHC9-related (MRXSZ) [MIM:300799]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Some MRXSZ patients have marfanoid habitus as an additional feature. {ECO:0000269|PubMed:17436253, ECO:0000269|PubMed:19377476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Turner Tipi, 300706 (X'e Bağlı Mental Retardasyon, Turner Tipi) (HUWE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked syndromic, Turner type, 300706 (X-linked intellectual disability, Turner type) (HUWE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HUWE1	HUWE1, UREB1, KIAA0312, LASU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 17 (MRX17) [MIM:300705]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:18252223}. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal microduplication involving HSD17B10 and HUWE1 has been found in patients with mental retardation.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı Sendromik, Turner Tipi, 300706 (X'e Bağlı Mental Retardasyon, Turner Tipi) (Prenatal) (HUWE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked syndromic, Turner type, 300706 (X-linked intellectual disability, Turner type) (HUWE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HUWE1	HUWE1, UREB1, KIAA0312, LASU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 17 (MRX17) [MIM:300705]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:18252223}. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal microduplication involving HSD17B10 and HUWE1 has been found in patients with mental retardation.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, 300495, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (NLGN4X geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, 300495, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asperger syndrome, X-linked, 2 (ASPGX2) [MIM:300497]: A syndrome with foatures similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:14963808}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, 300495, Sporadik Vakalar, X'e Bağlı, Multifaktöriyel (Prenatal) (NLGN4X geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, 300495, Isolated cases, X-linked, Multifactorial (NLGN4X gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLGN4X	NLGN4, KIAA1260, AUTSX2, ASPGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Asperger syndrome, X-linked, 2 (ASPGX2) [MIM:300497]: A syndrome with foatures similar to autism. Affected individuals exhibit qualitative impairment in social interaction, as manifest by impairment in the use of non-verbal behaviors such as eye-to- eye gaze, facial expression, body postures, and gestures, failure to develop appropriate peer relationships, and lack of social sharing or reciprocity. Patients also exhibit restricted, repetitive and stereotyped patterns of behavior, interests, and activities, including abnormal preoccupation with certain activities and inflexible adherence to routines or rituals. Asperger syndrome is primarily distinguished from autism by the higher cognitive abilities and a more normal and timely development of language and communicative phrases. {ECO:0000269	PubMed:14963808}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, FRAXE Tipi, 309548, X'e Bağlı Resesif (FRAXE Mental Retardasyon) (MLPA)	Mental retardation, X-linked, FRAXE type, 309548, X-linked recessive (FRAXE intellectual disability) (MLPA)	AFF2	AFF2, FMR2, FRAXE, MRX2	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, FRAXE Tipi, 309548, X'e Bağlı Resesif (FRAXE Mental Retardasyon) (Prenatal) (MLPA)	Mental retardation, X-linked, FRAXE type, 309548, X-linked recessive (FRAXE intellectual disability) (Prenatal) (MLPA)	AFF2	AFF2, FMR2, FRAXE, MRX2	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Snyder-Robinson Tipi, 309583, X'e Bağlı Resesif; MRXSSR (X'e Bağlı Mental Retardasyon, Snyder Tipi) (SMS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, Snyder-Robinson type, 309583, X-linked recessive; MRXSSR (X-linked intellectual disability, Snyder type) (SMS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMS	SMS, SRS, MRSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked syndromic mental retardation Snyder-Robinson type (MRXSSR) [MIM:309583]: Characterized by moderate intellectual deficit, hypotonia, an unsteady gait, osteoporosis, kyphoscoliosis and facial asymmetry. Transmission is X-linked recessive. {ECO:0000269|PubMed:14508504, ECO:0000269|PubMed:22612257}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Snyder-Robinson Tipi, 309583, X'e Bağlı Resesif; MRXSSR (X'e Bağlı Mental Retardasyon, Snyder Tipi) (Prenatal) (SMS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, Snyder-Robinson type, 309583, X-linked recessive; MRXSSR (X-linked intellectual disability, Snyder type) (SMS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMS	SMS, SRS, MRSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: X-linked syndromic mental retardation Snyder-Robinson type (MRXSSR) [MIM:309583]: Characterized by moderate intellectual deficit, hypotonia, an unsteady gait, osteoporosis, kyphoscoliosis and facial asymmetry. Transmission is X-linked recessive. {ECO:0000269|PubMed:14508504, ECO:0000269|PubMed:22612257}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 10, X'e Bağlı; Mrx10 (Prenatal) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC 10; MRXS10 (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 10 (MRXS10) [MIM:300220]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS10 patients manifest mild mental retardation, choreoathetosis and abnormal behavior. {ECO:0000269	PubMed:17236142}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 10, X'e Bağlı; Mrx10 (Prenatal) (HSD17B10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC 10; MRXS10 (HSD17B10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B10	HSD17B10, HADH2, ERAB, MRXS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 10 (MRXS10) [MIM:300220]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS10 patients manifest mild mental retardation, choreoathetosis and abnormal behavior. {ECO:0000269	PubMed:17236142}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 11, Shashi Tipi, 300238, X'e Bağlı Resesif; MRXS11 (X'e Bağlı Mental Retardasyon, Shashi Tipi) (RBMX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 11, Shashi type, 300238, X-linked recessive; MRXS11 (X-linked intellectual disability, Shashi type) (RBMX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBMX	RBMX, MRXS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 11 (MRXS11) [MIM:300238]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS11 patients manifest moderate intellectual disability and craniofacial dysmorphism. {ECO:0000269	PubMed:25256757}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 11, Shashi Tipi, 300238, X'e Bağlı Resesif; MRXS11 (X'e Bağlı Mental Retardasyon, Shashi Tipi) (Prenatal) (RBMX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 11, Shashi type, 300238, X-linked recessive; MRXS11 (X-linked intellectual disability, Shashi type) (RBMX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBMX	RBMX, MRXS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 11 (MRXS11) [MIM:300238]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXS11 patients manifest moderate intellectual disability and craniofacial dysmorphism. {ECO:0000269	PubMed:25256757}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 13, 300055, X'e Bağlı; MRXS13 (X'e Bağlı Entelektüal Yetersizlik-Psikoz-Makroorşidizm Sendromu) (MECP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 13, 300055, X-linked recessive; MRXS13 (X-linked intellectual disability-psychosis-macroorchidism syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 13, 300055, X'e Bağlı; MRXS13 (X'e Bağlı Entelektüal Yetersizlik-Psikoz-Makroorşidizm Sendromu) (MLPA)	Mental retardation, X-linked, syndromic 13, 300055, X-linked recessive; MRXS13 (X-linked intellectual disability-psychosis-macroorchidism syndrome) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 13, 300055, X'e Bağlı; MRXS13 (X'e Bağlı Entelektüal Yetersizlik-Psikoz-Makroorşidizm Sendromu) (Prenatal) (MECP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 13, 300055, X-linked recessive; MRXS13 (X-linked intellectual disability-psychosis-macroorchidism syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mental Retardasyon, X'e Bağlı, Sendromik 13, 300055, X'e Bağlı MRXS13 (X'e Bağlı Entelektüel Yetersizlik-Psikoz-Makroorgidizm Sendromu) (Prenatal) (MLPA)	Mental retardation, X-linked, syndromic 13, 300055, X-linked recessive; MRXS13 (X-linked intellectual disability-psychosis-macroorchidism syndrome) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, ALTS3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 14, 300676, X'e Bağlı Reseslf; MRXS14 (X'e Bağlı Mental Retardasyon- Marfanoid Habitus) (UPF3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 14, 300676, X-linked recessive; MRXS14 (X-linked intellectual disability with marfanoid habitus) (UPF3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UPF3B	UPF3B, RENT3B, MRXS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 14, 300676, X'e Bağlı Reseslf; MRXS14 (X'e Bağlı Mental Retardasyon- Marfanoid Habitus) (Prenatal) (UPF3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 14, 300676, X-linked recessive; MRXS14 (X-linked intellectual disability with marfanoid habitus) (UPF3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UPF3B	UPF3B, RENT3B, MRXS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 15 (Cabezas Tipi), 300354, X'e Bağlı Reseslf (X'e Bağlı Mental Retardasyon, Cabezas Tip) (CUL4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354, X-linked recessive (X-linked intellectual disability, Cabezas type) (CUL4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CUL4B	CUL4B, MRXSC, MRXHF2, SFM2, MRXS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 15 (MRXS15) [MIM:300354]: A syndromic form of X-linked mental retardation characterized by severe intellectual deficit associated with short stature, craniofacial dysmorphism, small testes, muscle wasting in lower legs, kyphosis, joint hyperextensibility, pes cavus, small feet, and abnormalities of the toes. Additional neurologic manifestations include speech delay and impairment, tremor, seizures, gait ataxia, hyperactivity and decreased attention span. {ECO:0000269|PubMed:17236139, ECO:0000269|PubMed:17273978, ECO:0000269|PubMed:19377476, ECO:0000269|PubMed:20002452}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 15 (Cabezas Tipi), 300354, X'e Bağlı Reseslf (X'e Bağlı Mental Retardasyon, Cabezas Tip) (CUL4B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 15 (Cabezas type), 300354, X-linked recessive (X-linked intellectual disability, Cabezas type) (CUL4B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CUL4B	CUL4B, MRXSC, MRXHF2, SFM2, MRXS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 15 (MRXS15) [MIM:300354]: A syndromic form of X-linked mental retardation characterized by severe intellectual deficit associated with short stature, craniofacial dysmorphism, small testes, muscle wasting in lower legs, kyphosis, joint hyperextensibility, pes cavus, small feet, and abnormalities of the toes. Additional neurologic manifestations include speech delay and impairment, tremor, seizures, gait ataxia, hyperactivity and decreased attention span. {ECO:0000269|PubMed:17236139, ECO:0000269|PubMed:17273978, ECO:0000269|PubMed:19377476, ECO:0000269|PubMed:20002452}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 32, 300886, X'e Bağlı Reseslf; MRXS32 (X'e Bağlı Mental Retardasyon-Kardiyomegali-Konjestif Kalp Yetmezliği Sendromu) (CLIC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 32, 300886, X-linked recessive; MRXS32 (X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome) (CLIC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLIC2	CLIC2, XAP121, MRXS32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 32 (MRXS32) [MIM:300886]: A mental retardation syndrome characterized by profound intellectual deficit, delayed psychomotor development beginning in infancy and little or no speech development. Additional features include seizures, large joint contractures, and abnormal positioning of the thumbs. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:22814392}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 32, 300886, X'e Bağlı Reseslf; MRXS32 (X'e Bağlı Mental Retardasyon-Kardiyomegali-Konjestif Kalp Yetmezliği Sendromu) (Prenatal) (CLIC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 32, 300886, X-linked recessive; MRXS32 (X-linked intellectual disability-cardiomegaly-congestive heart failure syndrome) (CLIC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLIC2	CLIC2, XAP121, MRXS32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 32 (MRXS32) [MIM:300886]: A mental retardation syndrome characterized by profound intellectual deficit, delayed psychomotor development beginning in infancy and little or no speech development. Additional features include seizures, large joint contractures, and abnormal positioning of the thumbs. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. {ECO:0000269	PubMed:22814392}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 33, 300966, X'e Bağlı Reseslf; MRXS33 (TAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 33, 300966, X-linked recessive; MRXS33 (TAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF1	TAF1, TAF2A, CCG1, BA2R, DYT3, MRXS33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 3, torsion, X-linked (DYT3) [MIM:314250]: A X- linked dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT3 is characterized by severe progressive torsion dystonia followed by parkinsonism. It has a well-defined pathology of extensive neuronal loss and mosaic gliosis in the striatum (caudate nucleus and putamen) which appears to resemble that in Huntington disease. {ECO:0000269|PubMed:12928496, ECO:0000269|PubMed:17273961}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 33, 300966, X'e Bağlı Reseslf; MRXS33 (TAF1 geni) (Prenatal) (TAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 33, 300966, X-linked recessive; MRXS33 (TAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAF1	TAF1, TAF2A, CCG1, BA2R, DYT3, MRXS33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dystonia 3, torsion, X-linked (DYT3) [MIM:314250]: A X- linked dystonia-parkinsonism disorder. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. DYT3 is characterized by severe progressive torsion dystonia followed by parkinsonism. It has a well-defined pathology of extensive neuronal loss and mosaic gliosis in the striatum (caudate nucleus and putamen) which appears to resemble that in Huntington disease. {ECO:0000269|PubMed:12928496, ECO:0000269|PubMed:17273961}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 34, 300967, X'e Bağlı; MRXS34 (Malreosefali-Mental Retardasyon-Sol Ventrikül Noncompaction Sendromu) (NONO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic 34, 300967, X-linked; MRXS34 (Macrocephaly-intellectual disability-left ventricular non compaction syndrome) (NONO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NONO	NONO, NRB54, MRXS34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NONO may be a cause of papillary renal cell carcinoma (PRCC). Translocation t(X;X)(p11.2;q13.1) with TFE3. {ECO:0000269	PubMed:9393982}.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik 34, 300967, X'e Bağlı; MRXS34 (Malreosefali-Mental Retardasyon-Sol Ventrikül Noncompaction Sendromu) (Prenatal) (NONO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic 34, 300967, X-linked; MRXS34 (Macrocephaly-intellectual disability-left ventricular non compaction syndrome) (NONO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NONO	NONO, NRB54, MRXS34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving NONO may be a cause of papillary renal cell carcinoma (PRCC). Translocation t(X;X)(p11.2;q13.1) with TFE3. {ECO:0000269	PubMed:9393982}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Bain Tipi, 300986, X'e Bağlı Dominant; MRXSB (HNRNPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic, Bain type, 300986, X-linked dominant; MRXSB (HNRNPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPH2	HNRPH2, MRXSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Bain Tipi, 300986, X'e Bağlı Dominant; MRXSB (HNRNPH2 geni) (Prenatal)	Mental retardation, X-linked, syndromic, Bain type, 300986, X-linked dominant; MRXSB (HNRNPH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HNRNPH2	HNRPH2, MRXSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Borck Tipi, 300987, X'e Bağlı Reseslf; MRXSBRK (EIF2S3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic, Borck type, 300987, X-linked recessive; MRXSBRK (EIF2S3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2S3	EIF2S3, EIF2G, MRXSBRK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Borck Tipi, 300987, X'e Bağlı Reseslf; MRXSBRK (Prenatal) (EIF2S3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic, Borck type, 300987, X-linked recessive; MRXSBRK (EIF2S3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2S3	EIF2S3, EIF2G, MRXSBRK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik Cabezas Tipi, Mrxsc (X'e Bağlı Mental Retardasyon, Cabezas Tip) (CUL4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE; MRXSC (X-linked intellectual disability, Cabezas type) (CUL4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CUL4B	CUL4B, MRXSC, MRXHF2, SFM2, MRXS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, 15 (MRXS15) [MIM:300354]: A syndromic form of X-linked mental retardation characterized by severe intellectual deficit associated with short stature, craniofacial dysmorphism, small testes, muscle wasting in lower legs, kyphosis, joint hyperextensibility, pes cavus, small feet, and abnormalities of the toes. Additional neurologic manifestations include speech delay and impairment, tremor, seizures, gait ataxia, hyperactivity and decreased attention span. {ECO:0000269|PubMed:17236139, ECO:0000269|PubMed:17273978, ECO:0000269|PubMed:19377476, ECO:0000269|PubMed:20002452}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik Cabezas Tipi, Mrxsc (X'e Bağlı Mental Retardasyon, Cabezas Tip) (Prenatal) (CUL4B geni) (Tüm Gen Dizi Analizi) (Prenatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC, CABEZAS TYPE; MRXSC (X-linked intellectual disability, Cabezas type) (CUL4B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CUL4B	CUL4B, MRXSC, MRXHF2, SFM2, MRXS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, 15 (MRXS15) [MIM:300354]: A syndromic form of X-linked mental retardation characterized by severe intellectual deficit associated with short stature, craniofacial dysmorphism, small testes, muscle wasting in lower legs, kyphosis, joint hyperextensibility, pes cavus, small feet, and abnormalities of the toes. Additional neurologic manifestations include speech delay and impairment, tremor, seizures, gait ataxia, hyperactivity and decreased attention span. {ECO:0000269|PubMed:17236139, ECO:0000269|PubMed:17273978, ECO:0000269|PubMed:19377476, ECO:0000269|PubMed:20002452}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Claes-Jensen Tipi, 300534, X'e Bağlı Reseslf; MRXSCJ (Sendromik X'e Bağlı Mental Retardasyon, JARID1C Mutasyonuna Bağlı, Claes-Jensen Tipi) (KDM5C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534, X-linked recessive; MRXSCJ (Syndromic X-linked intellectual disability due to JARID1C mutation) (KDM5C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KDM5C	KDM5C, JARID1C, SMCX, DXS1272E, XE169, MRXSCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, syndromic, Claes-Jensen type (MRXSCJ) [MIM:300534]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXSCJ patients manifest mental retardation associated with variable features such as slowly progressive spastic paraplegia, seizures, facial dysmorphism. {ECO:0000269|PubMed:15586325, ECO:0000269|PubMed:16538222, ECO:0000269|PubMed:16541399, ECO:0000269|PubMed:23356856, ECO:0000269|PubMed:25666439}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mental Retardasyon, X'e Bağlı, Sendromik, Claes–Jensen Tipi, 300534, X'e Bağlı Resesif; MRX5CJ (Sendromik X'e Bağlı Mental Retardasyon, JARID1C Mutasyonuna Bağlı, Claes–Jensen Tipi) (Prenatal) (KDM5C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic, Claes-Jensen type, 300534, X-linked recessive; MRX5CJ (Syndromic X-linked intellectual disability due to JARID1C mutation) (KDM5C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KDM5C	KDM5C, JARID1C, SMCX, DXS1272E, XE169, MRX5CJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, syndromic, Claes-Jensen type (MRX5CJ) [MIM:300534]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRX5CJ patients manifest mental retardation associated with variable features such as slowly progressive spastic paraplegia, seizures, facial dysmorphism. {ECO:0000269|PubMed:15586325, ECO:0000269|PubMed:16538222, ECO:0000269|PubMed:16541399, ECO:0000269|PubMed:23356856, ECO:0000269|PubMed:25666439}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Hedera Tipi, 300423, X'e Bağlı Resesif; MRX5H (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (ATP6AP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, syndromic, Hedera type, 300423, X-linked recessive; MRX5H (X-linked intellectual disability-epilepsy syndrome) (ATP6AP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6AP2	ATP6AP2, ATP6M8-9, XMRE, MRX5H, XPDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, with epilepsy (MRXE) [MIM:300423]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXE patients manifest mild to moderate mental retardation associated with epilepsy, delays in motor milestones and speech acquisition in infancy. {ECO:0000269	PubMed:15746149}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Hedera Tipi, 300423, X'e Bağlı Resesif; MRX5H (X'e Bağlı Mental Retardasyon Epilepsi Sendromu) (Prenatal) (ATP6AP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, syndromic, Hedera type, 300423, X-linked recessive; MRX5H (X-linked intellectual disability-epilepsy syndrome) (ATP6AP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP6AP2	ATP6AP2, ATP6M8-9, XMRE, MRX5H, XPDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, with epilepsy (MRXE) [MIM:300423]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXE patients manifest mild to moderate mental retardation associated with epilepsy, delays in motor milestones and speech acquisition in infancy. {ECO:0000269	PubMed:15746149}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Turner Tipi; Mxst (X'e Bağlı Mental Retardasyon, Turner Tipi) (HUWE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC, TURNER TYPE; MRX5T (X-linked intellectual disability, Turner type) (HUWE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HUWE1	HUWE1, UREB1, KIAA0312, LASU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked 17 (MRX17) [MIM:300705]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:18252223}. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal microduplication involving HSD17B10 and HUWE1 has been found in patients with mental retardation.;	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Sendromik, Turner Tipi; Mxst (X'e Bağlı Mental Retardasyon, Turner Tipi) (Prenatal) (HUWE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MENTAL RETARDATION, X-LINKED, SYNDROMIC, TURNER TYPE; MRX5T (X-linked intellectual disability, Turner type) (HUWE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HUWE1	HUWE1, UREB1, KIAA0312, LASU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked 17 (MRX17) [MIM:300705]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. Intellectual deficiency is the only primary symptom of non- syndromic X-linked mental retardation, while syndromic mental retardation presents with associated physical, neurological and/or psychiatric manifestations. {ECO:0000269	PubMed:18252223}. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal microduplication involving HSD17B10 and HUWE1 has been found in patients with mental retardation.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Serebellar Hipoplazi ve Belirgin Yüz Görünümü, 300486, X'e Bağlı Resesif (X'e Bağlı Entelektüel Yetersizlik-Serebellar Hipoplazi Sendromu) (OPHN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486, X-linked recessive (X-linked intellectual disability-cerebellar hypoplasia syndrome) (OPHN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPHN1	OPHN1, MRX60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, Serebellar Hipoplazi ve Belirgin Yüz Görünümü, 300486, X'e Bağlı Resesif (X'e Bağlı Entelektüel Yetersizlik-Serebellar Hipoplazi Sendromu) (Prenatal) (OPHN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, with cerebellar hypoplasia and distinctive facial appearance, 300486, X-linked recessive (X-linked intellectual disability-cerebellar hypoplasia syndrome) (OPHN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPHN1	OPHN1, MRX60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, İzole Büyüme Hormonu Eksikliği İle, 300123 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (SOX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (Non-acquired isolated growth hormone deficiency) (SOX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX3	SOX3, MRGH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, with isolated growth hormone deficiency (MRXGH) [MIM:300123]: A disorder characterized by the association of variable degrees of mental retardation with panhypopituitarism, variable combinations of hypothyroidism, delayed pubertal development, and short stature due to growth hormone deficiency. {ECO:0000269	PubMed:12428212}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mental Retardasyon, X'e Bağlı, İzole Büyüme Hormonu Eksikliği İle, 300123 (Non-Akkiz İzole Büyüme Hormonu Eksikliği) (Prenatal) (SOX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (Non-acquired isolated growth hormone deficiency) (SOX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX3	SOX3, MRGH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, X-linked, with isolated growth hormone deficiency (MRXGH) [MIM:300123]: A disorder characterized by the association of variable degrees of mental retardation with panhypopituitarism, variable combinations of hypothyroidism, delayed pubertal development, and short stature due to growth hormone deficiency. {ECO:0000269	PubMed:12428212}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mefenytoin Metabolizmasında Azalma, 609535, Otozomal Resesif (Klopidogrel Direnci) (CYP2C19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mephenytoin poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (CYP2C19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C19	CYP2C, CYP2C19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mefenytoin Metabolizmasında Azalma, 609535, Otozomal Resesif (Klopidogrel Direnci) (MLPA)	Mephenytoin poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (MLPA)	CYP2C19	CYP2C, CYP2C19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Merkel Hücreli Karsinom, Somatik (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Merkel cell carcinoma, somatic (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269	PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mezomeli-Sinostozis Sendromu, 600383, Otozomal Dominant (Mezomeli-Sinostozis Sendromu) (2200)	Mesomelia-synostoses syndrome, 600383, Autosomal dominant (Mesomelia-synostoses syndrome) (440)	.	DEL8q13, C8DELq13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Mezomeli-Sinostozis Sendromu, 600383, Otozomal Dominant (Mezomeli-Sinostozis Sendromu) (Prenatal)	Mesomelia-synostoses syndrome, 600383, Autosomal dominant (Mesomelia-synostoses syndrome) (Prenatal)	.	DEL8q13, C8DELq13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mezotelyoma, Somatik, 156240 (Plevral Mezotelyoma) (WT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mesothelioma, somatic, 156240 (Pleural mesothelioma) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPH54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Mezotelyoma, Somatik, 156240; MESOM (Plevral Mezotelyoma) BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mesothelioma, somatic, 156240; MESOM (Pleural mesothelioma) (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Metabolik Ensefalomiyopatik Kriz, Rekürren, Rabdomyoliz, Kardiyak Aritmiler ve Nörodejenerasyon, 616878, Otozomal Resesif; MECRCN (TANGO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878, Autosomal recessive; MECRCN (TANGO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TANGO2	TANGO2, C22orf25, MECRCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metabolik Ensefalomiyopatik Kriz, Rekürren, Rabdomyoliz, Kardiyak Aritmiler ve Nörodejenerasyon, 616878, Otozomal Resesif; MECRCN (TANGO2 geni) (Prenatal) (TANGO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metabolic encephalomyopathic crises, recurrent, with rhabdomyolysis, cardiac arrhythmias, and neurodegeneration, 616878, Autosomal recessive; MECRCN (TANGO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TANGO2	TANGO2, C22orf25, MECRCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metabolik Sendrom, Karışı Koruma, 605552, Otozomal Dominant (MTTP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metabolic syndrome, protection against, 605552, Autosomal dominant (MTTP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTTP	MTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Abetalipoproteinemia (ABL) [MIM:200100]: An autosomal recessive disorder of lipoprotein metabolism. Affected individuals produce virtually no circulating apolipoprotein B-containing lipoproteins (chylomicrons, VLDL, LDL, lipoprotein(a)). Malabsorption of the antioxidant vitamin E occurs, leading to spinocerebellar and retinal degeneration. {ECO:0000269|PubMed:10679949, ECO:0000269|PubMed:10946006, ECO:0000269|PubMed:22236406, ECO:0000269|PubMed:23475612, ECO:0000269|PubMed:25108285, ECO:0000269|PubMed:26224785, ECO:0000269|PubMed:8939939}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metakarpal 4-5 Füzyon, 309630, X'e Bağlı Resesif; MF4 (Sindaktili Tip 8) (FGF16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metacarpal 4-5 fusion, 309630, X-linked recessive; MF4 (Syndactyly type 8) (FGF16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF16	FGF16, MF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Metacarpal 4-5 fusion (MF4) [MIM:309630]: A rare congenital malformation of the hand characterized by the partial or complete fusion of the fourth and fifth metacarpals. The anomaly occurs as an isolated trait or part of a syndrome. {ECO:0000269|PubMed:23709756, ECO:0000269|PubMed:25333065}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metakarpal 4-5 Füzyon, 309630, X'e Bağlı Resesif; MF4 (Sindaktili Tip 8) (Prenatal) (FGF16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metacarpal 4-5 fusion, 309630, X-linked recessive; MF4 (Syndactyly type 8) (FGF16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGF16	FGF16, MF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Metacarpal 4-5 fusion (MF4) [MIM:309630]: A rare congenital malformation of the hand characterized by the partial or complete fusion of the fourth and fifth metacarpals. The anomaly occurs as an isolated trait or part of a syndrome. {ECO:0000269|PubMed:23709756, ECO:0000269|PubMed:25333065}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metakondromatozis, 156250, Otozomal Dominant; METCDS (Metakondromatozis) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metachondromatosis, 156250, Autosomal dominant; METCDS (Metachondromatosis) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. {ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Metakondromatozis, 156250, Otozomal Dominant; METCDS (Metakondromatozis) (Prenatal) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metachondromatosis, 156250, Autosomal dominant; METCDS (Metachondromatosis) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. {ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metakromatik Lökodistrofi, SAP-B Eksikliğine Bağlı, 249900, Otozomal Resesif (Metakromatik Lökodistrofi) (PSAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metachromatic leukodystrophy due to SAP-b deficiency, 249900, Autosomal recessive (Metachromatic leukodystrophy) (PSAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Leukodystrophy metachromatic due to saposin-B deficiency (MLD-SAPB) [MIM:249900]: An atypical form of metachromatic leukodystrophy. It is characterized by tissue accumulation of cerebroside-3-sulfate, demyelination, periventricular white matter abnormalities, peripheral neuropathy. Additional neurological features include dysarthria, ataxic gait, psychomotor regression, seizures, cognitive decline and spastic quadriparesis. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Metakromatik Lökodistrofi, SAP-B Eksikliğine Bağlı, 249900, Otozomal Resesif (Metakromatik Lökodistrofi) (Prenatal) (PSAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metachromatic leukodystrophy due to SAP-b deficiency, 249900, Autosomal recessive (Metachromatic leukodystrophy) (PSAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAP	PSAP, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy metachromatic due to saposin-B deficiency (MLD-SAPB) [MIM:249900]: An atypical form of metachromatic leukodystrophy. It is characterized by tissue accumulation of cerebroside-3-sulfate, demyelination, periventricular white matter abnormalities, peripheral neuropathy. Additional neurological features include dysarthria, ataxic gait, psychomotor regression, seizures, cognitive decline and spastic quadriparesis. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metakromatik Lökodistrofi, 250100, Otozomal Resesif; MLD (Metakromatik Lökodistrofi) (ARSA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metachromatic leukodystrophy, 250100, Autosomal recessive; MLD (Metachromatic leukodystrophy) (ARSA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARSA	ARSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Leukodystrophy metachromatic (MLD) [MIM:250100]: A leukodystrophy due to a lysosomal storage defect. Characterized by intralysosomal storage of cerebroside-3-sulfate in neural and non- neural tissues, with a diffuse loss of myelin in the central nervous system. Progressive demyelination causes a variety of neurological symptoms, including gait disturbances, ataxias, optical atrophy, dementia, seizures, and spastic tetraparesis. Three forms of the disease can be distinguished according to the age at onset: late-infantile, juvenile and adult. {ECO:0000269|PubMed:10220151, ECO:0000269|PubMed:10381328, ECO:0000269|PubMed:10477432, ECO:0000269|PubMed:10533072, ECO:0000269|PubMed:10751093, ECO:0000269|PubMed:11020646, ECO:0000269|PubMed:11061266, ECO:0000269|PubMed:11456299, ECO:0000269|PubMed:11941485, ECO:0000269|PubMed:1353340, ECO:0000269|PubMed:14517960, ECO:0000269|PubMed:14680985, ECO:0000269|PubMed:15026521, ECO:0000269|PubMed:15326627, ECO:0000269|PubMed:15710861, ECO:0000269|PubMed:1670590, ECO:0000269|PubMed:1673291, ECO:0000269|PubMed:1678251, ECO:0000269|PubMed:18693274, ECO:0000269|PubMed:19036494, ECO:0000269|PubMed:20339381, ECO:0000269|PubMed:21265945, ECO:0000269|PubMed:7581401	EDTA Blood Sample
Metakromatik Lökodistrofi, 250100, Otozomal Resesif; MLD (Metakromatik Lökodistrofi) (Prenatal) (ARSA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metachromatic leukodystrophy, 250100, Autosomal recessive; MLD (Metachromatic leukodystrophy) (ARSA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARSA	ARSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy metachromatic (MLD) [MIM:250100]: A leukodystrophy due to a lysosomal storage defect. Characterized by intralysosomal storage of cerebroside-3-sulfate in neural and non- neural tissues, with a diffuse loss of myelin in the central nervous system. Progressive demyelination causes a variety of neurological symptoms, including gait disturbances, ataxias, optical atrophy, dementia, seizures, and spastic tetraparesis. Three forms of the disease can be distinguished according to the age at onset: late-infantile, juvenile and adult. {ECO:0000269|PubMed:10220151, ECO:0000269|PubMed:10381328, ECO:0000269|PubMed:10477432, ECO:0000269|PubMed:10533072, ECO:0000269|PubMed:10751093, ECO:0000269|PubMed:11020646, ECO:0000269|PubMed:11061266, ECO:0000269|PubMed:11456299, ECO:0000269|PubMed:11941485, ECO:0000269|PubMed:1353340, ECO:0000269|PubMed:14517960, ECO:0000269|PubMed:14680985, ECO:0000269|PubMed:15026521, ECO:0000269|PubMed:15326627, ECO:0000269|PubMed:15710861, ECO:0000269|PubMed:1670590, ECO:0000269|PubMed:1673291, ECO:0000269|PubMed:1678251, ECO:0000269|PubMed:18693274, ECO:0000269|PubMed:19036494, ECO:0000269|PubMed:20339381, ECO:0000269|PubMed:21265945, ECO:0000269|PubMed:7581401	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafiziyal Anadisplazi 1, 602111, Otozomal Dominant (Metafiziyal Anadisplazi) (MMP13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal anadysplasia 1, 602111, Autosomal dominant (Metaphyseal anadysplasia) (MMP13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP13	MMP13, CLG3, MANDP1, MDST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Metaphyseal dysplasia, Spahr type (MDST) [MIM:250400]: An autosomal recessive, rare disease characterized by moderate short stature, mild genua vara, and radiographic signs of metaphyseal dysplasia, but no biochemical signs of rickets. {ECO:0000269|PubMed:24648384, ECO:0000269|PubMed:24781753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metafiziyal Anadisplazi 1, 602111, Otozomal Dominant (Metafiziyal Anadisplazi) (Prenatal) (MMP13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal anadysplasia 1, 602111, Autosomal dominant (Metaphyseal anadysplasia) (MMP13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP13	MMP13, CLG3, MANDP1, MDST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Metaphyseal dysplasia, Spahr type (MDST) [MIM:250400]: An autosomal recessive, rare disease characterized by moderate short stature, mild genua vara, and radiographic signs of metaphyseal dysplasia, but no biochemical signs of rickets. {ECO:0000269|PubMed:24648384, ECO:0000269|PubMed:24781753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafiziyal Anadisplazi 2, 613073; MANDP2 (Metafiziyal Anadisplazi) (MMP9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal anadysplasia 2, 613073; MANDP2 (Metaphyseal anadysplasia) (MMP9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP9	MMP9, CLG4B, MANDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Intervertebral disc disease (IDD) [MIM:603932]: A common musculo-skeletal disorder caused by degeneration of intervertebral disks of the lumbar spine. It results in low-back pain and unilateral leg pain. {ECO:0000269|PubMed:18455130}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Metafiziyal Anadisplazi 2, 613073; MANDP2 (Metafiziyal Anadisplazi) (Prenatal) (MMP9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal anadysplasia 2, 613073; MANDP2 (Metaphyseal anadysplasia) (MMP9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP9	MMP9, CLG4B, MANDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intervertebral disc disease (IDD) [MIM:603932]: A common musculo-skeletal disorder caused by degeneration of intervertebral disks of the lumbar spine. It results in low-back pain and unilateral leg pain. {ECO:0000269|PubMed:18455130}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafiziyel Kondrodisplazi, Murk Jansen Tipi, 156400, Otozomal Dominant (Metafiziyal Kondrodisplazi, Jansen Tipi) (PTH1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal chondrodysplasia, Murk Jansen type, 156400, Autosomal dominant (Metaphyseal chondrodysplasia, Jansen type) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Primary failure of tooth eruption (PFE) [MIM:125350]: Rare condition that has high penetrance and variable expressivity and in which tooth retention occurs without evidence of any obvious mechanical interference. Instead, malfunction of the eruptive mechanism itself appears to cause nonankylosed permanent teeth to fail to erupt, although the eruption pathway has been cleared by bone resorption. {ECO:0000269|PubMed:19061984}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metafiziyel Kondrodisplazi, Murk Jansen Tipi, 156400, Otozomal Dominant (Metafiziyal Kondrodisplazi, Jansen Tipi) (Prenatal) (PTH1R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal chondrodysplasia, Murk Jansen type, 156400, Autosomal dominant (Metaphyseal chondrodysplasia, Jansen type) (PTH1R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTH1R	PTHR1, PTHR, PFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Primary failure of tooth eruption (PFE) [MIM:125350]: Rare condition that has high penetrance and variable expressivity and in which tooth retention occurs without evidence of any obvious mechanical interference. Instead, malfunction of the eruptive mechanism itself appears to cause nonankylosed permanent teeth to fail to erupt, although the eruption pathway has been cleared by bone resorption. {ECO:0000269|PubMed:19061984}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafiziyel Kondrodisplazi, Schmid Tipi, 156500, Otozomal Dominant (Metafiziyal Kondrodisplazi, Schmid Tipi) (COL10A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal chondrodysplasia, Schmid type, 156500, Autosomal dominant (Metaphyseal chondrodysplasia, Schmid type) (COL10A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL10A1	COL10A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Schmid type metaphyseal chondrodysplasia (SMCD) [MIM:156500]: Dominantly inherited disorder of the ossaous skeleton. The cardinal features of the phenotype are mild short stature, coxa vara and a waddling gait. Radiography usually shows sclerosis of the ribs, flaring of the metaphyses, and a wide irregular growth plate, especially of the knees. A variant form of SMCD is spondylometaphyseal dysplasia Japanese type. It is characterized by spinal involvement comprising mild platyspondyly, vertebral body abnormalities, and end plate irregularity. {ECO:0000269|PubMed:15880705, ECO:0000269|PubMed:7607655, ECO:0000269|PubMed:7876225, ECO:0000269|PubMed:8004099, ECO:0000269|PubMed:8004336, ECO:0000269|PubMed:8782043, ECO:0000269|PubMed:9067753, ECO:0000269|PubMed:9852679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Metafizyel Kondrodisplazi, Schmid Tipi, 156500, Otozomal Dominant (Metafizyal Kondrodisplazi, Schmid Tipi) (Prenatal) (COL10A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal chondrodysplasia, Schmid type, 156500, Autosomal dominant (Metaphyseal chondrodysplasia, Schmid type) (COL10A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL10A1	COL10A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schmid type metaphyseal chondrodysplasia (SMCD) [MIM:156500]: Dominantly inherited disorder of the osseous skeleton. The cardinal features of the phenotype are mild short stature, coxa vara and a waddling gait. Radiography usually shows sclerosis of the ribs, flaring of the metaphyses, and a wide irregular growth plate, especially of the knees. A variant form of SMCD is spondylometaphyseal dysplasia Japanese type. It is characterized by spinal involvement comprising mild platyspondyly, vertebral body abnormalities, and end-plate irregularity. {ECO:0000269|PubMed:15880705, ECO:0000269|PubMed:7607655, ECO:0000269|PubMed:7876225, ECO:0000269|PubMed:8004099, ECO:0000269|PubMed:8304336, ECO:0000269|PubMed:8782043, ECO:0000269|PubMed:9067753, ECO:0000269|PubMed:9852679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafizyel Kondrodisplazi, Schmid Tipi, MCDS (Metafizyal Kondrodisplazi, Schmid Tipi) (COL10A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE; MCDS (Metaphyseal chondrodysplasia, Schmid type) (COL10A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL10A1	COL10A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schmid type metaphyseal chondrodysplasia (SMCD) [MIM:156500]: Dominantly inherited disorder of the osseous skeleton. The cardinal features of the phenotype are mild short stature, coxa vara and a waddling gait. Radiography usually shows sclerosis of the ribs, flaring of the metaphyses, and a wide irregular growth plate, especially of the knees. A variant form of SMCD is spondylometaphyseal dysplasia Japanese type. It is characterized by spinal involvement comprising mild platyspondyly, vertebral body abnormalities, and end-plate irregularity. {ECO:0000269|PubMed:15880705, ECO:0000269|PubMed:7607655, ECO:0000269|PubMed:7876225, ECO:0000269|PubMed:8004099, ECO:0000269|PubMed:8304336, ECO:0000269|PubMed:8782043, ECO:0000269|PubMed:9067753, ECO:0000269|PubMed:9852679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metafizyel Kondrodisplazi, Schmid Tipi; MCDS (Metafizyal Kondrodisplazi, Schmid Tipi) (Prenatal) (COL10A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE; MCDS (Metaphyseal chondrodysplasia, Schmid type) (COL10A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL10A1	COL10A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schmid type metaphyseal chondrodysplasia (SMCD) [MIM:156500]: Dominantly inherited disorder of the osseous skeleton. The cardinal features of the phenotype are mild short stature, coxa vara and a waddling gait. Radiography usually shows sclerosis of the ribs, flaring of the metaphyses, and a wide irregular growth plate, especially of the knees. A variant form of SMCD is spondylometaphyseal dysplasia Japanese type. It is characterized by spinal involvement comprising mild platyspondyly, vertebral body abnormalities, and end-plate irregularity. {ECO:0000269|PubMed:15880705, ECO:0000269|PubMed:7607655, ECO:0000269|PubMed:7876225, ECO:0000269|PubMed:8004099, ECO:0000269|PubMed:8304336, ECO:0000269|PubMed:8782043, ECO:0000269|PubMed:9067753, ECO:0000269|PubMed:9852679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafizyel Displazi-Maksiller Hipoplazi ± Brakidaktili, 156510, Otozomal Dominant; MDMHB (Metafizyal Displazi-Maksiller Hipoplazi-Brakidaktili Sendromu) (RUNX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510, Autosomal dominant; MDMHB (Metaphyseal dysplasia-maxillary hypoplasia-brachydactyly syndrome) (RUNX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metafizyel Displazi-Maksiller Hipoplazi ± Brakidaktili, 156510, Otozomal Dominant; MDMHB (Metafizyal Displazi-Maksiller Hipoplazi-Brakidaktili Sendromu) (Prenatal) (MLPA)	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510, Autosomal dominant; MDMHB (Metaphyseal dysplasia-maxillary hypoplasia-brachydactyly syndrome) (MLPA)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Metafizyel Displazi-Maksiller Hipoplazi ± Brakidaktili, 156510, Otozomal Dominant; MDMHB (Metafizyal Displazi-Maksiller Hipoplazi-Brakidaktili Sendromu) (Prenatal) (RUNX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510, Autosomal dominant; MDMHB (Metaphyseal dysplasia-maxillary hypoplasia-brachydactyly syndrome) (RUNX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafizyel Displazi-Maksiller Hipoplazi ± Brakidaktili, 156510, Otozomal Dominant; MDMHB (Metafizyal Displazi-Maksiller Hipoplazi-Brakidaktili Sendromu) (Prenatal) (MLPA)	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510, Autosomal dominant; MDMHB (Metaphyseal dysplasia-maxillary hypoplasia-brachydactyly syndrome) (Prenatal) (MLPA)	RUNX2	RUNX2, CBFA1, PEBP2A1, AML3, CCD, CLCD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cleidocranial dysplasia (CLCD) [MIM:119600]: Autosomal dominant skeletal disorder with high penetrance and variable expressivity. It is due to defective endochondral and intramembranous bone formation. Typical features include hypoplasia/aplasia of clavicles, patent fontanelles, wormian bones (additional cranial plates caused by abnormal ossification of the calvaria), supernumerary teeth, short stature, and other skeletal changes. In some cases defects in RUNX2 are exclusively associated with dental anomalies. {ECO:0000269|PubMed:10521292, ECO:0000269|PubMed:10545612, ECO:0000269|PubMed:10689183, ECO:0000269|PubMed:10980549, ECO:0000269|PubMed:11857736, ECO:0000269|PubMed:12081718, ECO:0000269|PubMed:12196916, ECO:0000269|PubMed:12424590, ECO:0000269|PubMed:16270353, ECO:0000269|PubMed:19744171, ECO:0000269|PubMed:20082269, ECO:0000269|PubMed:20648631, ECO:0000269|PubMed:9182765, ECO:0000269|PubMed:9207800}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafizyel Displazi, Hipotrikozis Olmadan, 250460, Otozomal Resesif; MDWH (Metafizyal Displazi-Hipotrikoz Olmadan) (RMRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal dysplasia without hypotrichosis, 250460, Autosomal recessive; MDWH (Metaphyseal dysplasia without hypotrichosis) (RMRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metafizyel Displazi, Hipotrikozis Olmadan, 250460, Otozomal Resesif; MDWH (Metafizyal Displazi-Hipotrikoz Olmadan) (Prenatal) (RMRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal dysplasia without hypotrichosis, 250460, Autosomal recessive; MDWH (Metaphyseal dysplasia without hypotrichosis) (RMRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RMRP	RMRP, RMRPR, CHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metafizyel Displazi, Spahr Tipi, 250400, Otozomal Resesif; MDST (Metafizyal Kondrodisplazi, Spahr Tipi) (MMP13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metaphyseal dysplasia, Spahr type, 250400, Autosomal recessive; MDST (Metaphyseal chondrodysplasia, Spahr type) (MMP13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP13	MMP13, CLG3, MANDP1, MDST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Metaphyseal dysplasia, Spahr type (MDST) [MIM:250400]: An autosomal recessive, rare disease characterized by moderate short stature, mild genua vara, and radiographic signs of metaphyseal dysplasia, but no biochemical signs of rickets. {ECO:0000269|PubMed:24648384, ECO:0000269|PubMed:24781753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Metafizyel Displazi, Spahr Tipi, 250400, Otozomal Resesif; MDST (Metafiyal Kondrodisplazi, Spahr Tipi) (Prenatal) (MMP13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metaphyseal dysplasia, Spahr type, 250400, Autosomal recessive; MDST (Metaphyseal chondrodysplasia, Spahr type) (MMP13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP13	MMP13, CLG3, MANDP1, MDST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Metaphyseal dysplasia, Spahr type (MDST) [MIM:250400]: An autosomal recessive, rare disease characterized by moderate short stature, mild genua vara, and radiographic signs of metaphyseal dysplasia, but no biochemical signs of rickets. (ECO:0000269|PubMed:24648394, ECO:0000269|PubMed:24781753). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metatropik Displazi, 156530, Otozomal Dominant (Metatropik Displazi) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Metatropic dysplasia, 156530, Autosomal dominant (Metatropic dysplasia) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. (ECO:0000269	PubMed:18587396). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Metatropik Displazi, 156530, Otozomal Dominant (Metatropik Displazi) (Prenatal) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Metatropic dysplasia, 156530, Autosomal dominant (Metatropic dysplasia) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. (ECO:0000269	PubMed:18587396). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Methemoglobinemi, Tip 1, 250800, Otozomal Resesif (Kalıtsal Methemoglobinemi) (CYB5R3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methemoglobinemia, type I, 250800, Autosomal recessive (Hereditary methemoglobinemia) (CYB5R3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYB5R3	CYB5R3, DIA1, BSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methemoglobinemia CYB5R3-related (METHB-CYB5R3) [MIM:250800]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. There are two types of methemoglobinemia CYB5R3-related. In type 1, the defect affects the soluble form of the enzyme, is restricted to red blood cells, and causes well-tolerated methemoglobinemia. In type 2, the defect affects both the soluble and microsomal forms of the enzyme and is thus generalized, affecting red cells, leukocytes and all body tissues. Type 2 methemoglobinemia is associated with mental deficiency and other neurologic symptoms. (ECO:0000269|PubMed:10807796, ECO:0000269|PubMed:12393396, ECO:0000269|PubMed:1400360, ECO:0000269|PubMed:15622768, ECO:0000269|PubMed:1707593, ECO:0000269|PubMed:1898726, ECO:0000269|PubMed:7718898, ECO:0000269|PubMed:8119939, ECO:0000269|PubMed:9695975, ECO:0000269|PubMed:9886302). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Methemoglobinemi, Tip 1, 250800, Otozomal Resesif (Kalıtsal Methemoglobinemi) (Prenatal) (CYB5R3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methemoglobinemia, type I, 250800, Autosomal recessive (Hereditary methemoglobinemia) (CYB5R3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYB5R3	CYB5R3, DIA1, BSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methemoglobinemia CYB5R3-related (METHB-CYB5R3) [MIM:250800]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. There are two types of methemoglobinemia CYB5R3-related. In type 1, the defect affects the soluble form of the enzyme, is restricted to red blood cells, and causes well-tolerated methemoglobinemia. In type 2, the defect affects both the soluble and microsomal forms of the enzyme and is thus generalized, affecting red cells, leukocytes and all body tissues. Type 2 methemoglobinemia is associated with mental deficiency and other neurologic symptoms. (ECO:0000269|PubMed:10807796, ECO:0000269|PubMed:12393396, ECO:0000269|PubMed:1400360, ECO:0000269|PubMed:15622768, ECO:0000269|PubMed:1707593, ECO:0000269|PubMed:1898726, ECO:0000269|PubMed:7718898, ECO:0000269|PubMed:8119939, ECO:0000269|PubMed:9695975, ECO:0000269|PubMed:9886302). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Methemoglobinemi, Tip 2, 250800, Otozomal Resesif (Kalıtsal Methemoglobinemi) (CYB5R3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methemoglobinemia, type II, 250800, Autosomal recessive (Hereditary methemoglobinemia) (CYB5R3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYB5R3	CYB5R3, DIA1, BSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methemoglobinemia CYB5R3-related (METHB-CYB5R3) [MIM:250800]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. There are two types of methemoglobinemia CYB5R3-related. In type 1, the defect affects the soluble form of the enzyme, is restricted to red blood cells, and causes well-tolerated methemoglobinemia. In type 2, the defect affects both the soluble and microsomal forms of the enzyme and is thus generalized, affecting red cells, leukocytes and all body tissues. Type 2 methemoglobinemia is associated with mental deficiency and other neurologic symptoms. (ECO:0000269|PubMed:10807796, ECO:0000269|PubMed:12393396, ECO:0000269|PubMed:1400360, ECO:0000269|PubMed:15622768, ECO:0000269|PubMed:1707593, ECO:0000269|PubMed:1898726, ECO:0000269|PubMed:7718898, ECO:0000269|PubMed:8119939, ECO:0000269|PubMed:9695975, ECO:0000269|PubMed:9886302). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Methemoglobinemi, Tip 2, 250800, Otozomal Resesif (Kalıtsal Methemoglobinemi) (Prenatal) (CYB5R3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methemoglobinemia, type II, 250800, Autosomal recessive (Hereditary methemoglobinemia) (CYB5R3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYB5R3	CYB5R3, DIA1, BSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methemoglobinemia CYB5R3-related (METHB-CYB5R3) [MIM:250800]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. There are two types of methemoglobinemia CYB5R3-related. In type 1, the defect affects the soluble form of the enzyme, is restricted to red blood cells, and causes well-tolerated methemoglobinemia. In type 2, the defect affects both the soluble and microsomal forms of the enzyme and is thus generalized, affecting red cells, leukocytes and all body tissues. Type 2 methemoglobinemia is associated with mental deficiency and other neurologic symptoms. (ECO:0000269|PubMed:10807796, ECO:0000269|PubMed:12393396, ECO:0000269|PubMed:1400360, ECO:0000269|PubMed:15622768, ECO:0000269|PubMed:1707593, ECO:0000269|PubMed:1898726, ECO:0000269|PubMed:7718898, ECO:0000269|PubMed:8119939, ECO:0000269|PubMed:9695975, ECO:0000269|PubMed:9886302). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Methemoglobinemi, Tip 4, 250790, Otozomal Resesif (Kalıtsal Methemoglobinemi) (CYB5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methemoglobinemia, type IV, 250790, Autosomal recessive (Hereditary methemoglobinemia) (CYB5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYB5A	CYB5A, MCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methemoglobinemia CYB5A-related (METHB-CYB5A) [MIM:250790]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. (ECO:0000269	PubMed:8168836). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Methemoglobinemi, Tip 4, 250790, Otozomal Resesif (Kalıtsal Methemoglobinemi) (Prenatal) (CYB5A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methemoglobinemia, type IV, 250790, Autosomal recessive (Hereditary methemoglobinemia) (CYB5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYB5A	CYB5A, MCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methemoglobinemia CYB5A-related (METHB-CYB5A) [MIM:250790]: A form of methemoglobinemia, a hematologic disease characterized by the presence of excessive amounts of methemoglobin in blood cells, resulting in decreased oxygen carrying capacity of the blood, cyanosis and hypoxia. (ECO:0000269	PubMed:8168836). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Methemoglobinemi, Alfa- (MLPA)	Methemoglobinemias, alpha- (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Methemoglobinemi, Alfa- (Prenatal) (MLPA)	Methemoglobinemias, alpha- (Prenatal) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Methemoglobinemi, Beta- (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methemoglobinemias, beta- (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Methemoglobinemi, Beta- (MLPA)	Methemoglobinemias, beta- (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Methemoglobinemi, Beta- (Prenatal) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methemoglobinemias, beta- (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Methemoglobinemi, Beta- (Prenatal) (MLPA)	Methemoglobinemias, beta- (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metionin Adenosiltransferaz Eksikliği- Metionin Adenosiltransferaz 1 / 3 Eksikliği, Otozomal Resesif, 250850, Otozomal Resesif, Otozomal Dominant (Beyin Demiyelinizasyonu, Metionin Adenosiltransferaz Eksikliğine Bağlı) (MAT1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methionine adenosyltransferase deficiency-METHIONINE ADENOSYLTRANSFERASE I/III DEFICIENCY, autosomal recessive, 250850, Autosomal recessive, Autosomal dominant (Brain demyelination due to methionine adenosyltransferase deficiency) (MAT1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAT1A	MAT1A, MATA1, SAMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methionine adenosyltransferase deficiency (MATD) [MIM:250850]: An inborn error of metabolism resulting in isolated hypermethioninemia. Most patients have no clinical abnormalities, although some neurologic symptoms may be present in rare cases with severe loss of methionine adenosyltransferase activity. (ECO:0000269|PubMed:10677294, ECO:0000269|PubMed:7560086, ECO:0000269|PubMed:8770875, ECO:0000269|PubMed:9042912). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metionin Adenosiltransferaz Eksikliği- Metionin Adenosiltransferaz 1 / 3 Eksikliği, Otozomal Resesif, 250850, Otozomal Resesif, Otozomal Dominant (Beyin Demiyelinizasyonu, Metionin Adenosiltransferaz Eksikliğine Bağlı) (Prenatal) (MAT1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methionine adenosyltransferase deficiency-METHIONINE ADENOSYLTRANSFERASE I/III DEFICIENCY, autosomal recessive, 250850, Autosomal recessive, Autosomal dominant (Brain demyelination due to methionine adenosyltransferase deficiency) (MAT1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAT1A	MAT1A, MATA1, SAMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methionine adenosyltransferase deficiency (MATD) [MIM:250850]: An inborn error of metabolism resulting in isolated hypermethioninemia. Most patients have no clinical abnormalities, although some neurologic symptoms may be present in rare cases with severe loss of methionine adenosyltransferase activity. (ECO:0000269|PubMed:10677294, ECO:0000269|PubMed:7560086, ECO:0000269|PubMed:8770875, ECO:0000269|PubMed:9042912). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonat Semialdehit Dehidrogenaz Eksikliği, 614105, Otozomal Resesif, MMSDHD (Gelişimsel Gecikme, Metilmalonat Semialdehit Dehidrogenaz Eksikliğine Bağlı) (ALDH6A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonate semialdehyde dehydrogenase deficiency, 614105, Autosomal recessive; MMSDHD (Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency) (ALDH6A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH6A1	ALDH6A1, MMSDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylmalonate semialdehyde dehydrogenase deficiency (MMSDHD) [MIM:614105]: A metabolic disorder characterized by elevated beta-alanine, 3-hydroxypropionic acid, and both isomers of 3-amino and 3-hydroxyisobutyric acids in urine organic acids. (ECO:0000269	PubMed:10947204). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metilmalonat Semialdehit Dehidrogenaz Eksikliği, 614105, Otozomal Resesif, MMSDHD (Gelişimsel Gecikme, Metilmalonat Semialdehit Dehidrogenaz Eksikliğine Bağlı) (Prenatal) (ALDH6A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonate semialdehyde dehydrogenase deficiency, 614105, Autosomal recessive; MMSDHD (Developmental delay due to methylmalonate semialdehyde dehydrogenase deficiency) (ALDH6A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH6A1	ALDH6A1, MMSDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylmalonate semialdehyde dehydrogenase deficiency (MMSDHD) [MIM:614105]: A metabolic disorder characterized by elevated beta-alanine, 3-hydroxypropionic acid, and both isomers of 3-amino and 3-hydroxyisobutyric acids in urine organic acids. (ECO:0000269	PubMed:10947204). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, , cblC Tipi, 277400, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (MMACHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria and homocystinuria, cblC type, 277400, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMACHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMACHC	MMACHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, , cblC Tipi, 277400, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (MMACHC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria and homocystinuria, cblC type, 277400, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMACHC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMACHC	MMACHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cbld Tipi, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (MMADHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria and homocystinuria, cblD type, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cbld Tipi, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (MMADHC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria and homocystinuria, cblD type, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cblf Tipi, 277380, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (LMBRD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria and homocystinuria, cblF type, 277380, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (LMBRD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBRD1	LMBRD1, LMBD1, NESI, MAHCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylmalonic aciduria and homocystinuria type cblF (MMAHCF) [MIM:277380]: A disorder of cobalamin metabolism characterized by decreased levels of the coenzymes adenosylcobalamin (AdoCbl) and methylcobalamin (MeCbl). It is due to accumulation of free cobalamin in lysosomes, thus hindering its conversion to cofactors. Clinical features include developmental delay, stomatitis, glossitis, seizures and methylmalonic aciduria responsive to vitamin B12. (ECO:0000269	PubMed:19136951). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cblf Tipi, 277380, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (LMBRD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria and homocystinuria, cblF type, 277380, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (LMBRD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMBRD1	LMBRD1, LMBD1, NESI, MAHCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylmalonic aciduria and homocystinuria type cblF (MMAHCF) [MIM:277380]: A disorder of cobalamin metabolism characterized by decreased levels of the coenzymes adenosylcobalamin (AdoCbl) and methylcobalamin (MeCbl). It is due to accumulation of free cobalamin in lysosomes, thus hindering its conversion to cofactors. Clinical features include developmental delay, stomatitis, glossitis, seizures and methylmalonic aciduria responsive to vitamin B12. (ECO:0000269	PubMed:19136951). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cblj Tipi, 614857, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (ABCD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria and homocystinuria, cblJ type, 614857, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (ABCD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCD4	ABCD4, PXMP1L, P79R, PMP69, MAHCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asidüri ve Homosistinüri, Cblj Tipi, 614857, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (ABCD4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria and homocystinuria, cblJ type, 614857, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (ABCD4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCD4	ABCD4, PXMP1L, P79R, PMP69, MAHCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri, Metilmalonil-CoA Mutaz Eksikliğine Bağlı (B12 Vitaminine Yanıtsız Metilmalonik Asidemi) (MUT geni) (Tüm Gen Dizi Analizi) (Postnatal)	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY (Vitamin B12-unresponsive methylmalonic acidemia) (MUT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUT	MUT, MCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylmalonic aciduria type mut (MMAM) [MIM:251000]: An often fatal disorder of organic acid metabolism. Common clinical features include lethargy, vomiting, failure to thrive, hypotonia, neurological deficit and early death. Two forms of the disease are distinguished by the presence (mut-) or absence (mut0) of residual enzyme activity. Mut0 patients have more severe neurological manifestations of the disease than do MUT- patients. MMAM is unresponsive to vitamin B12 therapy. (ECO:0000269|PubMed:10923046, ECO:0000269|PubMed:11350191, ECO:0000269|PubMed:1346616, ECO:0000269|PubMed:1351030, ECO:0000269|PubMed:15643616, ECO:0000269|PubMed:15781192, ECO:0000269|PubMed:16281286, ECO:0000269|PubMed:1670635, ECO:0000269|PubMed:1977311, ECO:0000269|PubMed:7909321, ECO:0000269|PubMed:7912889, ECO:0000269|PubMed:9285782, ECO:0000269|PubMed:9452100, ECO:0000269|PubMed:9554742). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Metilmalonik Asidüri, Metilmalonil-CoA Mutaz Eksikliğine Bağlı (B12 Vitaminine Yanıtsız Metilmalonik Asidemi) (Prenatal) (MUT geni) (Tüm Gen Dizi Analizi) (Prenatal)	METHYLMALONIC ACIDURIA DUE TO METHYLMALONYL-CoA MUTASE DEFICIENCY (Vitamin B12-unresponsive methylmalonic acidemia) (MUT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MUT	MUT, MCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylmalonic aciduria type mut (MMAM) [MIM:251000]: An often fatal disorder of organic acid metabolism. Common clinical features include lethargy, vomiting, failure to thrive, hypotonia, neurological deficit and early death. Two forms of the disease are distinguished by the presence (mut-) or absence (mut0) of residual enzyme activity. Mut0 patients have more severe neurological manifestations of the disease than do MUT- patients. MMAM is unresponsive to vitamin B12 therapy. {ECO:0000269|PubMed:10923046, ECO:0000269|PubMed:11350191, ECO:0000269|PubMed:1346616, ECO:0000269|PubMed:1351030, ECO:0000269|PubMed:15643616, ECO:0000269|PubMed:15781192, ECO:0000269|PubMed:16281286, ECO:0000269|PubMed:1670635, ECO:0000269|PubMed:1977311, ECO:0000269|PubMed:7909321, ECO:0000269|PubMed:7912889, ECO:0000269|PubMed:9285782, ECO:0000269|PubMed:9452100, ECO:0000269|PubMed:9554742}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri, Cbld Tip, Varyant 2, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (MMADHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria, cblD type, variant 2, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asidüri, Cbld Tip, Varyant 2, 277410, Otozomal Resesif (Metilmalonik Asidemi-Homosistinüri) (Prenatal) (MMADHC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria, cblD type, variant 2, 277410, Autosomal recessive (Methylmalonic acidemia with homocystinuria) (MMADHC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMADHC	C2orf25, MMADHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri, Mut (0) 251000, Otozomal Resesif Yazın (B12 Vitaminine Yanıtsız Metilmalonik Asidemi) (MUT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria, mut(0) type, 251000, Autosomal recessive (Vitamin B12-unresponsive methylmalonic acidemia) (MUT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUT	MUT, MCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylmalonic aciduria type mut (MMAM) [MIM:251000]: An often fatal disorder of organic acid metabolism. Common clinical features include lethargy, vomiting, failure to thrive, hypotonia, neurological deficit and early death. Two forms of the disease are distinguished by the presence (mut-) or absence (mut0) of residual enzyme activity. Mut0 patients have more severe neurological manifestations of the disease than do MUT- patients. MMAM is unresponsive to vitamin B12 therapy. {ECO:0000269|PubMed:10923046, ECO:0000269|PubMed:11350191, ECO:0000269|PubMed:1346616, ECO:0000269|PubMed:1351030, ECO:0000269|PubMed:15643616, ECO:0000269|PubMed:15781192, ECO:0000269|PubMed:16281286, ECO:0000269|PubMed:1670635, ECO:0000269|PubMed:1977311, ECO:0000269|PubMed:7909321, ECO:0000269|PubMed:7912889, ECO:0000269|PubMed:9285782, ECO:0000269|PubMed:9452100, ECO:0000269|PubMed:9554742}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metilmalonik Asidüri, Mut (0) 251000, Otozomal Resesif Yazın (B12 Vitaminine Yanıtsız Metilmalonik Asidemi) (Prenatal) (MUT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria, mut(0) type, 251000, Autosomal recessive (Vitamin B12-unresponsive methylmalonic acidemia) (MUT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MUT	MUT, MCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylmalonic aciduria type mut (MMAM) [MIM:251000]: An often fatal disorder of organic acid metabolism. Common clinical features include lethargy, vomiting, failure to thrive, hypotonia, neurological deficit and early death. Two forms of the disease are distinguished by the presence (mut-) or absence (mut0) of residual enzyme activity. Mut0 patients have more severe neurological manifestations of the disease than do MUT- patients. MMAM is unresponsive to vitamin B12 therapy. {ECO:0000269|PubMed:10923046, ECO:0000269|PubMed:11350191, ECO:0000269|PubMed:1346616, ECO:0000269|PubMed:1351030, ECO:0000269|PubMed:15643616, ECO:0000269|PubMed:15781192, ECO:0000269|PubMed:16281286, ECO:0000269|PubMed:1670635, ECO:0000269|PubMed:1977311, ECO:0000269|PubMed:7909321, ECO:0000269|PubMed:7912889, ECO:0000269|PubMed:9285782, ECO:0000269|PubMed:9452100, ECO:0000269|PubMed:9554742}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asitüri, Geçici, Transkobalamin Reseptör Defektine Bağlı, 613646 (Metilmalonik Asitüri, Transkobalamin Reseptör Defektine Bağlı) (CD320 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (Methylmalonic aciduria due to transcobalamin receptor defect) (CD320 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD320	CD320, 8D6, 8D6A, TCBLR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asitüri, Geçici, Transkobalamin Reseptör Defektine Bağlı, 613646 (Metilmalonik Asitüri, Transkobalamin Reseptör Defektine Bağlı) (Prenatal) (CD320 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria, transient, due to transcobalamin receptor defect, 613646 (Methylmalonic aciduria due to transcobalamin receptor defect) (CD320 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD320	CD320, 8D6, 8D6A, TCBLR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asidüri, Vitamin B12 Yanıtlı, 251100, Otozomal Resesif, cblB (Metilmalonik Asidemi, B12 Vitamini Yanıtlı) (MMAA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria, vitamin B12-responsive, 251100, Autosomal recessive, cblB (vitamin B12-responsive methylmalonic acidemia) (MMAA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMAA	MMAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asidüri, Vitamin B12 Yanıtlı, 251100, Otozomal Resesif, cblB (Metilmalonik Asidemi, B12 Vitamini Yanıtlı) (Prenatal) (MMAA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria, vitamin B12-responsive, 251100, Autosomal recessive, cblB (vitamin B12-responsive methylmalonic acidemia) (MMAA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMAA	MMAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonik Asitüri, Vitamin B12 Yanıtlı, Adenozilkobalamin Sentezindeki Defekte Bağlı, Cblb Komplementasyon Tipi, 251110, Otozomal Resesif (Metilmalonik Asidemi, B12 Vitamini Yanıtlı) (MMAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110, Autosomal recessive (vitamin B12-responsive methylmalonic acidemia) (MMAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMAB	MMAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Metilmalonik Asitüri, Vitamin B12 Yanıtlı, Adenozilkobalamin Sentezindeki Defekte Bağlı, Cblb Komplementasyon Tipi, 251110, Otozomal Resesif (Metilmalonik Asidemi, B12 Vitamini Yanıtlı) (Prenatal) (MMAB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110, Autosomal recessive (vitamin B12-responsive methylmalonic acidemia) (MMAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMAB	MMAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Metilmalonil-CoA Epimeraz Eksikliği, 251120, Otozomal Resesif (Metilmalonik Asidemi, Metilmalonil-CoA Epimerazi Eksikliğine Bağlı) (MCEE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Methylmalonyl-CoA epimerase deficiency, 251120, Autosomal recessive (Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency) (MCEE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCEE	MCEE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylmalonyl-CoA epimerase deficiency (MCEED) [MIM:251120]: Autosomal recessive inborn error of amino acid metabolism, involving valine, threonine, isoleucine and methionine. This organic aciduria may present in the neonatal period with life-threatening metabolic acidosis, hyperammonemia, feeding difficulties, pancytopenia and coma. {ECO:0000269	PubMed:16752391}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Metilmalonil-CoA Epimeraz Eksikliği, 251120, Otozomal Resesif (Metilmalonik Asidemi, Metilmalonil-CoA Epimerazi Eksikliğine Bağlı) (Prenatal) (MCEE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Methylmalonyl-CoA epimerase deficiency, 251120, Autosomal recessive (Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency) (MCEE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCEE	MCEE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Methylmalonyl-CoA epimerase deficiency (MCEED) [MIM:251120]: Autosomal recessive inborn error of amino acid metabolism, involving valine, threonine, isoleucine and methionine. This organic aciduria may present in the neonatal period with life-threatening metabolic acidosis, hyperammonemia, feeding difficulties, pancytopenia and coma. {ECO:0000269	PubMed:16752391}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mevalonik Asidüri, 610377, Otozomal Resesif; MEVA (Mevalonik Asitüri) (MVK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mevalonic aciduria, 610377, Autosomal recessive; MEVA (Mevalonic aciduria) (MVK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MVK	MVK, MVLK, POROK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porokeratosis 3, multiple types (POROK3) [MIM:175900]: A form of porokeratosis, a disorder of faulty keratinization characterized by one or more atrophic patches surrounded by a distinctive hyperkeratotic ridgelike border called the cornoid lamella. The keratotic lesions can progress to overt cutaneous neoplasms, typically squamous cell carcinomas. Multiple clinical variants of porokeratosis are recognized, including porokeratosis of Mibelli, linear porokeratosis, disseminated superficial actinic porokeratosis, palmoplantar porokeratosis, and punctate porokeratosis. Different clinical presentations can be observed among members of the same family. Individuals expressing more than one variant have also been reported. {ECO:0000269|PubMed:22983302, ECO:0000269|PubMed:24781643}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mevalonik Asidüri, 610377, Otozomal Resesif; MEVA (Mevalonik Asitüri) (Prenatal) (MVK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mevalonic aciduria, 610377, Autosomal recessive; MEVA (Mevalonic aciduria) (MVK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MVK	MVK, MVLK, POROK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Porokeratosis 3, multiple types (POROK3) [MIM:175900]: A form of porokeratosis, a disorder of faulty keratinization characterized by one or more atrophic patches surrounded by a distinctive hyperkeratotic ridgelike border called the cornoid lamella. The keratotic lesions can progress to overt cutaneous neoplasms, typically squamous cell carcinomas. Multiple clinical variants of porokeratosis are recognized, including porokeratosis of Mibelli, linear porokeratosis, disseminated superficial actinic porokeratosis, palmoplantar porokeratosis, and punctate porokeratosis. Different clinical presentations can be observed among members of the same family. Individuals expressing more than one variant have also been reported. {ECO:0000269|PubMed:22983302, ECO:0000269|PubMed:24781643}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

MHC Sınıf 2 Eksikliği, Komplementasyon Grubu B, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı) (RFXANK geni) (Tüm Gen Dizi Analizi) (Postnatal)	MHC class II deficiency, complementation group B, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFXANK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RFXANK	RFXANK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. {ECO:0000269|PubMed:10072068, ECO:0000269|PubMed:10725724, ECO:0000269|PubMed:9806546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MHC Sınıf 2 Eksikliği, Komplementasyon Grubu B, 209920, Otozomal Resesif (Immun Yetmezlik, HLA Sınıf 2 Eksikliğine Bağlı) (Prenatal) (RFXANK geni) (Tüm Gen Dizi Analizi) (Prenatal)	MHC class II deficiency, complementation group B, 209920, Autosomal recessive (Immunodeficiency by defective expression of HLA class 2) (RFXANK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFXANK	RFXANK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. {ECO:0000269	PubMed:10072068, ECO:0000269	PubMed:10725724, ECO:0000269	PubMed:9806546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefalik Osteodisplastik Primordial Dwarfizm, Tip I, 210720, Otozomal Resesif; MOPD2 (Mikrosefalik Osteodisplastik Primordial Dwarfizm Tip 2) (PCNT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephalic osteodysplastic primordial dwarfism, type II, 210720, Autosomal recessive; MOPD2 (Microcephalic osteodysplastic primordial dwarfism type II) (PCNT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCNT	PCNT, PCNT2, KEN, SCKL4, MOPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephalic osteodysplastic primordial dwarfism 2 (MOPD2) [MIM:210720]: Adults with this rare inherited condition have an average height of 100 centimeters and a brain size comparable to that of a 3-month-old baby, but are of near-normal intelligence. {ECO:0000269|PubMed:18157127, ECO:0000269|PubMed:18174396}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefalik Osteodisplastik Primordial Dwarfizm, Tip I, 210720, Otozomal Resesif; MOPD2 (Mikrosefalik Osteodisplastik Primordial Dwarfizm Tip 2) (Prenatal) (PCNT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephalic osteodysplastic primordial dwarfism, type II, 210720, Autosomal recessive; MOPD2 (Microcephalic osteodysplastic primordial dwarfism type II) (PCNT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCNT	PCNT, PCNT2, KEN, SCKL4, MOPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephalic osteodysplastic primordial dwarfism 2 (MOPD2) [MIM:210720]: Adults with this rare inherited condition have an average height of 100 centimeters and a brain size comparable to that of a 3-month-old baby, but are of near-normal intelligence. {ECO:0000269	PubMed:18157127, ECO:0000269	PubMed:18174396}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 1, Primer, Otozomal Resesif, 251200, Otozomal Resesif; MCPH1 (Otozomal Resesif Primer Mikrosefali) (MCPH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 1, primary, autosomal recessive, 251200, Autosomal recessive; MCPH1 (Autosomal recessive primary microcephaly) (MCPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCPH1	MCPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 1, primary, autosomal recessive (MCPH1) [MIM:251200]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. Some MCHP1 patients also present growth retardation, short stature, and misregulated chromosome condensation as indicated by a high number of prophase-like cells detected in routine cytogenetic preparations and poor-quality metaphase G-banding. {ECO:0000269|PubMed:12046007, ECO:0000269|PubMed:15199523, ECO:0000269|PubMed:16211557}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 1, Primer, Otozomal Resesif, 251200, Otozomal Resesif; MCPH1 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (MCPH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 1, primary, autosomal recessive, 251200, Autosomal recessive; MCPH1 (Autosomal recessive primary microcephaly) (MCPH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCPH1	MCPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 1, primary, autosomal recessive (MCPH1) [MIM:251200]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. Some MCHP1 patients also present growth retardation, short stature, and misregulated chromosome condensation as indicated by a high number of prophase-like cells detected in routine cytogenetic preparations and poor-quality metaphase G-banding. {ECO:0000269	PubMed:12046007, ECO:0000269	PubMed:15199523, ECO:0000269	PubMed:16211557}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 10, Primer, Otozomal Resesif, 615095, Otozomal Resesif; MCPH10 (Mikrosefalik Primordial Dwarfizm, ZNF335 Eksikliğine Bağlı) (ZNF335 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 10, primary, autosomal recessive, 615095, Autosomal recessive; MCPH10 (Microcephalic primordial dwarfism due to ZNF335 deficiency) (ZNF335 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF335	ZNF335, NIF1, NIF2, MCPH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 10, primary, autosomal recessive (MCPH10) [MIM:615095]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. MCPH10 is characterized by extremely small head size and death usually by 1 year of age. Neuropathologic examination shows severe loss of neurons as well as neuronal loss of polarity and abnormal dendritic maturation. {ECO:0000269	PubMed:23178126}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 10, Primer, Otozomal Resesif, 615095, Otozomal Resesif; MCPH10 (Mikrosefalik Primordial Dwarfizm, ZNF335 Eksikliğine Bağlı) (Prenatal) (ZNF335 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 10, primary, autosomal recessive, 615095, Autosomal recessive; MCPH10 (Microcephalic primordial dwarfism due to ZNF335 deficiency) (ZNF335 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNF335	ZNF335, NIF1, NIF2, MCPH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 10, primary, autosomal recessive (MCPH10) [MIM:615095]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. MCPH10 is characterized by extremely small head size and death usually by 1 year of age. Neuropathologic examination shows severe loss of neurons as well as neuronal loss of polarity and abnormal dendritic maturation. {ECO:0000269	PubMed:23178126}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 11, Primer, Otozomal Resesif, 615414, Otozomal Resesif; MCPH11 (Otozomal Resesif Primer Mikrosefali) (PHC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 11, primary, autosomal recessive, 615414, Autosomal recessive; MCPH11 (Autosomal recessive primary microcephaly) (PHC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHC1	PHC1, EDR1, HPH1, RAE28, MCPH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 11, primary, autosomal recessive (MCPH11) [MIM:615414]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:23418308}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 11, Primer, Otozomal Resesif, 615414, Otozomal Resesif; MCPH11 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (PHC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 11, primary, autosomal recessive, 615414, Autosomal recessive; MCPH11 (Autosomal recessive primary microcephaly) (PHC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHC1	PHC1, EDR1, HPH1, RAE28, MCPH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 11, primary, autosomal recessive (MCPH11) [MIM:615414]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:23418308}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 12, Primer, Otozomal Resesif, 616080, Otozomal Resesif; MCPH12 (Otozomal Resesif Primer Mikrosefali) (CDK6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 12, primary, autosomal recessive, 616080, Autosomal recessive; MCPH12 (Autosomal recessive primary microcephaly) (CDK6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDK6	CDK6, PLSTIRE, MCPH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 12, primary, autosomal recessive (MCPH12) [MIM:616080]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:23918663}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 12, Primer, Otozomal Resesif, 616080, Otozomal Resesif; MCPH12 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CDK6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 12, primary, autosomal recessive, 616080, Autosomal recessive; MCPH12 (Autosomal recessive primary microcephaly) (CDK6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDK6	CDK6, PLSTIRE, MCPH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 12, primary, autosomal recessive (MCPH12) [MIM:616080]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:23918663}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 13, Primer, Otozomal Resesif, 616051, Otozomal Resesif; MCPH13 (Seckel Sendromu) (CENPE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 13, primary, autosomal recessive, 616051, Autosomal recessive; MCPH13 (Seckel syndrome) (CENPE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CENPE	CENPE, MCPH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 13, primary, autosomal recessive (MCPH13) [MIM:616051]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:24748105}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 13, Primer, Otozomal Resesif, 616051, Otozomal Resesif; MCPH13 (Seckel Sendromu) (Prenatal) (CENPE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 13, primary, autosomal recessive, 616051, Autosomal recessive; MCPH13 (Seckel syndrome) (CENPE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CENPE	CENPE, MCPH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 13, primary, autosomal recessive (MCPH13) [MIM:616051]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age- related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:24748105}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 14, Primer, Otozomal Resesif, 616402, Otozomal Resesif; MCPH14 (Otozomal Resesif Primer Mikrosefali) (SASS6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 14, primary, autosomal recessive, 616402, Autosomal recessive; MCPH14 (Autosomal recessive primary microcephaly) (SASS6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SASS6	SASS6, SAS6, MCPH14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 14, primary, autosomal recessive (MCPH14) [MIM:616402]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age, sex and ethnically matched mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:24951542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 14, Primer, Otozomal Resesif, 616402, Otozomal Resesif; MCPH14 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (SASS6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 14, primary, autosomal recessive, 616402, Autosomal recessive; MCPH14 (Autosomal recessive primary microcephaly) (SASS6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SASS6	SASS6, SAS6, MCPH14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Microcephaly 14, primary, autosomal recessive (MCPH14) [MIM:616402]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age, sex and ethnically matched mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269	PubMed:24951542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 15, Primer, Otozomal Resesif, 616486, Otozomal Resesif; MCPH15 (Otozomal Resesif Primer Mikrosefali) (MFSD2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 15, primary, autosomal recessive, 616486, Autosomal recessive; MCPH15 (Autosomal recessive primary microcephaly) (MFSD2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFSD2A	MFSD2A, MCPH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 15, primary, autosomal recessive (MCPH15) [MIM:616486]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age, sex and ethnically matched mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269|PubMed:26005865, ECO:0000269|PubMed:26005868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mikrosefali 15, Primer, Otozomal Resesif, 616486, Otozomal Resesif; MCPH15 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (MFSD2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 15, primary, autosomal recessive, 616486, Autosomal recessive; MCPH15 (Autosomal recessive primary microcephaly) (MFSD2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MFSD2A	MFSD2A, MCPH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 15, primary, autosomal recessive (MCPH15) [MIM:616486]: A form of microcephaly, a disease defined as a head circumference more than 3 standard deviations below the age, sex and ethnically matched mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. {ECO:0000269|PubMed:26005865, ECO:0000269|PubMed:26005868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 16, Primer, Otozomal Resesif, 616681, Otozomal Resesif; MCPH16 (Otozomal Resesif Primer Mikrosefali) (ANKLE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 16, primary, autosomal recessive, 616681, Autosomal recessive; MCPH16 (Autosomal recessive primary microcephaly) (ANKLE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKLE2	ANKLE2, LEM4, KIAA0692, MCPH16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali 16, Primer, Otozomal Resesif, 616681, Otozomal Resesif; MCPH16 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (ANKLE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 16, primary, autosomal recessive, 616681, Autosomal recessive; MCPH16 (Autosomal recessive primary microcephaly) (ANKLE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANKLE2	ANKLE2, LEM4, KIAA0692, MCPH16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 17, Primer, Otozomal Resesif, 617090, Otozomal Resesif; MCPH17 (Otozomal Resesif Primer Mikrosefali) (CIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 17, primary, autosomal recessive, 617090, Autosomal recessive; MCPH17 (Autosomal recessive primary microcephaly) (CIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIT	CIT, STK21, CRIK, MCPH17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali 17, Primer, Otozomal Resesif, 617090, Otozomal Resesif; MCPH17 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CIT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 17, primary, autosomal recessive, 617090, Autosomal recessive; MCPH17 (Autosomal recessive primary microcephaly) (CIT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CIT	CIT, STK21, CRIK, MCPH17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 2, Primer, Otozomal Resesif, ± Kortikal Malformasyon, 604317, Otozomal Resesif; MCPH2 (Otozomal Resesif Primer Mikrosefali) (WDR62 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317, Autosomal recessive; MCPH2 (Autosomal recessive primary microcephaly) (WDR62 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR62	WDR62, C19orf14, MCPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali 2, Primer, Otozomal Resesif, ± Kortikal Malformasyon, 604317, Otozomal Resesif; MCPH2 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (WDR62 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 2, primary, autosomal recessive, with or without cortical malformations, 604317, Autosomal recessive; MCPH2 (Autosomal recessive primary microcephaly) (WDR62 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR62	WDR62, C19orf14, MCPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 3, Primer, Otozomal Resesif, 604804, Otozomal Resesif; MCPH3 (Otozomal Resesif Primer Mikrosefali) (CDK5RAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 3, primary, autosomal recessive, 604804, Autosomal recessive; MCPH3 (Autosomal recessive primary microcephaly) (CDK5RAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDK5RAP2	CDK5RAP2, KIAA1633, MCPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 3, primary, autosomal recessive (MCPH3) [MIM:604804]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269	PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 3, Primer, Otozomal Resesif, 604804, Otozomal Resesif; MCPH3 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CDK5RAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 3, primary, autosomal recessive, 604804, Autosomal recessive; MCPH3 (Autosomal recessive primary microcephaly) (CDK5RAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDK5RAP2	CDK5RAP2, KIAA1633, MCPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 3, primary, autosomal recessive (MCPH3) [MIM:604804]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269	PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 4, Primer, Otozomal Resesif, 604321, Otozomal Resesif; MCPH4 (Otozomal Resesif Primer Mikrosefali) (KNL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 4, primary, autosomal recessive, 604321, Autosomal recessive; MCPH4 (Autosomal recessive primary microcephaly) (KNL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KNL1	CASC5, AF15Q14, KIAA1570, D40, MCPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali 4, Primer, Otozomal Resesif, 604321, Otozomal Resesif; MCPH4 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (KNL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 4, primary, autosomal recessive, 604321, Autosomal recessive; MCPH4 (Autosomal recessive primary microcephaly) (KNL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KNL1	CASC5, AF15Q14, KIAA1570, D40, MCPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 5, Primer, Otozomal Resesif, 608716, Otozomal Resesif; MCPH5 (Otozomal Resesif Primer Mikrosefali) (ASPM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 5, primary, autosomal recessive, 608716, Autosomal recessive; MCPH5 (Autosomal recessive primary microcephaly) (ASPM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPM	ASPM, MCPH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 5, primary, autosomal recessive (MCPH5) [MIM:608716]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:12355089, ECO:0000269|PubMed:14574646, ECO:0000269|PubMed:16673149, ECO:0000269|PubMed:22989186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 5, Primer, Otozomal Resesif, 608716, Otozomal Resesif; MCPH5 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (ASPM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 5, primary, autosomal recessive, 608716, Autosomal recessive; MCPH5 (Autosomal recessive primary microcephaly) (ASPM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASPM	ASPM, MCPH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 5, primary, autosomal recessive (MCPH5) [MIM:608716]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:12355089, ECO:0000269|PubMed:14574646, ECO:0000269|PubMed:16673149, ECO:0000269|PubMed:22989186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 6, Primer, Otozomal Resesif, 608393, Otozomal Resesif; MCPH6 (Otozomal Resesif Primer Mikrosefali) (CENPJ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 6, primary, autosomal recessive, 608393, Autosomal recessive; MCPH6 (Autosomal recessive primary microcephaly) (CENPJ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CENPJ	CENPJ, CPAP, MCPH6, SCKL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 6, primary, autosomal recessive (MCPH6) [MIM:608393]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269	PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali 6, Primer, Otozomal Resesif, 608393, Otozomal Resesif; MCPH6 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CENPJ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 6, primary, autosomal recessive, 608393, Autosomal recessive; MCPH6 (Autosomal recessive primary microcephaly) (CENPJ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CENPJ	CENPJ, CPAP, MCPH6, SCKL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 6, primary, autosomal recessive (MCPH6) [MIM:608393]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269	PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 7, Primer, Otozomal Resesif, 612703, Otozomal Resesif; MCPH7 (Otozomal Resesif Primer Mikrosefali) (STIL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 7, primary, autosomal recessive, 612703, Autosomal recessive; MCPH7 (Autosomal recessive primary microcephaly) (STIL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STIL	STIL, SIL, MCPH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 7, primary, autosomal recessive (MCPH7) [MIM:612703]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:19215732, ECO:0000269|PubMed:22989186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali 7, Primer, Otozomal Resesif, 612703, Otozomal Resesif; MCPH7 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (STIL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 7, primary, autosomal recessive, 612703, Autosomal recessive; MCPH7 (Autosomal recessive primary microcephaly) (STIL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STIL	STIL, SIL, MCPH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 7, primary, autosomal recessive (MCPH7) [MIM:612703]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:19215732, ECO:0000269|PubMed:22989186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mikrosefali 8, Primer, Otozomal Resesif, 614673, Otozomal Resesif; MCPH8 (Otozomal Resesif Primer Mikrosefali) (CEP135 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 8, primary, autosomal recessive, 614673, Autosomal recessive; MCPH8 (Autosomal recessive primary microcephaly) (CEP135 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP135	CEP135, KIAA0635, MCPH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 8, primary, autosomal recessive (MCPH8) [MIM:614673]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:22521416}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali 8, Primer, Otozomal Resesif, 614673, Otozomal Resesif; MCPH8 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CEP135 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 8, primary, autosomal recessive, 614673, Autosomal recessive; MCPH8 (Autosomal recessive primary microcephaly) (CEP135 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP135	CEP135, KIAA0635, MCPH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 8, primary, autosomal recessive (MCPH8) [MIM:614673]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:22521416}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali 9, Primer, Otozomal Resesif, 614852, Otozomal Resesif; MCPH9 (Otozomal Resesif Primer Mikrosefali) (CEP152 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly 9, primary, autosomal recessive, 614852, Autosomal recessive; MCPH9 (Autosomal recessive primary microcephaly) (CEP152 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP152	CEP152, KIAA0912, MCPH9, SCKL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seckel syndrome 5 (SCKL5) [MIM:613823]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21131973}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali 9, Primer, Otozomal Resesif, 614852, Otozomal Resesif; MCPH9 (Otozomal Resesif Primer Mikrosefali) (Prenatal) (CEP152 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly 9, primary, autosomal recessive, 614852, Autosomal recessive; MCPH9 (Autosomal recessive primary microcephaly) (CEP152 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP152	CEP152, KIAA0912, MCPH9, SCKL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 5 (SCKL5) [MIM:613823]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21131973}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 1, 251270, Otozomal Resesif; MCCRP1 (Otozomal Resesif Koriyoretinopati-Mikrosefali Sendromu) (TUBGCP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270, Autosomal recessive; MCCRP1 (Autosomal recessive chorioretinopathy-microcephaly syndrome) (TUBGCP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBGCP6	TUBGCP6, GCP6, KIAA1669, MCCRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly and chorioretinopathy, autosomal recessive, 1 (MCCRP1) [MIM:251270]: A syndrome characterized by microcephaly, cognitive impairment, underdeveloped retina and choroid, and epilepsy in some patients. The more anterior parts of the retina, near the periphery and pars plana, have a grayish hue and diminutive vasculature similar to retinopathy of prematurity. Visual impairment becomes evident during the first year of life. {ECO:0000269|PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 1, 251270, Otozomal Resesif; MCCRP1 (Otozomal Resesif Koriyoretinopati-Mikrosefali Sendromu) (Prenatal) (TUBGCP6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly and chorioretinopathy, autosomal recessive, 1, 251270, Autosomal recessive; MCCRP1 (Autosomal recessive chorioretinopathy-microcephaly syndrome) (TUBGCP6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBGCP6	TUBGCP6, GCP6, KIAA1669, MCCRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly and chorioretinopathy, autosomal recessive, 1 (MCCRP1) [MIM:251270]: A syndrome characterized by microcephaly, cognitive impairment, underdeveloped retina and choroid, and epilepsy in some patients. The more anterior parts of the retina, near the periphery and pars plana, have a grayish hue and diminutive vasculature similar to retinopathy of prematurity. Visual impairment becomes evident during the first year of life. {ECO:0000269|PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 2, 616171, Otozomal Resesif; MCCRP2 (Seckel Sendromu) (PLK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171, Autosomal recessive; MCCRP2 (Seckel syndrome) (PLK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLK4	PLK4, STK18, SAK, MCCRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly and chorioretinopathy, autosomal recessive, 2 (MCCRP2) [MIM:616171]: A severe disorder characterized by microcephaly, delayed psychomotor development, growth retardation with dwarfism, and ocular abnormalities. {ECO:0000269|PubMed:25344692}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 2, 616171, Otozomal Resesif; MCCRP2 (Seckel Sendromu) (PLK4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly and chorioretinopathy, autosomal recessive, 2, 616171, Autosomal recessive; MCCRP2 (Seckel syndrome) (PLK4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLK4	PLK4, STK18, SAK, MCCRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly and chorioretinopathy, autosomal recessive, 2 (MCCRP2) [MIM:616171]: A severe disorder characterized by microcephaly, delayed psychomotor development, growth retardation with dwarfism, and ocular abnormalities. {ECO:0000269|PubMed:25344692}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 3, 616335, Otozomal Resesif; MCCRP3 (Otozomal Resesif Koriyoretinopati-Mikrosefali Sendromu) (TUBGCP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335, Autosomal recessive; MCCRP3 (Autosomal recessive chorioretinopathy-microcephaly syndrome) (TUBGCP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBGCP4	TUBGCP4, GCP4, MCCRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali ve Korioretinopati, Otozomal Resesif, 3, 616335, Otozomal Resesif; MCCRP3 (Otozomal Resesif Koriyoretinopati-Mikrosefali Sendromu) (Prenatal) (TUBGCP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly and chorioretinopathy, autosomal recessive, 3, 616335, Autosomal recessive; MCCRP3 (Autosomal recessive chorioretinopathy-microcephaly syndrome) (TUBGCP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBGCP4	TUBGCP4, GCP4, MCCRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, ± Korioretinopati, Lenfödem Veyamental Retardasyon, 152950, Otozomal Dominant; MCLMR (Mikrosefali Lenfödem-Korioretinopati Sendromu) (KIF11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950, Autosomal dominant; MCLMR (Microcephaly-lymphedema-chorioretinopathy syndrome) (KIF11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF11	KIF11, KNSL1, MCLMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation (MCLMR) [MIM:152950]: An autosomal dominant disorder that involves an overlapping but variable spectrum of central nervous system and ocular developmental anomalies. Microcephaly ranges from mild to severe and is often associated with mild to moderate developmental delay and a characteristic facial phenotype with upslanting palpebral fissures, broad nose with rounded tip, long philtrum with thin upper lip, prominent chin, and prominent ears. Chorioretinopathy is the most common eye abnormality, but retinal folds, microphthalmia, and myopic and hypermetropic astigmatism have also been reported, and some individuals have no overt ocular phenotype. Congenital lymphedema, when present, is typically confined to the dorsa of the feet, and lymphoscintigraphy reveals the absence of radioactive isotope uptake from the webspaces between the toes. {ECO:0000269|PubMed:22284827}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali, ± Korioretinopati, Lenfödem Veyamental Retardasyon, 152950, Otozomal Dominant; MCLMR (Mikrosefali Lenfödem-Korioretinopati Sendromu) (Prenatal) (KIF11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation, 152950, Autosomal dominant; MCLMR (Microcephaly-lymphedema-chorioretinopathy syndrome) (KIF11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF11	KIF11, KNSL1, MCLMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly with or without chorioretinopathy, lymphedema, or mental retardation (MCLMR) [MIM:152950]: An autosomal dominant disorder that involves an overlapping but variable spectrum of central nervous system and ocular developmental anomalies. Microcephaly ranges from mild to severe and is often associated with mild to moderate developmental delay and a characteristic facial phenotype with upslanting palpebral fissures, broad nose with rounded tip, long philtrum with thin upper lip, prominent chin, and prominent ears. Chorioretinopathy is the most common eye abnormality, but retinal folds, microphthalmia, and myopic and hypermetropic astigmatism have also been reported, and some individuals have no overt ocular phenotype. Congenital lymphedema, when present, is typically confined to the dorsa of the feet, and lymphoscintigraphy reveals the absence of radioactive isotope uptake from the webspaces between the toes. {ECO:0000269|PubMed:22284827}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali-Kapiller Malformasyon Sendromu, 614261, Otozomal Resesif; MICCAP (Mikrosefali Kapiller Malformasyon Sendromu) (STAMBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly-capillary malformation syndrome, 614261, Autosomal recessive; MICCAP (Microcephaly-capillary malformation syndrome) (STAMBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAMBP	STAMBP, AMSH, MICCAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly-capillary malformation syndrome (MICCAP) [MIM:614261]: A congenital disorder characterized by severe progressive microcephaly, early-onset refractory epilepsy, profound developmental delay, and multiple small capillary malformations spread diffusely on the body. Additional more variable features include dysmorphic facial features, distal limb abnormalities, and mild heart defects. {ECO:0000269|PubMed:23542699}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali-Kapiller Malformasyon Sendromu, 614261, Otozomal Resesif; MICCAP (Mikrosefali Kapiller Malformasyon Sendromu) (Prenatal) (STAMBP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly-capillary malformation syndrome, 614261, Autosomal recessive; MICCAP (Microcephaly-capillary malformation syndrome) (STAMBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STAMBP	STAMBP, AMSH, MICCAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly-capillary malformation syndrome (MICCAP) [MIM:614261]: A congenital disorder characterized by severe progressive microcephaly, early-onset refractory epilepsy, profound developmental delay, and multiple small capillary malformations spread diffusely on the body. Additional more variable features include dysmorphic facial features, distal limb abnormalities, and mild heart defects. {ECO:0000269|PubMed:23542699}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Amish Tipi, 607196, Otozomal Resesif; MCPHA (Amish Letal Mikrosefali) (SLC25A19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, Amish type, 607196, Autosomal recessive; MCPHA (Amish lethal microcephaly) (SLC25A19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A19	SLC25A19, DNC, MUP1, MCPHA, THMD3, THMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thiamine metabolism dysfunction syndrome 4, bilateral striatal degeneration and progressive polyneuropathy type (THMD4) [MIM:613710]: A disease characterized by recurrent episodes of flaccid paralysis and encephalopathy associated with bilateral striatal necrosis and chronic progressive polyneuropathy. {ECO:0000269|PubMed:19798730}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali, Amish Tipi, 607196, Otozomal Resesif; MCPHA (Amish Letal Mikrosefali) (Prenatal) (SLC25A19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, Amish type, 607196, Autosomal recessive; MCPHA (Amish lethal microcephaly) (SLC25A19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A19	SLC25A19, DNC, MUP1, MCPHA, THMD3, THMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Thiamine metabolism dysfunction syndrome 4, bilateral striatal degeneration and progressive polyneuropathy type (THMD4) [MIM:613710]: A disease characterized by recurrent episodes of flaccid paralysis and encephalopathy associated with bilateral striatal necrosis and chronic progressive polyneuropathy. {ECO:0000269|PubMed:19798730}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Konjenital Katarakt ve Psoriaziform Dermatit, 616834, Otozomal Resesif; MCCPD (MSMO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834, Autosomal recessive; MCCPD (MSMO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSMO1	MSMO1, SC4MOL, ERG25, MCCPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali, Konjenital Katarakt ve Psoriaziform Dermatit, 616834, Otozomal Resesif; MCCPD (Prenatal) (MSMO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, congenital cataract, and psoriasiform dermatitis, 616834, Autosomal recessive; MCCPD (MSMO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSMO1	MSMO1, SC4MOL, ERG25, MCCPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Epilepsi, Diyabet Sendromu, 614231, Otozomal Resesif; MEDS (Primer Mikrosefali-Epilepsi-Kalıcı Neonatal Diyabet Sendromu) (IER3IP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, epilepsy, and diabetes syndrome, 614231, Autosomal Resesif; MEDS (Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome) (ER3IP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IER3IP1	IER3IP1, MEDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Mikrosefali, Epilepsi, Diyabet Sendromu, 614231, Otozomal Resesif; MEDS (Primer Mikrosefali-Epilepsi-Kalıcı Neonatal Diyabet Sendromu) (Prenatal) (IER3IP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, epilepsy, and diabetes syndrome, 614231, Autosomal recessive; MEDS (Primary microcephaly-epilepsy-permanent neonatal diabetes syndrome) (IER3IP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IER3IP1	IER3IP1, MEDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Postnatal, Progresif, Nöbetler ve Beyin Atrofisi, 613668, Otozomal Resesif (Infantil Serebral ve Serebellar Atrofi-Postnatal Progresif Mikrosefali) (MED17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668, Autosomal recessive (Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly) (MED17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED17	MED17, CRSP6, CRSP77, TRAP80, DRIP80	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali, Postnatal, Progresif, Nöbetler ve Beyin Atrofisi, 613668, Otozomal Resesif (Infantil Serebral ve Serebellar Atrofi-Postnatal Progresif Mikrosefali) (MED17 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, postnatal progressive, with seizures and brain atrophy, 613668, Autosomal recessive (Infantile cerebral and cerebellar atrophy with postnatal progressive microcephaly) (MED17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED17	MED17, CRSP6, CRSP77, TRAP80, DRIP80	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Progresif, Nöbetler ve Serebral ve Beyin Atrofisi, 615760, Otozomal Resesif; MSCCA (Diffüz Serebral ve Serebellar Atrofi-Direngli Nöbetler-Progresif Mikrosefali Sendromu) (QARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760, Autosomal recessive; MSCCA (Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome) (QARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	QARS	QARS, GLNRS, MSCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali, Progresif, Nöbetler ve Serebral ve Beyin Atrofisi, 615760, Otozomal Resesif; MSCCA (Diffüz Serebral ve Serebellar Atrofi-Direngli Nöbetler-Progresif Mikrosefali Sendromu) (Prenatal) (QARS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, progressive, seizures, and cerebral and cerebellar atrophy, 615760, Autosomal recessive; MSCCA (Diffuse cerebral and cerebellar atrophy-intractable seizures-progressive microcephaly syndrome) (QARS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	QARS	QARS, GLNRS, MSCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Nöbetler, ve Gelişimsel Gecikme, 613402, Otozomal Resesif; MCSZ (Erken İnfantil Epileptik Ensefalopati) (PNKP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, seizures, and developmental delay, 613402, Autosomal recessive; MCSZ (Early infantile epileptic encephalopathy) (PNKP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNKP	PNKP, PNK, MCSZ, EIEE10, MCSZ, AOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly, seizures, and developmental delay (MCSZ) [MIM:613402]: A disease characterized by infantile-onset seizures, microcephaly, severe intellectual disability and delayed motor milestones with absent speech or only achieving a few words. Most patients also have behavioral problems with hyperactivity. Microcephaly is progressive and without neuronal migration or structural abnormalities, consistent with primary microcephaly. {ECO:0000269	PubMed:20189933}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali, Nöbetler, ve Gelişimsel Gecikme, 613402, Otozomal Resesif; MCSZ (Erken İnfantil Epileptik Ensefalopati) (PNKP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, seizures, and developmental delay, 613402, Autosomal recessive; MCSZ (Early infantile epileptic encephalopathy) (PNKP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNKP	PNKP, PNK, MCSZ, EIEE10, MCSZ, AOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly, seizures, and developmental delay (MCSZ) [MIM:613402]: A disease characterized by infantile-onset seizures, microcephaly, severe intellectual disability and delayed motor milestones with absent speech or only achieving a few words. Most patients also have behavioral problems with hyperactivity. Microcephaly is progressive and without neuronal migration or structural abnormalities, consistent with primary microcephaly. {ECO:0000269	PubMed:20189933}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Kısa Boy, ve Bozulmuş Glikoz Metabolizması 1, 616033, Otozomal Resesif; MSSGM1 (Primer Mikrosefali-Hafif Mental Retardasyon-Geniş-Başlangıçlı Diyabet Sendromu) (TRMT10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, short stature, and impaired glucose metabolism 1, 616033, Autosomal recessive; MSSGM1 (Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome) (TRMT10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRMT10A	TRMT10A, RG9MTD2, MSSGM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali, Kısa Boy, ve Bozulmuş Glikoz Metabolizması 1, 616033, Otozomal Resesif; MSSGM1 (Primer Mikrosefali-Hafif Mental Retardasyon-Geniş-Başlangıçlı Diyabet Sendromu) (Prenatal) (TRMT10A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, short stature, and impaired glucose metabolism 1, 616033, Autosomal recessive; MSSGM1 (Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome) (TRMT10A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRMT10A	TRMT10A, RG9MTD2, MSSGM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Kısa Boy, ve Bozulmuş Glikoz Metabolizması 2, 616817, Otozomal Resesif; MSSGM2 (Primer Mikrosefali-Hafif Mental Retardasyon-Geniş-Başlangıçlı Diyabet Sendromu) (PPP1R15B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, short stature, and impaired glucose metabolism 2, 616817, Autosomal recessive; MSSGM2 (Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome) (PPP1R15B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP1R15B	PPP1R15B, CREP, MSSGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosefali, Kısa Boy, ve Bozulmuş Glikoz Metabolizması 2, 616817, Otozomal Resesif; MSSGM2 (Primer Mikrosefali-Hafif Mental Retardasyon-Geniş-Başlangıçlı Diyabet Sendromu) (Prenatal) (PPP1R15B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, short stature, and impaired glucose metabolism 2, 616817, Autosomal recessive; MSSGM2 (Primary microcephaly-mild intellectual disability-young-onset diabetes syndrome) (PPP1R15B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPP1R15B	PPP1R15B, CREP, MSSGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrosefali, Kısa Boy, Polimikrogiri ve Nöbet, 614833, Otozomal Resesif; MSSP (Mikrosefalik Primordial Dwarfizm, RTTN Eksikliğine Bağlı) (RTTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcephaly, short stature, and polymicrogyria with seizures, 614833, Autosomal recessive; MSSP (Microcephalic primordial dwarfism due to RTTN deficiency) (RTTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTTN	RTTN, MSSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polymicrogyria with seizures (PMGYS) [MIM:614833]: A disease characterized by many irregular small gyri in the brain surface and fusion of the molecular layer over multiple small gyri, which gives a festooned appearance to the cortical surface, without abnormal neuronal migration. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. PMGYS patients have moderate to severe mental retardation, poor speech, dysarthria and seizures. {ECO:0000269	PubMed:22939636}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrosefali, Kısa Boy, Polimikrogiri ve Nöbet, 614833, Otozomal Resesif; MSSP (Mikrosefalik Primordial Dwarfizm, RTTN Eksikliğine Bağlı) (Prenatal) (RTTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microcephaly, short stature, and polymicrogyria with seizures, 614833, Autosomal recessive; MSSP (Microcephalic primordial dwarfism due to RTTN deficiency) (RTTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RTTN	RTTN, MSSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polymicrogyria with seizures (PMGYS) [MIM:614833]: A disease characterized by many irregular small gyri in the brain surface and fusion of the molecular layer over multiple small gyri, which gives a festooned appearance to the cortical surface, without abnormal neuronal migration. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. PMGYS patients have moderate to severe mental retardation, poor speech, dysarthria and seizures. {ECO:0000269	PubMed:22939636}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrokori, Konjenital, 156600, Otozomal Dominant (Konjenital Mikrokori) (22000)	Microcoria, congenital, 156600, Autosomal dominant (Congenital microcoria) (Prenatal)	.	MCOR, C13DEL q32, DEL13q32	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Mikrokori, Konjenital, 156600, Otozomal Dominant (Konjenital Mikrokori) (Prenatal)	Microcoria, congenital, 156600, Autosomal dominant (Congenital microcoria) (Prenatal)	.	MCOR, C13DEL q32, DEL13q32	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikrokornea, Miyopi Korioretinal Atrofi ve Telekantüsü 615458, Otozomal Resesif; MMCAT (Mikrokornea-Miyopi Koryoretinal Atrofi-Telekantus Sendromu) (ADAMTS18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcornea, myopic chorioretinal atrophy, and telecanthus, 615458, Autosomal recessive; MMCAT (Microcornea-myopic chorioretinal atrophy-telecanthus syndrome) (ADAMTS18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTS18	ADAMTS18, MMCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcornea, myopic chorioretinal atrophy, and telecanthus (MMCAT) [MIM:615458]: A ocular syndrome characterized by microcornea and myopic chorioretinal atrophy. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. In addition to ocular findings, some patients have telecanthus and posteriorly rotated ears. {ECO:0000269	PubMed:23818446}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikrokornea, Rod-Kon Distrofisi, Kataralt ve Posterior Stafilom, 193220, Otozomal Dominant; RP50 (MRCS Sendromu) (BEST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microcornea, rod-cone dystrophy, cataract, and posterior staphyloma, 193220, Autosomal dominant; RP50 (MRCS syndrome) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy, vitelliform, 2 (VMD2) [MIM:153700]: An autosomal dominant form of macular degeneration that usually begins in childhood or adolescence. VMD2 is characterized by typical 'egg-yolk' macular lesions due to abnormal accumulation of lipofuscin within and beneath the retinal pigment epithelium cells. Progression of the disease leads to destruction of the retinal pigment epithelium and vision loss. {ECO:0000269|PubMed:10331951, ECO:0000269|PubMed:10394929, ECO:0000269|PubMed:10453731, ECO:0000269|PubMed:10682987, ECO:0000269|PubMed:10798642, ECO:0000269|PubMed:11241846, ECO:0000269|PubMed:11449320, ECO:0000269|PubMed:12187431, ECO:0000269|PubMed:12324875, ECO:0000269|PubMed:13129869, ECO:0000269|PubMed:14517959, ECO:0000269|PubMed:15176385, ECO:0000269|PubMed:18766995, ECO:0000269|PubMed:19357557, ECO:0000269|PubMed:9662395, ECO:0000269|PubMed:9700209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrohidransefali, 605013, Otozomal Resesif; MHAC (NDE1 İle İlişkili Mikrohidranensefali) (NDE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microhydranencephaly, 605013, Autosomal recessive; MHAC (NDE1-related microhydranencephaly) (NDE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDE1	NDE1, NUDE, LIS4, MHAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microhydranencephaly (MHAC) [MIM:605013]: A severe neurodevelopmental disorder characterized by microcephaly, severe motor and mental retardation, spasticity, and brain malformations that include gross dilation of the ventricles with complete absence of the cerebral hemispheres or severe delay in their development. {ECO:0000269|PubMed:22576350}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi-Kolobom 3, 610092 İle Mikroftalmi (Kolobomatöz Mikroftalmi) (VSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia with coloboma 3, 610092 (Colobomatous microphthalmia) (VSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VSX2	CHX10, HOX10, MCOP2, MCOPCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia with cataracts and iris abnormalities (MCOPCTI) [MIM:610092]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, cataract and other abnormalities like cataract may also be present. {ECO:0000269|PubMed:10932181}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi-Kolobom 3, 610092 İle Mikroftalmi (Kolobomatöz Mikroftalmi) (Prenatal) (VSX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia with coloboma 3, 610092 (Colobomatous microphthalmia) (VSX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VSX2	CHX10, HOX10, MCOP2, MCOPCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microphthalmia with cataracts and iris abnormalities (MCOPCTI) [MIM:610092]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, cataract and other abnormalities like cataract may also be present. {ECO:0000269|PubMed:10932181}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mikroftalmi-Kolobom 5, 611638, Otozomal Dominant; MCOPCB5 (Kolobomatöz Mikroftalmi) (SHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia with coloboma 5, 611638, Autosomal dominant; MCOPCB5 (Coloboma tous microphthalmia) (SHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Mikroftalmi-Kolobom 5, 611638, Otozomal Dominant; MCOPCB5 (Kolobomatöz Mikroftalmi) (MLPA)	Microphthalmia with coloboma 5, 611638, Autosomal dominant; MCOPCB5 (Coloboma tous microphthalmia) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Mikroftalmi-Kolobom 5, 611638, Otozomal Dominant; MCOPCB5 (Kolobomatöz Mikroftalmi) (Prenatal) (SHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia with coloboma 5, 611638, Autosomal dominant; MCOPCB5 (Coloboma tous microphthalmia) (SHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi-Kolobom 5, 611638, Otozomal Dominant; MCOPCB5 (Kolobomatöz Mikroftalmi) (MLPA)	Microphthalmia with coloboma 5, 611638, Autosomal dominant; MCOPCB5 (Coloboma tous microphthalmia) (Prenatal) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi-Kolobom 6, 613703 (Kolobomatöz Mikroftalmi) (GDF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia with coloboma 6, 613703 (Coloboma tous microphthalmia) (GDF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF3	GDF3, KFS3, MCOPCB6, MCOP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 3, autosomal dominant (KFS3) [MIM:613702]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. (ECO:0000269	PubMed:19864492). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi-Kolobom 6, 613703 (Kolobomatöz Mikroftalmi) (Prenatal) (GDF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia with coloboma 6, 613703 (Coloboma tous microphthalmia) (GDF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF3	GDF3, KFS3, MCOPCB6, MCOP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Klippel-Feil syndrome 3, autosomal dominant (KFS3) [MIM:613702]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. (ECO:0000269	PubMed:19864492). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi-Kolobom 6, Dijenik, 613703 (Kolobomatöz Mikroftalmi) (GDF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia with coloboma 6, digenic, 613703 (Coloboma tous microphthalmia) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. (ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi-Kolobom 6, Dijenik, 613703 (Kolobomatöz Mikroftalmi) (Prenatal) (GDF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia with coloboma 6, digenic, 613703 (Coloboma tous microphthalmia) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. (ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi, Ekstremite Anomalileri, 206920, Otozomal Resesif; MLA (Mikroftalmi-Ekstremite Anomalisi) (SMOC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia with limb anomalies, 206920, Autosomal recessive; MLA (Microphthalmia with limb anomalies) (SMOC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMOC1	SMOC1, OAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ophthalmoacromelic syndrome (OAS) [MIM:206920]: A rare disorder presenting with ocular anomalies, ranging from mild microphthalmia to true anophthalmia, and limb anomalies. Limb malformations include fused 4th and 5th metacarpals and short 5th finger in hands, and oligodactyly in foot (four toes). Most patients have bilateral anophthalmia/ microphthalmia, but unilateral abnormality is also noted. Other malformations are rare, but venous or vertebral anomaly was recognized each in single cases. (ECO:0000269|PubMed:21194678, ECO:0000269|PubMed:21194680, ECO:0000269|PubMed:21750680, ECO:0000269|PubMed:23646827). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Ekstremite Anomalileri, 206920, Otozomal Resesif; MLA (Mikroftalmi-Ekstremite Anomalisi) (Prenatal) (SMOC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia with limb anomalies, 206920, Autosomal recessive; MLA (Microphthalmia with limb anomalies) (SMOC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMOC1	SMOC1, OAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ophthalmoacromelic syndrome (OAS) [MIM:206920]: A rare disorder presenting with ocular anomalies, ranging from mild microphthalmia to true anophthalmia, and limb anomalies. Limb malformations include fused 4th and 5th metacarpals and short 5th finger in hands, and oligodactyly in foot (four toes). Most patients have bilateral anophthalmia/ microphthalmia, but unilateral abnormality is also noted. Other malformations are rare, but venous or vertebral anomaly was recognized each in single cases. (ECO:0000269|PubMed:21194678, ECO:0000269|PubMed:21194680, ECO:0000269|PubMed:21750680, ECO:0000269|PubMed:23646827). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi, İzole 2, 610093; MCOP2 (İzole Anoftalmi-Mikroftalmi Sendromu) (VSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 2, 610093; MCOP2 (Isolated anophthalmia-microphthalmia syndrome) (VSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VSX2	CHX10, HOX10, MCOP2, MCOPCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia with cataracts and iris abnormalities (MCOPCTI) [MIM:610092]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scarring of the retina and choroid, cataract and other abnormalities like cataract may also be present. (ECO:0000269	PubMed:10932181). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, İzole 3, 611038, Otozomal Resesif; MCOP3 (İzole Anoftalmi-Mikroftalmi Sendromu) (RAX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 3, 611038, Autosomal recessive; MCOP3 (Isolated anophthalmia-microphthalmia syndrome) (RAX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAX	RAX, RX, MCOP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mikroftalmi, İzole 4, 613094; MCOP4 (İzole Anoftalmi-Mikroftalmi Sendromu) (GDF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 4, 613094; MCOP4 (Isolated anophthalmia-microphthalmia syndrome) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. (ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, İzole 5, 611040, Otozomal Resesif; MCOP5 (Mikroftalmi-Retinitis Pigmentoza-Foveoşizis-Optik Disk Drusenli Sendromu) (MFRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 5, 611040, Autosomal recessive; MCOP5 (Microphthalmia-retinitis pigmentosa-foveoschisis-optic disc drusen syndrome) (MFRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFRP	MFRP, MCOP5, NNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nanophthalmos 2 (NNO2) [MIM:609549]: Rare autosomal recessive disorder of eye development characterized by extreme hyperopia and small functional eyes. (ECO:0000269	PubMed:15976036). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, İzole 6, 613517, Otozomal Resesif; MCOP6 (İzole Anoftalmi-Mikroftalmi Sendromu) (PRSS56 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 6, 613517, Autosomal recessive; MCOP6 (Isolated anophthalmia-microphthalmia syndrome) (PRSS56 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRSS56	PRSS56, MCOP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, isolated, 6 (MCOP6) [MIM:613517]: A developmental ocular disorder characterized by small malformed eyes. Clinical features are extreme hyperopia due to short axial length with essentially normal anterior segment, steep corneal curvatures, shallow anterior chamber, thick lenses, and thickened scleral wall. Palpebral fissures appear narrow because of relatively deep-set eyes, visual acuity is mildly to moderately reduced, and anisometropic or strabismic amblyopia is common. The fundus of the eye shows crowded optical disks, tortuous vessels, and an abnormal foveal avascular zone. (ECO:0000269|PubMed:21397065, ECO:0000269|PubMed:21532570, ECO:0000269|PubMed:21850159). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, İzole 7, 613704; MCOP7 (İzole Anoftalmi-Mikroftalmi Sendromu) (GDF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 7, 613704; MCOP7 (Isolated anophthalmia-microphthalmia syndrome) (GDF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF3	GDF3, KFS3, MCOPCB6, MCOP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 3, autosomal dominant (KFS3) [MIM:613702]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. (ECO:0000269	PubMed:19864492). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mikroftalmi, İzole 8, 615113, Otozomal Resesif; MCOP8 (İzole Anoftalmi-Mikroftalmi Sendromu) (ALDH1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated 8, 615113, Autosomal recessive; MCOP8 (Isolated anophthalmia-microphthalmia syndrome) (ALDH1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH1A3	ALDH1A3, ALDH6, MCOP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikroftalmi,İzole, Koloboın 10, 616428, Otozomal Dominant; MCOPCB10 (Kolobomatöz Mikroftalmi) (RBP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated, with coloboma 10, 616428, Autosomal dominant; MCOPCB10 (Colobomatous microphthalmia) (RBP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBP4	RBP4, RDCCAS, MCOPCB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal dystrophy, iris coloboma, and comedogenic acne syndrome (RDCCAS) [MIM:615147]: A disease characterized by retinal degeneration, ocular colobomas involving both the anterior and posterior segment, impaired night vision and loss of visual acuity. Additional characteristic features include developmental abnormalities and severe acne. {ECO:0000269|PubMed:23189188, ECO:0000269|PubMed:9888420}. Note=The disease is caused by mutations affecting the gene represented in this entry. Loss of functional RBP4 protein results in serum retinol deficiency. Lack of normal levels of retinol impairs the visual cycle leading to night blindness at early stages; prolonged deficiency may lead to retinal degeneration. Additionally, retinol deficiency may result in dry skin, increased susceptibility to infection and acne (PubMed:23189188). {ECO:0000269|PubMed:23189188}.	EDTA Blood Sample
Mikroftalmi, İzole-Koloboma 3, İzole Edinilmiş, Mcopcb3 (Kolobomatöz Mikroftalmi) (VSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 3; MCOPCB3 (Colobomatous microphthalmia) (VSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VSX2	CHX10, HOX10, MCOP2, MCOPCB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia with cataracts and iris abnormalities (MCOPCTI) [MIM:610092]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, cataract and other abnormalities like cataract may also be present. {ECO:0000269	PubMed:10932181}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, İzole-Koloboma 6, İzole Edinilmiş, Mcopcb6 (Kolobomatöz Mikroftalmi) (GDF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MICROPHTHALMIA, ISOLATED, WITH COLOBOMA 6; MCOPCB6 (Colobomatous microphthalmia) (GDF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF6	GDF6, MCOP4, KFS1, MCOPCB6, LCA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Klippel-Feil syndrome 1, autosomal dominant (KFS1) [MIM:118100]: A skeletal disorder characterized by congenital fusion of cervical vertebrae. It is due to a failure in the normal segmentation of vertebrae during the early weeks of fetal development. The clinical triad consists of short neck, low posterior hairline, and limited neck movement. Deafness is a feature in some cases and may be of sensorineural, conductive, or mixed type. {ECO:0000269|PubMed:18425797, ECO:0000269|PubMed:19129173}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, Koloborn 7, İzole, 614497, Otozomal Dominant; MCOPCB7 (Kolobomatöz Mikroftalmi) (ABCB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated, with coloboma 7, 614497, Autosomal dominant; MCOPCB7 (Colobomatous microphthalmia) (ABCB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB6	ABCB6, MTABC3, MCOPCB7, LAN, DUH3, PSHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, isolated, with coloboma, 7 (MCOPCB7) [MIM:614497]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, and other abnormalities may also be present. Ocular colobomas are a set of malformations resulting from abnormal morphogenesis of the optic cup and stalk, and the fusion of the fetal fissure (optic fissure). {ECO:0000269	PubMed:22226084}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, Koloborn 8, 601186, Otozomal Resesif İle İzole (Matthew-Wood Sendromu) (STRA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated, with coloboma 8, 601186, Autosomal recessive (Matthew-Wood syndrome) (STRA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STRA6	STRA6, MCOPS9, MCOPCB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 9 (MCOPS9) [MIM:601186]: A rare clinical entity including as main characteristics anophthalmia or severe microphthalmia, and pulmonary hypoplasia or aplasia. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269|PubMed:17273977, ECO:0000269|PubMed:17503335, ECO:0000269|PubMed:21901792}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, Koloborn 9, 615145, Otozomal Resesif İle İzole; MCOPCB9 (Kolobomatöz Mikroftalmi) (TENM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, isolated, with coloboma 9, 615145, Autosomal recessive; MCOPCB9 (Colobomatous microphthalmia) (TENM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TENM3	TENM3, ODZ3, TNM3, KIAA1455, MCOPCB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, isolated, with coloboma, 9 (MCOPCB9) [MIM:615145]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, and other abnormalities may also be present. Ocular colobomas are a set of malformations resulting from abnormal morphogenesis of the optic cup and stalk, and the fusion of the fetal fissure (optic fissure). {ECO:0000269	PubMed:22766609}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendrom 1, 309800, X'e Bağlı; MCOPS1 (Mikroftalmi, Lenz Tipi) (NAA10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 1, 309800, X-linked; MCOPS1 (Microphthalmia, Lenz type) (NAA10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAA10	NAA10, ARD1A, ARD1, TE2, NATD, OGDNS, MCOPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: N-terminal acetyltransferase deficiency (NATD) [MIM:300855]: An enzymatic deficiency resulting in postnatal growth failure with severe delays and dysmorphic features. It is clinically characterized by wrinkled forehead, prominent eyes, widely opened anterior and posterior fontanels, downsloping palpebral fissures, thickened lids, large ears, flared nares, hypoplastic alae, short columella, protruding upper lip, and microretrognathia. There are also delayed closing of fontanels and broad great toes. Skin is characterized by redundancy or laxity with minimal subcutaneous fat, cutaneous capillary malformations, and very fine hair and eyebrows. Death results from cardiogenic shock following arrhythmia. {ECO:0000269	PubMed:21700266}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, Sendromik 11, 614402, Otozomal Resesif (VAX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 11, 614402, Autosomal recessive (VAX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VAX1	VAX1, MCOPS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 11 (MCOPS11) [MIM:614402]: A rare clinical entity including as main characteristics microphthalmia and small optic nerves, cleft lip and palate, absence of corpus callosum, hippocampal malformations, and absence of the pineal gland. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:22095910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendromik 12, 615524, Otozomal Resesif, Otozomal Dominant; MCOPS12 (Matthew-Wood Sendromu) (RARB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 12, 615524, Autosomal recessive, Autosomal dominant; MCOPS12 (Matthew-Wood syndrome) (RARB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RARB	RARB, HAP, MCOPS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 12 (MCOPS12) [MIM:615524]: A form of microphthalmia, a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. MCOPS12 patients manifest variable features, including diaphragmatic hernia, pulmonary hypoplasia, and cardiac abnormalities. {ECO:0000269	PubMed:24075189}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendromik 13, 300915, X'e Bağlı; MCOPS13 (X'e Bağlı Kolobomatöz Mikroftalmi-Mikrosefali-Mental Retardasyon-Kısa Boy Sendromu) (HMGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 13, 300915, X-linked; MCOPS13 (X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome) (HMGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMGB3	HMGB3, HMG4, HMG2A, MCOPS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 13 (MCOPS13) [MIM:300915]: A form of microphthalmia, a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. MCOPS13 patients exhibit colobomatous microphthalmia with microcephaly, short stature, and psychomotor retardation. {ECO:0000269	PubMed:24993872}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendromik 14, 615877, Otozomal Resesif, Otozomal Dominant; MCOPS14 (Kolobomatöz Mikroftalmi-Rizomelik Displazi Sendromu) (MAB21L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 14, 615877, Autosomal recessive, Autosomal dominant; MCOPS14 (Colobomatous microphthalmia-rhizomelic dysplasia syndrome) (MAB21L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAB21L2	MAB21L2, MCOPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 14 (MCOPS14) [MIM:615877]: A form of microphthalmia, a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. MCOPS14 patients exhibit bilateral colobomatous microphthalmia or bilateral anophthalmia. Intellectual disability and rhizomelic skeletal dysplasia is present in some affected individuals. {ECO:0000269	PubMed:24906020}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendromik 14, 615877, Otozomal Resesif, Otozomal Dominant; MCOPS14 (Kolobomatöz Mikroftalmi-Rizomelik Displazi Sendromu) (Prenatal) (MAB21L2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microphthalmia, syndromic 14, 615877, Autosomal recessive, Autosomal dominant; MCOPS14 (Colobomatous microphthalmia-rhizomelic dysplasia syndrome) (MAB21L2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAB21L2	MAB21L2, MCOPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microphthalmia, syndromic, 14 (MCOPS14) [MIM:615877]: A form of microphthalmia, a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. MCOPS14 patients exhibit bilateral colobomatous microphthalmia or bilateral anophthalmia. Intellectual disability and rhizomelic skeletal dysplasia is present in some affected individuals. {ECO:0000269	PubMed:24906020}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mikroftalmi, Sendromik 2, 300166, X'e Bağlı Dominant; MCOPS2 (Mikroftalmi, Lenz Tip) (BCOR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 2, 300166, X-linked dominant; MCOPS2 (Microphthalmia, Lenz type) (BCOR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCOR	BCOR, KIAA1575, MCOPS2, MAA2, ANOP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikroftalmi, Sendromik 3, 206900, Otozomal Dominant; MCOPS3 (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (SOX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 3, 206900, Autosomal dominant; MCOPS3 (Anophthalmia/microphthalmia-esophageal atresia syndrome) (SOX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX2	SOX2, MCOPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mikroftalmi, Sendromik 3, 206900, Otozomal Dominant; MCOPS3 (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (MLPA)	Microphthalmia, syndromic 3, 206900, Autosomal dominant; MCOPS3 (Anophthalmia/microphthalmia-esophageal atresia syndrome) (MLPA)	SOX2	SOX2, MCOPS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendromik 5, 610125, Otozomal Dominant; MCOPS5 (Sendromik Mikroftalmi Tip 5) (OTX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 5, 610125, Autosomal dominant; MCOPS5 (Syndromic microphthalmia type 5) (OTX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTX2	OTX2, MCOPS5, CPHD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 5 (MCOPS5) [MIM:610125]: Patients manifest unilateral or bilateral microphthalmia/clinical anophthalmia and variable additional features including pituitary dysfunction, coloboma, microcornea, cataract, retinal dystrophy, hypoplasia or agenesis of the optic nerve, agenesis of the corpus callosum, developmental delay, joint laxity, hypotonia, and seizures. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269|PubMed:15946561, ECO:0000269|PubMed:20396904, ECO:0000269|PubMed:22577225, ECO:0000269|PubMed:24167467}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroftalmi, Sendromik 6, 607932, Otozomal Dominant; MCOPS6 (Mikroftalmi-Beyin ve Parmak Anomalileri) (BMP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 6, 607932, Autosomal dominant; MCOPS6 (Microphthalmia with brain and digit anomalies) (BMP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP4	BMP4, BMP2B1, BMP2B, MCOPS6, OFC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-syndromic orofacial cleft 11 (OFC11) [MIM:600625]: A birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum. {ECO:0000269	PubMed:10240007}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mikroftalmi, Sendrom 9, 601186, Otozomal Resesif; MCOPS9 (Kolobomatöz Mikroftalmi) (Matthew-Wood Sendromu) (STRA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microphthalmia, syndromic 9, 601186, Autosomal recessive; MCOPS9 (Colobomatous microphthalmia) (Matthew-Wood syndrome) (STRA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STRA6	STRA6, MCOPS9, MCOPCB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 9 (MCOPS9) [MIM:601186]: A rare clinical entity including as main characteristics anophthalmia or severe microphthalmia, and pulmonary hypoplasia or aplasia. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269|PubMed:17273977, ECO:0000269|PubMed:17503335, ECO:0000269|PubMed:21901792}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosferofaki ve / Veya Megalokornea, Ektopi Lentis ± Sekonder Glokom, 251750, Otozomal Resesif; MSPKA (Sekonder Glokom-Sferofaki / Ektopi Lentis ve Megalokornea) (LTBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma, 251750, Autosomal recessive; MSPKA (Glaucoma secondary to spherophakia/ectopia lentis and megalocornea) (LTBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTBP2	LTBP2, LTBP3, GLC3D, MSPKA, WMS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microspherophakia and/or megalocornea, with ectopia lentis and with or without secondary glaucoma (MSPKA) [MIM:251750]: A rare disease characterized by smaller and more spherical lenses than normal bilaterally, an increased anteroposterior thickness of the lens, and highly myopic eyes. Lens dislocation or subluxation may occur, leading to defective accommodation. {ECO:0000269	PubMed:20617341}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroti, ± İşitme Kaybı (OD), 612290, Otozomal Resesif, Otozomal Dominant (Bilateral Mikroti-Sağırlık-Yarık Damak Sendromu) (HOXA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microtia with or without hearing impairment (AD), 612290, Autosomal recessive, Autosomal dominant (Bilateral microtia-deafness-cleft palate syndrome) (HOXA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA2	HOXA2, MCOHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microtia, hearing impairment, and cleft palate (MHCP) [MIM:612290]: A disease characterized by microtia, mixed symmetric severe to profound hearing impairment, and partial cleft palate. Microtia is a congenital deformity of the outer ear that is small and abnormally shaped. In classic microtia, the pinna is essentially absent, except for a vertical sausage-shaped skin remnant. The superior aspect of this sausage-shaped skin remnant consists of underlying unorganized cartilage, and the inferior aspect of this remnant consists of a relatively well-formed lobule. Syndromic forms of microtia occur in conjunction with other abnormalities including cleft palate, a congenital fissure of the soft and/or hard palate due to faulty fusion. {ECO:0000269	PubMed:18394579}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikroti, İşitme Engeli ve Yarık Damak (OR), 612290, Otozomal Resesif, Otozomal Dominant (Bilateral Mikroti-Sağırlık-Yarık Damak Sendromu) (HOXA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microtia, hearing impairment, and cleft palate (AR), 612290, Autosomal recessive, Autosomal dominant (Bilateral microtia-deafness-cleft palate syndrome) (HOXA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA2	HOXA2, MCOHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microtia, hearing impairment, and cleft palate (MHCP) [MIM:612290]: A disease characterized by microtia, mixed symmetric severe to profound hearing impairment, and partial cleft palate. Microtia is a congenital deformity of the outer ear that is small and abnormally shaped. In classic microtia, the pinna is essentially absent, except for a vertical sausage-shaped skin remnant. The superior aspect of this sausage-shaped skin remnant consists of underlying unorganized cartilage, and the inferior aspect of this remnant consists of a relatively well-formed lobule. Syndromic forms of microtia occur in conjunction with other abnormalities including cleft palate, a congenital fissure of the soft and/or hard palate due to faulty fusion. {ECO:0000269	PubMed:18394579}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
(Diyabetin Mikrovasküler Komplikasyonları 1), 603933 (VEGFA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 1, 603933 (VEGFA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VEGFA	VEGF, MVCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microvascular complications of diabetes 1 (MVCD1) [MIM:603933]: Pathological conditions that develop in numerous tissues and organs as a consequence of diabetes mellitus. They include diabetic retinopathy, diabetic nephropathy leading to end-stage renal disease, and diabetic neuropathy. Diabetic retinopathy remains the major cause of new-onset blindness among diabetic adults. It is characterized by vascular permeability and increased tissue ischemia and angiogenesis. {ECO:0000269	PubMed:11978667}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabetin Mikrovasküler Komplikasyonları 2, 612623 (EPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 2, 612623 (EPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPO	EPO, MVCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
(Diyabetin Mikrovasküler Komplikasyonları 3), 612624 (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 3, 612624 (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diyabetin Mikrovasküler Komplikasyonları 4, 612628 (IL1RN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 4, 612628 (IL1RN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL1RN	IL1RN, MVCD4, DIRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Interleukin 1 receptor antagonist deficiency (DIRA) [MIM:612852]: A rare autoinflammatory disease of skin and bone resulting in sterile multifocal osteomyelitis, periostitis, and pustulosis from birth. The term autoinflammatory disease describes a group of disorders characterized by attacks of seemingly unprovoked inflammation without significant levels of autoantibodies and autoreactive T-cells. {ECO:0000269	PubMed:19494218}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabetin Mikrovasküler Komplikasyonları 5, 612633 (PON1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 5, 612633 (PON1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PON1	PON1, PON, ESA, MVCD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microvascular complications of diabetes 5 (MVCD5) [MIM:612633]: Pathological conditions that develop in numerous tissues and organs as a consequence of diabetes mellitus. They include diabetic retinopathy, diabetic nephropathy leading to end-stage renal disease, and diabetic neuropathy. Diabetic retinopathy remains the major cause of new-onset blindness among diabetic adults. It is characterized by vascular permeability and increased tissue ischemia and angiogenesis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Homozygosity for the Leu-55 allele is strongly associated with the development of retinal disease in diabetic patients.;	EDTA Blood Sample
(Diyabetin Mikrovasküler Komplikasyonları 6), 612634 (SOD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 6, 612634 (SOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOD2	SOD2, MNSOD, MVCD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microvascular complications of diabetes 6 (MVCD6) [MIM:612634]: Pathological conditions that develop in numerous tissues and organs as a consequence of diabetes mellitus. They include diabetic retinopathy, diabetic nephropathy leading to end-stage renal disease, and diabetic neuropathy. Diabetic retinopathy remains the major cause of new-onset blindness among diabetic adults. It is characterized by vascular permeability and increased tissue ischemia and angiogenesis. {ECO:0000269	PubMed:12624725}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Diyabetin Mikrovasküler Komplikasyonları 7, 612635 (HFE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvascular complications of diabetes 7, 612635 (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Variegate porphyria (VP) [MIM:176200]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Variegate porphyria is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Iron overload due to HFE variants is a precipitating or exacerbating factor in variegate porphyria.	EDTA Blood Sample
Mikrovillus İnklüzyon Hastalığı, 251850, Otozomal Resesif (Mikrovillus İnklüzyon Hastalığı) (MYO5B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Microvillus inclusion disease, 251850, Autosomal recessive (Microvillus inclusion disease) (MYO5B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO5B	MYO5B, KIAA1119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diarrhea 2, with microvillus atrophy (DIAR2) [MIM:251850]: A disease characterized by onset of intractable life-threatening watery diarrhea during infancy. Two forms are recognized: early-onset microvillus inclusion disease with diarrhea beginning in the neonatal period, and late-onset, with first symptoms appearing after 3 or 4 months of life. {ECO:0000269|PubMed:18724368, ECO:0000269|PubMed:19006234, ECO:0000269|PubMed:20186687, ECO:0000269|PubMed:21206382, ECO:0000269|PubMed:24138727, ECO:0000269|PubMed:24892806}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mikrovillus İnklüzyon Hastalığı, 251850, Otozomal Resesif (Mikrovillus İnklüzyon Hastalığı) (Prenatal) (MYO5B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Microvillus inclusion disease, 251850, Autosomal recessive (Microvillus inclusion disease) (MYO5B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYO5B	MYO5B, KIAA1119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diarrhea 2, with microvillus atrophy (DIAR2) [MIM:251850]: A disease characterized by onset of intractable life-threatening watery diarrhea during infancy. Two forms are recognized: early-onset microvillus inclusion disease with diarrhea beginning in the neonatal period, and late-onset, with first symptoms appearing after 3 or 4 months of life. {ECO:0000269|PubMed:18724368, ECO:0000269|PubMed:19006234, ECO:0000269|PubMed:20186687, ECO:0000269|PubMed:21206382, ECO:0000269|PubMed:24138727, ECO:0000269|PubMed:24892806}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orta Yüzey Hipoplazisi, İşitme Bozukluğu, Eliptositoz ve Nefrokalsinosis, 300990, X'e Bağlı Resesif; MFHEN (Aport Sendromu-Mental Retardasyon-Orta Yüz Fasiyal Hipoplazisi-Eliptositozis Sendromu) (AMMECR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990, X-linked recessive; MFHEN (Aport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome) (AMMECR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMMECR1	AMMECR1, MFHEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aport syndrome with mental retardation, midface hypoplasia and elliptocytosis (ATS-MR) [MIM:300194]: A X-linked contiguous gene deletion syndrome characterized by glomerulonephritis, sensorineural hearing loss, mental retardation, midface hypoplasia and elliptocytosis. {ECO:0000269	PubMed:10049589}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	EDTA Blood Sample
Orta Yüzey Hipoplazisi, İşitme Bozukluğu, Eliptositoz ve Nefrokalsinosis, 300990, X'e Bağlı Resesif; MFHEN (Aport Sendromu-Mental Retardasyon-Orta Yüz Fasiyal Hipoplazisi-Eliptositozis Sendromu) (Prenatal) (AMMECR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990, X-linked recessive; MFHEN (Aport syndrome-intellectual disability-midface hypoplasia-elliptocytosis syndrome) (AMMECR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMMECR1	AMMECR1, MFHEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Aport syndrome with mental retardation, midface hypoplasia and elliptocytosis (ATS-MR) [MIM:300194]: A X-linked contiguous gene deletion syndrome characterized by glomerulonephritis, sensorineural hearing loss, mental retardation, midface hypoplasia and elliptocytosis. {ECO:0000269	PubMed:10049589}. Note=The gene represented in this entry may be involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Migren Ailesel Tip, Yatkının 157300, Otozomal Dominant (TNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine without aura, susceptibility to, 157300, Autosomal dominant (TNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNF	TNF, TNFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet; arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). {ECO:0000269	PubMed:12746914}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Bazilar, 602481, Otozomal Dominant; FHM2 (Ailesel Veya Sporadik Hemiplejik Migren) (ATP1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, familial basilar, 602481, Autosomal dominant; FHM2 (Familial or sporadic hemiplegic migraine) (ATP1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP1A2	ATP1A2, FHM2, MHP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alternating hemiplegia of childhood 1 (AHC1) [MIM:104290]: A rare syndrome of episodic hemi- or quadriplegia lasting minutes to days. Most cases are accompanied by dystonic posturing, choreoathetoid movements, nystagmus, other ocular motor abnormalities, autonomic disturbances, and progressive cognitive impairment. It is typically distinguished from familial hemiplegic migraine by infantile onset and high prevalence of associated neurological deficits that become increasingly obvious with age. {ECO:0000269	PubMed:15174025}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Hemiplejik, 1, 141500, Otozomal Dominant (Ailesel Veya Sporadik Hemiplejik Migren) (CACNA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, familial hemiplegic, 1, 141500, Autosomal dominant (Familial or sporadic hemiplegic migraine) (CACNA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Hemiplejik, 1, 141500, Otozomal Dominant (Ailesel Veya Sporadik Hemiplejik Migren) (MLPA)	Migraine, familial hemiplegic, 1, 141500, Autosomal dominant (Familial or sporadic hemiplegic migraine) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Hemiplejik, 1, Progresif Serebellar Ataksi, 141500, Otozomal Dominant; FHM1 (Spinoserebellar Ataksi Tip 6) (CACNA1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500, Autosomal dominant; FHM1 (Spinocerebellar ataxia type 6) (CACNA1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Hemiplejik, 1, Progresif Serebellar Ataksi, 141500, Otozomal Dominant; FHM1 (Spinoserebellar Ataksi Tip 6) (MLPA)	Migraine, familial hemiplegic, 1, with progressive cerebellar ataxia, 141500, Autosomal dominant; FHM1 (Spinocerebellar ataxia type 6) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, Ailesel Hemiplejik, 3, 609634, Otozomal Dominant; FHM3 (Ailesel Veya Sporadik Hemiplejik Migren) (SCN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, familial hemiplegic, 3, 609634, Autosomal dominant; FHM3 (Familial or sporadic hemiplegic migraine) (SCN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18568737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117252,	EDTA Blood Sample

Migren, Ailesel Hemiplejik, 3, 609634, Otozomal Dominant, FHM3 (Ailesel Veya Sporadik Hemiplejik Migren) (MLPA)	Migraine, familial hemiplegic, 3, 609634, Autosomal dominant; FHM3 (Familial or sporadic hemiplegic migraine) (MLPA)	SCN1A	SCN1A, GEFSP2, SMEI, FEB3A, EIEE6, FHM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Generalized epilepsy with febrile seizures plus 2 (GEFS+2) [MIM:604403]: A rare autosomal dominant, familial condition with incomplete penetrance and large intrafamilial variability. Patients display febrile seizures persisting sometimes beyond the age of 6 years and/or a variety of afebrile seizure types. This disease combines febrile seizures, generalized seizures often precipitated by fever at age 6 years or more, and partial seizures, with a variable degree of severity. {ECO:0000269|PubMed:10742094, ECO:0000269|PubMed:11254444, ECO:0000269|PubMed:11254445, ECO:0000269|PubMed:11524484, ECO:0000269|PubMed:11756608, ECO:0000269|PubMed:12535936, ECO:0000269|PubMed:12576172, ECO:0000269|PubMed:12919402, ECO:0000269|PubMed:14672992, ECO:0000269|PubMed:15525788, ECO:0000269|PubMed:15694566, ECO:0000269|PubMed:15715999, ECO:0000269|PubMed:16525050, ECO:0000269|PubMed:17347258, ECO:0000269|PubMed:17507202, ECO:0000269|PubMed:17561957, ECO:0000269|PubMed:17927801, ECO:0000269|PubMed:18251839, ECO:0000269|PubMed:18413471, ECO:0000269|PubMed:18566737, ECO:0000269|PubMed:19339291, ECO:0000269|PubMed:19464195, ECO:0000269|PubMed:19522081, ECO:0000269|PubMed:20117752,	EDTA Blood Sample
Migren, Direnç, 157300, Otozomal Dominant (EDNRA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, resistance to, 157300, Autosomal dominant (EDNRA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDNRA	EDNRA, MFDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Migren, Yatkınlık, 157300, Otozomal Dominant (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, susceptibility to, 157300, Autosomal dominant (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Estrogen resistance (ESTRR) [MIM:615363]: A disorder characterized by partial or complete resistance to estrogens, in the presence of elevated estrogen serum levels. Clinical features include absence of the pubertal growth spurt, delayed bone maturation, unfused epiphyses, reduced bone mineral density, osteoporosis, continued growth into adulthood and very tall adult stature. Glucose intolerance, hyperinsulinemia and lipid abnormalities may also be present. {ECO:0000269|PubMed:23841731, ECO:0000269|PubMed:8961262}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Migren, ± Aura, Yatkınlık 12, 13, 613656 (KCNK18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Migraine, with or without aura, susceptibility to, 13, 613656 (KCNK18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNK18	KCNK18, TRESK, TRIK, MGR13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Migraine with or without aura 13 (MGR13) [MIM:613656]: A form of migraine transmitted in an autosomal dominant pattern. Migraine is a disabling symptom complex of periodic headaches, usually temporal and unilateral. Headaches are often accompanied by irritability, nausea, vomiting and photophobia, preceded by constriction of the cranial arteries. The two major subtypes are common migraine (migraine without aura) and classic migraine (migraine with aura). Classic migraine is characterized by recurrent attacks of reversible neurological symptoms (aura) that precede or accompany the headache. Aura may include a combination of sensory disturbances, such as blurred vision, hallucinations, vertigo, numbness and difficulty in concentrating and speaking. {ECO:0000269|PubMed:20871611}. Note=The disease is caused by mutations affecting the gene represented in this entry. Susceptibility to migraine has been shown to be conferred by a frameshift mutation that segregates with the disorder in a large multigenerational family. Migraine was associated with sensitivity to lights, sounds, and smells, as well as nausea and occasional vomiting. Triggers included fatigue, alcohol and bright lights. Mutations in KCNK18 are a rare cause of migraine.;	EDTA Blood Sample
Miller Sendromu, 263750, Otozomal Resesif; POADS (Postaksiyel Akrofasiyal Disostozi (DHODH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miller syndrome, 263750, Autosomal recessive; POADS (Postaxial acrofacial dysostosis) (DHODH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHODH	DHODH, URA1, POADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Postaxial acrofacial dysostosis (POADS) [MIM:263750]: POADS is characterized by severe micrognathia, cleft lip and/or palate, hypoplasia or aplasia of the posterior elements of the limbs, coloboma of the eyelids and supernumerary nipples. POADS is a very rare disorder: only 2 multiplex families, each consisting of 2 affected siblings born to unaffected, nonconsanguineous parents, have been described among a total of around 30 reported cases. {ECO:0000269|PubMed:19915526}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miller Sendromu, 263750, Otozomal Resesif; POADS (Postaksiyel Akrofasiyal Disostozi (Prenatal) (DHODH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miller syndrome, 263750, Autosomal recessive; POADS (Postaxial acrofacial dysostosis) (DHODH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHODH	DHODH, URA1, POADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Postaxial acrofacial dysostosis (POADS) [MIM:263750]: POADS is characterized by severe micrognathia, cleft lip and/or palate, hypoplasia or aplasia of the posterior elements of the limbs, coloboma of the eyelids and supernumerary nipples. POADS is a very rare disorder: only 2 multiplex families, each consisting of 2 affected siblings born to unaffected, nonconsanguineous parents, have been described among a total of around 30 reported cases. {ECO:0000269|PubMed:19915526}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miller-Dieker Lizensefali Sendromu, 247200, Otozomal Dominant (Miller-Dieker Sendromu) (FISH)	Miller-Dieker lissencephaly syndrome, 247200, Autosomal dominant (Miller-Dieker syndrome) (FISH)	.	MDLS, MDS, MDCR, DEL17p13.3, C17DELp13.3	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Miller-Dieker Lizensefali Sendromu, 247200, Otozomal Dominant (Miller-Dieker Sendromu) (MLPA)	Miller-Dieker lissencephaly syndrome, 247200, Autosomal dominant (Miller-Dieker syndrome) (MLPA)	.	MDLS, MDS, MDCR, DEL17p13.3, C17DELp13.3	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Miller-Dieker Lizensefali Sendromu, 247200, Otozomal Dominant (Miller-Dieker Sendromu) (Prenatal) (FISH)	Miller-Dieker lissencephaly syndrome, 247200, Autosomal dominant (Miller-Dieker syndrome) (Prenatal) (FISH)	.	MDLS, MDS, MDCR, DEL17p13.3, C17DELp13.3	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miller-Dieker Lizensefali Sendromu, 247200, Otozomal Dominant (Miller-Dieker Sendromu) (Prenatal) (MLPA)	Miller-Dieker lissencephaly syndrome, 247200, Autosomal dominant (Miller-Dieker syndrome) (Prenatal) (MLPA)	.	MDLS, MDS, MDCR, DEL17p13.3, C17DELp13.3	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Minicore Miyopati-Eksternal Oftalmopleji, 255320, Otozomal Resesif (Multiminikore Miyopati) (RYR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Minicore myopathy with external ophthalmoplegia, 255320, Autosomal recessive (Multiminicore myopathy) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095,	EDTA Blood Sample
Minicore Miyopati-Eksternal Oftalmopleji, 255320, Otozomal Resesif (Multiminikore Miyopati) (Prenatal) (RYR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Minicore myopathy with external ophthalmoplegia, 255320, Autosomal recessive (Multiminicore myopathy) (RYR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MIRAGE Sendromu, 617053, Otozomal Dominant; MIRAGE (SAMD9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MIRAGE syndrome, 617053, Autosomal dominant; MIRAGE (SAMD9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAMD9	SAMD9, NFTC, MIRAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tumoral calcinosis, normophosphatemic, familial (NFTC) [MIM:610455]: An uncommon, life-threatening disorder characterized by progressive deposition of calcified masses in cutaneous and subcutaneous tissues. Serum phosphate levels are normal. Clinical features include painful calcified ulcerative lesions and massive calcium deposition in the mid- and lower dermis, severe skin and bone infections, erythematous papular skin eruption in infancy, conjunctivitis, and gingivitis. NFTC shows a striking resemblance to acquired dystrophic calcinosis, in which tissue calcification occurs as a consequence of tissue injury/inflammation. {ECO:0000269|PubMed:16960814, ECO:0000269|PubMed:18094730}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

MIRAGE Sendromu, 617053, Otozomal Dominant; MIRAGE (Prenatal) (SAMD9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MIRAGE syndrome, 617053, Autosomal dominant; MIRAGE (SAMD9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SAMD9	SAMD9, NFTC, MIRAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tumoral calcinosis, normophosphatemic, familial (NFTC) [MIM:610455]: An uncommon, life-threatening disorder characterized by progressive deposition of calcified masses in cutaneous and subcutaneous tissues. Serum phosphate levels are normal. Clinical features include painful calcified ulcerative lesions and massive calcium deposition in the mid- and lower dermis, severe skin and bone infections, erythematous papular skin eruption in infancy, conjunctivitis, and gingivitis. NFTC shows a striking resemblance to acquired dystrophic calcinosis, in which tissue calcification occurs as a consequence of tissue injury/inflammation. {ECO:0000269|PubMed:16960814, ECO:0000269|PubMed:18094730}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ayna Hareketleri 1, 157600, Otozomal Dominant; MRMV1 (Ailesel Konjenital Ayna Hareketleri) (DCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mirror movements 1, 157600, Autosomal dominant; MRMV1 (Familial congenital mirror movements) (DCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCC	DCC, MRMV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mirror movements 1 (MRMV1) [MIM:157600]: A disorder characterized by contralateral involuntary movements that mirror voluntary ones. While mirror movements are occasionally found in young children, persistence beyond the age of 10 is abnormal. Mirror movements occur more commonly in the upper extremities. {ECO:0000269	PubMed:20431009}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ayna Hareketleri 2, 614508, Otozomal Dominant; MRMV2 (Ailesel Konjenital Ayna Hareketleri) (RAD51 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mirror movements 2, 614508, Autosomal dominant; MRMV2 (Familial congenital mirror movements) (RAD51 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD51	RAD51A, RECA, MRMV2, FANCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:10807537}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Ayna Hareketleri 3, 616059, Otozomal Resesif; MRMV3 (Ailesel Konjenital Ayna Hareketleri) (DNAL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mirror movements 3, 616059, Autosomal recessive; MRMV3 (Familial congenital mirror movements) (DNAL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAL4	DNAL4, MRMV3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mirror movements 3 (MRMV3) [MIM:616059]: A disorder characterized by contralateral involuntary movements that mirror voluntary ones. While mirror movements are occasionally found in young children, persistence beyond the age of 10 is abnormal. Mirror movements occur more commonly in the upper extremities. {ECO:0000269	PubMed:25985561}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MLPA)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MLPA)	MSH6	MSH6, GTBP, HNPCC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mismatch repair cancer syndrome (MMRCS) [MIM:276300]: An autosomal recessive, rare, childhood cancer predisposition syndrome encompassing a broad tumor spectrum. This includes hematological malignancies, central nervous system tumors, Lynch syndrome-associated malignancies such as colorectal tumors as well as multiple intestinal polyps, embryonic tumors and rhabdomyosarcoma. Multiple cafe-au-lait macules, a feature reminiscent of neurofibromatosis type 1, are often found as first manifestation of the underlying cancer. Areas of skin hypopigmentation have also been reported in MMRCS patients. {ECO:0000269	PubMed:17557300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MLPA)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MLPA)	MSH2	MSH2, COCA1, FCC1, HNPCC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11920458	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MLPA)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MLPA)	MLH1	MLH1, COCA2, HNPCC2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muir-Torre syndrome (MRTES) [MIM:158320]: Rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy. {ECO:0000269|PubMed:8751876}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MSH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MSH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH6	MSH6, GTBP, HNPCC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mismatch repair cancer syndrome (MMRCS) [MIM:276300]: An autosomal recessive, rare, childhood cancer predisposition syndrome encompassing a broad tumor spectrum. This includes hematological malignancies, central nervous system tumors, Lynch syndrome-associated malignancies such as colorectal tumors as well as multiple intestinal polyps, embryonic tumors and rhabdomyosarcoma. Multiple cafe-au-lait macules, a feature reminiscent of neurofibromatosis type 1, are often found as first manifestation of the underlying cancer. Areas of skin hypopigmentation have also been reported in MMRCS patients. {ECO:0000269|PubMed:17557300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MSH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MSH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH2	MSH2, COCA1, FCC1, HNPCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. {ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11920458	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MLH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mismatch repair cancer syndrome, 276300, Autosomal recessive (Constitutional mismatch repair deficiency syndrome) (MLH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH1	MLH1, COCA2, HNPCC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muir-Torre syndrome (MRTES) [MIM:158320]: Rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy. {ECO:0000269|PubMed:8751876}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif; MMRCS (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (PMS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mismatch repair cancer syndrome, 276300, Autosomal recessive; MMRCS (Constitutional mismatch repair deficiency syndrome) (PMS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMS2	PMS2, PMSL2, HNPCC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mismatch repair cancer syndrome (MMRCS) [MIM:276300]: An autosomal recessive, rare, childhood cancer predisposition syndrome encompassing a broad tumor spectrum. This includes hematological malignancies, central nervous system tumors, Lynch syndrome-associated malignancies such as colorectal tumors as well as multiple intestinal polyps, embryonic tumors and rhabdomyosarcoma. Multiple cafe-au-lait macules, a feature reminiscent of neurofibromatosis type 1, are often found as first manifestation of the underlying cancer. Areas of skin hypopigmentation have also been reported in MMRCS patients. {ECO:0000269|PubMed:15077197, ECO:0000269|PubMed:17557300, ECO:0000269|PubMed:7661935, ECO:0000269|PubMed:9419979}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mismatch Repair Kanser Sendromu, 276300, Otozomal Resesif; MMRCS (Konstitüsyonel Mismatch Onarım Eksikliği Sendromu) (MLPA)	Mismatch repair cancer syndrome, 276300, Autosomal recessive; MMRCS (Constitutional mismatch repair deficiency syndrome) (MLPA)	PMS2	PMS2, PMSL2, HNPCC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mismatch repair cancer syndrome (MMRCS) [MIM:276300]: An autosomal recessive, rare, childhood cancer predisposition syndrome encompassing a broad tumor spectrum. This includes hematological malignancies, central nervous system tumors, Lynch syndrome-associated malignancies such as colorectal tumors as well as multiple intestinal polyps, embryonic tumors and rhabdomyosarcoma. Multiple cafe-au-lait macules, a feature reminiscent of neurofibromatosis type 1, are often found as first manifestation of the underlying cancer. Areas of skin hypopigmentation have also been reported in MMRCS patients. {ECO:0000269|PubMed:15077197, ECO:0000269|PubMed:17557300, ECO:0000269|PubMed:7661935, ECO:0000269|PubMed:9419979}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mitchell Riley Sendromu, 615710, Otozomal Resesif, MTCHRS (Hipoplastik Pankreas-Bağırsak Atrezisi-Hipoplastik Gallbalder Sendromu) (RFX6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitchell-Riley syndrome, 615710, Autosomal recessive; MTCHRS (Hypoplastic pancreas-intestinal atresia-hypoplastic gallbalder syndrome) (RFX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFX6	RFX6, RFXDC1, MTCHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitchell Riley Sendromu, 615710, Otozomal Resesif, MTCHRS (Hipoplastik Pankreas-Bağırsak Atrezisi-Hipoplastik Gallbalder Sendromu) (Prenatal) (RFX6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitchell-Riley syndrome, 615710, Autosomal recessive; MTCHRS (Hypoplastic pancreas-intestinal atresia-hypoplastic gallbalder syndrome) (RFX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RFX6	RFX6, RFXDC1, MTCHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks (ATP Sentaz) Eksikliği, Nükleer Tip 4, 615228, Otozomal Resesif (Izole ATP Sentaz Eksikliği) (ATP5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228, Autosomal recessive (isolated ATP synthase deficiency) (ATP5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP5A1	ATP5A1, ATPM, ATP5A, ORM, MCSDN4, COXPD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex V deficiency, nuclear 4 (MCSDN4) [MIM:615228]: A mitochondrial disorder with heterogeneous clinical manifestations including dysmorphic features, psychomotor retardation, hypotonia, growth retardation, cardiomyopathy, enlarged liver, hypoplastic kidneys and elevated lactate levels in urine, plasma and cerebrospinal fluid. {ECO:0000269|PubMed:23599390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks (ATP Sentaz) Eksikliği, Nükleer Tip 4, 615228, Otozomal Resesif (Izole ATP Sentaz Eksikliği) (Prenatal) (ATP5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex (ATP synthase) deficiency, nuclear type 4, 615228, Autosomal recessive (isolated ATP synthase deficiency) (ATP5A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP5A1	ATP5A1, ATPM, ATP5A, ORM, MCSDN4, COXPD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex V deficiency, nuclear 4 (MCSDN4) [MIM:615228]: A mitochondrial disorder with heterogeneous clinical manifestations including dysmorphic features, psychomotor retardation, hypotonia, growth retardation, cardiomyopathy, enlarged liver, hypoplastic kidneys and elevated lactate levels in urine, plasma and cerebrospinal fluid. {ECO:0000269|PubMed:23599390}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFAF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex 1 deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF5	NDUFAF5, C20orf7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:18940309}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (Prenatal) (NDUFAF5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex 1 deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF5	NDUFAF5, C20orf7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:18940309}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, ACAD9 Eksikliğine Bağlı, 611126, Otozomal Resesif (Leigh Sendromu ve Mitokondriyal Ensefalopati (Açil-CoA Dehidrojenaz 9 Eksikliği) (ACAD9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126, Autosomal recessive (Leigh syndrome and mitochondrial encephalopathy) (Acyl-CoA dehydrogenase 9 deficiency) (ACAD9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAD9	ACAD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, ACAD9 Eksikliğine Bağlı, 611126, Otozomal Resesif (Leigh Sendromu ve Mitokondriyal Ensefalopati (Açil-CoA Dehidrojenaz 9 Eksikliği) (Prenatal) (ACAD9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency due to ACAD9 deficiency, 611126, Autosomal recessive (Leigh syndrome and mitochondrial encephalopathy) (Acyl-CoA dehydrogenase 9 deficiency) (ACAD9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACAD9	ACAD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (TMEM126B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (TMEM126B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM126B	TMEM126B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NUBPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NUBPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUBPL	NUBPL, IND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFV2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFV2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFV2	NDUFV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFV1	NDUFV1, UQOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:10080174, ECO:0000269|PubMed:11340233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFS6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS6	NDUFS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFS4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS4	NDUFS4, AQDQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:9463323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS3	NDUFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS2	NDUFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:11220739}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFS1	NDUFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:11340233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal Resesif, X'e Bağlı Dominant, Mitokondriyal (Izole Kompleks 1 Eksikliği) (NDUFB9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFB9	NDUFB9, UQOR22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFB3	NDUFB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal Resesif, X'e Bağlı Dominant, Mitokondriyal (Izole Kompleks 1 Eksikliği) (NDUFB11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFB11	NDUFB11, LSDMCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFAF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF4	NDUFAF4, HRPAP20, C6orf66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:18179882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (Izole Kompleks 1 Eksikliği) (NDUFAF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF3	NDUFAF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:19463981}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (NDUFAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF2	NDUFAF2, NDUFA12L, MMTN, B17.2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:16200211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (NDUFAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFAF1	NDUFAF1, CIA30, CGI65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (NDUFA11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFA11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA11	NDUFA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:18306244}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (NDUFA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA1	NDUFA1, MWFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (FOXRED1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (FOXRED1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXRED1	FOXRED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:20818383, ECO:0000269|PubMed:20858599}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal Resesif, X'e Bağlı Dominant, Mitokondriyal (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFB9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFB9	NDUFB9, UQOR22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal Resesif, X'e Bağlı Dominant, Mitokondriyal (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFB11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFB11	NDUFB11, LSDMCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (TMEM126B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (TMEM126B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM126B	TMEM126B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NUBPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NUBPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUBPL	NUBPL, IND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFV2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFV2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFV2	NDUFV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFV1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFV1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFV1	NDUFV1, UQOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:10080174, ECO:0000269|PubMed:11349233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFS6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS6	NDUFS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFS4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS4	NDUFS4, AQDQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:9463323}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFS3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS3	NDUFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS2	NDUFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:11220739}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFS1	NDUFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:11349233}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFB3	NDUFB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFAF4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF4	NDUFAF4, HRPAP20, C5orf66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:18179882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFAF3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF3	NDUFAF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:19463981}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFAF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF2	NDUFAF2, NDUFA12L, MMTN, B17.2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269	PubMed:16200211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (isolated complex I deficiency) (NDUFAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFAF1	NDUFAF1, CIA30, CGI65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFA11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (Isolated complex I deficiency) (NDUFA11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA11	NDUFA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:18306244}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (NDUFA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (Isolated complex I deficiency) (NDUFA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDUFA1	NDUFA1, MWFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 1 Eksikliği, 252010, Otozomal, Mitokondriyal, X'e Bağlı Dominant (İzole Kompleks 1 Eksikliği) (Prenatal) (FOXRED1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex I deficiency, 252010, Autosomal recessive, X-linked dominant, Mitochondrial (Isolated complex I deficiency) (FOXRED1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXRED1	FOXRED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex I deficiency (MT-C1D) [MIM:252010]: A disorder of the mitochondrial respiratory chain that causes a wide range of clinical manifestations from lethal neonatal disease to adult-onset neurodegenerative disorders. Phenotypes include macrocephaly with progressive leukodystrophy, non-specific encephalopathy, cardiomyopathy, myopathy, liver disease, Leigh syndrome, Leber hereditary optic neuropathy, and some forms of Parkinson disease. {ECO:0000269|PubMed:20818383, ECO:0000269|PubMed:20858599}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği (İzole Süksinat-Coq Redüktaz Eksikliği) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	MITOCHONDRIAL COMPLEX II DEFICIENCY (Isolated succinate-CoQ reductase deficiency) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği (İzole Süksinat-Coq Redüktaz Eksikliği) (Prenatal) (SDHD geni) (Tüm Gen Dizi Analizi) (Prenatal)	MITOCHONDRIAL COMPLEX II DEFICIENCY (Isolated succinate-CoQ reductase deficiency) (SDHD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği, 252011, Otozomal Resesif (İzole Süksinat-Coq Redüktaz Eksikliği) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex II deficiency, 252011, Autosomal recessive (Isolated succinate-CoQ reductase deficiency) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği, 252011, Otozomal Resesif (İzole Süksinat-Coq Redüktaz Eksikliği) (SDHAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex II deficiency, 252011, Autosomal recessive (Isolated succinate-CoQ reductase deficiency) (SDHAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHAF1	SDHAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği, 252011, Otozomal Resesif (İzole Süksinat-Coq Redüktaz Eksikliği) (Prenatal) (SDHD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex II deficiency, 252011, Autosomal recessive (Isolated succinate-CoQ reductase deficiency) (SDHD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 2 Eksikliği, 252011, Otozomal Resesif (İzole Süksinat-Coq Redüktaz Eksikliği) (Prenatal) (SDHAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex II deficiency, 252011, Autosomal recessive (Isolated succinate-CoQ reductase deficiency) (SDHAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SDHAF1	SDHAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 1, 124000, Otozomal Resesifv (Tübülopati-Ensefalopati-Karaciğer Yetmezliği Sendromu) (BCS1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 1, 124000, Autosomal recessive; MC3DN1 (Renal tubulopathy-encephalopathy-liver failure syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 1 (MC3DN1) [MIM:124000]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:11528392, ECO:0000269|PubMed:12910490, ECO:0000269|PubMed:17314340, ECO:0000269|PubMed:17403714, ECO:0000269|PubMed:19162478, ECO:0000269|PubMed:22991165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 1, 124000, Otozomal Resesifv (Tübülopati-Ensefalopati-Karaciğer Yetmezliği Sendromu) (Prenatal) (BCS1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 1, 124000, Autosomal recessive; MC3DN1 (Renal tubulopathy-encephalopathy-liver failure syndrome) (BCS1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BCS1L	BCS1L, FLNMS, GRACILE, BJS, PTD, MC3DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 1 (MC3DN1) [MIM:124000]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:11528392, ECO:0000269|PubMed:12910490, ECO:0000269|PubMed:17314340, ECO:0000269|PubMed:17403714, ECO:0000269|PubMed:19162478, ECO:0000269|PubMed:22991165}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 2, 615157, Otozomal Resesif; MC3DN2 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (TTC19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 2, 615157, Autosomal recessive; MC3DN2 (Isolated CoQ-cytochrome C reductase deficiency) (TTC19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC19	TTC19, MC3DN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 2 (MC3DN2) [MIM:615157]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:21278747}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 2, 615157, Otozomal Resesif; MC3DN2 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (TTC19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 2, 615157, Autosomal recessive; MC3DN2 (Isolated CoQ-cytochrome C reductase deficiency) (TTC19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTC19	TTC19, MC3DN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 2 (MC3DN2) [MIM:615157]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:21278747}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 3, 615158, Otozomal Resesif; MC3DN3 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (UQCRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 3, 615158, Autosomal recessive; MC3DN3 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UQCRB	UQCRB, UQBP, QPC, MC3DN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 3 (MC3DN3) [MIM:615158]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:12709789}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 3, 615158, Otozomal Resesif; MC3DN3 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (UQCRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 3, 615158, Autosomal recessive; MC3DN3 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UQCRB	UQCRB, UQBP, QPC, MC3DN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 3 (MC3DN3) [MIM:615158]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:12709789}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 4, 615159, Otozomal Resesif; MC3DN4 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (UQCRQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 4, 615159, Autosomal recessive; MC3DN4 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UQCRQ	UQCRQ, QPC, MC3DN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 4 (MC3DN4) [MIM:615159]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:18439546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 4, 615159, Otozomal Resesif; MC3DN4 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (UQCRQ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 4, 615159, Autosomal recessive; MC3DN4 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRQ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UQCRQ	UQCRQ, QPC, MC3DN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 4 (MC3DN4) [MIM:615159]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:18439546}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 5, 615160, Otozomal Resesif; MC3DN5 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (UQCRC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 5, 615160, Autosomal recessive; MC3DN5 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UQCRC2	UQCRC2, MC3DN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 5 (MC3DN5) [MIM:615160]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:23281071}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 5, 615160, Otozomal Resesif; MC3DN5 (İzole Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (UQCRC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 5, 615160, Autosomal recessive; MC3DN5 (Isolated CoQ-cytochrome C reductase deficiency) (UQCRC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UQCRC2	UQCRC2, MC3DN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 5 (MC3DN5) [MIM:615160]: A disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269|PubMed:23281071}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 6, 615453, Otozomal Resesif; MC3DN6 (özele Coq-Sitokrom C Redüktaz Eksikliği) (CYC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 6, 615453, Autosomal recessive; MC3DN6 (isolated CoQ-cytochrome C reductase deficiency) (CYC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYC1	CYC1, MC3DN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 6 (MC3DN6) [MIM:615453]: An autosomal recessive disorder caused by mitochondrial dysfunction. It is characterized by onset in early childhood of episodic acute lactic acidosis, ketoacidosis, and insulin-responsive hyperglycemia, usually associated with infection. Laboratory studies show decreased activity of mitochondrial complex III. Psychomotor development is normal. {ECO:0000269	PubMed:23910460}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 6, 615453, Otozomal Resesif; MC3DN6 (özele Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (CYC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 6, 615453, Autosomal recessive; MC3DN6 (isolated CoQ-cytochrome C reductase deficiency) (CYC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYC1	CYC1, MC3DN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 6 (MC3DN6) [MIM:615453]: An autosomal recessive disorder caused by mitochondrial dysfunction. It is characterized by onset in early childhood of episodic acute lactic acidosis, ketoacidosis, and insulin-responsive hyperglycemia, usually associated with infection. Laboratory studies show decreased activity of mitochondrial complex III. Psychomotor development is normal. {ECO:0000269	PubMed:23910460}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 7, 615824, Otozomal Resesif; MC3DN7 (özele Coq-Sitokrom C Redüktaz Eksikliği) (UQCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 7, 615824, Autosomal recessive; MC3DN7 (isolated CoQ-cytochrome C reductase deficiency) (UQCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UQCC2	UQCC2, C6orf126, M19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 7, 615824, Otozomal Resesif; MC3DN7 (özele Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (UQCC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 7, 615824, Autosomal recessive; MC3DN7 (isolated CoQ-cytochrome C reductase deficiency) (UQCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UQCC2	UQCC2, C6orf126, M19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 8, 615838, Otozomal Resesif; MC3DN8 (özele Coq-Sitokrom C Redüktaz Eksikliği) (LYRM7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 8, 615838, Autosomal recessive; MC3DN8 (isolated CoQ-cytochrome C reductase deficiency) (LYRM7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LYRM7	LYRM7, MZM1L, MC3DN8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 8 (MC3DN8) [MIM:615838]: A form of mitochondrial complex III deficiency, a disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269	PubMed:24014394}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 8, 615838, Otozomal Resesif; MC3DN8 (özele Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (LYRM7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 8, 615838, Autosomal recessive; MC3DN8 (isolated CoQ-cytochrome C reductase deficiency) (LYRM7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LYRM7	LYRM7, MZM1L, MC3DN8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 8 (MC3DN8) [MIM:615838]: A form of mitochondrial complex III deficiency, a disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. Clinical features include mitochondrial encephalopathy, psychomotor retardation, ataxia, severe failure to thrive, liver dysfunction, renal tubulopathy, muscle weakness and exercise intolerance. {ECO:0000269	PubMed:24014394}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 9, 616111, Otozomal Resesif; MC3DN9 (özele Coq-Sitokrom C Redüktaz Eksikliği) (UQCC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex III deficiency, nuclear type 9, 616111, Autosomal recessive; MC3DN9 (isolated CoQ-cytochrome C reductase deficiency) (UQCC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UQCC3	UQCC3, C11orf83, MC3DN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex III deficiency, nuclear 9 (MC3DN9) [MIM:616111]: A form of mitochondrial complex III deficiency, a disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. MC3DN9 clinical features include feeding difficulties, hypoglycemia, severe lactic acidosis, and delayed psychomotor development. {ECO:0000269	PubMed:25008109}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 3 Eksikliği, Nükleer Tip 9, 616111, Otozomal Resesif; MC3DN9 (özele Coq-Sitokrom C Redüktaz Eksikliği) (Prenatal) (UQCC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex III deficiency, nuclear type 9, 616111, Autosomal recessive; MC3DN9 (isolated CoQ-cytochrome C reductase deficiency) (UQCC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UQCC3	UQCC3, C11orf83, MC3DN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex III deficiency, nuclear 9 (MC3DN9) [MIM:616111]: A form of mitochondrial complex III deficiency, a disorder of the mitochondrial respiratory chain resulting in a highly variable phenotype depending on which tissues are affected. MC3DN9 clinical features include feeding difficulties, hypoglycemia, severe lactic acidosis, and delayed psychomotor development. {ECO:0000269	PubMed:25008109}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (TACO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (TACO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TACO1	TACO1, CCDC44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (SCO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (SCO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCO1	SCO1, SCOD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (PET100 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (PET100 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PET100	PET100, C19orf79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (FASTKD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (FASTKD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FASTKD2	FASTKD2, KIAA0971	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (COX8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX8A	COX8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (COX6B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX6B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX6B1	COX6B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. {ECO:0000269	PubMed:18490582}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (COX20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX20	COX20, FAM36A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (COX14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX14	COX14, C12orf62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in COX14 may be a cause of a mitochondrial disorder presenting with severe congenital lactic acidosis and dysmorphic features associated with a COX assembly defect. Other features include brain hypertrophy, diffuse alteration of the white-matter myelination, and numerous cavities in the parieto- occipital region, brainstem, and cerebellum, as well as hepatomegaly, hypertrophic cardiomyopathy, renal hypoplasia, and adrenal-gland hyperplasia. {ECO:0000269	PubMed:22243966}.;	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (COX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COX10	COX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. {ECO:0000269|PubMed:10767350, ECO:0000269|PubMed:12928494}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (APOPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (APOPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOPT1	APOPT1, APOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (Prenatal) (TACO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (TACO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TACO1	TACO1, CCDC44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leigh syndrome (LS) [MIM:256000]: An early-onset progressive neurodegenerative disorder characterized by the presence of focal, bilateral lesions in one or more areas of the central nervous system including the brainstem, thalamus, basal ganglia, cerebellum and spinal cord. Clinical features depend on which areas of the central nervous system are involved and include subacute onset of psychomotor retardation, hypotonia, ataxia, weakness, vision loss, eye movement abnormalities, seizures, and dysphagia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (Prenatal) (SCO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (SCO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCO1	SCO1, SCOD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (Prenatal) (PET100 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (PET100 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PET100	PET100, C19orf79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (Prenatal) (FASTKD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (FASTKD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FASTKD2	FASTKD2, KIAA0971	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (özele Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (isolated cytochrome C oxidase deficiency) (COX8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX8A	COX8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (Izole Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX6B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (Isolated cytochrome C oxidase deficiency) (COX6B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX6B1	COX6B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. (ECO:0000269	PubMed:18490682). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (Izole Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (Isolated cytochrome C oxidase deficiency) (COX20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX20	COX20, FAM36A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (Izole Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (Isolated cytochrome C oxidase deficiency) (COX14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX14	COX14, C12orf62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Defects in COX14 may be a cause of a mitochondrial disorder presenting with severe congenital lactic acidosis and dysmorphic features associated with a COX assembly defect. Other features include brain hypertrophy, diffuse alteration of the white-matter myelination, and numerous cavities in the parieto- occipital region, brainstem, and cerebellum, as well as hepatomegaly, hypertrophic cardiomyopathy, renal hypoplasia, and adrenal-gland hyperplasia. (ECO:0000269	PubMed:22423661.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (Izole Sitokrom C Oksidaz Eksikliği) (Prenatal) (COX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (Isolated cytochrome C oxidase deficiency) (COX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COX10	COX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex IV deficiency (MT-C4D) [MIM:220110]: A disorder of the mitochondrial respiratory chain with heterogeneous clinical manifestations, ranging from isolated myopathy to severe multisystem disease affecting several tissues and organs. Features include hypertrophic cardiomyopathy, hepatomegaly and liver dysfunction, hypotonia, muscle weakness, exercise intolerance, developmental delay, delayed motor development and mental retardation. Some affected individuals manifest a fatal hypertrophic cardiomyopathy resulting in neonatal death. A subset of patients manifest Leigh syndrome. (ECO:0000269|PubMed:10767350, ECO:0000269|PubMed:12928484). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 4 Eksikliği, 220110, Otozomal Resesif, Mitokondriyal (Izole Sitokrom C Oksidaz Eksikliği) (Prenatal) (APOPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex IV deficiency, 220110, Autosomal recessive, Mitochondrial (Isolated cytochrome C oxidase deficiency) (APOPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	APOPT1	APOPT1, APOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 1, 604273, Otozomal Resesif, MC5DN1 (Izole ATP Sentaz Eksikliği) (ATPAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273, Autosomal recessive; MC5DN1 (Isolated ATP synthase deficiency) (ATPAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATPAF2	ATPAF2, ATP12, MC5DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 1, 604273, Otozomal Resesif, MC5DN1 (Izole ATP Sentaz Eksikliği) (Prenatal) (ATPAF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 1, 604273, Autosomal recessive; MC5DN1 (Isolated ATP synthase deficiency) (ATPAF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATPAF2	ATPAF2, ATP12, MC5DN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 1, 604273, Otozomal Resesif, MC5DN2 (Izole ATP Sentaz Eksikliği) (TMEM70 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052, Autosomal recessive; MC5DN2 (Isolated ATP synthase deficiency) (TMEM70 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM70	TMEM70, MC5DN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial complex V deficiency, nuclear 2 (MC5DN2) [MIM:614052]: A mitochondrial disorder with heterogeneous clinical manifestations including dysmorphic features, psychomotor retardation, hypotonia, growth retardation, cardiomyopathy, enlarged liver, hypoplastic kidneys and elevated lactate levels in urine, plasma and cerebrospinal fluid. (ECO:0000269|PubMed:18953340, ECO:0000269|PubMed:22986587). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 1, 604273, Otozomal Resesif, MC5DN2 (Izole ATP Sentaz Eksikliği) (Prenatal) (TMEM70 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 2, 614052, Autosomal recessive; MC5DN2 (Isolated ATP synthase deficiency) (TMEM70 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM70	TMEM70, MC5DN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial complex V deficiency, nuclear 2 (MC5DN2) [MIM:614052]: A mitochondrial disorder with heterogeneous clinical manifestations including dysmorphic features, psychomotor retardation, hypotonia, growth retardation, cardiomyopathy, enlarged liver, hypoplastic kidneys and elevated lactate levels in urine, plasma and cerebrospinal fluid. (ECO:0000269|PubMed:18953340, ECO:0000269|PubMed:22986587). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 3, 614053; MC5DN3 (Izole ATP Sentaz Eksikliği) (ATP5E geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053; MC5DN3 (Isolated ATP synthase deficiency) (ATP5E gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP5E	ATP5E, MC5DN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kompleks 5 (ATP Sentaz) Eksikliği, Nükleer Tip 3, 614053; MC5DN3 (Izole ATP Sentaz Eksikliği) (Prenatal) (ATP5E geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial complex V (ATP synthase) deficiency, nuclear type 3, 614053; MC5DN3 (Isolated ATP synthase deficiency) (ATP5E gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP5E	ATP5E, MC5DN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 1 (MNGIE Tip), 603041, Otozomal Resesif, MTDPS1 (Mitochondrial Neurogastrointestinal Encephalomyopathy) (TYMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041, Autosomal recessive; MTDPS1 (Mitochondrial neurogastrointestinal encephalomyopathy) (TYMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYMP	TYMP, ECGF1, MNGIE, PDECGF, MEDPS1, MTDPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 1, MNGIE type (MTDPS1) [MIM:603041]: A multisystem disease associated with mitochondrial dysfunction. It is clinically characterized by onset between the second and fifth decades of life, ptosis, progressive external ophthalmoplegia, gastrointestinal dysmotility (often pseudoobstruction), diffuse leukoencephalopathy, cachexia, peripheral neuropathy, and myopathy. (ECO:0000269|PubMed:12177387, ECO:0000269|PubMed:9924029). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 1 (MNGIE Tip), 603041, Otozomal Resesif, MTDPS1 (Mitochondrial Neurogastrointestinal Encephalomyopathy) (Prenatal) (TYMP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 1 (MNGIE type), 603041, Autosomal recessive; MTDPS1 (Mitochondrial neurogastrointestinal encephalomyopathy) (TYMP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TYMP	TYMP, ECGF1, MNGIE, PDECGF, MEDPS1, MTDPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 1, MNGIE type (MTDPS1) [MIM:603041]: A multisystem disease associated with mitochondrial dysfunction. It is clinically characterized by onset between the second and fifth decades of life, ptosis, progressive external ophthalmoplegia, gastrointestinal dysmotility (often pseudoobstruction), diffuse leukoencephalopathy, cachexia, peripheral neuropathy, and myopathy. (ECO:0000269|PubMed:12177387, ECO:0000269|PubMed:9924029). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 11, 615084, Otozomal Resesif, MTDPS11 (Progresif Eksternal Oftalmoplejі-Miyopati-Zayflama Sendromu) (MGME1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 11, 615084, Autosomal recessive; MTDPS11 (Progressive external ophthalmoplegia-myopathy-emaciation syndrome) (MGME1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MGME1	MGME1, C20orf72, MTDPS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 11 (MTDPS11) [MIM:615084]: An autosomal recessive mitochondrial disorder characterized by onset in childhood or adulthood of progressive external ophthalmoplegia, muscle weakness and atrophy, exercise intolerance, and respiratory insufficiency due to muscle weakness. More variable features include spinal deformity, emaciation, and cardiac abnormalities. Skeletal muscle biopsies show deletion and depletion of mitochondrial DNA (mtDNA) with variable defects in respiratory chain enzyme activities. (ECO:0000269	PubMed:23313956). Note=The disease may be caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 11, 615084, Otozomal Resesif, MTDPS11 (Progresif Eksternal Oftalmoplejі-Miyopati-Zayflama Sendromu) (Prenatal) (MGME1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 11, 615084, Autosomal recessive; MTDPS11 (Progressive external ophthalmoplegia-myopathy-emaciation syndrome) (MGME1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MGME1	MGME1, C20orf72, MTDPS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 11 (MTDPS11) [MIM:615084]: An autosomal recessive mitochondrial disorder characterized by onset in childhood or adulthood of progressive external ophthalmoplegia, muscle weakness and atrophy, exercise intolerance, and respiratory insufficiency due to muscle weakness. More variable features include spinal deformity, emaciation, and cardiac abnormalities. Skeletal muscle biopsies show deletion and depletion of mitochondrial DNA (mtDNA) with variable defects in respiratory chain enzyme activities. (ECO:0000269	PubMed:23313956). Note=The disease may be caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12A (Kardiyomiyopatik Tip) AD, 617184, Otozomal Dominant; MTDPS12A (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184, Autosomal dominant; MTDPS12A (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18579922). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12A (Kardiyomiyopatik Tip) AD, 617184, Otozomal Dominant; MTDPS12A (MLPA)	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184, Autosomal dominant; MTDPS12A (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18579922). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mitokondriyal DNA Deplesyon Sendromu 12A (Kardiyomiyopatik Tipi) AD, 617184, Otozomal Dominant; MTDPS12A (Prenatal) (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184, Autosomal dominant; MTDPS12A (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12A (Kardiyomiyopatik Tipi) AD, 617184, Otozomal Dominant; MTDPS12A (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 12A (cardiomyopathic type) AD, 617184, Autosomal dominant; MTDPS12A (Prenatal) (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12B (Kardiyomiyopatik Tipi) AR, 615418, Otozomal Resesif; MTDPS12B (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418, Autosomal recessive; MTDPS12B (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12B (Kardiyomiyopatik Tipi) AR, 615418, Otozomal Resesif; MTDPS12B (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (MLPA)	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418, Autosomal recessive; MTDPS12B (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12B (Kardiyomiyopatik Tipi) AR, 615418, Otozomal Resesif; MTDPS12B (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (Prenatal) (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418, Autosomal recessive; MTDPS12B (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 12B (Kardiyomiyopatik Tipi) AR, 615418, Otozomal Resesif; MTDPS12B (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 12B (cardiomyopathic type) AR, 615418, Autosomal recessive; MTDPS12B (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (Prenatal) (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 13 (Ensefalomiyopatik Tipi), 615471, Otozomal Resesif; MTDPS13 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form-Değişken Kraniofasiyal Anomaliler) (FBXL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471, Autosomal recessive; MTDPS13 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies) (FBXL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBXL4	FBXL4, FBL4, MTDPS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 13 (Ensefalomiyopatik Tipi), 615471, Otozomal Resesif; MTDPS13 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form-Değişken Kraniofasiyal Anomaliler) (Prenatal) (FBXL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 13 (encephalomyopathic type), 615471, Autosomal recessive; MTDPS13 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies) (FBXL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBXL4	FBXL4, FBL4, MTDPS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 14 (Ensefalokardiyomiyopatik Tipi), 616896; MTDPS14 (Otozomal Dominant Optik Atrofi Plus Sendromu) (OPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896; MTDPS14 (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 14 (Ensefalokardiyomiyopatik Tipi), 616896; MTDPS14 (Otozomal Dominant Optik Atrofi Plus Sendromu) (Prenatal) (OPA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type), 616896; MTDPS14 (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 15 (Hepatoserebral Tipi), 617156, Otozomal Resesif; MTDPS15 (TFAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156, Autosomal recessive; MTDPS15 (TFAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFAM	TFAM, TCF6L2, TCF6L1, TCF6L3, MTTF1, TCF6, MTDPS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 15 (Hepatoserebral Tipi), 617156, Otozomal Resesif; MTDPS15 (Prenatal) (TFAM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type), 617156, Autosomal recessive; MTDPS15 (TFAM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TFAM	TFAM, TCF6L2, TCF6L1, TCF6L3, MTTF1, TCF6, MTDPS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal DNA Deplesyon Sendromu 2 (Miyopatik Tip), 609560, Otozomal Resesif; MTDPS2 (Mitokondriyal DNA Deplesyon Sendromu, Miyopatik Şekilde) (TK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560, Autosomal recessive; MTDPS2 (Mitochondrial DNA depletion syndrome, myopathic form) (TK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TK2	TK2, MTDPS2, PEOB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 2 (Miyopatik Tip), 609560, Otozomal Resesif; MTDPS2 (Mitokondriyal DNA Deplesyon Sendromu, Miyopatik Şekilde) (MLPA)	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560, Autosomal recessive; MTDPS2 (Mitochondrial DNA depletion syndrome, myopathic form) (MLPA)	TK2	TK2, MTDPS2, PEOB3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 2 (Miyopatik Tip), 609560, Otozomal Resesif; MTDPS2 (Mitokondriyal DNA Deplesyon Sendromu, Miyopatik Şekilde) (Prenatal) (TK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560, Autosomal recessive; MTDPS2 (Mitochondrial DNA depletion syndrome, myopathic form) (TK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TK2	TK2, MTDPS2, PEOB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 2 (Miyopatik Tip), 609560, Otozomal Resesif; MTDPS2 (Mitokondriyal DNA Deplesyon Sendromu, Miyopatik Şekilde) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 2 (myopathic type), 609560, Autosomal recessive; MTDPS2 (Mitochondrial DNA depletion syndrome, myopathic form) (Prenatal) (MLPA)	TK2	TK2, MTDPS2, PEOB3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 3 (Hepatoserebral Tip), 251880, Otozomal Resesif; MTDPS3 (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (DGUOK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880, Autosomal recessive; MTDPS3 (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 3 (Hepatoserebral Tip), 251880, Otozomal Resesif; MTDPS3 (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (MLPA)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880, Autosomal recessive; MTDPS3 (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 3 (Hepatoserebral Tip), 251880, Otozomal Resesif; MTDPS3 (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (Prenatal) (DGUOK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880, Autosomal recessive; MTDPS3 (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 3 (Hepatoserebral Tip), 251880, Otozomal Resesif; MTDPS3 (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type), 251880, Autosomal recessive; MTDPS3 (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (Prenatal) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4A (Alpers Tipi), 203700, Otozomal Resesif; MTDPS4A (Alpers-Huttenlocher Sendromu) (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700, Autosomal recessive; MTDPS4A (Alpers-Huttenlocher syndrome) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4A (Alpers Tipi), 203700, Otozomal Resesif; MTDPS4A (Alpers-Huttenlocher Sendromu) (MLPA)	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700, Autosomal recessive; MTDPS4A (Alpers-Huttenlocher syndrome) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4A (Alpers Tipi), 203700, Otozomal Resesif; MTDPS4A (Alpers-Huttenlocher Sendromu) (Prenatal) (POLG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700, Autosomal recessive; MTDPS4A (Alpers-Huttenlocher syndrome) (POLG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4A (Alpers Tipi), 203700, Otozomal Resesif; MTDPS4A (Alpers-Huttenlocher Sendromu) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700, Autosomal recessive; MTDPS4A (Alpers-Huttenlocher syndrome) (Prenatal) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4B (MNGIE Tip), 613662, Otozomal Resesif; MTDPS4B (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662, Autosomal recessive; MTDPS4B (Mitochondrial neurogastrointestinal encephalomyopathy) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4B (MNGIE Tip), 613662, Otozomal Resesif; MTDPS4B (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (MLPA)	Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662, Autosomal recessive; MTDPS4B (Mitochondrial neurogastrointestinal encephalomyopathy) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mitokondriyal DNA Deplesyon Sendromu 4B (MNGIE Tipi), 613662, Otozomal Resesif; MTDPS4B (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (Prenatal) (POLG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662, Autosomal recessive; MTDPS4B (Mitochondrial neurogastrointestinal encephalomyopathy) (POLG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18579522}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 4B (MNGIE Tipi), 613662, Otozomal Resesif; MTDPS4B (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662, Autosomal recessive; MTDPS4B (Mitochondrial neurogastrointestinal encephalomyopathy) (Prenatal) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18579522}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 5 (Ensefalomiyopatik ± Metilmalonik Asidüri), 612073, Otozomal Resesif; MTDPS5 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form - Metilmalonik Asidüri) (SUCLA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073, Autosomal recessive; MTDPS5 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria) (SUCLA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUCLA2	SUCLA2, MTDPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 5 (Ensefalomiyopatik ± Metilmalonik Asidüri), 612073, Otozomal Resesif; MTDPS5 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form - Metilmalonik Asidüri) (MLPA)	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073, Autosomal recessive; MTDPS5 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria) (MLPA)	SUCLA2	SUCLA2, MTDPS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 5 (Ensefalomiyopatik ± Metilmalonik Asidüri), 612073, Otozomal Resesif; MTDPS5 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form - Metilmalonik Asidüri) (Prenatal) (SUCLA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073, Autosomal recessive; MTDPS5 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria) (SUCLA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUCLA2	SUCLA2, MTDPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 5 (Ensefalomiyopatik ± Metilmalonik Asidüri), 612073, Otozomal Resesif; MTDPS5 (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyopatik Form - Metilmalonik Asidüri) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria), 612073, Autosomal recessive; MTDPS5 (Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria) (Prenatal) (MLPA)	SUCLA2	SUCLA2, MTDPS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 6 (Hepatoserebral Tip), 256810, Otozomal Resesif; MTDPS6 (Navajo Nörohepatopati) (MPV17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810, Autosomal recessive; MTDPS6 (Navajo neurohepatopathy) (MPV17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPV17	MPV17, MTDPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 6 (Hepatoserebral Tip), 256810, Otozomal Resesif; MTDPS6 (Navajo Nörohepatopati) (MLPA)	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810, Autosomal recessive; MTDPS6 (Navajo neurohepatopathy) (MLPA)	MPV17	MPV17, MTDPS6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 6 (Hepatoserebral Tip), 256810, Otozomal Resesif; MTDPS6 (Navajo Nörohepatopati) (Prenatal) (MPV17 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810, Autosomal recessive; MTDPS6 (Navajo neurohepatopathy) (MPV17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPV17	MPV17, MTDPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 6 (Hepatoserebral Tip), 256810, Otozomal Resesif; MTDPS6 (Navajo Nörohepatopati) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 6 (hepatocerebral type), 256810, Autosomal recessive; MTDPS6 (Navajo neurohepatopathy) (Prenatal) (MLPA)	MPV17	MPV17, MTDPS6	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 7 (Hepatoserebral Tip), 271245, Otozomal Resesif; MTDPS7 (İnfantil Başlangıçlı Spinoserebellar Ataksi) (TWNK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245, Autosomal recessive; MTDPS7 (Infantile onset spinocerebellar ataxia) (TWNK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 7 (Hepatoserebral Tip), 271245, Otozomal Resesif; MTDPS7 (İnfantil Başlangıçlı Spinoserebellar Ataksi) (MLPA)	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245, Autosomal recessive; MTDPS7 (Infantile onset spinocerebellar ataxia) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 7 (Hepatoserebral Tip), 271245, Otozomal Resesif; MTDPS7 (İnfantil Başlangıçlı Spinoserebellar Ataksi) (Prenatal) (TWNK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245, Autosomal recessive; MTDPS7 (Infantile onset spinocerebellar ataxia) (TWNK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 7 (Hepatoserebral Tip), 271245, Otozomal Resesif; MTDPS7 (İnfantil Başlangıçlı Spinoserebellar Ataksi) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245, Autosomal recessive; MTDPS7 (Infantile onset spinocerebellar ataxia) (Prenatal) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8A (Tübülopati İle Ensefalomiyopatik Tip), 612075, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyelopatik Form) (RRM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075, Autosomal recessive (Mitochondrial DNA depletion syndrome, encephalomyopathic form) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8A (Tübülopati İle Ensefalomiyopatik Tip), 612075, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyelopatik Form) (MLPA)	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075, Autosomal recessive (Mitochondrial DNA depletion syndrome, encephalomyopathic form) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8A (Tübülopati İle Ensefalomiyopatik Tip), 612075, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyelopatik Form) (Prenatal) (RRM2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075, Autosomal recessive (Mitochondrial DNA depletion syndrome, encephalomyopathic form) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8A (Tübülopati İle Ensefalomiyopatik Tip), 612075, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, Ensefalomiyelopatik Form) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 8A (encephalomyopathic type with renal tubulopathy), 612075, Autosomal recessive (Mitochondrial DNA depletion syndrome, encephalomyopathic form) (Prenatal) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8B (MNGIE Tipi), 612075, Otozomal Resesif (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (RRM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075, Autosomal recessive (Mitochondrial neurogastrointestinal encephalomyopathy) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8B (MNGIE Tipi), 612075, Otozomal Resesif (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (MLPA)	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075, Autosomal recessive (Mitochondrial neurogastrointestinal encephalomyopathy) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8B (MNGIE Tipi), 612075, Otozomal Resesif (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (Prenatal) (RRM2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075, Autosomal recessive (Mitochondrial neurogastrointestinal encephalomyopathy) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 8B (MNGIE Tipi), 612075, Otozomal Resesif (Mitokondriyal Nörogastrointestinal Ensefalomiyopati) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 8B (MNGIE type), 612075, Autosomal recessive (Mitochondrial neurogastrointestinal encephalomyopathy) (Prenatal) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPS8A, MTDPS8B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPS8A) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal DNA Deplesyon Sendromu 9 (Metilmalonik Asidüri Ensefalomiyopatik Tip), 245400, Otozomal Resesif; MTDPS9 (Fatal Infantil Laktik Asidoz-Metilmalonik Asidüri) (SUCLG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400, Autosomal recessive; MTDPS9 (Fatal infantile lactic acidosis with methylmalonic aciduria) (SUCLG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUCLG1	SUCLG1, SUCLA1, MTDPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 9 (MTDPS9) [MIM:245400]: A severe disorder due to mitochondrial dysfunction. It is characterized by infantile onset of hypotonia, lactic acidosis, severe psychomotor retardation, progressive neurologic deterioration, and excretion of methylmalonic acid. {ECO:0000269|PubMed:17668387, ECO:0000269|PubMed:19526370, ECO:0000269|PubMed:20453710, ECO:0000269|PubMed:20693550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 9 (Metilmalonik Asidüri Ensefalomiyopatik Tip), 245400, Otozomal Resesif; MTDPS9 (Fatal Infantil Laktik Asidoz-Metilmalonik Asidüri) (MLPA)	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400, Autosomal recessive; MTDPS9 (Fatal infantile lactic acidosis with methylmalonic aciduria) (MLPA)	SUCLG1	SUCLG1, SUCLA1, MTDPS9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 9 (MTDPS9) [MIM:245400]: A severe disorder due to mitochondrial dysfunction. It is characterized by infantile onset of hypotonia, lactic acidosis, severe psychomotor retardation, progressive neurologic deterioration, and excretion of methylmalonic acid. {ECO:0000269|PubMed:17668387, ECO:0000269|PubMed:19526370, ECO:0000269|PubMed:20453710, ECO:0000269|PubMed:20693550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 9 (Metilmalonik Asidüri Ensefalomiyopatik Tip), 245400, Otozomal Resesif; MTDPS9 (Fatal Infantil Laktik Asidoz-Metilmalonik Asidüri) (Prenatal) (SUCLG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400, Autosomal recessive; MTDPS9 (Fatal infantile lactic acidosis with methylmalonic aciduria) (SUCLG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUCLG1	SUCLG1, SUCLA1, MTDPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 9 (MTDPS9) [MIM:245400]: A severe disorder due to mitochondrial dysfunction. It is characterized by infantile onset of hypotonia, lactic acidosis, severe psychomotor retardation, progressive neurologic deterioration, and excretion of methylmalonic acid. {ECO:0000269|PubMed:17668387, ECO:0000269|PubMed:19526370, ECO:0000269|PubMed:20453710, ECO:0000269|PubMed:20693550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Deplesyon Sendromu 9 (Metilmalonik Asidüri Ensefalomiyopatik Tip), 245400, Otozomal Resesif; MTDPS9 (Fatal Infantil Laktik Asidoz-Metilmalonik Asidüri) (Prenatal) (MLPA)	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria), 245400, Autosomal recessive; MTDPS9 (Fatal infantile lactic acidosis with methylmalonic aciduria) (Prenatal) (MLPA)	SUCLG1	SUCLG1, SUCLA1, MTDPS9	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 9 (MTDPS9) [MIM:245400]: A severe disorder due to mitochondrial dysfunction. It is characterized by infantile onset of hypotonia, lactic acidosis, severe psychomotor retardation, progressive neurologic deterioration, and excretion of methylmalonic acid. {ECO:0000269|PubMed:17668387, ECO:0000269|PubMed:19526370, ECO:0000269|PubMed:20453710, ECO:0000269|PubMed:20693550}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA deplesyon sendromu paneli (TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1) (MLPA)	Mitochondrial DNA depletion syndrome panel (TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1) (MLPA)	TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA deplesyon sendromu paneli (TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1) (MLPA) (Prenatal)	Mitochondrial DNA depletion syndrome panel (TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1) (MLPA) (Prenatal)	TK2, MPV17, DGUOK, RRM2B, SUCLA2, SUCLG1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal bakım genleri (POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)) (MLPA)	Mitochondrial maintenance genes (POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)) (MLPA)	POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal bakım genleri (POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)) (MLPA) (Prenatal)	Mitochondrial maintenance genes (POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)) (MLPA) (Prenatal)	POLG, POLG2, TWNK (C10orf2; PEO1), SLC25A4 (ANT1)	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Miyopati, Laktik Asidoz, 251950, Otozomal Resesif; MMLA (Mitokondriyal Miyopati-Laktik Asidoz-Sağırlık Sendromu) (PNPLA8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial myopathy with lactic acidosis, 251950, Autosomal recessive; MMLA (Mitochondrial myopathy-lactic acidosis-deafness syndrome) (PNPLA8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA8	PNPLA8, MMLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Miyopati, Laktik Asidoz, 251950, Otozomal Resesif; MMLA (Mitokondriyal Miyopati-Laktik Asidoz-Sağırlık Sendromu) (Prenatal) (PNPLA8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial myopathy with lactic acidosis, 251950, Autosomal recessive; MMLA (Mitochondrial myopathy-lactic acidosis-deafness syndrome) (PNPLA8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPLA8	PNPLA8, MMLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Fosfat Taşıyıcı Eksikliği, 610773 (Kardiyomyopati-Hipotoni-Laktik Asidoz Sendromu) (SLC25A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial phosphate carrier deficiency, 610773 (Cardiomyopathy-hypotonia-lactic acidosis syndrome) (SLC25A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A3	SLC25A3, PHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial phosphate carrier deficiency (MPCD) [MIM:610773]: Fatal disorder of oxidative phosphorylation. Patients have lactic acidosis, hypertrophic cardiomyopathy and muscular hypotonia and die within the first year of life. {ECO:0000269	PubMed:17273968}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Fosfat Taşıyıcı Eksikliği, 610773 (Kardiyomyopati-Hipotoni-Laktik Asidoz Sendromu) (Prenatal) (SLC25A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial phosphate carrier deficiency, 610773 (Cardiomyopathy-hypotonia-lactic acidosis syndrome) (SLC25A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A3	SLC25A3, PHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial phosphate carrier deficiency (MPCD) [MIM:610773]: Fatal disorder of oxidative phosphorylation. Patients have lactic acidosis, hypertrophic cardiomyopathy and muscular hypotonia and die within the first year of life. {ECO:0000269	PubMed:17273968}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Piruvat Taşıyıcı Eksikliği, 614741, Otozomal Resesif; MPYCD (Mitokondriyal Piruvat Taşıyıcı Eksikliği) (MPC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial pyruvate carrier deficiency, 614741, Autosomal recessive; MPYCD (Mitochondrial pyruvate carrier deficiency) (MPC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPC1	BRP44L, MPC1, MPYCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial pyruvate carrier deficiency (MPYCD) [MIM:614741]: An autosomal recessive metabolic disorder characterized by severely delayed psychomotor development, mild dysmorphic features, hepatomegaly, marked metabolic acidosis, hyperlactacidemia with normal lactate/pyruvate, and encephalopathy. Some patients have epilepsy and peripheral neuropathy. {ECO:0000269	PubMed:22628558}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mitokondriyal Piruvat Taşıyıcı Eksikliği, 614741, Otozomal Resesif; MPYCD (Mitokondriyal Piruvat Taşıyıcı Eksikliği) (Prenatal) (MPC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial pyruvate carrier deficiency, 614741, Autosomal recessive; MPYCD (Mitochondrial pyruvate carrier deficiency) (MPC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPC1	BRP44L, MPC1, MPYCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial pyruvate carrier deficiency (MPYCD) [MIM:614741]: An autosomal recessive metabolic disorder characterized by severely delayed psychomotor development, mild dysmorphic features, hepatomegaly, marked metabolic acidosis, hyperlactacidemia with normal lactate/pyruvate, and encephalopathy. Some patients have epilepsy and peripheral neuropathy. {ECO:0000269	PubMed:22628558}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Resesif Ataksi Sendromu (Sando ve SCAEy'i İçerir), 607459, Otozomal Resesif (Epilepsi-Spinoserebellar Ataksi) (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459, Autosomal recessive (Spinocerebellar ataxia with epilepsy) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Resesif Ataksi Sendromu (Sando ve SCAEy'i İçerir), 607459, Otozomal Resesif (Epilepsi-Spinoserebellar Ataksi) (MLPA)	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459, Autosomal recessive (Spinocerebellar ataxia with epilepsy) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mitokondriyal Resesif Ataksi Sendromu (Sando ve SCAEy'i İçerir), 607459, Otozomal Resesif (Epilepsi-Spinoserebellar Ataksi) (Prenatal) (POLG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459, Autosomal recessive (Spinocerebellar ataxia with epilepsy) (POLG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mitokondriyal Resesif Ataksi Sendromu (Sando ve SCAE'yi İçerir), 607459, Otozomal Resesif (Epilepsi Spinoserebellar Ataksi) (Prenatal) (MLPA)	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459, Autosomal recessive (Spinocerebellar ataxia with epilepsy) (Prenatal) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal Kısa Zincirli Enoil-CoA 1 Hidratlaz Eksikliği, 616277, Otozomal Resesif, ECHS1D (Lökodistrofi-Leigh Sendromu) (ECHS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277, Autosomal recessive; ECHS1D (Leigh syndrome with leukodystrophy) (ECHS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ECHS1	ECHS1, SCEH, ECHS1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal Kısa Zincirli Enoil-CoA 1 Hidratlaz Eksikliği, 616277, Otozomal Resesif, ECHS1D (Lökodistrofi-Leigh Sendromu) (Prenatal) (ECHS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mitochondrial short-chain enoyl-CoA hydratase 1 deficiency, 616277, Autosomal recessive; ECHS1D (Leigh syndrome with leukodystrophy) (ECHS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ECHS1	ECHS1, SCEH, ECHS1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitral Kapak Prolapsusu 2, 607829, Otozomal Dominant; MVP2 (Ailesel Mitral Kapak Prolapsusu) (DCHS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mitral valve prolapse 2, 607829, Autosomal dominant; MVP2 (Familial mitral valve prolapse) (DCHS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCHS1	DCHS1, PCDH16, FIB1, CDH19, VMLDS1, MVP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitral valve prolapse 2 (MVP2) [MIM:607829]: A form of mitral valve prolapse, a valvular hearth disease characterized by abnormally elongated and thickened mitral valve leaflets, that typically show myxomatous degeneration with increased leaflet compliance. It is associated with mitral regurgitation. Myxomatous mitral valves have an abnormal layered architecture characterized by loose collagen in fibrosa, expanded spongiosa strongly positive for proteoglycans, and disrupted elastin in atrialis. In classic mitral valve prolapse, leaflets are at least 5 mm thick, whereas in the non-classic form, they are less than 5 mm thick. Severe classic mitral valve prolapse is strongly associated with arrhythmias, endocarditis, heart failure, and need for valve surgery. {ECO:0000269	PubMed:26258302}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyoshi Müskiler Distrofi 1, 254130, Otozomal Resesif; MMD1 (Miyoshi Miyopati) (DYSF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyoshi muscular dystrophy 1, 254130, Autosomal recessive; MMD1 (Miyoshi myopathy) (DYSF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYSF	DYSF, LGMD2B, MMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvifemoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. {ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoshi Müskiler Distrofi 1, 254130, Otozomal Resesif; MMD1 (Miyoshi Miyopati) (MLPA)	Miyoshi muscular dystrophy 1, 254130, Autosomal recessive; MMD1 (Miyoshi myopathy) (MLPA)	DYSF	DYSF, LGMD2B, MMD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvifemoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. {ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoshi Müskiler Distrofi 1, 254130, Otozomal Resesif; MMD1 (Miyoshi Miyopati) (Prenatal) (DYSF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyoshi muscular dystrophy 1, 254130, Autosomal recessive; MMD1 (Miyoshi myopathy) (DYSF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYSF	DYSF, LGMD2B, MMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvifemoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. {ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyoshi Müskiler Distrofi 1, 254130, Otozomal Resesif; MMD1 (Miyoshi Miyopati) (Prenatal) (MLPA)	Miyoshi muscular dystrophy 1, 254130, Autosomal recessive; MMD1 (Miyoshi myopathy) (Prenatal) (MLPA)	DYSF	DYSF, LGMD2B, MMD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvifemoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. {ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyoshi Müskiler Distrofi 3, 613319, Otozomal Resesif; MMD3 (Distal Anoktaminopati) (ANO5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyoshi muscular dystrophy 3, 613319, Autosomal recessive; MMD3 (Distal anoctaminopathy) (ANO5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO5	ANO5, TMEM16E, GDD1, LGMD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2L (LGMD2L) [MIM:611307]: An autosomal recessive degenerative myopathy characterized by proximal weakness, weakness of the hip and shoulder girdles and prominent asymmetrical quadriceps femoris and biceps brachii atrophy. {ECO:0000269	PubMed:20096397}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoshi Müskiler Distrofi 3, 613319, Otozomal Resesif; MMD3 (Distal Anoktaminopati) (Prenatal) (ANO5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyoshi muscular dystrophy 3, 613319, Autosomal recessive; MMD3 (Distal anoctaminopathy) (ANO5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANO5	ANO5, TMEM16E, GDD1, LGMD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Limb-girdle muscular dystrophy 2L (LGMD2L) [MIM:611307]: An autosomal recessive degenerative myopathy characterized by proximal weakness, weakness of the hip and shoulder girdles and prominent asymmetrical quadriceps femoris and biceps brachii atrophy. {ECO:0000269	PubMed:20096397}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
MLL/MLLT1 t(11;19)(q23;p13.3) (FISH)	MLL/MLLT1 t(11;19)(q23;p13.3) (FISH)	11q23-19p13.3	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
MLL/MLLT3 t(9;11)(p22;q23) (FISH)	MLL/MLLT3 t(9;11)(p22;q23) (FISH)	9p22-11q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
MODY tip 1, 2, 3, 5 paneli (HNF4A, GCK, HNF1A, HNF1B) (MLPA)	MODY type 1, 2, 3, 5 panel (HNF4A, GCK, HNF1A, HNF1B) (MLPA)	HNF4A, GCK, HNF1A, HNF1B	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
MODY, Tip 1, 125850, Otozomal Dominant; MODY1 (MODY) (HNF4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	MODY, type I, 125850, Autosomal dominant; MODY1 (MODY) (HNF4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Maturity-onset diabetes of the young 1 (MODY1) [MIM:125850]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269|PubMed:17407387, ECO:0000269|PubMed:9243109, ECO:0000269|PubMed:9313765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MODY, Tip 1, 125850, Otozomal Dominant; MODY1 (MODY) (MLPA)	MODY, type I, 125850, Autosomal dominant; MODY1 (MODY) (MLPA)	HNF4A	HNF4A, TCF14, MODY1, FRTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Maturity-onset diabetes of the young 1 (MODY1) [MIM:125850]: A form of diabetes that is characterized by an autosomal dominant mode of inheritance, onset in childhood or early adulthood (usually before 25 years of age), a primary defect in insulin secretion and frequent insulin-independence at the beginning of the disease. {ECO:0000269|PubMed:17407387, ECO:0000269|PubMed:9243109, ECO:0000269|PubMed:9313765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

MODY, Tip 2, 125851, Otozomal Dominant; MODY2 (MODY) (GCK geni) (Tüm Gen Dizi Analizi) (Postnatal)	MODY, type II, 125851, Autosomal dominant; MODY2 (MODY) (GCK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCK	GCK, HHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269	PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MODY, Tip 2, 125851, Otozomal Dominant; MODY2 (MODY) (MLPA)	MODY, type II, 125851, Autosomal dominant; MODY2 (MODY) (MLPA)	GCK	GCK, HHF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 3 (HHF3) [MIM:602485]: Most common cause of persistent hypoglycemia in infancy. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. {ECO:0000269	PubMed:9435328}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MODY, Tip 3, 600496, Otozomal Dominant; MODY3 (MODY) (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	MODY, type III, 600496, Autosomal dominant; MODY3 (MODY) (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic adenomas familial (HEPAF) [MIM:142330]: Rare benign liver tumors of presumable epithelial origin that develop in an otherwise normal liver. Hepatic adenomas may be single or multiple. They consist of sheets of well-differentiated hepatocytes that contain fat and glycogen and can produce bile. Bile ducts or portal areas are absent. Kupffer cells, if present, are reduced in number and are non-functional. Conditions associated with adenomas are insulin-dependent diabetes mellitus and glycogen storage diseases (types 1 and 3). Note=The disease is caused by mutations affecting the gene represented in this entry. Bi-allelic inactivation of HNF1A, whether sporadic or associated with MODY3, may be an early step in the development of some hepatocellular carcinomas.	EDTA Blood Sample
MODY, Tip 3, 600496, Otozomal Dominant; MODY3 (MODY) (MLPA)	MODY, type III, 600496, Autosomal dominant; MODY3 (MODY) (MLPA)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hepatic adenomas familial (HEPAF) [MIM:142330]: Rare benign liver tumors of presumable epithelial origin that develop in an otherwise normal liver. Hepatic adenomas may be single or multiple. They consist of sheets of well-differentiated hepatocytes that contain fat and glycogen and can produce bile. Bile ducts or portal areas are absent. Kupffer cells, if present, are reduced in number and are non-functional. Conditions associated with adenomas are insulin-dependent diabetes mellitus and glycogen storage diseases (types 1 and 3). Note=The disease is caused by mutations affecting the gene represented in this entry. Bi-allelic inactivation of HNF1A, whether sporadic or associated with MODY3, may be an early step in the development of some hepatocellular carcinomas.	EDTA Blood Sample
MODY, Tip 4, 606392, MODY4 (MODY) (PDX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MODY, type IV, 606392, MODY4 (MODY) (PDX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDX1	PDX1, IPF1, MODY4, PAGEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Mohr-Tranebjaerg Sendromu, 304700, X'e Bağlı Resesif; MTS (Mohr-Tranebjaerg Sendromu) (TIMM8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mohr-Tranebjaerg syndrome, 304700, X-linked recessive; MTS (Mohr-Tranebjaerg syndrome) (TIMM8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIMM8A	TIMM8A, DFN1, DDP, MTS, DDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mohr-Tranebjaerg syndrome (MTS) [MIM:304700]: Recessive neurodegenerative syndrome characterized by postlingual progressive sensorineural deafness as the first presenting symptom in early childhood, followed by progressive dystonia, spasticity, dysphagia, mental deterioration, paranoia and cortical blindness. {ECO:0000269|PubMed:10878869, ECO:0000269|PubMed:11875042, ECO:0000269|PubMed:11956200}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mohr-Tranebjaerg Sendromu, 304700, X'e Bağlı Resesif; MTS (Mohr-Tranebjaerg Sendromu) (Prenatal) (TIMM8A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mohr-Tranebjaerg syndrome, 304700, X-linked recessive; MTS (Mohr-Tranebjaerg syndrome) (TIMM8A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TIMM8A	TIMM8A, DFN1, DDP, MTS, DDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mohr-Tranebjaerg syndrome (MTS) [MIM:304700]: Recessive neurodegenerative syndrome characterized by postlingual progressive sensorineural deafness as the first presenting symptom in early childhood, followed by progressive dystonia, spasticity, dysphagia, mental deterioration, paranoia and cortical blindness. {ECO:0000269|PubMed:10878869, ECO:0000269|PubMed:11875042, ECO:0000269|PubMed:11956200}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Molibden Kofaktör Eksikliği A, 252150, Otozomal Resesif; MOCODA (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (MOCS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Molybdenum cofactor deficiency A, 252150, Autosomal recessive; MOCODA (Encephalopathy due to sulfite oxidase deficiency) (MOCS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MOCS1	MOCS1, MOCODA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Molybdenum cofactor deficiency, complementation group A (MOCODA) [MIM:252150]: An autosomal recessive metabolic disorder leading to the pleiotropic loss of molybdoenzyme activities. It is clinically characterized by onset in infancy of poor feeding, intractable seizures, severe psychomotor retardation, and death in early childhood in most patients. {ECO:0000269|PubMed:12754701, ECO:0000269|PubMed:16021469, ECO:0000269|PubMed:9731530, ECO:0000269|PubMed:9921896}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Molibden Kofaktör Eksikliği A, 252150, Otozomal Resesif; MOCODA (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (Prenatal) (MOCS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Molybdenum cofactor deficiency A, 252150, Autosomal recessive; MOCODA (Encephalopathy due to sulfite oxidase deficiency) (MOCS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MOCS1	MOCS1, MOCODA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Molybdenum cofactor deficiency, complementation group A (MOCODA) [MIM:252150]: An autosomal recessive metabolic disorder leading to the pleiotropic loss of molybdoenzyme activities. It is clinically characterized by onset in infancy of poor feeding, intractable seizures, severe psychomotor retardation, and death in early childhood in most patients. {ECO:0000269|PubMed:12754701, ECO:0000269|PubMed:16021469, ECO:0000269|PubMed:9731530, ECO:0000269|PubMed:9921896}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Molibden Kofaktör Eksikliği B, 252160, Otozomal Resesif; MOCODB (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (MOCS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Molybdenum cofactor deficiency B, 252160, Autosomal recessive; MOCODB (Encephalopathy due to sulfite oxidase deficiency) (MOCS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MOCS2	MOCS2, MPTS, MOCODB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Molibden Kofaktör Eksikliği B, 252160, Otozomal Resesif; MOCODB (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (Prenatal) (MOCS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Molybdenum cofactor deficiency B, 252160, Autosomal recessive; MOCODB (Encephalopathy due to sulfite oxidase deficiency) (MOCS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MOCS2	MOCS2, MPTS, MOCODB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Molibden Kofaktör Eksikliği C 615501; MOCODC (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (GPHN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Molybdenum cofactor deficiency C, 615501; MOCODC (Encephalopathy due to sulfite oxidase deficiency) (GPHN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPHN	GPHN, GPH, KIAA1385, GEPH, MOCODC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Molybdenum cofactor deficiency, complementation group C (MOCODC) [MIM:615501]: A form of molybdenum cofactor deficiency, an autosomal recessive metabolic disorder leading to the pleiotropic loss of molybdoenzyme activities. It is clinically characterized by onset in infancy of poor feeding, intractable seizures, severe psychomotor retardation, and death in early childhood in most patients. {ECO:0000269|PubMed:11095995, ECO:0000269|PubMed:22040219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Molibden Kofaktör Eksikliği C 615501; MOCODC (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (Prenatal) (GPHN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Molybdenum cofactor deficiency C, 615501; MOCODC (Encephalopathy due to sulfite oxidase deficiency) (GPHN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPHN	GPHN, GPH, KIAA1385, GEPH, MOCODC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Molybdenum cofactor deficiency, complementation group C (MOCODC) [MIM:615501]: A form of molybdenum cofactor deficiency, an autosomal recessive metabolic disorder leading to the pleiotropic loss of molybdoenzyme activities. It is clinically characterized by onset in infancy of poor feeding, intractable seizures, severe psychomotor retardation, and death in early childhood in most patients. {ECO:0000269|PubMed:11095995, ECO:0000269|PubMed:22040219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Monilethrix, 158000, Otozomal Dominant (Monilethrix) (KRT83 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Monilethrix, 158000, Autosomal dominant (Monilethrix) (KRT83 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT83	KRT83, KRTHB3, HB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Monilethrix, 158000, Otozomal Dominant (Monilethrix) (KRTB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Monilethrix, 158000, Autosomal dominant (Monilethrix) (KRTB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRTB1	KRTB1, KRTHB1, HB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Monilethrix, 158000, Otozomal Dominant; MNLIX (Monilethrix) (KRT86 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Monilethrix, 158000, Autosomal dominant; MNLIX (Monilethrix) (KRT86 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT86	KRT86, KRTHB6, HB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Monokarboksilat Taşıyıcı 1 Eksikliği, 616095, Otozomal Resesif, Otozomal Dominant; MCT1D (Ketoasidoz, Monokarboksilat Transporter 1 Eksikliğine Bağlı) (SLC16A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Monocarboxylate transporter 1 deficiency, 616095, Autosomal recessive, Autosomal dominant; MCT1D (Ketoacidosis due to monocarboxylate transporter-1 deficiency) (SLC16A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC16A1	SLC16A1, MCT1, HHF7, MCT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hyperinsulinemic hypoglycemia 7 (HHF7) [MIM:610021]: Dominantly inherited hypoglycemic disorder characterized by inappropriate insulin secretion during anaerobic exercise or on pyruvate load. {ECO:0000269	PubMed:17701893}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Monokarboksilat Taşıyıcı 1 Eksikliği, 616095, Otozomal Resesif, Otozomal Dominant; MCT1D (Ketoasidoz, Monokarboksilat Transporter 1 Eksikliğine Bağlı) (Prenatal) (SLC16A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Monocarboxylate transporter 1 deficiency, 616095, Autosomal recessive, Autosomal dominant; MCT1D (Ketoacidosis due to monocarboxylate transporter-1 deficiency) (SLC16A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC16A1	SLC16A1, MCT1, HHF7, MCT1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial hyperinsulinemic hypoglycemia 7 (HHF7) [MIM:610021]: Dominantly inherited hypoglycemic disorder characterized by inappropriate insulin secretion during anaerobic exercise or on pyruvate load. {ECO:0000269	PubMed:17701893}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Medyan Sinirin Mononöropatisi, Hafif, 613353, Otozomal Dominant; MNMN (Charcot-Marie-Tooth Hastalığı Tip 4C) (SH3TC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mononeuropathy of the median nerve, mild, 613353, Autosomal dominant; MNMN (Charcot-Marie-Tooth disease type 4C) (SH3TC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH3TC2	SH3TC2, KIAA1985, MNMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mononeuropathy of the median nerve mild (MNMN) [MIM:613353]: A disease characterized by median nerve mononeuropathy at the wrist. The clinical presentation ranges from a mild phenotype, consistent with carpal tunnel syndrome, to a severe median nerve mononeuropathy at the wrist associated with evidence of a more widespread axonal polyneuropathy. The latter phenotype is similar to that of patients with hereditary neuropathy with liability to pressure palsies. {ECO:0000269	PubMed:20230177}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Medyan Sinirin Mononöropatisi, Hafif, 613353, Otozomal Dominant; MNMN (Charcot-Marie-Tooth Hastalığı Tip 4C) (MLPA)	Mononeuropathy of the median nerve, mild, 613353, Autosomal dominant; MNMN (Charcot-Marie-Tooth disease type 4C) (MLPA)	SH3TC2	SH3TC2, KIAA1985, MNMN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mononeuropathy of the median nerve mild (MNMN) [MIM:613353]: A disease characterized by median nerve mononeuropathy at the wrist. The clinical presentation ranges from a mild phenotype, consistent with carpal tunnel syndrome, to a severe median nerve mononeuropathy at the wrist associated with evidence of a more widespread axonal polyneuropathy. The latter phenotype is similar to that of patients with hereditary neuropathy with liability to pressure palsies. {ECO:0000269	PubMed:20230177}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Monozomi 4 (FISH)	Monosomy 4 (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow

Monozomi 5 (FISH)	Monosomy 5 (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.		Heparinized blood sample / Heparinized bone marrow
Monozomi 7/ Delesyon 7q (-7/ Del (7q)) (FISH)	Monosomy 7/ deletion 7q (-7/del7q) (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)	*	10	30	.	.	20	Postnatal	.		Heparinized blood sample / Heparinized bone marrow
Morbid Obezite VeSpermatojenik Yetmezlik, 615703, Otozomal Resesif; MOSPGF (Obezite, CEP19 Eksikliğine Bağlı) (CEP19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Morbid obesity and spermatogenic failure, 615703, Autosomal recessive; MOSPGF (Obesity due to CEP19 deficiency) (CEP19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP19	CEP19, C3orf34, MOSPGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Morbid obesity and spermatogenic failure (MOSPGF) [MIM:615703]: An autosomal recessive morbid obesity syndrome characterized by hypertension, fatty liver disease, insulin resistance, and decreased sperm counts. Variable clinical manifestations are early coronary artery disease with myocardial infarction before 45 years of age, type II diabetes mellitus, and intellectual disability. Morbid obese individuals are defined as having a BMI greater than 40. [ECO:0000269	PubMed:24268657]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Morning Glory Disk Anomalisi, 120430, Otozomal Dominant (Morning Glory Sendromu) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Morning glory disc anomaly, 120430, Autosomal dominant (Morning glory syndrome) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Morning Glory Disk Anomalisi, 120430, Otozomal Dominant (Morning Glory Sendromu) (MLPA)	Morning glory disc anomaly, 120430, Autosomal dominant (Morning glory syndrome) (MLPA)	PAX6	PAX6, AN2, MGDA, FVHL, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mozaik Variagated Anöploidi Sendromu 1, 257300, Otozomal Resesif; MVA1 (Mozaik Değişken Anöploidi Sendromu) (BUB1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mosaic variegated aneuploidy syndrome 1, 257300, Autosomal recessive; MVA1 (Mosaic variegated aneuploidy syndrome) (BUB1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BUB1B	BUB1B, BUBR1, MVA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature chromatid separation trait (PCS) [MIM:176430]: Consists of separate and splayed chromatids with discernible centromeres and involves all or most chromosomes of a metaphase. It is found in up to 2% of metaphases in cultured lymphocytes from approximately 40% of normal individuals. When PCS is present in 5% or more of cells, it is known as the heterozygous PCS trait and has no obvious phenotypic effect, although some have reported decreased fertility. Inheritance is autosomal dominant. {ECO:0000269	PubMed:16411201}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mozaik Variagated Anöploidi Sendromu 2, 614114, Otozomal Resesif; MVA2 (Mozaik Değişken Anöploidi Sendromu) (CEP57 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mosaic variegated aneuploidy syndrome 2, 614114, Autosomal recessive; MVA2 (Mosaic variegated aneuploidy syndrome) (CEP57 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP57	CEP57, PIG8, TSP57, KIAA0092, MVA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mosaic variegated aneuploidy syndrome 2 (MVA2) [MIM:614114]: A severe developmental disorder characterized by mosaic aneuploidies, predominantly trisomies and monosomies, involving multiple different chromosomes and tissues. Affected individuals typically present with severe intrauterine growth retardation and microcephaly. Eye anomalies, mild dysmorphism, variable developmental delay, and a broad spectrum of additional congenital abnormalities and medical conditions may also occur. The risk of malignancy is high, with rhabdomyosarcoma, Wilms tumor and leukemia reported in several cases. {ECO:0000269	PubMed:21552266}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mowat-Wilson Sendromu, 235730, Otozomal Dominant; MOWS (Mowat-Wilson Sendromu) (ZEB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mowat-Wilson syndrome, 235730, Autosomal dominant; MOWS (Mowat-Wilson syndrome) (ZEB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZEB2	ZEB2, ZFHX1B, SMADIP1, SIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mowat-Wilson syndrome (MOWS) [MIM:235730]: A complex developmental disorder characterized by mental retardation, delayed motor development, epilepsy, microcephaly and a wide spectrum of clinically heterogeneous features suggestive of neurocristopathies at the cephalic, cardiac, and vagal levels. Affected patients show an easily recognizable facial appearance with deep set eyes and hypertelorism, medially divergent, broad eyebrows, prominent columella, pointed chin and uplifted, notched ear lobes. Some patients manifest Hirschsprung disease. {ECO:0000269|PubMed:11448942, ECO:0000269|PubMed:12451214, ECO:0000269|PubMed:15384097, ECO:0000269|PubMed:16688751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mowat-Wilson Sendromu, 235730, Otozomal Dominant; MOWS (Mowat-Wilson Sendromu) (Prenatal) (ZEB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mowat-Wilson syndrome, 235730, Autosomal dominant; MOWS (Mowat-Wilson syndrome) (ZEB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZEB2	ZEB2, ZFHX1B, SMADIP1, SIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mowat-Wilson syndrome (MOWS) [MIM:235730]: A complex developmental disorder characterized by mental retardation, delayed motor development, epilepsy, microcephaly and a wide spectrum of clinically heterogeneous features suggestive of neurocristopathies at the cephalic, cardiac, and vagal levels. Affected patients show an easily recognizable facial appearance with deep set eyes and hypertelorism, medially divergent, broad eyebrows, prominent columella, pointed chin and uplifted, notched ear lobes. Some patients manifest Hirschsprung disease. {ECO:0000269|PubMed:11448942, ECO:0000269|PubMed:12451214, ECO:0000269|PubMed:15384097, ECO:0000269|PubMed:16688751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Moyamoya 6, Akalazyada İle 615750, Otozomal Resesif (Moyamoya Hastalığı-Erken Başlangıçlı Akalazya) (GUCY1A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Moyamoya 6 with achalasia, 615750, Autosomal recessive (Moyamoya disease with early-onset achalasia) (GUCY1A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCY1A3	GUCY1A3, GUC1A3, GUCSA3, MYMY6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Moyamoya Hastalığı 2'ye Yatkınlık, 607151; MYMY2 (Moyamoya Hastalığı) (RNF213 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Moyamoya disease 2, susceptibility to, 607151; MYMY2 (Moyamoya disease) (RNF213 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF213	RNF213, ALO17, KIAA1618, MYMY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Moyamoya disease 2 (MYMY2) [MIM:607151]: A progressive cerebral angiopathy characterized by bilateral intracranial carotid artery stenosis and telangiectatic vessels in the region of the basal ganglia. The abnormal vessels resemble a 'puff of smoke' (moyamoya) on cerebral angiogram. Affected individuals can develop transient ischemic attacks and/or cerebral infarction, and rupture of the collateral vessels can cause intracranial hemorrhage. Hemiplegia of sudden onset and epileptic seizures constitute the prevailing presentation in childhood, while subarachnoid bleeding occurs more frequently in adults. {ECO:0000269|PubMed:21048783, ECO:0000269|PubMed:21799892}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Moyamoya Hastalığı 4, 300845, X'e Bağlı Resesif (Moyamoya Anjiyopati-Boy Kısalığı-Fasiyal Dismorfizm-Hipergonadotropik Hipogonadizm Sendromu) (2200)	Moyamoya disease 4, 300845, X-linked recessive (Moyamoya angiopathy-short stature-facial dysmorphism-hypergonadotropic hypogonadism syndrome) (440)	.	MYMY4, CXDELq38	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Moyamoya Hastalığı 5, 614042; MYMY5 (Moyamoya Hastalığı) (ACTA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Moyamoya disease 5, 614042; MYMY5 (Moyamoya disease) (ACTA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA2	ACTA2, ACTSA, AAT6, MYMY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 6 (AAT6) [MIM:611788]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269|PubMed:17994018, ECO:0000269|PubMed:19409525, ECO:0000269|PubMed:19639654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
MTHFR 1298 Polimorfizmi (1298A>C) (geni) (Dizi Analizi) (Postnatal)	MTHFR 1298 polymorphism (1298A>C) (gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
MTHFR 677 Polimorfizmi (677C>T) (geni) (Dizi Analizi) (Postnatal)	MTHFR 677 polymorphism (677C>T) (gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Muckle-Wells Sendromu, 191900, Otozomal Dominant; MWS (Muckle-Wells Sendromu) (NLRP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muckle-Wells syndrome, 191900, Autosomal dominant; MWS (Muckle-Wells syndrome) (NLRP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP3	NLRP3, CIAS1, FCU, FCAS1, NALP3, PYPAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial cold autoinflammatory syndrome 1 (FCAS1) [MIM:120100]: A rare autosomal dominant systemic inflammatory disease characterized by recurrent episodes of maculopapular rash associated with arthralgias, myalgias, fever and chills, swelling of the extremities, and conjunctivitis after generalized exposure to cold. Rarely, some patients may also develop late-onset renal amyloidosis. {ECO:0000269|PubMed:11687797, ECO:0000269|PubMed:11992256, ECO:0000269|PubMed:12355493, ECO:0000269|PubMed:12522564, ECO:0000269|PubMed:15593220, ECO:0000269|PubMed:17294928, ECO:0000269|PubMed:24952504}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Muckle-Wells Sendromu, 191900, Otozomal Dominant; MWS (Muckle-Wells Sendromu) (Prenatal) (NLRP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muckle-Wells syndrome, 191900, Autosomal dominant; MWS (Muckle-Wells syndrome) (NLRP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NLRP3	NLRP3, CIAS1, FCU, FCAS1, NALP3, PYPAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial cold autoinflammatory syndrome 1 (FCAS1) [MIM:120100]: A rare autosomal dominant systemic inflammatory disease characterized by recurrent episodes of maculopapular rash associated with arthralgias, myalgias, fever and chills, swelling of the extremities, and conjunctivitis after generalized exposure to cold. Rarely, some patients may also develop late-onset renal amyloidosis. {ECO:0000269|PubMed:11687797, ECO:0000269|PubMed:11992256, ECO:0000269|PubMed:12355493, ECO:0000269|PubMed:12522564, ECO:0000269|PubMed:15593220, ECO:0000269|PubMed:17284928, ECO:0000269|PubMed:24925504}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukoepidermoid Tükürük Bezi Karsinması (MAML2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucoepidermoid salivary gland carcinoma (MAML2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAML2	MAML2, MAM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MAML2 is found in mucoepidermoid carcinomas, benign Warthin tumors and clear cell hidradenomas. Translocation t(11;19)(q21;p13) with CRTC1. The fusion protein consists of the N-terminus of CRTC1 joined to the C-terminus of MAML2. The reciprocal fusion protein consisting of the N-terminus of MAML2 joined to the C-terminus of CRTC1 has been detected in a small number of mucoepidermoid carcinomas. {ECO:0000269	PubMed:15729701}.	EDTA Blood Sample
Mukoepidermoid Tükürük Bezi Karsinması (CRTC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucoepidermoid salivary gland carcinoma (CRTC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRTC1	CRTC1, MECT1, KIAA0616, FLJ14027	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving CRTC1 is found in mucoepidermoid carcinomas, benign Warthin tumors and clear cell hidradenomas. Translocation t(11;19)(q21;p13) with MAML2. The fusion protein consists of the N-terminus of CRTC1 joined to the C-terminus of MAML2. The reciprocal fusion protein consisting of the N-terminus of MAML2 joined to the C-terminus of CRTC1 has been detected in a small number of mucoepidermoid carcinomas..	EDTA Blood Sample
Mukolipidoz 2 Alfa / Beta, 252500, Otozomal Resesif (Mukolipidoz Tip 2) (GNPTAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucolipidosis II alpha/beta, 252500, Autosomal recessive (Mucolipidosis type II) (GNPTAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNPTAB	GNPTAB, GNPTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucolipidosis type II (MLII) [MIM:252500]: Fatal, autosomal recessive, lysosomal storage disorder characterized by severe clinical and radiologic features, peculiar fibroblast inclusions, and no excessive mucopolysacchariduria. Congenital dislocation of the hip, thoracic deformities, hernia, and hyperplastic gums are evident soon after birth. {ECO:0000269|PubMed:16200072, ECO:0000269|PubMed:16835905, ECO:0000269|PubMed:19197337, ECO:0000269|PubMed:19617216, ECO:0000269|PubMed:19634183, ECO:0000269|PubMed:19938078, ECO:0000269|PubMed:22495880, ECO:0000269|PubMed:23566849, ECO:0000269|PubMed:23733939, ECO:0000269|PubMed:23773965, ECO:0000269|PubMed:24375680, ECO:0000269|PubMed:24798265, ECO:0000269|PubMed:25505245, ECO:0000269|PubMed:25788519}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukolipidoz 3 Alfa / Beta, 252600, Otozomal Resesif (Mukolipidoz Tip 3) (GNPTAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucolipidosis III alpha/beta, 252600, Autosomal recessive (Mucolipidosis type III) (GNPTAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNPTAB	GNPTAB, GNPTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucolipidosis type II (MLII) [MIM:252500]: Fatal, autosomal recessive, lysosomal storage disorder characterized by severe clinical and radiologic features, peculiar fibroblast inclusions, and no excessive mucopolysacchariduria. Congenital dislocation of the hip, thoracic deformities, hernia, and hyperplastic gums are evident soon after birth. {ECO:0000269|PubMed:16200072, ECO:0000269|PubMed:16835905, ECO:0000269|PubMed:19197337, ECO:0000269|PubMed:19617216, ECO:0000269|PubMed:19634183, ECO:0000269|PubMed:19938078, ECO:0000269|PubMed:22495880, ECO:0000269|PubMed:23566849, ECO:0000269|PubMed:23733939, ECO:0000269|PubMed:23773965, ECO:0000269|PubMed:24375680, ECO:0000269|PubMed:24798265, ECO:0000269|PubMed:25505245, ECO:0000269|PubMed:25788519}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukolipidoz 3 Alfa / Beta, 252600, Otozomal Resesif (Mukolipidoz Tip 3) (Prenatal) (GNPTAB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucolipidosis III alpha/beta, 252600, Autosomal recessive (Mucolipidosis type III) (GNPTAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNPTAB	GNPTAB, GNPTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucolipidosis type II (MLII) [MIM:252500]: Fatal, autosomal recessive, lysosomal storage disorder characterized by severe clinical and radiologic features, peculiar fibroblast inclusions, and no excessive mucopolysacchariduria. Congenital dislocation of the hip, thoracic deformities, hernia, and hyperplastic gums are evident soon after birth. {ECO:0000269|PubMed:16200072, ECO:0000269|PubMed:16835905, ECO:0000269|PubMed:19197337, ECO:0000269|PubMed:19617216, ECO:0000269|PubMed:19634183, ECO:0000269|PubMed:19938078, ECO:0000269|PubMed:22495880, ECO:0000269|PubMed:23566849, ECO:0000269|PubMed:23733939, ECO:0000269|PubMed:23773965, ECO:0000269|PubMed:24375680, ECO:0000269|PubMed:24798265, ECO:0000269|PubMed:25505245, ECO:0000269|PubMed:25788519}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukolipidoz 3 Gama, 252605, Otozomal Resesif (Mukolipidoz Tip 3) (GNPTG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucolipidosis III gamma, 252605, Autosomal recessive (Mucolipidosis type III) (GNPTG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNPTG	GNPTAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucolipidosis type III complementation group C (ML IIIC) [MIM:252605]: Autosomal recessive disease of lysosomal hydrolase trafficking. Unlike the related diseases, mucolipidosis II and IIIA, the enzyme affected in mucolipidosis IIIC (GlcNAc- phosphotransferase) retains full transferase activity on synthetic substrates but lacks activity on lysosomal hydrolases. Typical clinical findings include stiffness of the hands and shoulders, claw-hand deformity, scoliosis, short stature, coarse facies, and mild mental retardation. Radiographically, severe dysostosis multiplex of the hip is characteristic and frequently disabling. The clinical diagnosis can be confirmed by finding elevated serum lysosomal enzyme levels and/or decreased lysosomal enzyme levels in cultured fibroblasts. {ECO:0000269|PubMed:10712439, ECO:0000269|PubMed:15532026, ECO:0000269|PubMed:24316125}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukolipidoz 3 Gama, 252605, Otozomal Resesif (Mukolipidoz Tip 3) (Prenatal) (GNPTG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucolipidosis III gamma, 252605, Autosomal recessive (Mucolipidosis type III) (GNPTG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNPTG	GNPTAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucolipidosis type III complementation group C (ML IIIC) [MIM:252605]: Autosomal recessive disease of lysosomal hydrolase trafficking. Unlike the related diseases, mucolipidosis II and IIIA, the enzyme affected in mucolipidosis IIIC (GlcNAc- phosphotransferase) retains full transferase activity on synthetic substrates but lacks activity on lysosomal hydrolases. Typical clinical findings include stiffness of the hands and shoulders, claw-hand deformity, scoliosis, short stature, coarse facies, and mild mental retardation. Radiographically, severe dysostosis multiplex of the hip is characteristic and frequently disabling. The clinical diagnosis can be confirmed by finding elevated serum lysosomal enzyme levels and/or decreased lysosomal enzyme levels in cultured fibroblasts. {ECO:0000269|PubMed:10712439, ECO:0000269|PubMed:15532026, ECO:0000269|PubMed:24316125}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukolipidoz 4, 252650, Otozomal Resesif; ML4 (Mukolipidoz Tip 4) (MCOLN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucolipidosis IV, 252650, Autosomal recessive; ML4 (Mucolipidosis type IV) (MCOLN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCOLN1	MCOLN1, ML4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mukolipidoz 4, 252650, Otozomal Resesif; ML4 (Mukolipidoz Tip 4) (Prenatal) (MCOLN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucolipidosis IV, 252650, Autosomal recessive; ML4 (Mucolipidosis type IV) (MCOLN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCOLN1	MCOLN1, ML4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Ih 607014, Otozomal Resesif (Mukopolisakaridozisis Tip 1), MPS1 (IDUA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis Ih, 607014, Autosomal recessive (Mucopolysaccharidosis type 1), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15309847, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7502232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mukopolisakkaridoz IH 607014, Otozomal Resesif (Mukopolisakaridozisis Tip 1), MPS1 (Prenatal) (IDUA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis Ih, 607014, Autosomal recessive (Mucopolysaccharidosis type 1), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz 1HS, 607015, Otozomal Resesif (Mukopolisakaridozisis Tip 1) (Hurler-Scheie Sendromu), MPS1 (IDUA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis Ih/s, 607015, Autosomal recessive (Mucopolysaccharidosis type 1) (HURLER-SCHEIE SYNDROME), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukopolisakkaridoz 1HS, 607015, Otozomal Resesif (Mukopolisakaridozisis Tip 1) (Hurler-Scheie Sendromu), MPS1 (Prenatal) (IDUA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis Ih/s, 607015, Autosomal recessive (Mucopolysaccharidosis type 1) (HURLER-SCHEIE SYNDROME), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300947, ECO:0000269|PubMed:21394825, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz 2 309900, X'e Bağlı; MPS2 (Mukopolisakaridozisis Tip 2)(Hunter sendromu) (IDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis II, 309900, X-linked recessive; MPS2 (Mucopolysaccharidosis type 2) (Hunter syndrome) (IDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDS	IDS, MPS2, SIDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 2 (MPS2) [MIM:309900]: An X-linked lysosomal storage disease characterized by intracellular accumulation of heparan sulfate and dermatan sulfate and their excretion in urine. Most children with MPS2 have a severe form with early somatic abnormalities including skeletal deformities, hepatosplenomegaly, and progressive cardiopulmonary deterioration. A prominent feature is neurological damage that presents as developmental delay and hyperactivity but progresses to mental retardation and dementia. They die before 15 years of age, usually as a result of obstructive airway disease or cardiac failure. In contrast, those with a mild form of MPS2 may survive into adulthood, with attenuated somatic complications and often without mental retardation. {ECO:0000269|PubMed:10215411, ECO:0000269|PubMed:10220152, ECO:0000269|PubMed:10447264, ECO:0000269|PubMed:10671065, ECO:0000269|PubMed:11015461, ECO:0000269|PubMed:11683780, ECO:0000269|PubMed:11731225, ECO:0000269|PubMed:12655569, ECO:0000269|PubMed:12794697, ECO:0000269|PubMed:1284597, ECO:0000269|PubMed:1303211, ECO:0000269|PubMed:16699754, ECO:0000269|PubMed:7581397, ECO:0000269|PubMed:7599640, ECO:0000269|PubMed:7728156, ECO:0000269|PubMed:7866405, ECO:0000269|PubMed:7887413, ECO:0000269|PubMed:7981716, ECO:0000269|PubMed:8281149,	EDTA Blood Sample
Mukopolisakkaridoz 2 309900, X'e Bağlı; MPS2 (Mukopolisakaridozisis Tip 2)(Hunter sendromu) (MLPA)	Mucopolysaccharidosis II, 309900, X-linked recessive; MPS2 (Mucopolysaccharidosis type 2) (Hunter syndrome) (MLPA)	IDS	IDS, MPS2, SIDS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mucopolysaccharidosis 2 (MPS2) [MIM:309900]: An X-linked lysosomal storage disease characterized by intracellular accumulation of heparan sulfate and dermatan sulfate and their excretion in urine. Most children with MPS2 have a severe form with early somatic abnormalities including skeletal deformities, hepatosplenomegaly, and progressive cardiopulmonary deterioration. A prominent feature is neurological damage that presents as developmental delay and hyperactivity but progresses to mental retardation and dementia. They die before 15 years of age, usually as a result of obstructive airway disease or cardiac failure. In contrast, those with a mild form of MPS2 may survive into adulthood, with attenuated somatic complications and often without mental retardation. {ECO:0000269|PubMed:10215411, ECO:0000269|PubMed:10220152, ECO:0000269|PubMed:10447264, ECO:0000269|PubMed:10671065, ECO:0000269|PubMed:11015461, ECO:0000269|PubMed:11683780, ECO:0000269|PubMed:11731225, ECO:0000269|PubMed:12655569, ECO:0000269|PubMed:12794697, ECO:0000269|PubMed:1284597, ECO:0000269|PubMed:1303211, ECO:0000269|PubMed:16699754, ECO:0000269|PubMed:7581397, ECO:0000269|PubMed:7599640, ECO:0000269|PubMed:7728156, ECO:0000269|PubMed:7866405, ECO:0000269|PubMed:7887413, ECO:0000269|PubMed:7981716, ECO:0000269|PubMed:8281149,	EDTA Blood Sample
Mukopolisakkaridoz 2 309900, X'e Bağlı; MPS2 (Mukopolisakaridozisis Tip 2)(Hunter sendromu) (IDS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis II, 309900, X-linked recessive; MPS2 (Mucopolysaccharidosis type 2) (Hunter syndrome) (IDS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDS	IDS, MPS2, SIDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 2 (MPS2) [MIM:309900]: An X-linked lysosomal storage disease characterized by intracellular accumulation of heparan sulfate and dermatan sulfate and their excretion in urine. Most children with MPS2 have a severe form with early somatic abnormalities including skeletal deformities, hepatosplenomegaly, and progressive cardiopulmonary deterioration. A prominent feature is neurological damage that presents as developmental delay and hyperactivity but progresses to mental retardation and dementia. They die before 15 years of age, usually as a result of obstructive airway disease or cardiac failure. In contrast, those with a mild form of MPS2 may survive into adulthood, with attenuated somatic complications and often without mental retardation. {ECO:0000269|PubMed:10215411, ECO:0000269|PubMed:10220152, ECO:0000269|PubMed:10447264, ECO:0000269|PubMed:10671065, ECO:0000269|PubMed:11015461, ECO:0000269|PubMed:11683780, ECO:0000269|PubMed:11731225, ECO:0000269|PubMed:12655569, ECO:0000269|PubMed:12794697, ECO:0000269|PubMed:1284597, ECO:0000269|PubMed:1303211, ECO:0000269|PubMed:16699754, ECO:0000269|PubMed:7581397, ECO:0000269|PubMed:7599640, ECO:0000269|PubMed:7728156, ECO:0000269|PubMed:7866405, ECO:0000269|PubMed:7887413, ECO:0000269|PubMed:7981716, ECO:0000269|PubMed:8281149,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Mukopolisakkaridoz 2 309900, X'e Bağlı; MPS2 (Mukopolisakaridozis Tip 2)(Hunter sendromu) (Prenatal) (MLPA)	Mucopolysaccharidosis II, 309900, X-linked recessive; MPS2 (Mucopolysaccharidosis type 2) (Hunter syndrome) (Prenatal) (MLPA)	IDS	IDS, MPS2, SIDS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 2 (MPS2) [MIM:309900]: An X-linked lysosomal storage disease characterized by intracellular accumulation of heparan sulfate and dermatan sulfate and their excretion in urine. Most children with MPS2 have a severe form with early somatic abnormalities including skeletal deformities, hepatosplenomegaly, and progressive cardiopulmonary deterioration. A prominent feature is neurological damage that presents as developmental delay and hyperactivity but progresses to mental retardation and dementia. They die before 15 years of age, usually as a result of obstructive airway disease or cardiac failure. In contrast, those with a mild form of MPS2 may survive into adulthood, with attenuated somatic complications and often without mental retardation. {ECO:0000269|PubMed:10215411, ECO:0000269|PubMed:10220152, ECO:0000269|PubMed:10447254, ECO:0000269|PubMed:10671065, ECO:0000269|PubMed:11015461, ECO:0000269|PubMed:11683780, ECO:0000269|PubMed:11731225, ECO:0000269|PubMed:12655569, ECO:0000269|PubMed:12794697, ECO:0000269|PubMed:1284597, ECO:0000269|PubMed:1303211, ECO:0000269|PubMed:16699754, ECO:0000269|PubMed:7581397, ECO:0000269|PubMed:7599640, ECO:0000269|PubMed:7728156, ECO:0000269|PubMed:7866405, ECO:0000269|PubMed:7937413, ECO:0000269|PubMed:7981716, ECO:0000269|PubMed:8281149,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz, 607016, Otozomal Resesif (Mukopolisakaridozisis Tip 1s) (Scheie Sendromu), MPS1 (IDUA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis Is, 607016, Autosomal recessive (Mucopolysaccharidosis type 1s) (SCHEIE SYNDROME), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300847, ECO:0000269|PubMed:2139482S, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukopolisakkaridoz, 607016, Otozomal Resesif (Mukopolisakaridozisis Tip 1s) (Scheie Sendromu), MPS1 (Prenatal) (IDUA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis Is, 607016, Autosomal recessive (Mucopolysaccharidosis type 1s) (SCHEIE SYNDROME), MPS1 (IDUA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IDUA	IDUA, IDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 1H (MPS1H) [MIM:607014]: A severe form of mucopolysaccharidosis type 1, a rare lysosomal storage disease characterized by progressive physical deterioration with urinary excretion of dermatan sulfate and heparan sulfate. Patients with MPS1H usually present, within the first year of life, a combination of hepatosplenomegaly, skeletal deformities, corneal clouding and severe mental retardation. Obstructive airways disease, respiratory infection and cardiac complications usually result in death before 10 years of age. {ECO:0000269|PubMed:10466419, ECO:0000269|PubMed:10735634, ECO:0000269|PubMed:12559846, ECO:0000269|PubMed:1301941, ECO:0000269|PubMed:15300847, ECO:0000269|PubMed:2139482S, ECO:0000269|PubMed:7550232, ECO:0000269|PubMed:7550242, ECO:0000269|PubMed:7951228, ECO:0000269|PubMed:8019563, ECO:0000269|PubMed:8328452, ECO:0000269|PubMed:8401515, ECO:0000269|Ref.20}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz 4A, 253000, Otozomal Resesif; MPS4A (Mukopolisakkaridoz Tip 4) (GALNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis IVA, 253000, Autosomal recessive; MPS4A (Mucopolysaccharidosis type 4) (GALNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALNS	GALNS, MPS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 4A (MPS4A) [MIM:253000]: A form of mucopolysaccharidosis type 4, an autosomal recessive lysosomal storage disease characterized by intracellular accumulation of keratan sulfate and chondroitin-6-sulfate. Key clinical features include short stature, skeletal dysplasia, dental anomalies, and corneal clouding. Intelligence is normal and there is no direct central nervous system involvement, although the skeletal changes may result in neurologic complications. There is variable severity, but patients with the severe phenotype usually do not survive past the second or third decade of life. {ECO:0000269|PubMed:1522213, ECO:0000269|PubMed:16287098, ECO:0000269|PubMed:24726177, ECO:0000269|PubMed:7581409, ECO:0000269|PubMed:7633425, ECO:0000269|PubMed:7668283, ECO:0000269|PubMed:7795586, ECO:0000269|PubMed:8651279, ECO:0000269|PubMed:8826435, ECO:0000269|PubMed:9298823, ECO:0000269|PubMed:9375852, ECO:0000269|PubMed:9521421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mukopolisakkaridoz 4A, 253000, Otozomal Resesif; MPS4A (Mukopolisakkaridoz Tip 4) (Prenatal) (GALNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis IVA, 253000, Autosomal recessive; MPS4A (Mucopolysaccharidosis type 4) (GALNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GALNS	GALNS, MPS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 4A (MPS4A) [MIM:253000]: A form of mucopolysaccharidosis type 4, an autosomal recessive lysosomal storage disease characterized by intracellular accumulation of keratan sulfate and chondroitin-6-sulfate. Key clinical features include short stature, skeletal dysplasia, dental anomalies, and corneal clouding. Intelligence is normal and there is no direct central nervous system involvement, although the skeletal changes may result in neurologic complications. There is variable severity, but patients with the severe phenotype usually do not survive past the second or third decade of life. {ECO:0000269|PubMed:1522213, ECO:0000269|PubMed:16287098, ECO:0000269|PubMed:24726177, ECO:0000269|PubMed:7581409, ECO:0000269|PubMed:7633425, ECO:0000269|PubMed:7668283, ECO:0000269|PubMed:7795586, ECO:0000269|PubMed:8651279, ECO:0000269|PubMed:8826435, ECO:0000269|PubMed:9298823, ECO:0000269|PubMed:9375852, ECO:0000269|PubMed:9521421}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Tip 3A (Sanfilippo A), 252900, Otozomal Resesif; MPS3A (Mukopolisakkaridozis Tip 3) (SGSH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900, Autosomal recessive; MPS3A (Mucopolysaccharidosis type 3) (SGSH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGSH	SGSH, MPS3A, SFMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 3A (MPS3A) [MIM:252900]: A severe form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. MPS3A is characterized by earlier onset, rapid progression of symptoms and shorter survival. {ECO:0000269|PubMed:11182930, ECO:0000269|PubMed:11793481, ECO:0000269|PubMed:12000360, ECO:0000269|PubMed:12702166, ECO:0000269|PubMed:15146460, ECO:0000269|PubMed:15637719, ECO:0000269|PubMed:15902564, ECO:0000269|PubMed:16311287, ECO:0000269|PubMed:17128482, ECO:0000269|PubMed:18407553, ECO:0000269|PubMed:21671392, ECO:0000269|PubMed:9158154, ECO:0000269|PubMed:9285796, ECO:0000269|PubMed:9401012, ECO:0000269|PubMed:9554748, ECO:0000269|PubMed:9744479}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Mukopolisakkaridoz Tip 3A (Sanfilippo A), 252900, Otozomal Resesif; MPS3A (Mukopolisakkaridozis Tip 3) (Prenatal) (SGSH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IIIA (Sanfilippo A), 252900, Autosomal recessive; MPS3A (Mucopolysaccharidosis type 3) (SGSH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SGSH	SGSH, MPS3A, SFMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 3A (MPS3A) [MIM:252900]: A severe form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. MPS3A is characterized by earlier onset, rapid progression of symptoms and shorter survival. {ECO:0000269|PubMed:11182930, ECO:0000269|PubMed:11793481, ECO:0000269|PubMed:12000360, ECO:0000269|PubMed:12702166, ECO:0000269|PubMed:15146460, ECO:0000269|PubMed:15637719, ECO:0000269|PubMed:15902564, ECO:0000269|PubMed:16311287, ECO:0000269|PubMed:17128482, ECO:0000269|PubMed:18407553, ECO:0000269|PubMed:21671382, ECO:0000269|PubMed:9158154, ECO:0000269|PubMed:9285796, ECO:0000269|PubMed:9401012, ECO:0000269|PubMed:9554748, ECO:0000269|PubMed:9744479}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridozis Tip 3B (Sanfilippo B), 252920, Otozomal Resesif; MPS3B (Mukopolisakkaridozis Tip 3) (NAGLU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920, Autosomal recessive; MPS3B (Mucopolysaccharidosis type 3) (NAGLU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGLU	NAGLU, MPS3B, CMT2V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2V (CMT2V) [MIM:616491]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. CMT2V is an autosomal dominant sensory neuropathy with late onset. The main clinical feature is recurrent leg pain that progresses to constant painful paraesthesias in the feet and later the hands. As it evolves, some patients develop a mild sensory ataxia. {ECO:0000269	PubMed:25818867}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mukopolisakkaridozis Tip 3B (Sanfilippo B), 252920, Otozomal Resesif; MPS3B (Mukopolisakkaridozis Tip 3) (Prenatal) (NAGLU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IIIB (Sanfilippo B), 252920, Autosomal recessive; MPS3B (Mucopolysaccharidosis type 3) (NAGLU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAGLU	NAGLU, MPS3B, CMT2V	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Charcot-Marie-Tooth disease 2V (CMT2V) [MIM:616491]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. CMT2V is an autosomal dominant sensory neuropathy with late onset. The main clinical feature is recurrent leg pain that progresses to constant painful paraesthesias in the feet and later the hands. As it evolves, some patients develop a mild sensory ataxia. {ECO:0000269	PubMed:25818867}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Tip 3C (Sanfilippo C), 252930, Otozomal Resesif; MPS3C (Mukopolisakkaridozis Tip 3) (HGSNAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930, Autosomal recessive; MPS3C (Mucopolysaccharidosis type 3) (HGSNAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HGSNAT	HGSNAT, TMEM76, MPS3C, RP73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 3C (MPS3C) [MIM:252930]: A form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. {ECO:0000269|PubMed:16960811, ECO:0000269|PubMed:17033958, ECO:0000269|PubMed:17397050, ECO:0000269|PubMed:18024218, ECO:0000269|PubMed:19479962}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mukopolisakkaridoz Tip 3C (Sanfilippo C), 252930, Otozomal Resesif; MPS3C (Mukopolisakkaridozis Tip 3) (Prenatal) (HGSNAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IIIC (Sanfilippo C), 252930, Autosomal recessive; MPS3C (Mucopolysaccharidosis type 3) (HGSNAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HGSNAT	HGSNAT, TMEM76, MPS3C, RP73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 3C (MPS3C) [MIM:252930]: A form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. {ECO:0000269|PubMed:16960811, ECO:0000269|PubMed:17033958, ECO:0000269|PubMed:17397050, ECO:0000269|PubMed:18024218, ECO:0000269|PubMed:19479962}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridozis (MSP 3), Otozomal Resesif 3D, 252940 Tip; MPS3D (Mukopolisakkaridozis Tip 3) (GNS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IIID, 252940, Autosomal recessive; MPS3D (Mucopolysaccharidosis type 3) (GNS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNS	GNS, G6S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 3D (MPS3D) [MIM:252940]: A form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. {ECO:0000269|PubMed:12573255, ECO:0000269|PubMed:20232353}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mukopolisakkaridozis (MSP 3), Otozomal Resesif 3D, 252940 Tip; MPS3D (Mukopolisakkaridozis Tip 3) (Prenatal) (GNS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IIID, 252940, Autosomal recessive; MPS3D (Mucopolysaccharidosis type 3) (GNS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNS	GNS, G6S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 3D (MPS3D) [MIM:252940]: A form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. {ECO:0000269|PubMed:12573255, ECO:0000269|PubMed:20232353}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Tip 4B (Morquio), 253010, Otozomal Resesif; MPS4B (Mukopolisakkaridoz Tip 4) (GLB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IVB (Morquio), 253010, Autosomal recessive; MPS4B (Mucopolysaccharidosis type 4) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM1-gangliosidosis 1 (GM1G1) [MIM:230500]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM1 gangliosides, glycoproteins and keratan sulfate primarily in neurons of the central nervous system. GM1-gangliosidosis type 1 is characterized by onset within the first three months of life, central nervous system degeneration, coarse facial features, hepatosplenomegaly, skeletal dysmorphology reminiscent of Hurler syndrome, and rapidly progressive psychomotor deterioration. Urinary oligosaccharide levels are high. It leads to death usually between the first and second year of life. {ECO:0000269|PubMed:10338095, ECO:0000269|PubMed:10737981, ECO:0000269|PubMed:10839995, ECO:0000269|PubMed:1487238, ECO:0000269|PubMed:15365997, ECO:0000269|PubMed:15714521, ECO:0000269|PubMed:15791924, ECO:0000269|PubMed:16538002, ECO:0000269|PubMed:16941474, ECO:0000269|PubMed:17309651, ECO:0000269|PubMed:1907800, ECO:0000269|PubMed:1909089, ECO:0000269|PubMed:1928092, ECO:0000269|PubMed:19472408, ECO:0000269|PubMed:25936995, ECO:0000269|PubMed:8213816, ECO:0000269|Ref.24, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Mukopolisakkaridoz Tip 4B (Morquio), 253010, Otozomal Resesif; MPS4B (Mukopolisakkaridoz Tip 4) (Prenatal) (GLB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IVB (Morquio), 253010, Autosomal recessive; MPS4B (Mucopolysaccharidosis type 4) (GLB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLB1	GLB1, MPS4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM1-gangliosidosis 1 (GM1G1) [MIM:230500]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM1 gangliosides, glycoproteins and keratan sulfate primarily in neurons of the central nervous system. GM1-gangliosidosis type 1 is characterized by onset within the first three months of life, central nervous system degeneration, coarse facial features, hepatosplenomegaly, skeletal dysmorphology reminiscent of Hurler syndrome, and rapidly progressive psychomotor deterioration. Urinary oligosaccharide levels are high. It leads to death usually between the first and second year of life. {ECO:0000269|PubMed:10338095, ECO:0000269|PubMed:10737981, ECO:0000269|PubMed:10839995, ECO:0000269|PubMed:1487238, ECO:0000269|PubMed:15365997, ECO:0000269|PubMed:15714521, ECO:0000269|PubMed:15791924, ECO:0000269|PubMed:16538002, ECO:0000269|PubMed:16941474, ECO:0000269|PubMed:17309651, ECO:0000269|PubMed:1907800, ECO:0000269|PubMed:1909089, ECO:0000269|PubMed:1928092, ECO:0000269|PubMed:19472468, ECO:0000269|PubMed:25936995, ECO:0000269|PubMed:8213816, ECO:0000269|Ref.24, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakaridozis Tip 1X, 601492, Otozomal Resesif; MPS9 (Hyalüronidaz Eksikliği) (HYAL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type IX, 601492, Autosomal recessive; MPS9 (Hyaluronidase deficiency) (HYAL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HYAL1	HYAL1, MPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mukopolisakaridozis Tip 1X, 601492, Otozomal Resesif; MPS9 (Hyalüronidaz Eksikliği) (Prenatal) (HYAL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type IX, 601492, Autosomal recessive; MPS9 (Hyaluronidase deficiency) (HYAL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HYAL1	HYAL1, MPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Tip 6 (Maroteaux-Lamy), 253200, Otozomal Resesif; MPS6 (Mukopolisakkaridozsis Tip 6) (ARSB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200, Autosomal recessive; MPS6 (Mucopolysaccharidosis type 6) (ARSB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARSB	ARSB, MPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 6 (MPS6) [MIM:253200]: An autosomal recessive lysosomal storage disease characterized by intracellular accumulation of dermatan sulfate. Clinical features can include abnormal growth, short stature, stiff joints, skeletal malformations, corneal clouding, hepatosplenomegaly, and cardiac abnormalities. A wide variation in clinical severity is observed. {ECO:0000269|PubMed:10036316, ECO:0000269|PubMed:10738004, ECO:0000269|PubMed:11802522, ECO:0000269|PubMed:14974081, ECO:0000269|PubMed:1550123, ECO:0000269|PubMed:1718978, ECO:0000269|PubMed:8116615, ECO:0000269|PubMed:8125475, ECO:0000269|PubMed:8541342, ECO:0000269|PubMed:8651289}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mukopolisakkaridoz Tip 6 (Maroteaux-Lamy), 253200, Otozomal Resesif; MPS6 (Mukopolisakkaridozsis Tip 6) (Prenatal) (ARSB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis type VI (Maroteaux-Lamy), 253200, Autosomal recessive; MPS6 (Mucopolysaccharidosis type 6) (ARSB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARSB	ARSB, MPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 6 (MPS6) [MIM:253200]: An autosomal recessive lysosomal storage disease characterized by intracellular accumulation of dermatan sulfate. Clinical features can include abnormal growth, short stature, stiff joints, skeletal malformations, corneal clouding, hepatosplenomegaly, and cardiac abnormalities. A wide variation in clinical severity is observed. {ECO:0000269|PubMed:10036316, ECO:0000269|PubMed:10738004, ECO:0000269|PubMed:11802522, ECO:0000269|PubMed:14974081, ECO:0000269|PubMed:1550123, ECO:0000269|PubMed:1718978, ECO:0000269|PubMed:8116615, ECO:0000269|PubMed:8125475, ECO:0000269|PubMed:8541342, ECO:0000269|PubMed:8651289}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz 7 253220, Otozomal Resesif; MPS7 (Mukopolisakkaridoz Tip 7, MPS7) (GUSB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis VII, 253220, Autosomal recessive; MPS7 (Mucopolysaccharidosis type 7) (GUSB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUSB	GUSB, MPS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 7 (MPS7) [MIM:253220]: An autosomal recessive lysosomal storage disease characterized by inability to degrade glucuronic acid-containing glycosaminoglycans. The phenotype is highly variable, ranging from severe lethal hydrops fetalis to mild forms with survival into adulthood. Most patients with the intermediate phenotype show hepatomegaly, skeletal anomalies, coarse facies, and variable degrees of mental impairment. {ECO:0000269|PubMed:12522561, ECO:0000269|PubMed:12859417, ECO:0000269|PubMed:1702266, ECO:0000269|PubMed:7573038, ECO:0000269|PubMed:7633414, ECO:0000269|PubMed:7680524, ECO:0000269|PubMed:8089138, ECO:0000269|PubMed:8111412, ECO:0000269|PubMed:8111413, ECO:0000269|PubMed:8644704, ECO:0000269|PubMed:8707294, ECO:0000269|PubMed:9099834, ECO:0000269|PubMed:9490302}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Mukopolisakkaridoz 7 253220, Otozomal Resesif; MPS7 (Mukopolisakkaridoz Tip 7, MPS7) (Prenatal) (GUSB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis VII, 253220, Autosomal recessive; MPS7 (Mucopolysaccharidosis type 7) (GUSB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GUSB	GUSB, MPS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mucopolysaccharidosis 7 (MPS7) [MIM:253220]: An autosomal recessive lysosomal storage disease characterized by inability to degrade glucuronic acid-containing glycosaminoglycans. The phenotype is highly variable, ranging from severe lethal hydrops fetalis to mild forms with survival into adulthood. Most patients with the intermediate phenotype show hepatomegaly, skeletal anomalies, coarse facies, and variable degrees of mental impairment. {ECO:0000269|PubMed:12522561, ECO:0000269|PubMed:12859417, ECO:0000269|PubMed:1702266, ECO:0000269|PubMed:7573038, ECO:0000269|PubMed:7633414, ECO:0000269|PubMed:7680524, ECO:0000269|PubMed:8089138, ECO:0000269|PubMed:8111412, ECO:0000269|PubMed:8111413, ECO:0000269|PubMed:8644704, ECO:0000269|PubMed:8707294, ECO:0000269|PubMed:9099834, ECO:0000269|PubMed:9490302}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mukopolisakkaridoz Arti Sendromu, 617303, Otozomal Resesif (VPS33A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mucopolysaccharidosis-plus syndrome, 617303, Autosomal recessive (VPS33A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS33A	VPS33A, MPSPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Mukopolisakkaridoz Arti Sendromu, 617303, Otozomal Resesif (Prenatal) (VPS33A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mucopolysaccharidosis-plus syndrome, 617303, Autosomal recessive (VPS33A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS33A	VPS33A, MPSPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Muenke Sendromu, 602849, Otozomal Dominant; MNKES (Muenke Sendromu) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muenke syndrome, 602849, Autosomal dominant; MNKES (Muenke syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Muenke Sendromu, 602849, Otozomal Dominant; MNKES (Muenke Sendromu) (MLPA)	Muenke syndrome, 602849, Autosomal dominant; MNKES (Muenke syndrome) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Muenke Sendromu, 602849, Otozomal Dominant; MNKES (Muenke Sendromu) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muenke syndrome, 602849, Autosomal dominant; MNKES (Muenke syndrome) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Muenke Sendromu, 602849, Otozomal Dominant; MNKES (Muenke Sendromu) (Prenatal) (MLPA)	Muenke syndrome, 602849, Autosomal dominant; MNKES (Muenke syndrome) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Muir-Torre Sendromu, 158320, Otozomal Dominant (Muir-Torre Sendromu) (MSH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muir-Torre syndrome, 158320, Autosomal dominant (Muir-Torre syndrome) (MSH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSH2	MSH2, COCA1, FCC1, HNPCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. (ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11920458,	EDTA Blood Sample
Muir-Torre Sendromu, 158320, Otozomal Dominant (Muir-Torre Sendromu) (MLH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muir-Torre syndrome, 158320, Autosomal dominant (Muir-Torre syndrome) (MLH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MLH1	MLH1, COCA2, HNPCC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muir-Torre syndrome (MRTES) [MIM:158320]: Rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy. (ECO:0000269	PubMed:8751876). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Muir-Torre Sendromu, 158320, Otozomal Dominant (Muir-Torre Sendromu) (MLPA)	Muir-Torre syndrome, 158320, Autosomal dominant (Muir-Torre syndrome) (MLPA)	MSH2	MSH2, COCA1, FCC1, HNPCC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hereditary non-polyposis colorectal cancer 1 (HNPCC1) [MIM:120435]: An autosomal dominant disease associated with marked increase in cancer susceptibility. It is characterized by a familial predisposition to early-onset colorectal carcinoma (CRC) and extra-colonic tumors of the gastrointestinal, urological and female reproductive tracts. HNPCC is reported to be the most common form of inherited colorectal cancer in the Western world. Clinically, HNPCC is often divided into two subgroups. Type I is characterized by hereditary predisposition to colorectal cancer, a young age of onset, and carcinoma observed in the proximal colon. Type II is characterized by increased risk for cancers in certain tissues such as the uterus, ovary, breast, stomach, small intestine, skin, and larynx in addition to the colon. Diagnosis of classical HNPCC is based on the Amsterdam criteria: 3 or more relatives affected by colorectal cancer, one a first degree relative of the other two; 2 or more generation affected; 1 or more colorectal cancers presenting before 50 years of age; exclusion of hereditary polyposis syndromes. The term 'suspected HNPCC' or 'incomplete HNPCC' can be used to describe families who do not or only partially fulfill the Amsterdam criteria, but in whom a genetic basis for colon cancer is strongly suspected. (ECO:0000269|PubMed:10375096, ECO:0000269|PubMed:10386556, ECO:0000269|PubMed:10528862, ECO:0000269|PubMed:10573010, ECO:0000269|PubMed:10612836, ECO:0000269|PubMed:10777691, ECO:0000269|PubMed:10829038, ECO:0000269|PubMed:11726306, ECO:0000269|PubMed:11870161, ECO:0000269|PubMed:11920458,	EDTA Blood Sample
Muir-Torre Sendromu, 158320, Otozomal Dominant (Muir-Torre Sendromu) (MLPA)	Muir-Torre syndrome, 158320, Autosomal dominant (Muir-Torre syndrome) (MLPA)	MLH1	MLH1, COCA2, HNPCC2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muir-Torre syndrome (MRTES) [MIM:158320]: Rare autosomal dominant disorder characterized by sebaceous neoplasms and visceral malignancy. (ECO:0000269	PubMed:8751876). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mulibrey Nanizm, 253250, Otozomal Resesif (MULIBREY Nanizmi) (Prenatal) (TRIM37 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mulibrey nanism, 253250, Autosomal recessive (MULIBREY nanism) (TRIM37 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIM37	TRIM37, MUL, KIAA0898	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mulibrey nanism (MUL) [MIM:253250]: An autosomal recessive growth disorder characterized by severe growth failure of prenatal onset, constrictive pericardium and progressive cardiomyopathy, facial dysmorphism, and failure of sexual maturation. Additional clinical features include hepatomegaly, muscle hypotonia, J-shaped sella turcica, yellowish dots in the ocular fundi, hypoplasia of various endocrine glands, insulin resistance with type 2 diabetes, and an increased risk for Wilms' tumor. (ECO:0000269|PubMed:10888877, ECO:0000269|PubMed:12754710, ECO:0000269|PubMed:15108285, ECO:0000269|PubMed:15885686, ECO:0000269|PubMed:17100991, ECO:0000269|PubMed:17551331, ECO:0000269|PubMed:21865362, ECO:0000269|PubMed:23385855). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mulibrey Nanizm, 253250, Otozomal Resesif (MULIBREY Nanizmi) (Prenatal) (TRIM37 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Mulibrey nanism, 253250, Autosomal recessive (MULIBREY nanism) (TRIM37 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIM37	TRIM37, MUL, KIAA0898	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Mulibrey nanism (MUL) [MIM:253250]: An autosomal recessive growth disorder characterized by severe growth failure of prenatal onset, constrictive pericardium and progressive cardiomyopathy, facial dysmorphism, and failure of sexual maturation. Additional clinical features include hepatomegaly, muscle hypotonia, J-shaped sella turcica, yellowish dots in the ocular fundi, hypoplasia of various endocrine glands, insulin resistance with type 2 diabetes, and an increased risk for Wilms' tumor. (ECO:0000269|PubMed:10888877, ECO:0000269|PubMed:12754710, ECO:0000269|PubMed:15108285, ECO:0000269|PubMed:15885686, ECO:0000269|PubMed:17100991, ECO:0000269|PubMed:17551331, ECO:0000269|PubMed:21865362, ECO:0000269|PubMed:23385855). Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müllerian Aplazi ve Hiperandrojenizm, 158330, Otozomal Dominant (Müller Aplazisi ve Hiperandrogenizmi) (WNT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mullerian aplasia and hyperandrogenism, 158330, Autosomal dominant (Müllerian aplasia and hyperandrogenism) (WNT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT4	WNT4, SERKAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Müllerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Müllerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Müllerian Aplazi ve Hiperandrojenizm, 158330, Otozomal Dominant (Müller Aplazisi ve Hiperandrogenizmi) (MLPA)	Mullerian aplasia and hyperandrogenism, 158330, Autosomal dominant (Müllerian aplasia and hyperandrogenism) (MLPA)	WNT4	WNT4, SERKAL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Müllerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Müllerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Multisentrik Karpotarsal Osteoliz Sendromu, 166300, Otozomal Dominant; MCTO (Multisentrik Karpo-Tarsal Osteoliz ± Nefropati) (MAFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multicentric carpotarsal osteolysis syndrome, 166300, Autosomal dominant; MCTO (Multicentric carpo-tarsal osteolysis with or without nephropathy) (MAFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAFB	MAFB, KRML, MCTO, DURS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multicentric carpotarsal osteolysis syndrome (MCTO) [MIM:166300]: A rare skeletal disorder, usually presenting in early childhood with a clinical picture mimicking juvenile rheumatoid arthritis. Progressive destruction of the carpal and tarsal bone usually occurs and other bones may also be involved. Chronic renal failure is a frequent component of the syndrome. Mental retardation and minor facial anomalies have been noted in some patients. [ECO:0000269	PubMed:22387013]. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Multisentrik Osteoliz, Nodüloz ve Artropati, 259600, Otozomal Resesif; MONA (Torg-Winchester Sendromu) (MMP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multicentric osteolysis, nodulosis, and arthropathy, 259600, Autosomal recessive; MONA (Torg-Winchester syndrome) (MMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP2	MMP2, CLG4A, MONA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multicentric osteolysis, nodulosis, and arthropathy (MONA) [MIM:259600]: An autosomal recessive syndrome characterized by severe multicentric osteolysis with predominant involvement of the hands and feet. Additional features include coarse face, corneal opacities, patches of thickened, hyperpigmented skin, hypertrichosis and gum hypertrophy. (ECO:0000269|PubMed:11431697, ECO:0000269|PubMed:15691365, ECO:0000269|PubMed:16542393). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Multipl Myeloma Panel (Geniş Panel) • CKS1B/CDKN2C (P18) (1q21/ 1p32.3) • Amplifikasyon/Delesyon, • IGH (14q32) breakapart, • t(11;14) (q13;q32) (IgH/ CCND1), • t(11;14) (IGH/ MYEOV), • P53 Gen (TP53) (17p13.1), • t(4;14) (IgH/ FGRF3), • t(14;16) (IGH/MAF), • 9, 11 ve 15 anöploidi, • 13q14.3 Delesyonu (RB1) (FISH)	Multipl Myeloma Panel (Advanced Panel) • CKS1B/CDKN2C (P18) (1q21/ 1p32.3) • Amplification/Deletion, • IGH (14q32) breakapart, • t(11;14) (q13;q32) (IgH/ CCND1), • t(11;14) (IGH/ MYEOV), • P53 Gene (TP53) (17p13.1), • t(4;14) (IgH/ FGRF3), • t(14;16) (IGH/MAF), • 9, 11 and 15 aneuploidy, • 13q14.3 Deletion (RB1) (FISH) (PANEL)	"CKS1B/CDKN2C (P18) (1q21/ 1p32.3) • IGH (14q32) • t(11;14) (q13;q32) (IgH/ CCND1), • t(11;14) (IGH/ MYEOV), • P53 (TP53) (17p13.1), • t(4;14) (IgH/ FGRF3), • t(14;16) (IGH/MAF), • chr 9, 11, 15, • 13q14.3 "	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample / Heparinized bone marrow
Multipl Konjenital Anomali-Hipotoni-Nöbetler Sendromu 1, 614080, Otozomal Resesif; MCAHS1 (Multipl Konjenital Anomaliler-Hipotoni-Nöbet Sendromu) (PIGN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080, Autosomal recessive; MCAHS1 (Multiple congenital anomalies-hypotonia-seizures syndrome) (PIGN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGN	PIGN, MCAHS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) [MIM:614080]: An autosomal recessive disorder characterized by neonatal hypotonia, lack of psychomotor development, seizures, dysmorphic features, and variable congenital anomalies involving the cardiac, urinary, and gastrointestinal systems. Most affected individuals die before 3 years of age. (ECO:0000269	PubMed:21493957). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Multipl Konjenital Anomali-Hipotoni-Nöbetler Sendromu 1, 614080, Otozomal Resesif; MCAHS1 (Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu) (Prenatal) (PIGN gen) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080, Autosomal recessive; MCAHS1 (Multiple congenital anomalies-hypotonia-seizures syndrome) (PIGN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGN	PIGN, MCAHS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) [MIM:614080]: An autosomal recessive disorder characterized by neonatal hypotonia, lack of psychomotor development, seizures, dysmorphic features, and variable congenital anomalies involving the cardiac, urinary, and gastrointestinal systems. Most affected individuals die before 3 years of age. {ECO:0000269|PubMed:21493957}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu 2, 300868, X'e Bağlı Resesif; MCAHS2 (Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu Tip 2) (PIGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868, X-linked recessive; MCAHS2 (Multiple congenital anomalies-hypotonia-seizures syndrome type 2) (PIGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGA	PIGA, PNH1, MCAHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paroxysmal nocturnal hemoglobinuria 1 (PNH1) [MIM:308818]: A disorder characterized by hemolytic anemia with hemoglobinuria, thromboses in large vessels, and a deficiency in hematopoiesis. Red blood cell breakdown with release of hemoglobin into the urine is manifested most prominently by dark-colored urine in the morning. {ECO:0000269|PubMed:10087994, ECO:0000269|PubMed:12037021, ECO:0000269|PubMed:8167330, ECO:0000269|PubMed:8306954, ECO:0000269|PubMed:8500164}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu 2, 300868, X'e Bağlı Resesif; MCAHS2 (Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu Tip 2) (Prenatal) (PIGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868, X-linked recessive; MCAHS2 (Multiple congenital anomalies-hypotonia-seizures syndrome type 2) (PIGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGA	PIGA, PNH1, MCAHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paroxysmal nocturnal hemoglobinuria 1 (PNH1) [MIM:308818]: A disorder characterized by hemolytic anemia with hemoglobinuria, thromboses in large vessels, and a deficiency in hematopoiesis. Red blood cell breakdown with release of hemoglobin into the urine is manifested most prominently by dark-colored urine in the morning. {ECO:0000269|PubMed:10087994, ECO:0000269|PubMed:12037021, ECO:0000269|PubMed:8167330, ECO:0000269|PubMed:8306954, ECO:0000269|PubMed:8500164}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu 3, 615398, Otozomal Resesif; MCAHS3 (Mental Retardasyon-Nöbet-Hipotoni-Oftalmolojik-Iskelet Anomalileri Sendromu) (PIGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398, Autosomal recessive; MCAHS3 (Intellectual disability-seizures-hypotonia-ophthalmologic-skeletal anomalies syndrome) (PIGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGT	PIGT, NDAP, PNH2, MCAHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple congenital anomalies-hypotonia-seizures syndrome 3 (MCAHS3) [MIM:615398]: An autosomal recessive syndrome characterized by distinct facial features, intellectual disability, hypotonia and seizures, in combination with abnormal skeletal, endocrine, and ophthalmologic findings including impaired vision, as well as abnormal motility of the eyes. {ECO:0000269|PubMed:23636107}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Konjenital Anomaller-Hipotoni-Nöbet Sendromu 3, 615398, Otozomal Resesif; MCAHS3 (Mental Retardasyon-Nöbet-Hipotoni-Oftalmolojik-Iskelet Anomalileri Sendromu) (Prenatal) (PIGT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple congenital anomalies-hypotonia-seizures syndrome 3, 615398, Autosomal recessive; MCAHS3 (Intellectual disability-seizures-hypotonia-ophthalmologic-skeletal anomalies syndrome) (PIGT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIGT	PIGT, NDAP, PNH2, MCAHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple congenital anomalies-hypotonia-seizures syndrome 3 (MCAHS3) [MIM:615398]: An autosomal recessive syndrome characterized by distinct facial features, intellectual disability, hypotonia and seizures, in combination with abnormal skeletal, endocrine, and ophthalmologic findings including impaired vision, as well as abnormal motility of the eyes. {ECO:0000269|PubMed:23636107}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Endokrin Neoplazi 1, 131100, Otozomal Dominant; MEN1 (Multipl Endokrin Neoplazi Tip 1) (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple endocrine neoplasia 1, 131100, Autosomal dominant; MEN1 (Multiple endocrine neoplasia type 1) (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial isolated hyperparathyroidism (FIHP) [MIM:145000]: Autosomal dominant disorder characterized by hypercalcemia, elevated parathyroid hormone (PTH) levels, and unilgandular or multiglandular parathyroid tumors. {ECO:0000269|PubMed:10634381, ECO:0000269|PubMed:10664521, ECO:0000269|PubMed:12016470, ECO:0000269|PubMed:12699448, ECO:0000269|PubMed:9792884, ECO:0000269|PubMed:9843042, ECO:0000269|PubMed:9888389}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Endokrin Neoplazi 1, 131100, Otozomal Dominant; MEN1 (Multipl Endokrin Neoplazi Tip 1) (MLPA)	Multiple endocrine neoplasia 1, 131100, Autosomal dominant; MEN1 (Multiple endocrine neoplasia type 1) (MLPA)	MEN1	MEN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial isolated hyperparathyroidism (FIHP) [MIM:145000]: Autosomal dominant disorder characterized by hypercalcemia, elevated parathyroid hormone (PTH) levels, and unilgandular or multiglandular parathyroid tumors. {ECO:0000269|PubMed:10634381, ECO:0000269|PubMed:10664521, ECO:0000269|PubMed:12016470, ECO:0000269|PubMed:12699448, ECO:0000269|PubMed:9792884, ECO:0000269|PubMed:9843042, ECO:0000269|PubMed:9888389}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Endokrin Neoplazi 2A, 171400, Otozomal Dominant; MEN2A (Multipl Endokrin Neoplazi Tip 2) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple endocrine neoplasia IIA, 171400, Autosomal dominant; MEN2A (Multiple endocrine neoplasia type 2) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Endokrin Neoplazi 2A, 171400, Otozomal Dominant; MEN2A (Multipl Endokrin Neoplazi Tip 2) (MLPA)	Multiple endocrine neoplasia IIA, 171400, Autosomal dominant; MEN2A (Multiple endocrine neoplasia type 2) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Endokrin Neoplazi IIB, 162300, Otozomal Dominant (Multipl Endokrin Neoplazi Tip 2) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple endocrine neoplasia IIB, 162300, Autosomal dominant (Multiple endocrine neoplasia type 2) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Endokrin Neoplazi IIB, 162300, Otozomal Dominant (Multipl Endokrin Neoplazi Tip 2) (MLPA)	Multiple endocrine neoplasia IIB, 162300, Autosomal dominant (Multiple endocrine neoplasia type 2) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl endokrin neoplazi paneli (AIP 11q13, MEN1 11q13, CDKN1B 12p13) (MLPA)	Multiple endocrine neoplasia panel (AIP 11q13, MEN1 11q13, CDKN1B 12p13) (MLPA)	AIP 11q13, MEN1 11q13, CDKN1B 12p13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Multipl Endokrin Neoplazi, Tip 2b; MEN2B (Multipl Endokrin Neoplazi Tip 2) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB; MEN2B (Multiple endocrine neoplasia type 2) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Endokrin Neoplazi, Tip 2b; MEN2B (Multipl Endokrin Neoplazi Tip 2) (MLPA)	MULTIPLE ENDOCRINE NEOPLASIA, TYPE IIB; MEN2B (Multiple endocrine neoplasia type 2) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multiple Endokrin Neoplazi, Tip 4, 610755, Otozomal Dominant; MEN4 (Multipl Endokrin Neoplazi Tip 4) (CDKN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple endocrine neoplasia, type IV, 610755, Autosomal dominant; MEN4 (Multiple endocrine neoplasia type 4) (CDKN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN1B	CDKN1B, KIP1, CDKN4, MEN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple endocrine neoplasia 4 (MEN4) [MIM:610755]: Multiple endocrine neoplasia (MEN) syndromes are inherited cancer syndromes of the thyroid. MEN4 is a MEN-like syndrome with a phenotypic overlap of both MEN1 and MEN2. {ECO:0000269|PubMed:17030811}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multiple Endokrin Neoplazi, Tip 4, 610755, Otozomal Dominant; MEN4 (Multipl Endokrin Neoplazi Tip 4) (CDKN1B geni) (MLPA)	Multiple endocrine neoplasia, type IV, 610755, Autosomal dominant; MEN4 (Multiple endocrine neoplasia type 4) (MLPA)	CDKN1B	CDKN1B, KIP1, CDKN4, MEN4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Multiple endocrine neoplasia 4 (MEN4) [MIM:610755]: Multiple endocrine neoplasia (MEN) syndromes are inherited cancer syndromes of the thyroid. MEN4 is a MEN-like syndrome with a phenotypic overlap of both MEN1 and MEN2. {ECO:0000269|PubMed:17030811}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl ekzostoz paneli (EXT1 8q24, EXT2 11p11) (MLPA)	Multiple exocytosis panel (EXT1 8q24, EXT2 11p11) (MLPA)	EXT1 8q24, EXT2 11p11	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Multipl Meme Fibroadenomu, 615554, Otozomal Dominant; MFAB (Memenin Multipl Fibroadenomu) (PRLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple fibroadenomas of the breast, 615554, Autosomal dominant; MFAB (Multiple fibroadenoma of the breast) (PRLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRLR	PRLR, MFAB, HPRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple fibroadenomas of the breast (MFAB) [MIM:615554]: A benign breast disease marked by lobuloalveolar growth with abnormally high proliferation of the epithelium, and characterized by the presence of more than 3 fibroadenomas in one breast. Fibroadenomas are adenomas containing fibrous tissue. {ECO:0000269|PubMed:18779591}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Eklem Çıkıkları, Kısa Boy, Kraniofasiyal Dismorfizm, e Konjenital Kalp Kusurları, 245600, Otozomal Resesif; JDSCD (Larsen Benzeri Sendrom, B3GAT3 Tipi) (B3GAT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600, Autosomal recessive; JDSCD (Larsen-like syndrome, B3GAT3 type) (B3GAT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GAT3	B3GAT3, GLCAT1, JDSCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple joint dislocations short stature craniofacial dysmorphism and congenital heart defects (JDSSDHD) [MIM:245600]: An autosomal recessive disease characterized by dysmorphic facies, bilateral dislocations of the elbows, hips, and knees, clubfeet, and short stature, as well as cardiovascular defects. {ECO:0000269|PubMed:21763480}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Multipl Eklem Çıkıkları, Kısa Boy, Kraniofasiyal Dismorfizm, ± Konjenital Kalp Kusurları, 245600, Otozomal Resesif; JDSCD (Larsen Benzeri Sendrom, B3GAT3 Tipi) (Prenatal) (B3GAT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects, 245600, Autosomal recessive; JDSCD (Larsen-like syndrome, B3GAT3 type) (B3GAT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B3GAT3	B3GAT3, GLCATI, JDSCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple joint dislocations short stature craniofacial dysmorphism and congenital heart defects (JDSSDHD) [MIM:245600]: An autosomal recessive disease characterized by dysmorphic facies, bilateral dislocations of the elbows, hips, and knees, clubfeet, and short stature, as well as cardiovascular defects. (ECO:0000269	PubMed:21763480). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 1, 605711, Otozomal Resesif; MMDS1 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 1) (NFU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple mitochondrial dysfunctions syndrome 1, 605711, Autosomal recessive; MMDS1 (Fatal multiple mitochondrial dysfunctions syndrome type 1) (NFU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFU1	NFU1, HIRIP, MMDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple mitochondrial dysfunctions syndrome 1 (MMDS1) [MIM:605711]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. Some patients show failure to thrive, pulmonary hypertension, hypotonia and irritability. Biochemical features include severe combined deficiency of the 2-oxoacid dehydrogenases, defective lipoic acid synthesis and reduction in activity of mitochondrial respiratory chain complexes. (ECO:0000269|PubMed:21944046, ECO:0000269|PubMed:22077971). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 1, 605711, Otozomal Resesif; MMDS1 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 1) (Prenatal) (NFU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple mitochondrial dysfunctions syndrome 1, 605711, Autosomal recessive; MMDS1 (Fatal multiple mitochondrial dysfunctions syndrome type 1) (NFU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFU1	NFU1, HIRIP, MMDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple mitochondrial dysfunctions syndrome 1 (MMDS1) [MIM:605711]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. Some patients show failure to thrive, pulmonary hypertension, hypotonia and irritability. Biochemical features include severe combined deficiency of the 2-oxoacid dehydrogenases, defective lipoic acid synthesis and reduction in activity of mitochondrial respiratory chain complexes. (ECO:0000269|PubMed:21944046, ECO:0000269|PubMed:22077971). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 2, Hiperglisinemi, 614299, Otozomal Resesif; MMDS2 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 2) (BOLA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299, Autosomal recessive; MMDS2 (Fatal multiple mitochondrial dysfunctions syndrome type 2) (BOLA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BOLA3	BOLA3, MMDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 2, Hiperglisinemi, 614299, Otozomal Resesif; MMDS2 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 2) (Prenatal) (BOLA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple mitochondrial dysfunctions syndrome 2 with hyperglycinemia, 614299, Autosomal recessive; MMDS2 (Fatal multiple mitochondrial dysfunctions syndrome type 2) (BOLA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BOLA3	BOLA3, MMDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 3, 615330, Otozomal Resesif; MMDS3 (Fatal Multipl Mitokondriyal Disfonksiyon Sendromu Tip 3) (IBA57 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple mitochondrial dysfunctions syndrome 3, 615330, Autosomal recessive; MMDS3 (Fatal multiple mitochondrial dysfunctions syndrome type 3) (IBA57 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IBA57	IBA57, C1orf69, MMDS3, SPG74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 74, autosomal recessive (SPG74) [MIM:616451]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG74 is characterized by a combination of spastic paraplegia, optic atrophy, and peripheral neuropathy with childhood-onset and slow progression into late adulthood. (ECO:0000269	PubMed:25609768). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 3, 615330, Otozomal Resesif; MMDS3 (Fatal Multipl Mitokondriyal Disfonksiyon Sendromu Tip 3) (Prenatal) (IBA57 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple mitochondrial dysfunctions syndrome 3, 615330, Autosomal recessive; MMDS3 (Fatal multiple mitochondrial dysfunctions syndrome type 3) (IBA57 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IBA57	IBA57, C1orf69, MMDS3, SPG74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 74, autosomal recessive (SPG74) [MIM:616451]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG74 is characterized by a combination of spastic paraplegia, optic atrophy, and peripheral neuropathy with childhood-onset and slow progression into late adulthood. (ECO:0000269	PubMed:25609768). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 4, 616370, Otozomal Resesif; MMDS4 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 4) (ISCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple mitochondrial dysfunctions syndrome 4, 616370, Autosomal recessive; MMDS4 (Fatal multiple mitochondrial dysfunctions syndrome type 4) (ISCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ISCA2	ISCA2, MMDS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple mitochondrial dysfunctions syndrome 4 (MMDS4) [MIM:616370]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. (ECO:0000269	PubMed:25539947). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Mitokondriyal İşlev Bozukluğu Sendromu 4, 616370, Otozomal Resesif; MMDS4 (Fatal Multipl Mitokondriyal İşlev Bozukluğu Sendromu Tip 4) (Prenatal) (ISCA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple mitochondrial dysfunctions syndrome 4, 616370, Autosomal recessive; MMDS4 (Fatal multiple mitochondrial dysfunctions syndrome type 4) (ISCA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ISCA2	ISCA2, MMDS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple mitochondrial dysfunctions syndrome 4 (MMDS4) [MIM:616370]: A severe disorder of systemic energy metabolism, resulting in weakness, respiratory failure, lack of neurologic development, lactic acidosis, hyperglycinemia and early death. (ECO:0000269	PubMed:25539947). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Myeloma, Direnç, 254500, Somatik Mutasyon (Multipl Miyelom) (LIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple myeloma, resistance to, 254500, Somatic mutation (Multiple myeloma) (LIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIG4	LIG4, LIG4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Multipl Myeloma, Yatkınlık, 254500, Somatik Mutasyon (CCND1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple myeloma, susceptibility to, 254500, Somatic mutation (CCND1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCND1	CCND1, PRAD1, BCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple myeloma (MM) [MIM:254500]: A malignant tumor of plasma cells usually arising in the bone marrow and characterized by diffuse involvement of the skeletal system, hyperglobulinemia, Bence-Jones proteinuria and anemia. Complications of multiple myeloma are bone pain, hypercalcemia, renal failure and spinal cord compression. The aberrant antibodies that are produced lead to impaired humoral immunity and patients have a high prevalence of infection. Amyloidosis may develop in some patients. Multiple myeloma is part of a spectrum of diseases ranging from monoclonal gammopathy of unknown significance (MGUS) to plasma cell leukemia. (ECO:0000269	PubMed:8695815). Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration involving CCND1 is found in multiple myeloma. Translocation t(11;14)(q13;q32) with the IgH locus.;	EDTA blood sample / EDTA bone marrow
Multipl Prerjum Sendromu, Letal Tip, 253290, Otozomal Resesif (Letal Multipl Pterjum Sendromu) (CHRNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive (Lethal multiple pterygium syndrome) (CHRNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNG	CHRNG, ACHRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. (ECO:0000269|PubMed:16826520, ECO:0000269|PubMed:16826531). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Prerjum Sendromu, Letal Tip, 253290, Otozomal Resesif (Letal Multipl Pterjum Sendromu) (CHRND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive (Lethal multiple pterygium syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. (ECO:0000269	PubMed:18252226). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Prerjum Sendromu, Letal Tip, 253290, Otozomal Resesif (Letal Multipl Pterjum Sendromu) (Prenatal) (CHRNG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive (Lethal multiple pterygium syndrome) (CHRNG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNG	CHRNG, ACHRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. (ECO:0000269|PubMed:16826520, ECO:0000269|PubMed:16826531). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Prerjum Sendromu, Letal Tip, 253290, Otozomal Resesif (Letal Multipl Pterjum Sendromu) (Prenatal) (CHRND geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive (Lethal multiple pterygium syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. (ECO:0000269	PubMed:18252226). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Multipl Pterjium Sendromu, Letal Tip, 253290, Otozomal Resesif; LMPS (Letal Multipl Pterjium Sendromu) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive; LMPS (Lethal multiple pterygium syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Pterjium Sendromu, Letal Tip, 253290, Otozomal Resesif; LMPS (Letal Multipl Pterjium Sendromu) (Prenatal) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple pterygium syndrome, lethal type, 253290, Autosomal recessive; LMPS (Lethal multiple pterygium syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Skleroz, Hastalık Progresyonu ve Modifiye Edici, 126200, Multifaktoryel (PDCD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple sclerosis, disease progression, modifier of, 126200, Multifactorial (PDCD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDCD1	PDCD1, SLEB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 2 (SLEB2) [MIM:605218]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:12402038}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Skleroz, 1; Yatkınlık, 126200, Multifaktöryel (HLA-DRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple sclerosis, susceptibility to, 1, 126200, Multifactorial (HLA-DRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DRB1	HLA-DRB1, SS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Multipl Skleroz, 1; Yatkınlık, 126200, Multifaktöryel (Sporadik Creutzfeldt-Jakob Hastalığı) (HLA-DQB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple sclerosis, susceptibility to, 1, 126200, Multifactorial (Sporadic Creutzfeldt-Jakob disease) (HLA-DQB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DQB1	HLA-DQB1, CELIAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Multipl Skleroz, Yatkınlık, 5, 614810; MS5 (TNFRSF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple sclerosis, susceptibility to, 5, 614810; MS5 (TNFRSF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF1A	TNFRSF1A, TNFR1, TNFAR, FPF, MS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hibernian fever (FHF) [MIM:142680]: A hereditary periodic fever syndrome characterized by recurrent fever, abdominal pain, localized tender skin lesions and myalgia. Reactive amyloidosis is the main complication and occurs in 25% of cases. {ECO:0000269|PubMed:10199409, ECO:0000269|PubMed:10902757, ECO:0000269|PubMed:11443543, ECO:0000269|PubMed:13130484, ECO:0000269|PubMed:14610673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Kendi İyileşen Skuamöz Epiteliyoma, Yatkınlık, 132800, Otozomal Dominant; MSSE (Multipl Kendiliğinden İyileşen Skuamöz Epiteliyoma) (TGFBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple self-healing squamous epithelioma, susceptibility to, 132800, Autosomal dominant; MSSE (Multiple self-healing squamous epithelioma) (TGFBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGFBR1	TGFBR1, ALK5, AAT5, LDS1, MSSE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple self-healing squamous epithelioma (MSSE) [MIM:132800]: A disorder characterized by multiple skin tumors that undergo spontaneous regression. Tumors appear most often on sun-exposed regions, are locally invasive, and undergo spontaneous resolution over a period of months leaving pitted scars. {ECO:0000269|PubMed:21358634}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Sülfataz Eksikliği, 272200, Otozomal Resesif; MSD (Multipl Sülfataz Eksikliği) (SUMF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple sulfatase deficiency, 272200, Autosomal recessive; MSD (Multiple sulfatase deficiency) (SUMF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUMF1	SUMF1, FGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple sulfatase deficiency (MSD) [MIM:272200]: A clinically and biochemically heterogeneous disorder caused by the simultaneous impairment of all sulfatases, due to defective post- translational modification and activation. It combines features of individual sulfatase deficiencies such as metachromatic leukodystrophy, mucopolysaccharidosis, chondrodysplasia punctata, hydrocephalus, ichthyosis, neurologic deterioration and developmental delay. {ECO:0000269|PubMed:12757706, ECO:0000269|PubMed:12757706, ECO:0000269|PubMed:15144642}. Note=The disease is caused by mutations affecting the gene represented in this entry. SUMF1 mutations result in defective post-translational modification of sulfatases. {ECO:0000269|PubMed:12757706}.;	EDTA Blood Sample
Multipl Sülfataz Eksikliği, 272200, Otozomal Resesif; MSD (Multipl Sülfataz Eksikliği) (Prenatal) (SUMF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Multiple sulfatase deficiency, 272200, Autosomal recessive; MSD (Multiple sulfatase deficiency) (SUMF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUMF1	SUMF1, FGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Multiple sulfatase deficiency (MSD) [MIM:272200]: A clinically and biochemically heterogeneous disorder caused by the simultaneous impairment of all sulfatases, due to defective post- translational modification and activation. It combines features of individual sulfatase deficiencies such as metachromatic leukodystrophy, mucopolysaccharidosis, chondrodysplasia punctata, hydrocephalus, ichthyosis, neurologic deterioration and developmental delay. {ECO:0000269|PubMed:12757706, ECO:0000269|PubMed:12757706, ECO:0000269|PubMed:15144642}. Note=The disease is caused by mutations affecting the gene represented in this entry. SUMF1 mutations result in defective post-translational modification of sulfatases. {ECO:0000269|PubMed:12757706}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Multipl Sinostozis Sendromu 1, 186500, Otozomal Dominant; SYNS1 (Multipl Sinostozis Sendromu) (NOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple synostoses syndrome 1, 186500, Autosomal dominant; SYNS1 (Multiple synostoses syndrome) (NOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOG	NOG, SYM1, SYNS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stapes ankylosis with broad thumb and toes (SABTS) [MIM:184460]: Congenital autosomal dominant disorder that includes hypermobile, a hemicylindrical nose, broad thumbs, great toes, and other minor skeletal anomalies but lacked carpal and tarsal fusion and symphalangism. {ECO:0000269|PubMed:12089654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Sinostozis Sendromu 2, 610017, Otozomal Dominant; SYNS2 (Multipl Sinostozis Sendromu) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple synostoses syndrome 2, 610017, Autosomal dominant; SYNS2 (Multiple synostoses syndrome) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:16127465, ECO:0000269|PubMed:18203755, ECO:0000269|PubMed:21976273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Multipl Sinostozis Sendromu 3, 612961, Otozomal Dominant; SYNS3 (Multipl Sinostozis Sendromu) (FGF9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple synostoses syndrome 3, 612961, Autosomal dominant; SYNS3 (Multiple synostoses syndrome) (FGF9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF9	FGF9, SYNS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple synostoses syndrome 3 (SYNS3) [MIM:612961]: A bone disease characterized by multiple progressive joint fusions that commonly involve proximal interphalangeal, tarsal-carpal, humeroradial and cervical spine joints. Additional features can include progressive conductive deafness and facial dysmorphism. {ECO:0000269|PubMed:19589401}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Multipl Sistem Atrofisi, Yatkınlık, 146500, Otozomal Resesif, Otozomal Dominant (Multipl Sistem Atrofisi) (COQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multiple system atrophy, susceptibility to, 146500, Autosomal recessive, Autosomal dominant (Multiple system atrophy (COQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ2	COQ2, COQ10D1, MSA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple system atrophy 1 (MSA1) [MIM:146500]: A progressive neurodegenerative disorder clinically characterized by parkinsonism, cerebellar ataxia, and autonomic, urogenital, and pyramidal dysfunction in various combinations. Pathologically, it is characterized by degeneration of striatonigral and olivopontocerebellar structures, and glial cytoplasmic inclusions that consist of abnormally phosphorylated alpha-synuclein or tau. {ECO:0000269|PubMed:23758206}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Multisistemik Düz Kas İşlev Bozukluğu Sendromu, 613834, Otozomal Dominant (Multisistemik Düz Kas Fonksiyon Bozukluğu Sendromu) (ACTA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Multisystemic smooth muscle dysfunction syndrome, 613834, Autosomal dominant (Multisystemic smooth muscle dysfunction syndrome) (ACTA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA2	ACTA2, ACTSA, AAT6, MYMY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aortic aneurysm, familial thoracic 6 (AAT6) [MIM:611788]: A disease characterized by permanent dilation of the thoracic aorta usually due to degenerative changes in the aortic wall. It is primarily associated with a characteristic histologic appearance known as 'medial necrosis' or 'Erdheim cystic medial necrosis' in which there is degeneration and fragmentation of elastic fibers, loss of smooth muscle cells, and an accumulation of basophilic ground substance. {ECO:0000269|PubMed:17994018, ECO:0000269|PubMed:19409525, ECO:0000269|PubMed:19639654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kas Glikojenozu, 300559, X'e Bağlı Resesif; GSD9D (Glikojen Depo Hastalığı, Kas Fosforilaz Kinaz Eksikliğine Bağlı) (PHKA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscle glycogenosis, 300559, X-linked recessive; GSD9D (Glycogen storage disease due to muscle phosphorylase kinase deficiency) (PHKA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKA1	PHKA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9D (GSD9D) [MIM:300559]: A metabolic disorder characterized by slowly progressive, predominantly distal muscle weakness and atrophy. Clinical features include exercise intolerance with early fatigability, pain, cramps and occasionally myoglobinuria. {ECO:0000269|PubMed:12825073}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kas Glikojenozu, 300559, X'e Bağlı Resesif; GSD9D (Glikojen Depo Hastalığı, Kas Fosforilaz Kinaz Eksikliğine Bağlı) (Prenatal) (PHKA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscle glycogenosis, 300559, X-linked recessive; GSD9D (Glycogen storage disease due to muscle phosphorylase kinase deficiency) (PHKA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKA1	PHKA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9D (GSD9D) [MIM:300559]: A metabolic disorder characterized by slowly progressive, predominantly distal muscle weakness and atrophy. Clinical features include exercise intolerance with early fatigability, pain, cramps and occasionally myoglobinuria. {ECO:0000269	PubMed:12825073}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kas Hipertrofisi, 614160; MSLHP (Miyostatin İlişkili Kas Hipertrofisi) (MSTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscle hypertrophy, 614160; MSLHP (Myostatin-related muscle hypertrophy) (MSTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSTN	GDF8, MSTN, MSLHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscle hypertrophy (MSLHP) [MIM:614160]: A condition characterized by increased muscle bulk and strength. Affected individuals are exceptionally strong. {ECO:0000269	PubMed:15215484}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kas Hipertrofisi, 614160; MSLHP (Miyostatin İlişkili Kas Hipertrofisi) (Prenatal) (MSTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscle hypertrophy, 614160; MSLHP (Myostatin-related muscle hypertrophy) (MSTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSTN	GDF8, MSTN, MSLHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscle hypertrophy (MSLHP) [MIM:614160]: A condition characterized by increased muscle bulk and strength. Affected individuals are exceptionally strong. {ECO:0000269	PubMed:15215484}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Göz ve Beyin Anomalileri), Tip A, 11, 615181, Otozomal Resesif; MDDGA11 (Kas-Göz-Beyin Hastalığı) (B3GALNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181, Autosomal recessive; MDDGA11 (Muscle-eye-brain disease) (B3GALNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GALNT2	B3GALNT2, MGC39558, MDDGA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A11 (MDDGA11) [MIM:615181]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269	PubMed:23453667}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Göz ve Beyin Anomalileri), Tip A, 11, 615181, Otozomal Resesif; MDDGA11 (Kas-Göz-Beyin Hastalığı) (Prenatal) (B3GALNT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11, 615181, Autosomal recessive; MDDGA11 (Muscle-eye-brain disease) (B3GALNT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B3GALNT2	B3GALNT2, MGC39558, MDDGA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A11 (MDDGA11) [MIM:615181]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269	PubMed:23453667}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Göz ve Beyin Anomalileri), Tip A, 8, 614830, Otozomal Resesif; MDDGA8 (Walker-Warburg Sendromu) (POMGNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830, Autosomal recessive; MDDGA8 (Walker-Warburg syndrome) (POMGNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMGNT2	POMGNT2, GTDC2, C3orf39, AGO61, MDDGA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Göz ve Beyin Anomalileri), Tip A, 8, 614830, Otozomal Resesif; MDDGA8 (Walker-Warburg Sendromu) (Prenatal) (POMGNT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 8, 614830, Autosomal recessive; MDDGA8 (Walker-Warburg syndrome) (POMGNT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMGNT2	POMGNT2, GTDC2, C3orf39, AGO61, MDDGA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif; MDDGA1 (Kas-Göz-Beyin Hastalığı) (POMT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670, Autosomal recessive; MDDGA1 (Muscle-eye-brain disease) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif; MDDGA1 (Kas-Göz-Beyin Hastalığı) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670, Autosomal recessive; MDDGA1 (Muscle-eye-brain disease) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif; MDDGA1 (Kas-Göz-Beyin Hastalığı) (Prenatal) (POMT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670, Autosomal recessive; MDDGA1 (Muscle-eye-brain disease) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif; MDDGA1 (Kas-Göz-Beyin Hastalığı) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670, Autosomal recessive; MDDGA1 (Muscle-eye-brain disease) (Prenatal) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalili), A Tipi, 10, 615041, Otozomal Resesif; MDDGA10 (Walker-Warburg Sendromu) (TMEM5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041, Autosomal recessive; MDDGA10 (Walker-Warburg syndrome) (TMEM5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM5	TMEM5, MDDGA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A10 (MDDGA10) [MIM:615041]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:23217329, ECO:0000269|PubMed:23519211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalili), A Tipi, 10, 615041, Otozomal Resesif; MDDGA10 (Walker-Warburg Sendromu) (Prenatal) (TMEM5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 10, 615041, Autosomal recessive; MDDGA10 (Walker-Warburg syndrome) (TMEM5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM5	TMEM5, MDDGA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A10 (MDDGA10) [MIM:615041]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:23217329, ECO:0000269|PubMed:23519211}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), Tip A, 12, 615249, Otozomal Resesif; MDDGA12 (Walker-Warburg Sendromu) (POMK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249, Autosomal recessive; MDDGA12 (Walker-Warburg syndrome) (POMK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMK	POMK, SGK196, MDDGA12, MDDGC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C12 (MDDGC12) [MIM:616094]: An autosomal recessive limb-girdle congenital muscular dystrophy, characterized by muscle weakness and delayed motor development in association with cognitive impairment. {ECO:0000269	PubMed:24925318}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), Tip A, 12, 615249, Otozomal Resesif; MDDGA12 (Walker-Warburg Sendromu) (Prenatal) (POMK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12, 615249, Autosomal recessive; MDDGA12 (Walker-Warburg syndrome) (POMK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMK	POMK, SGK196, MDDGA12, MDDGC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C12 (MDDGC12) [MIM:616094]: An autosomal recessive limb-girdle congenital muscular dystrophy, characterized by muscle weakness and delayed motor development in association with cognitive impairment. {ECO:0000269	PubMed:24925318}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), Tip A, 13, 615287, Otozomal Resesif; MDDGA13 (Walker-Warburg Sendromu) (B4GAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287, Autosomal recessive; MDDGA13 (Walker-Warburg syndrome) (B4GAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B4GAT1	B3GNT1, IGNT, IGAT, MDDGA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), Tip A, 13, 615287, Otozomal Resesif; MDDGA13 (Walker-Warburg Sendromu) (Prenatal) (B4GAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13, 615287, Autosomal recessive; MDDGA13 (Walker-Warburg syndrome) (B4GAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B4GAT1	B3GNT1, IGNT, IGAT, MDDGA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalili) Tip A, 14, 615350, Otozomal Resesif; MDDGA14 (Kas-Göz-Beyin Hastalığı) (GMPPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350, Autosomal recessive; MDDGA14 (Muscle-eye-brain disease) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768513}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalili) Tip A, 14, 615350, Otozomal Resesif; MDDGA14 (Kas-Göz-Beyin Hastalığı) (Prenatal) (GMPPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 14, 615350, Autosomal recessive; MDDGA14 (Muscle-eye-brain disease) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768512}, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif (Kas-Göz-Beyin Hastalığı) (POMT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150, Autosomal recessive (Muscle-eye-brain disease) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:15894594, ECO:0000269|PubMed:16701995, ECO:0000269|PubMed:17878207, ECO:0000269|PubMed:19138766, ECO:0000269|PubMed:22958903}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 1, 236670, Otozomal Resesif (Kas-Göz-Beyin Hastalığı) (Prenatal) (POMT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 2, 613150, Autosomal recessive (Muscle-eye-brain disease) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269	PubMed:15894594, ECO:0000269	PubMed:16701995, ECO:0000269	PubMed:17878207, ECO:0000269	PubMed:19138766, ECO:0000269	PubMed:22958903}, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 3, 253280, Otozomal Resesif; MDDGA3 (Kas-Göz-Beyin Hastalığı) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280, Autosomal recessive; MDDGA3 (Muscle-eye-brain disease) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 3, 253280, Otozomal Resesif; MDDGA3 (Kas-Göz-Beyin Hastalığı) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280, Autosomal recessive; MDDGA3 (Muscle-eye-brain disease) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 3, 253280, Otozomal Resesif; MDDGA3 (Kas-Göz-Beyin Hastalığı) (Prenatal) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280, Autosomal recessive; MDDGA3 (Muscle-eye-brain disease) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglycanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 3, 253280, Otozomal Resesif; MDDGA3 (Kas-Göz-Beyin Hastalığı) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 3, 253280, Autosomal recessive; MDDGA3 (Muscle-eye-brain disease) (Prenatal) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DiSEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalileri İle Konjenital), A Tipi, 4, 253800, Otozomal Resesif; MDDGA4 (Kas-Göz-Beyin Hastalığı) (FKTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800, Autosomal recessive; MDDGA4 (Muscle-eye-brain disease) (FKTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DiSEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269|PubMed:17044012, ECO:0000269|PubMed:16342235}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalileri İle Konjenital), A Tipi, 4, 253800, Otozomal Resesif; MDDGA4 (Kas-Göz-Beyin Hastalığı) (Prenatal) (FKTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 4, 253800, Autosomal recessive; MDDGA4 (Muscle-eye-brain disease) (FKTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DiSEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269|PubMed:17044012, ECO:0000269|PubMed:16342235}, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 5,613153, Otozomal Resesif; MDDGA5 (Kas-Göz-Beyin Hastalığı) (FKRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153, Autosomal recessive; MDDGA5 (Muscle-eye-brain disease) (FKRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 5,613153, Otozomal Resesif; MDDGA5 (Kas-Göz-Beyin Hastalığı) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153, Autosomal recessive; MDDGA5 (Muscle-eye-brain disease) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 5,613153, Otozomal Resesif; MDDGA6 (Kas-Göz-Beyin Hastalığı) (Prenatal) (FKRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153, Autosomal recessive; MDDGA5 (Muscle-eye-brain disease) (FKRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 5,613153, Otozomal Resesif; MDDGA5 (Kas-Göz-Beyin Hastalığı) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 5, 613153, Autosomal recessive; MDDGA5 (Muscle-eye-brain disease) (Prenatal) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 6,613154, Otozomal Resesif; MDDGA6 (Walker Warburg Sendromu) (Kas-Göz-Beyin Hastalığı) (LARGE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154, Autosomal recessive; MDDGA6 (Walker Warburg Syndrome) (Muscle-eye-brain disease) (LARGE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARGE1	LARGE, KIAA0609, MDC1D, MDDGA6, MDDGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Beyin ve Göz Anomalileri), A Tipi, 6,613154, Otozomal Resesif; MDDGA6 (Walker Warburg Sendromu) (Kas-Göz-Beyin Hastalığı) (Prenatal) (LARGE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 6, 613154, Autosomal recessive; MDDGA6 (Walker Warburg Syndrome) (Muscle-eye-brain disease) (LARGE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARGE1	LARGE, KIAA0609, MDC1D, MDDGA6, MDDGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati, (Konjenital Beyin ve Göz Anomali), Tip A, 7, 614643, Otozomal Resesif; MDDGA7 (Walker-Warburg Sendromu) (ISPD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643, Autosomal recessive; MDDGA7 (Walker-Warburg syndrome) (ISPD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ISPD	ISPD, MDDGA7, MDDGC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C7 (MDDGC7) [MIM:616052]: A form of muscular dystrophy resulting from defective glycosylation of alpha-dystroglycan, and characterized by a limb-girdle phenotype with muscular weakness apparent after ambulation is achieved. MDDGC7 individuals do not show epilepsy, mental retardation, structural eye/brain abnormalities, or white matter changes. {ECO:0000269	PubMed:23390185}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati, (Konjenital Beyin ve Göz Anomali), Tip A, 7, 614643, Otozomal Resesif; MDDGA7 (Walker-Warburg Sendromu) (Prenatal) (ISPD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7, 614643, Autosomal recessive; MDDGA7 (Walker-Warburg syndrome) (ISPD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ISPD	ISPD, MDDGA7, MDDGC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C7 (MDDGC7) [MIM:616052]: A form of muscular dystrophy resulting from defective glycosylation of alpha-dystroglycan, and characterized by a limb-girdle phenotype with muscular weakness apparent after ambulation is achieved. MDDGC7 individuals do not show epilepsy, mental retardation, structural eye/brain abnormalities, or white matter changes. {ECO:0000269	PubMed:23390185}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 9, 616538, Otozomal Resesif; MDDGA9 (Kas-Göz-Beyin Hastalığı- Bilateral Multikistik Lökodistrofi) (DAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538, Autosomal recessive; MDDGA9 (Muscle-eye-brain disease with bilateral multicystic leucodystrophy) (DAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAG1	DAG1, DAG, MDDGC9, MDDGA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C9 (MDDGC9) [MIM:613818]: An autosomal recessive muscular dystrophy showing onset in early childhood, and associated with mental retardation without structural brain anomalies. {ECO:0000269	PubMed:21388311}. Note=The disease is caused by mutations affecting the gene represented in this entry. MDDGC7 is caused by DAG1 mutations that interfere with normal post- translational processing, resulting in defective DAG1 glycosylation and impaired interactions with extracellular-matrix components. Other muscular dystrophy-dystroglycanopathies are caused by defects in enzymes involved in protein O-glycosylation.;	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Beyin ve Göz Anomalili Konjenital), Tip A, 9, 616538, Otozomal Resesif; MDDGA9 (Kas-Göz-Beyin Hastalığı- Bilateral Multikistik Lökodistrofi) (Prenatal) (DAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 9, 616538, Autosomal recessive; MDDGA9 (Muscle-eye-brain disease with bilateral multicystic leucodystrophy) (DAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DAG1	DAG1, DAG, MDDGC9, MDDGA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C9 (MDDGC9) [MIM:613818]: An autosomal recessive muscular dystrophy showing onset in early childhood, and associated with mental retardation without structural brain anomalies. {ECO:0000269	PubMed:21388311}. Note=The disease is caused by mutations affecting the gene represented in this entry. MDDGC7 is caused by DAG1 mutations that interfere with normal post- translational processing, resulting in defective DAG1 glycosylation and impaired interactions with extracellular-matrix components. Other muscular dystrophy-dystroglycanopathies are caused by defects in enzymes involved in protein O-glycosylation.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 1, 613155, Otozomal Resesif; MDDGB1 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (POMT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155, Autosomal recessive; MDDGB1 (Congenital muscular dystrophy with cerebellar involvement) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 1, 613155, Otozomal Resesif; MDDGB1 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155, Autosomal recessive; MDDGB1 (Congenital muscular dystrophy with cerebellar involvement) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 1, 613155, Otozomal Resesif; MDDGB1 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (Prenatal) (POMT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155, Autosomal recessive; MDDGB1 (Congenital muscular dystrophy with cerebellar involvement) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 1, 613155, Otozomal Resesif; MDDGB1 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155, Autosomal recessive; MDDGB1 (Congenital muscular dystrophy with cerebellar involvement) (Prenatal) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 14, 615351, Otozomal Resesif; MDDGB14 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (GMPPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351, Autosomal recessive; MDDGB14 (Congenital muscular dystrophy with cerebellar involvement) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon Ile), Tip B, 14, 615351, Otozomal Resesif; MDDGB14 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (Prenatal) (GMPPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 14, 615351, Autosomal recessive; MDDGB14 (Congenital muscular dystrophy with cerebellar involvement) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon İle), Tip B, 2, 613156, Otozomal Resesif; MDDGA2 (Konjenital Musküler Distrofi-Serebellar Tutulum) (POMT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156, Autosomal recessive; MDDGA2 (Congenital muscular dystrophy with cerebellar involvement) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:15894594, ECO:0000269|PubMed:16701995, ECO:0000269|PubMed:17878207, ECO:0000269|PubMed:19138766, ECO:0000269|PubMed:22958903}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital, Mental Retardasyon İle), Tip B, 2, 613156, Otozomal Resesif; MDDGA2 (Konjenital Musküler Distrofi-Serebellar Tutulum) (Prenatal) (POMT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 2, 613156, Autosomal recessive; MDDGA2 (Congenital muscular dystrophy with cerebellar involvement) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:15894594, ECO:0000269|PubMed:16701995, ECO:0000269|PubMed:17878207, ECO:0000269|PubMed:19138766, ECO:0000269|PubMed:22958903}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital-Mental Retardasyon İle), Tip B, 3, 613151, Otozomal Resesif; MDDGB3 (Konjenital Musküler Distrofi-Serebellar Tutulum) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151, Autosomal recessive; MDDGB3 (Congenital muscular dystrophy with cerebellar involvement) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital-Mental Retardasyon İle), Tip B, 3, 613151, Otozomal Resesif; MDDGB3 (Konjenital Musküler Distrofi-Serebellar Tutulum) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151, Autosomal recessive; MDDGB3 (Congenital muscular dystrophy with cerebellar involvement) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital-Mental Retardasyon İle), Tip B, 3, 613151, Otozomal Resesif; MDDGB3 (Konjenital Musküler Distrofi-Serebellar Tutulum) (Prenatal) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151, Autosomal recessive; MDDGB3 (Congenital muscular dystrophy with cerebellar involvement) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital-Mental Retardasyon İle), Tip B, 3, 613151, Otozomal Resesif; MDDGB3 (Konjenital Musküler Distrofi-Serebellar Tutulum) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 3, 613151, Autosomal recessive; MDDGB3 (Congenital muscular dystrophy with cerebellar involvement) (Prenatal) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Mental Retardasyon, Konjenital), Tip B, 6, 608840, Otozomal Resesif; MDDGB6 (Walker Warburg Sendromu) (Konjenital Musküler Distrofi-Mental Retardasyon) (LARGE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840, Autosomal recessive; MDDGB6 (Walker Warburg Syndrome (Congenital muscular dystrophy with intellectual disability) (LARGE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARGE1	LARGE, KIAA0609, MDC1D, MDDGA6, MDDGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Mental Retardasyon, Konjenital), Tip B, 6, 608840, Otozomal Resesif; MDDGB6 (Walker Warburg Sendromu) (Konjenital Musküler Distrofi-Mental Retardasyon) (Prenatal) (LARGE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 6, 608840, Autosomal recessive; MDDGB6 (Walker Warburg Syndrome (Congenital muscular dystrophy with intellectual disability) (LARGE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARGE1	LARGE, KIAA0609, MDC1D, MDDGA6, MDDGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital ± Mental Retardasyon), Tip B, 5, 606612, Otozomal Resesif; MDDGB5 (Konjenital Musküler Distrofi-Serebellar Tutulum) (FKRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612, Autosomal recessive; MDDGB5 (Congenital muscular dystrophy with cerebellar involvement) (FKRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital ± Mental Retardasyon), Tip B, 5, 606612, Otozomal Resesif; MDDGB5 (Konjenital Musküler Distrofi-Serebellar Tutulum) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612, Autosomal recessive; MDDGB5 (Congenital muscular dystrophy with cerebellar involvement) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital ± Mental Retardasyon), Tip B, 5, 606612, Otozomal Resesif; MDDGB5 (Konjenital Musküler Distrofi-Serebellar Tutulum) (Prenatal) (FKRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612, Autosomal recessive; MDDGB5 (Congenital muscular dystrophy with cerebellar involvement) (FKRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Konjenital ± Mental Retardasyon), Tip B, 5, 606612, Otozomal Resesif; MDDGB5 (Konjenital Müsküler Distrofi-Serebellar Tutulum) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (congenital with or without mental retardation), type B, 5, 606612, Autosomal recessive; MDDGB5 (Congenital muscular dystrophy with cerebellar involvement) (Prenatal) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Olmayan Mental Retardasyon), Tip B, 4, 613152, Otozomal Resesif; MDDGB4 (Konjenital Müsküler Distrofi-Mental Retardasyon Olmadan) (FKTN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152, Autosomal recessive; MDDGB4 (Congenital muscular dystrophy without intellectual disability) (FKTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum-creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269	PubMed:17044012, ECO:0000269	PubMed:19342235}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Konjenital Olmayan Mental Retardasyon), Tip B, 4, 613152, Otozomal Resesif; MDDGB4 (Konjenital Müsküler Distrofi-Mental Retardasyon Olmadan) (Prenatal) (FKTN gen) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (congenital without mental retardation), type B, 4, 613152, Autosomal recessive; MDDGB4 (Congenital muscular dystrophy without intellectual disability) (FKTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum-creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269	PubMed:17044012, ECO:0000269	PubMed:19342235}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 1, 609308, Otozomal Resesif; MDDGC1 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2K) (POMT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308, Autosomal recessive; MDDGC1 (Autosomal recessive limb-girdle muscular dystrophy type 2K) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 1, 609308, Otozomal Resesif; MDDGC1 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2K) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308, Autosomal recessive; MDDGC1 (Autosomal recessive limb-girdle muscular dystrophy type 2K) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 1, 609308, Otozomal Resesif; MDDGC1 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2K) (Prenatal) (POMT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308, Autosomal recessive; MDDGC1 (Autosomal recessive limb-girdle muscular dystrophy type 2K) (POMT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 1, 609308, Otozomal Resesif; MDDGC1 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2K) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308, Autosomal recessive; MDDGC1 (Autosomal recessive limb-girdle muscular dystrophy type 2K) (Prenatal) (MLPA)	POMT1	POMT1, MDDGA1, MDDGB1, MDDGC1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A1 (MDDGA1) [MIM:236670]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:12369018, ECO:0000269|PubMed:15037715, ECO:0000269|PubMed:15637732, ECO:0000269|PubMed:16575835}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 12, 616094, Otozomal Resesif; MDDGC12 (Limb-Girdle Müsküler Distrofi, POMK Eksikliğine Bağlı) (POMK gen) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094, Autosomal recessive; MDDGC12 (Limb-girdle muscular dystrophy due to POMK deficiency) (POMK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMK	POMK, SGK196, MDDGA12, MDDGC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C12 (MDDGC12) [MIM:616094]: An autosomal recessive limb-girdle congenital muscular dystrophy, characterized by muscle weakness and delayed motor development in association with cognitive impairment. {ECO:0000269	PubMed:24925318}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 12, 616094, Otozomal Resesif; MDDGC12 (Limb-Girdle Müsküler Distrofi, POMK Eksikliğine Bağlı) (Prenatal) (POMK gen) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 12, 616094, Autosomal recessive; MDDGC12 (Limb-girdle muscular dystrophy due to POMK deficiency) (POMK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMK	POMK, SGK196, MDDGA12, MDDGC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C12 (MDDGC12) [MIM:616094]: An autosomal recessive limb-girdle congenital muscular dystrophy, characterized by muscle weakness and delayed motor development in association with cognitive impairment. {ECO:0000269	PubMed:24925318}. Note=The disease is caused by mutations affecting the gene represented in this entry. ;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 14, 615352, Otozomal Resesif; MDDGC14 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2T) (GMPPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352, Autosomal recessive; MDDGC14 (Autosomal recessive limb-girdle muscular dystrophy type 2T) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 14, 615352, Otozomal Resesif; MDDGC14 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2T) (Prenatal) (GMPPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14, 615352, Autosomal recessive; MDDGC14 (Autosomal recessive limb-girdle muscular dystrophy type 2T) (GMPPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GMPPB	GMPPB, KIAA1851, MDDGA14, MDDGB14, MDDGC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A14 (MDDGA14) [MIM:615350]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with brain anomalies, eye malformations, and profound mental retardation. The disorder includes a severe form designated as Walker-Warburg syndrome and a less severe phenotype known as muscle-eye-brain disease. MDDGA14 features include increased muscle tone, microcephaly, cleft palate, feeding difficulties, severe muscle weakness, sensorineural hearing loss, cerebellar hypoplasia, ataxia, and retinal dysfunction. {ECO:0000269	PubMed:23768512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 2, 613158, Otozomal Resesif; MDDGC2 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2N) (POMT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158, Autosomal recessive; MDDGC2 (Autosomal recessive limb-girdle muscular dystrophy type 2N) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:15894594, ECO:0000269|PubMed:16701995, ECO:0000269|PubMed:17878207, ECO:0000269|PubMed:19138766, ECO:0000269|PubMed:22958903}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 2, 613158, Otozomal Resesif; MDDGC2 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2N) (Prenatal) (POMT2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 2, 613158, Autosomal recessive; MDDGC2 (Autosomal recessive limb-girdle muscular dystrophy type 2N) (POMT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMT2	POMT2, MDDGA2, MDDGB2, MDDGC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A2 (MDDGA2) [MIM:613150]: An autosomal recessive disorder characterized by congenital muscular dystrophy associated with cobblestone lissencephaly and other brain anomalies, eye malformations, profound mental retardation, and death usually in the first years of life. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:15894594, ECO:0000269|PubMed:16701995, ECO:0000269|PubMed:17878207, ECO:0000269|PubMed:19138766, ECO:0000269|PubMed:22958903}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 3, 613157, Otozomal Resesif; MDDGC3 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2O) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157, Autosomal recessive; MDDGC3 (Autosomal recessive limb-girdle muscular dystrophy type 2O) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 3, 613157, Otozomal Resesif; MDDGC3 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2O) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157, Autosomal recessive; MDDGC3 (Autosomal recessive limb-girdle muscular dystrophy type 2O) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 3, 613157, Otozomal Resesif; MDDGC3 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2O) (Prenatal) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157, Autosomal recessive; MDDGC3 (Autosomal recessive limb-girdle muscular dystrophy type 2O) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 3, 613157, Otozomal Resesif; MDDGC3 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2O) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 3, 613157, Autosomal recessive; MDDGC3 (Autosomal recessive limb-girdle muscular dystrophy type 2O) (Prenatal) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 4, 611588, Otozomal Resesif; MDDGC4 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2M) (FKTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588, Autosomal recessive; MDDGC4 (Autosomal recessive limb-girdle muscular dystrophy type 2M) (FKTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269|PubMed:17044012, ECO:0000269|PubMed:19342235}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 4, 611588, Otozomal Resesif; MDDGC4 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2M) (Prenatal) (FKTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 4, 611588, Autosomal recessive; MDDGC4 (Autosomal recessive limb-girdle muscular dystrophy type 2M) (FKTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKTN	FKTN, FCMD, CMD1X, LGMD2M, MDDGA4, MDDGB4, MDDGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C4 (MDDGC4) [MIM:611588]: An autosomal recessive degenerative myopathy characterized by progressive weakness of the pelvic and shoulder girdle muscles, and elevated serum creatine kinase. MDDGC4 has no brain involvement and a remarkable clinical response to corticosteroids. {ECO:0000269|PubMed:17044012, ECO:0000269|PubMed:19342235}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 5, 607155, Otozomal Resesif; MDDGC5 (Otozomal Resesif Limb-Girdle Tipi Müsküler Distrofi Tip 2I) (FKRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155, Autosomal recessive; MDDGC5 (Autosomal recessive limb-girdle muscular dystrophy type 2I) (FKRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 5, 607155, Otozomal Resesif; MDDGC5 (Otozomal Resesif Limb-Girdle Tipi Müsküler Distrofi Tip 2I) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155, Autosomal recessive; MDDGC5 (Autosomal recessive limb-girdle muscular dystrophy type 2I) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 5, 607155, Otozomal Resesif; MDDGC5 (Otozomal Resesif Limb-Girdle Tipi Müsküler Distrofi Tip 2I) (Prenatal) (FKRP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155, Autosomal recessive; MDDGC5 (Autosomal recessive limb-girdle muscular dystrophy type 2I) (FKRP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 5, 607155, Otozomal Resesif; MDDGC5 (Otozomal Resesif Limb-Girdle Tipi Müsküler Distrofi Tip 2I) (Prenatal) (MLPA)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5, 607155, Autosomal recessive; MDDGC5 (Autosomal recessive limb-girdle muscular dystrophy type 2I) (Prenatal) (MLPA)	FKRP	FKRP, MDC1C, LGMD2I, MDDGA5, MDDGB5, MDDGC5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with or without mental retardation B5 (MDDGB5) [MIM:606612]: A congenital muscular dystrophy characterized by a severe phenotype with inability to walk, muscle hypertrophy, marked elevation of serum creatine kinase, secondary deficiency of laminin alpha2, and a marked reduction in alpha-dystroglycan expression. Only a subset of affected individuals have brain involvements. {ECO:0000269|PubMed:11592034, ECO:0000269|PubMed:12654965, ECO:0000269|PubMed:12666124, ECO:0000269|PubMed:14652796, ECO:0000269|PubMed:17336067}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 7, 616052, Otozomal Resesif; MDDGC7 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2U) (ISPD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052, Autosomal recessive; MDDGC7 (Autosomal recessive limb-girdle muscular dystrophy type 2U) (ISPD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ISPD	ISPD, MDDGA7, MDDGC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C7 (MDDGC7) [MIM:616052]: A form of muscular dystrophy resulting from defective glycosylation of alpha-dystroglycan, and characterized by a limb-girdle phenotype with muscular weakness apparent after ambulation is achieved. MDDGC7 individuals do not show epilepsy, mental retardation, structural eye/brain abnormalities, or white matter changes. {ECO:0000269|PubMed:23390185}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), C, 7, 616052, Otozomal Resesif; MDDGC7 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2U) (Prenatal) (ISPD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 7, 616052, Autosomal recessive; MDDGC7 (Autosomal recessive limb-girdle muscular dystrophy type 2U) (ISPD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ISPD	ISPD, MDDGA7, MDDGC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C7 (MDDGC7) [MIM:616052]: A form of muscular dystrophy resulting from defective glycosylation of alpha-dystroglycan, and characterized by a limb-girdle phenotype with muscular weakness apparent after ambulation is achieved. MDDGC7 individuals do not show epilepsy, mental retardation, structural eye/brain abnormalities, or white matter changes. {ECO:0000269|PubMed:23390185}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 9, 613818, Otozomal Resesif; MDDGC9 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2P) (LGMD2P) (DAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818, Autosomal recessive; MDDGC9 (Autosomal recessive limb-girdle muscular dystrophy type 2P) (LGMD2P) (DAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAG1	DAG1, DAG, MDDGC9, MDDGA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C9 (MDDGC9) [MIM:613818]: An autosomal recessive muscular dystrophy showing onset in early childhood, and associated with mental retardation without structural brain anomalies. {ECO:0000269	PubMed:21388311}. Note=The disease is caused by mutations affecting the gene represented in this entry. MDDGC7 is caused by DAG1 mutations that interfere with normal post- translational processing, resulting in defective DAG1 glycosylation and impaired interactions with extracellular-matrix components. Other muscular dystrophy-dystroglycanopathies are caused by defects in enzymes involved in protein O-glycosylation.	EDTA Blood Sample
Müsküler Distrofi-Distroglikanopati (Limb-Girdle), Tip C, 9, 613818, Otozomal Resesif; MDDGC9 (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2P) (LGMD2P) (Prenatal) (DAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 9, 613818, Autosomal recessive; MDDGC9 (Autosomal recessive limb-girdle muscular dystrophy type 2P) (LGMD2P) (DAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DAG1	DAG1, DAG, MDDGC9, MDDGA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy-dystroglycanopathy limb-girdle C9 (MDDGC9) [MIM:613818]: An autosomal recessive muscular dystrophy showing onset in early childhood, and associated with mental retardation without structural brain anomalies. {ECO:0000269	PubMed:21388311}. Note=The disease is caused by mutations affecting the gene represented in this entry. MDDGC7 is caused by DAG1 mutations that interfere with normal post- translational processing, resulting in defective DAG1 glycosylation and impaired interactions with extracellular-matrix components. Other muscular dystrophy-dystroglycanopathies are caused by defects in enzymes involved in protein O-glycosylation.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Becker Tipi; BMD (Becker Müsküler Distrofi) (DMD geni) (Tüm Gen Dizi Analizi) (Postnatal)	MUSCULAR DYSTROPHY, BECKER TYPE; BMD (Becker muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Becker Tipi; BMD (Becker Müsküler Distrofi) (MLPA)	MUSCULAR DYSTROPHY, BECKER TYPE; BMD (Becker muscular dystrophy) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Becker Tipi; BMD (Becker Müsküler Distrofi) (Prenatal) (DMD geni) (Tüm Gen Dizi Analizi) (Prenatal)	MUSCULAR DYSTROPHY, BECKER TYPE; BMD (Becker muscular dystrophy) (DMD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DMD	DMD, BMD, CMD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Becker Tipi; BMD (Becker Müsküler Distrofi) (Prenatal) (MLPA)	MUSCULAR DYSTROPHY, BECKER TYPE; BMD (Becker muscular dystrophy) (Prenatal) (MLPA)	DMD	DMD, BMD, CMD3B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Becker muscular dystrophy (BMD) [MIM:300376]: A neuromuscular disorder characterized by dystrophin deficiency. It appears between the age of 5 and 15 years with a proximal motor deficiency of variable progression. Heart involvement can be the initial sign. Becker muscular dystrophy has a more benign course than Duchenne muscular dystrophy. {ECO:0000269	PubMed:10573008}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Konjenital Merosin Eksikliği, 607855, Otozomal Resesif (Konjenital Müsküler Distrofi Tip 1A) (LAMA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, congenital merosin-deficient, 607855, Autosomal recessive (Congenital muscular dystrophy type 1A) (LAMA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA2	LAMA2, LAMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Merosin-deficient congenital muscular dystrophy 1A (MDC1A) [MIM:607855]: Characterized by difficulty walking, hypotonia, proximal weakness, hyporeflexia, and white matter hypodensity on MRI. {ECO:0000269|PubMed:11591858, ECO:0000269|PubMed:12552556}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Konjenital Merosin Eksikliği, 607855, Otozomal Resesif (Konjenital Müsküler Distrofi Tip 1A) (Prenatal) (LAMA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, congenital merosin-deficient, 607855, Autosomal recessive (Congenital muscular dystrophy type 1A) (LAMA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA2	LAMA2, LAMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Merosin-deficient congenital muscular dystrophy 1A (MDC1A) [MIM:607855]: Characterized by difficulty walking, hypotonia, proximal weakness, hyporeflexia, and white matter hypodensity on MRI. {ECO:0000269|PubMed:11591858, ECO:0000269|PubMed:12552556}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Konjenital, Davignon-Chauveau Tipi, 617066, Otozomal Resesif; MDCDC (TRIP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, congenital, Davignon-Chauveau type, 617066, Autosomal recessive; MDCDC (TRIP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIP4	TRIP4, ASC1, SMABF1, MDCDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Konjenital, Davignon-Chauveau Tipi, 617066, Otozomal Resesif; MDCDC (Prenatal) (TRIP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, congenital, Davignon-Chauveau type, 617066, Autosomal recessive; MDCDC (TRIP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIP4	TRIP4, ASC1, SMABF1, MDCDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Konjenital, ITGA7 Eksikliğine Bağlı, 613204, Otozomal Resesif (Konjenital Müsküler Distrofi, Integrin Alfa-7 Eksikliğine Bağlı) (ITGA7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204, Autosomal recessive (Congenital muscular dystrophy with integrin alpha-7 deficiency) (ITGA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA7	ITGA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy congenital due to integrin alpha-7 deficiency (MDCI) [MIM:613204]: A form of congenital muscular dystrophy. Patients present at birth, or within the first few months of life, with hypotonia, muscle weakness and often with joint contractures. {ECO:0000269	PubMed:9590299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Konjenital, ITGA7 Eksikliğine Bağlı, 613204, Otozomal Resesif (Konjenital Müsküler Distrofi, Integrin Alfa-7 Eksikliğine Bağlı) (Prenatal) (ITGA7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, congenital, due to ITGA7 deficiency, 613204, Autosomal recessive (Congenital muscular dystrophy with integrin alpha-7 deficiency) (ITGA7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGA7	ITGA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Muscular dystrophy congenital due to integrin alpha-7 deficiency (MDCI) [MIM:613204]: A form of congenital muscular dystrophy. Patients present at birth, or within the first few months of life, with hypotonia, muscle weakness and often with joint contractures. {ECO:0000269	PubMed:9590299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Konjenital, Parsiyel LAMA2 Eksikliğine Bağlı, 607855, Otozomal Resesif (Konjenital Müsküler Distrofi Tip 1A) (LAMA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855, Autosomal recessive (Congenital muscular dystrophy type 1A) (LAMA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA2	LAMA2, LAMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Merosin-deficient congenital muscular dystrophy 1A (MDC1A) [MIM:607855]: Characterized by difficulty walking, hypotonia, proximal weakness, hyporeflexia, and white matter hypodensity on MRI. {ECO:0000269|PubMed:11591858, ECO:0000269|PubMed:12552556}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Konjenital, Parsiyel LAMA2 Eksikliğine Bağlı, 607855, Otozomal Resesif (Konjenital Müsküler Distrofi Tip 1A) (Prenatal) (LAMA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855, Autosomal recessive (Congenital muscular dystrophy type 1A) (LAMA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA2	LAMA2, LAMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Merosin-deficient congenital muscular dystrophy 1A (MDC1A) [MIM:607855]: Characterized by difficulty walking, hypotonia, proximal weakness, hyporeflexia, and white matter hypodensity on MRI. {ECO:0000269|PubMed:11591858, ECO:0000269|PubMed:12552556}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Konjenital, Megalorial Tip, 602541, Otozomal Resesif; MDCMC (Konjenital Müsküler Distrofi, Fosfatidilkolin Biyosentez Defektine Bağlı) (CHKB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, congenital, megaconial type, 602541, Autosomal recessive; MDCMC (Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect) (CHKB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHKB	CHKB, CHKL, CKEKB, EKB, MDCMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Konjenital, Megalorial Tip, 602541, Otozomal Resesif; MDCMC (Konjenital Müsküler Distrofi, Fosfatidilkolin Biyosentez Defektine Bağlı) (Prenatal) (CHKB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, congenital, megaconial type, 602541, Autosomal recessive; MDCMC (Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect) (CHKB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHKB	CHKB, CHKL, CKEKB, EKB, MDCMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1A, 159000, Otozomal Dominant; LGMD1A (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1A) (MYOT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 1A, 159000, Autosomal dominant; LGMD1A (Autosomal dominant limb-girdle muscular dystrophy type 1A) (MYOT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYOT	MYOT, TTOD, MFM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1A, 159000, Otozomal Dominant; LGMD1A (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1A) (MLPA)	Muscular dystrophy, limb-girdle, type 1A, 159000, Autosomal dominant; LGMD1A (Autosomal dominant limb-girdle muscular dystrophy type 1A) (MLPA)	MYOT	MYOT, TTOD, MFM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1b; Lgmd1b (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1B) (LMNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B; LGMD1B (Autosomal dominant limb-girdle muscular dystrophy type 1B) (LMNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. {ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11561226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21846512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Müsküler Distrofi, Limb-Girdle, Tip 1b; Lgmd1b (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1B) (MLPA)	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B; LGMD1B (Autosomal dominant limb-girdle muscular dystrophy type 1B) (MLPA)	LMNA	LMNA, LMN1, EMD2, FPLD2, CMD1A, HGPS, LGMD1B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cardiomyopathy, dilated 1A (CMD1A) [MIM:115200]: A disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death. (ECO:0000269|PubMed:10580070, ECO:0000269|PubMed:11501226, ECO:0000269|PubMed:11897440, ECO:0000269|PubMed:12486434, ECO:0000269|PubMed:12628721, ECO:0000269|PubMed:12920062, ECO:0000269|PubMed:14684700, ECO:0000269|PubMed:15140538, ECO:0000269|PubMed:15219508, ECO:0000269|PubMed:16061563, ECO:0000269|PubMed:20160190, ECO:0000269|PubMed:21845121). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1E, 603511, Otozomal Dominant; LGMD1E (Otozomal Dominant Limb-Girdle Tipi Müsküler Distrofi Tip 1D) (DNAJB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 1E, 603511, Autosomal dominant; LGMD1E (Autosomal dominant limb-girdle muscular dystrophy type 1D) (DNAJB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJB6	DNAJB6, MRJ, DJ4, LGMD1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 1E (LGMD1E) [MIM:603511]: An autosomal dominant myopathy characterized by adult onset of proximal muscle weakness, beginning in the hip girdle region and later progressing to the shoulder girdle region. (ECO:0000269|PubMed:22334415, ECO:0000269|PubMed:22366786). Note=The disease is caused by mutations affecting the gene represented in this entry. There is evidence that LGMD1E is caused by dysfunction of isoform B (PubMed:22366786). (ECO:0000269|PubMed:22366786).;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1F, 608423, Otozomal Dominant; LGMD1F (Otozomal Dominant Limb-Girdle Tipi Müsküler Distrofi Tip 1F) (TNPO3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 1F, 608423, Autosomal dominant; LGMD1F (Autosomal dominant limb-girdle muscular dystrophy type 1F) (TNPO3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNPO3	TNPO3, TRNSR, LGMD1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1G, 609115, Otozomal Dominant; LGMD1G (Otozomal Dominant Limb-Girdle Tipi Müsküler Distrofi Tip 1G) (HNRNPDL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 1G, 609115, Autosomal dominant; LGMD1G (Autosomal dominant limb-girdle muscular dystrophy type 1G) (HNRNPDL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNRNPDL	HNRNPDL, HNRPDL, JKTBP, LGMD1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 1G (LGMD1G) [MIM:609115]: An autosomal dominant degenerative myopathy characterized by slowly progressive wasting and weakness of the proximal muscles of arms and legs around the pelvic or shoulder girdles, elevated creatine kinase levels and dystrophic features on muscle biopsy. LGMD1G is characterized by a mild late-onset and is associated with progressive fingers and toes flexion limitation. Affected individuals may also develop cataracts before age 50. (ECO:0000269	PubMed:24647604). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2A, 253600, Otozomal Resesif; LGMD2A (Limb-Girdle Tipi Müsküler Distrofi, Tip 2A, Otozomal Resesif) (CAPN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2A, 253600, Autosomal recessive; LGMD2A (Autosomal recessive limb-girdle muscular dystrophy type 2A) (CAPN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAPN3	CAPN3, CANP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2A, 253600, Otozomal Resesif; LGMD2A (Limb-Girdle Tipi Müsküler Distrofi, Tip 2A, Otozomal Resesif) (MLPA)	Muscular dystrophy, limb-girdle, type 2A, 253600, Autosomal recessive; LGMD2A (Autosomal recessive limb-girdle muscular dystrophy type 2A) (MLPA)	CAPN3	CAPN3, CANP3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2B, 253601, Otozomal Resesif; LGMD2B (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2B) (DYSF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2B, 253601, Autosomal recessive; LGMD2B (Autosomal recessive limb-girdle muscular dystrophy type 2B) (DYSF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYSF	DYSF, LGMD2B, MMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2B, 253601, Otozomal Resesif; LGMD2B (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2B) (MLPA)	Muscular dystrophy, limb-girdle, type 2B, 253601, Autosomal recessive; LGMD2B (Autosomal recessive limb-girdle muscular dystrophy type 2B) (MLPA)	DYSF	DYSF, LGMD2B, MMD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2C, 253700, Otozomal Resesif; LGMD2C (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2C) (SGCG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2C, 253700, Autosomal recessive; LGMD2C (Autosomal recessive limb-girdle muscular dystrophy type 2C) (SGCG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCG	SGCG, LGMD2C, DMDA1, SCG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2C, 253700, Otozomal Resesif; LGMD2C (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2C) (MLPA)	Muscular dystrophy, limb-girdle, type 2C, 253700, Autosomal recessive; LGMD2C (Autosomal recessive limb-girdle muscular dystrophy type 2C) (MLPA)	SGCG	SGCG, LGMD2C, DMDA1, SCG3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2D, 608099, Otozomal Resesif; LGMD2D (Limb-Girdle Tipi Müsküler Distrofi, Tip 2D, Otozomal Resesif) (SGCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2D, 608099, Autosomal recessive; LGMD2D (Autosomal recessive limb-girdle muscular dystrophy type 2D) (SGCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCA	SGCA, ADL, DAG2, LGMD2D, DMDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2D, 608099, Otozomal Resesif; LGMD2D (Limb-Girdle Tipi Müsküler Distrofi, Tip 2D, Otozomal Resesif) (MLPA)	Muscular dystrophy, limb-girdle, type 2D, 608099, Autosomal recessive; LGMD2D (Autosomal recessive limb-girdle muscular dystrophy type 2D) (MLPA)	SGCA	SGCA, ADL, DAG2, LGMD2D, DMDA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2E, 604286, Otozomal Resesif; LGMD2E (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2E) (SGCB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2E, 604286, Autosomal recessive; LGMD2E (Autosomal recessive limb-girdle muscular dystrophy type 2E) (SGCB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCB	SGCB, LGMD2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2E (LGMD2E) [MIM:604286]: An autosomal recessive degenerative myopathy characterized by pelvic and shoulder muscle wasting, onset usually in childhood and variable progression rate. (ECO:0000269|PubMed:8968749, ECO:0000269|PubMed:9565988, ECO:0000269|PubMed:9631401). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2E, 604286, Otozomal Resesif; LGMD2E (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2E) (MLPA)	Muscular dystrophy, limb-girdle, type 2E, 604286, Autosomal recessive; LGMD2E (Autosomal recessive limb-girdle muscular dystrophy type 2E) (MLPA)	SGCB	SGCB, LGMD2E	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2E (LGMD2E) [MIM:604286]: An autosomal recessive degenerative myopathy characterized by pelvic and shoulder muscle wasting, onset usually in childhood and variable progression rate. (ECO:0000269|PubMed:8968749, ECO:0000269|PubMed:9565988, ECO:0000269|PubMed:9631401). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2F, 601287, Otozomal Resesif; LGMD2F (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2F) (SGCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2F, 601287, Autosomal recessive; LGMD2F (Autosomal recessive limb-girdle muscular dystrophy type 2F) (SGCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SGCD	SGCD, SGD, LGMD2F, CMD1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2F (LGMD2F) [MIM:601287]: An autosomal recessive degenerative myopathy initially affecting the proximal limb girdle musculature. Muscle from patients shows a complete loss of delta-sarcoglycan as well as of the others components of the sarcoglycan complex. (ECO:0000269	PubMed:9832045). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2F, 601287, Otozomal Resesif; LGMD2F (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 2F) (MLPA)	Muscular dystrophy, limb-girdle, type 2F, 601287, Autosomal recessive; LGMD2F (Autosomal recessive limb-girdle muscular dystrophy type 2F) (MLPA)	SGCD	SGCD, SGD, LGMD2F, CMD1L	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2F (LGMD2F) [MIM:601287]: An autosomal recessive degenerative myopathy initially affecting the proximal limb girdle musculature. Muscle from patients shows a complete loss of delta-sarcoglycan as well as of the others components of the sarcoglycan complex. (ECO:0000269	PubMed:9832045). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2G, 601954, Otozomal Resesif; LGMD2G (Otozomal Resesif Limb-Girdle Tipi Müsküler Distrofi Tip 2G) (TCAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2G, 601954, Autosomal recessive; LGMD2G (Autosomal recessive limb-girdle muscular dystrophy type 2G) (TCAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCAP	TCAP, LGMD2G, CMH2S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2G (LGMD2G) [MIM:601954]: An autosomal recessive degenerative myopathy characterized by proximal and distal muscle weakness and atrophy in the limbs, dystrophic changes on muscle biopsy, and absence of telethonin. Cardiac muscle is involved in a subset of patients. (ECO:0000269	PubMed:10655062). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2H, 254110, Otozomal Resesif; LGMD2H (Limb-Girdle Tipi Müsküler Distrofi, Tip 2H, Otozomal Resesif) (TRIM32 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2H, 254110, Autosomal recessive; LGMD2H (Autosomal recessive limb-girdle muscular dystrophy type 2H) (TRIM32 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIM32	TRIM32, HT2A, LGMD2H, BBS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2H (LGMD2H) [MIM:254110]: An autosomal recessive degenerative myopathy characterized by pelvic girdle, shoulder girdle and quadriceps muscle weakness. Clinical phenotype and severity are highly variable. Disease progression is slow and most patients remain ambulatory into the sixth decade of life. . (ECO:0000269|PubMed:11822024, ECO:0000269|PubMed:17994549). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Otozomal Resesif Tip 2J, 608807; LGMD2J (Limb-Girdle Tipi Müsküler Distrofi, Tip 2J, Otozomal Resesif) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2J, 608807, Autosomal recessive; LGMD2J (Autosomal recessive limb-girdle muscular dystrophy type 2J) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. (ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Müsküler Distrofi, Limb-Girdle, Tip 2 L, 611307, Otozomal Resesif; LGMD2L (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2L) (ANO5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2L, 611307, Autosomal recessive; LGMD2L (Autosomal recessive limb-girdle muscular dystrophy type 2L) (ANO5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO5	ANO5, TMEM16E, GDD1, LGMD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2L (LGMD2L) [MIM:611307]: An autosomal recessive degenerative myopathy characterized by proximal weakness, weakness of the hip and shoulder girdles and prominent asymmetrical quadriceps femoris and biceps brachii atrophy. {ECO:0000269	PubMed:20096397}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2Q, 613723, Otozomal Resesif; LGMD2Q (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2Q) (PLEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2Q, 613723, Autosomal recessive; LGMD2Q (Autosomal recessive limb-girdle muscular dystrophy type 2Q) (PLEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEC	PLEC1, PLEC, PLTN, EBS1, LGMD2Q, EBSOG, EBSPA, EBSMD, EBSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolysis bullosa simplex, Ogna type (O-EBS) [MIM:131950]: A form of intraepidermal epidermolysis bullosa characterized by generalized skin bruising, skin fragility with non-scarring blistering and small hemorrhagic blisters on hands. At the ultrastructural level, it is differentiated from classical cases of K-EBS, WC-EBS and DM-EBS, by the occurrence of blisters originating in basal cells above hemidesmosomes, and abnormal hemidesmosome intracellular attachment plates. {ECO:0000269	PubMed:11851880}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2R, 615325, Otozomal Resesif; LGMD2R (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2R) (DES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2R, 615325, Autosomal recessive; LGMD2R (Autosomal recessive limb-girdle muscular dystrophy type 2R) (DES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DES	DES, CMD1I, MFM1, SCPNK, ARVD7, ARVC7, LGMD2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurogenic scapuloperoneal syndrome Kaeser type (Kaeser syndrome) [MIM:181400]: Autosomal dominant disorder with a peculiar scapuloperoneal distribution of weakness and atrophy. A large clinical variability is observed ranging from scapuloperoneal, limb grindle and distal phenotypes with variable cardiac or respiratory involvement. Facial weakness, dysphagia and gynaecomastia are frequent additional symptoms. Affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Histological and immunohistochemical examination of muscle biopsy specimens reveal a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin. {ECO:0000269|PubMed:17439987, ECO:0000269|PubMed:25394388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2S, 615356, Otozomal Resesif; LGMD2S (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2S) (TRAPPC11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2S, 615356, Autosomal recessive; LGMD2S (Autosomal recessive limb-girdle muscular dystrophy type 2S) (TRAPPC11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAPPC11	TRAPPC11, C4orf41, LGMD2S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2S (LGMD2S) [MIM:615356]: A form of limb-girdle muscular dystrophy characterized by proximal muscle weakness with childhood onset, resulting in gait abnormalities and scapular winging. Serum creatine kinase is increased. A subset of patients may show a hyperkinetic movement disorder with chorea, ataxia, or dystonia and global developmental delay. {ECO:0000269	PubMed:23830518}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle,Tip 2W, 616827, Otozomal Resesif; LGMD2W (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2W) (LIMS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2W, 616827, Autosomal recessive; LGMD2W (Autosomal recessive limb-girdle muscular dystrophy type 2W) (LIMS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIMS2	LIMS2, PINCH2, LGMD2W	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2X, 616812, Otozomal Resesif; LGMD2X (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2X) (BVES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2X, 616812, Autosomal recessive; LGMD2X (Autosomal recessive limb-girdle muscular dystrophy type 2X) (BVES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BVES	BVES, HBVES, POPDC1, LGMD2X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2Y, 617072, Otozomal Resesif; LGMD2Y (Otozomal Resesif Limb-Girdle Müsküler Distrofi Tip 2Y) (TOR1AIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2Y, 617072, Autosomal recessive; LGMD2Y (Muscular dystrophy with progressive weakness, distal contractures and rigid spine) (TOR1AIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOR1AIP1	TOR1AIP1, LAP1, LAP1B, LGMD2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 2Z, 617232, Otozomal Resesif; LGMD2Z (Dowling-Degos Hastalığı) (POGLUT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type 2Z, 617232, Autosomal recessive; LGMD2Z (Dowling-Degos disease) (POGLUT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POGLUT1	POGLUT1, CLP46, KTELC1, RUMI, C3orf9, DDD4, LGMD2Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1C, 607801, Otozomal Resesif, Otozomal Dominant (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1C) (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, limb-girdle, type IC, 607801, Autosomal recessive, Autosomal dominant (Autosomal dominant limb-girdle muscular dystrophy type 1C) (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Limb-Girdle, Tip 1C, 607801, Otozomal Resesif, Otozomal Dominant (Otozomal Dominant Limb-Girdle Müsküler Distrofi Tip 1C) (MLPA)	Muscular dystrophy, limb-girdle, type IC, 607801, Autosomal recessive, Autosomal dominant (Autosomal dominant limb-girdle muscular dystrophy type 1C) (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Sert Omurga, 1, 602771, Otozomal Resesif; RSMD1 (Multiminikore Miyopati) (SELENON geni) (Tüm Gen Dizi Analizi) (Postnatal)	Muscular dystrophy, rigid spine, 1, 602771, Autosomal recessive; RSMD1 (Multiminicore myopathy) (SELENON gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SELENON	SELENON, SEPN1, SELN, RSMD1, CFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Müsküler Distrofi, Sert Omurga, 1, 602771, Otozomal Resesif; RSMD1 (Multiminikore Miyopati) (Prenatal) (SELENON geni) (Tüm Gen Dizi Analizi) (Prenatal)	Muscular dystrophy, rigid spine, 1, 602771, Autosomal recessive; RSMD1 (Multiminicore myopathy) (SELENON gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SELENON	SELENON, SEPN1, SELN, RSMD1, CFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyasteni, Konjenital, 12, Tübüler Agregatlar İle, 610542, Otozomal Resesif; CMS12 (Konjenital Miyastenik Sendrom, Glikozilasyon Defekti) (GFPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenia, congenital, 12, with tubular aggregates, 610542, Autosomal recessive; CMS12 (Congenital myasthenic syndromes with glycosylation defect) (GFPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFPT1	GFPT1, GFAT1, GFPT1L, MSLG, CMS12, CMSTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyasteni, Konjenital, 12, Tübüler Agregatlar İle, 610542, Otozomal Resesif; CMS12 (Konjenital Miyastenik Sendrom, Glikozilasyon Defekti) (Prenatal) (GFPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenia, congenital, 12, with tubular aggregates, 610542, Autosomal recessive; CMS12 (Congenital myasthenic syndromes with glycosylation defect) (GFPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFPT1	GFPT1, GFAT1, GFPT1L, MSLG, CMS12, CMSTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 10, 254300, Otozomal Resesif; CMS10 (Konjenital Miyastenik Sendrom) (DOK7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 10, 254300, Autosomal recessive; CMS10 (Congenital myasthenic syndrome) (DOK7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DOK7	DOK7, C4orf25, CMS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 10 (CMS10) [MIM:254300]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS10 is an autosomal recessive, post-synaptic form characterized by a typical 'limb girdle' pattern of muscle weakness with small, simplified neuromuscular junctions but normal acetylcholine receptor and acetylcholinesterase function. {ECO:0000269|PubMed:16917026, ECO:0000269|PubMed:17439981, ECO:0000269|PubMed:20012313, ECO:0000269|PubMed:22661499}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 10, 254300, Otozomal Resesif; CMS10 (Konjenital Miyastenik Sendrom) (Prenatal) (DOK7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 10, 254300, Autosomal recessive; CMS10 (Congenital myasthenic syndrome) (DOK7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DOK7	DOK7, C4orf25, CMS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 10 (CMS10) [MIM:254300]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS10 is an autosomal recessive, post-synaptic form characterized by a typical 'limb girdle' pattern of muscle weakness with small, simplified neuromuscular junctions but normal acetylcholine receptor and acetylcholinesterase function. {ECO:0000269|PubMed:16917026, ECO:0000269|PubMed:17439981, ECO:0000269|PubMed:20012313, ECO:0000269|PubMed:22661499}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 11, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616326, Otozomal Resesif; CMS11 (Konjenital Miyastenik Sendrom) (RAPSN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326, Autosomal recessive; CMS11 (Congenital myasthenic syndrome) (RAPSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAPSN	RAPSN, CMS1D, CMS11, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:18179903, ECO:0000269|PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 11, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616326, Otozomal Resesif; CMS11 (Konjenital Miyastenik Sendrom) (Prenatal) (RAPSN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 11, associated with acetylcholine receptor deficiency, 616326, Autosomal recessive; CMS11 (Congenital myasthenic syndrome) (RAPSN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAPSN	RAPSN, CMS1D, CMS11, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:18179903, ECO:0000269|PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyasenik Sendrom, Konjenital, 13, Tübüler Agregatlar, 614750, Otozomal Resesif; CMS13 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (DPAGT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750, Autosomal recessive; CMS13 (Congenital myasthenic syndromes with glycosylation defect) (DPAGT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPAGT1	DPAGT1, DPAGT2, DGPT, CDG1J, CMSTA2, CMS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1J (CDG1J) [MIM:608093]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12872255). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyasenik Sendrom, Konjenital, 13, Tübüler Agregatlar, 614750, Otozomal Resesif; CMS13 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (Prenatal) (DPAGT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 13, with tubular aggregates, 614750, Autosomal recessive; CMS13 (Congenital myasthenic syndromes with glycosylation defect) (DPAGT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DPAGT1	DPAGT1, DPAGT2, DGPT, CDG1J, CMSTA2, CMS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1J (CDG1J) [MIM:608093]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12872255). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyasenik Sendrom, Konjenital, 14, Tübüler Agregatlar, 616228, Otozomal Resesif; CMS14 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (ALG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228, Autosomal recessive; CMS14 (Congenital myasthenic syndromes with glycosylation defect) (ALG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG2	ALG2, CDG1I, CMSTA3, CMS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 1I (CDG1I) [MIM:607906]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12684507). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyasenik Sendrom, Konjenital, 14, Tübüler Agregatlar, 616228, Otozomal Resesif; CMS14 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (Prenatal) (ALG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 14, with tubular aggregates, 616228, Autosomal recessive; CMS14 (Congenital myasthenic syndromes with glycosylation defect) (ALG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG2	ALG2, CDG1I, CMSTA3, CMS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 1I (CDG1I) [MIM:607906]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. (ECO:0000269	PubMed:12684507). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyasenik Sendrom, Konjenital, 15, Tübüler Agregatlar Olmadan, 616227; CMS15 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (ALG14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227; CMS15 (Congenital myasthenic syndromes with glycosylation defect) (ALG14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALG14	ALG14, CMS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 15 (CMS15) [MIM:616227]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. {ECO:0000269	PubMed:23404334}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Miyasenik Sendrom, Konjenital, 15, Tübüler Agregatlar Olmadan, 616227; CMS15 (Konjenital Miyasenik Sendrom-Glikozilasyon Defekti) (Prenatal) (ALG14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 15, without tubular aggregates, 616227; CMS15 (Congenital myasthenic syndromes with glycosylation defect) (ALG14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALG14	ALG14, CMS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 15 (CMS15) [MIM:616227]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. {ECO:0000269	PubMed:23404334}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 16, 614198, Otozomal Resesif; CMS16 (Konjenital Miyastenik Sendrom) (SCN4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 16, 614198, Autosomal recessive; CMS16 (Congenital myasthenic syndrome) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 16, 614198, Otozomal Resesif; CMS16 (Konjenital Miyastenik Sendrom) (SCN4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 16, 614198, Autosomal recessive; CMS16 (Congenital myasthenic syndrome) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 16, 614198, Otozomal Resesif; CMS16 (Konjenital Miyastenik Sendrom) (Prenatal) (MLPA)	Myasthenic syndrome, congenital, 16, 614198, Autosomal recessive; CMS16 (Congenital myasthenic syndrome) (Prenatal) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyastenik Sendrom, Konjenital, 17, 616304, Otozomal Resesif; CMS17 (Konjenital Miyastenik Sendrom) (LRP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 17, 616304, Autosomal recessive; CMS17 (Congenital myasthenic syndrome) (LRP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP4	LRP4, MEGF7, CLSS, SOST2, CMS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sclerosteosis 2 (SOST2) [MIM:614305]: A sclerosing bone dysplasia characterized by a generalized hyperostosis and sclerosis leading to a markedly thickened skull, with mandible, ribs, clavicles and all long bones also being affected. Due to narrowing of the foramina of the cranial nerves, facial nerve palsy, hearing loss and atrophy of the optic nerves can occur. Sclerosteosis is clinically and radiologically very similar to van Buchem disease, mainly differentiated by hand malformations and a large stature in sclerosteosis patients. {ECO:0000269|PubMed:21471202, ECO:0000269|PubMed:24234652}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 17, 616304, Otozomal Resesif; CMS17 (Konjenital Miyastenik Sendrom) (Prenatal) (LRP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 17, 616304, Autosomal recessive; CMS17 (Congenital myasthenic syndrome) (LRP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP4	LRP4, MEGF7, CLSS, SOST2, CMS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sclerosteosis 2 (SOST2) [MIM:614305]: A sclerosing bone dysplasia characterized by a generalized hyperostosis and sclerosis leading to a markedly thickened skull, with mandible, ribs, clavicles and all long bones also being affected. Due to narrowing of the foramina of the cranial nerves, facial nerve palsy, hearing loss and atrophy of the optic nerves can occur. Sclerosteosis is clinically and radiologically very similar to van Buchem disease, mainly differentiated by hand malformations and a large stature in sclerosteosis patients. {ECO:0000269|PubMed:21471202, ECO:0000269|PubMed:24234652}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 18, 616330, Otozomal Dominant; CMS18 (Konjenital Miyastenik Sendrom) (SNAP25 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 18, 616330, Autosomal dominant; CMS18 (Congenital myasthenic syndrome) (SNAP25 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNAP25	SNAP25, CMS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 18 (CMS18) [MIM:616330]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS18 is an autosomal dominant presynaptic disorder clinically characterized by early-onset muscle weakness and easy fatigability associated with delayed psychomotor development and ataxia. {ECO:0000269|PubMed:25381298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 18, 616330, Otozomal Dominant; CMS18 (Konjenital Miyastenik Sendrom) (Prenatal) (SNAP25 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 18, 616330, Autosomal dominant; CMS18 (Congenital myasthenic syndrome) (SNAP25 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNAP25	SNAP25, CMS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 18 (CMS18) [MIM:616330]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS18 is an autosomal dominant presynaptic disorder clinically characterized by early-onset muscle weakness and easy fatigability associated with delayed psychomotor development and ataxia. {ECO:0000269|PubMed:25381298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 19, 616720, Otozomal Resesif; CMS19 (Konjenital Miyastenik Sendrom) (COL13A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 19, 616720, Autosomal recessive; CMS19 (Congenital myasthenic syndrome) (COL13A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL13A1	COL13A1, CMS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 19, 616720, Otozomal Resesif; CMS19 (Konjenital Miyastenik Sendrom) (Prenatal) (COL13A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 19, 616720, Autosomal recessive; CMS19 (Congenital myasthenic syndrome) (COL13A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL13A1	COL13A1, CMS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 1A, Yavaş Kanal, 601462, Otozomal Dominant; CMS1A (Konjenital Miyastenik Sendrom) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 1A, slow-channel, 601462, Autosomal dominant; CMS1A (Congenital myasthenic syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 1A, Yavaş Kanal, 601462, Otozomal Dominant; CMS1A (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 1A, slow-channel, 601462, Autosomal dominant; CMS1A (Congenital myasthenic syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 1B, Hızlı Kanal, 608930, Otozomal Resesif, Otozomal Dominant; CMS1B (Konjenital Miyastenik Sendrom) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 1B, fast-channel, 608930, Autosomal recessive, Autosomal dominant; CMS1B (Congenital myasthenic syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 1B, Hızlı Kanal, 608930, Otozomal Resesif, Otozomal Dominant; CMS1B (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 1B, fast-channel, 608930, Autosomal recessive, Autosomal dominant; CMS1B (Congenital myasthenic syndrome) (CHRNA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNA1	CHRNA1, ACHRD, CMS1B, CMS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 1A, slow-channel (CMS1A) [MIM:601462]: A common congenital myasthenic syndrome. Congenital myasthenic syndromes are characterized by muscle weakness affecting the axial and limb muscles (with hypotonia in early- onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS1A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:16685696, ECO:0000269|PubMed:7619526, ECO:0000269|PubMed:8872460, ECO:0000269|PubMed:9158151, ECO:0000269|PubMed:9221765}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 20, Presinaptik, 617143, Otozomal Resesif; CMS20 (Konjenital Miyastenik Sendrom) (SLC5A7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 20, presynaptic, 617143, Autosomal recessive; CMS20 (Congenital myasthenic syndrome) (SLC5A7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC5A7	SLC5A7, CHT1, HMN7A, CMS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 7A (HMN7A) [MIM:158580]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN7A is characterized by onset in the second decade of progressive distal muscle wasting and weakness affecting the upper and lower limbs and resulting in walking difficulties and hand grip. There is significant muscle atrophy of the hands and lower limbs. The disorder is associated with vocal cord paresis due to involvement of the tenth cranial nerve. {ECO:0000269|PubMed:23141292}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Miyastenik Sendrom, Konjenital, 20, Presinaptik, 617143, Otozomal Resesif; CMS20 (Konjenital Miyastenik Sendrom) (Prenatal) (SLC5A7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 20, presynaptic, 617143, Autosomal recessive; CMS20 (Congenital myasthenic syndrome) (SLC5A7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC5A7	SLC5A7, CHT1, HMN7A, CMS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuronopathy, distal hereditary motor, 7A (HMN7A) [MIM:158580]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN7A is characterized by onset in the second decade of progressive distal muscle wasting and weakness affecting the upper and lower limbs and resulting in walking difficulties and hand grip. There is significant muscle atrophy of the hands and lower limbs. The disorder is associated with vocal cord paresis due to involvement of the tenth cranial nerve. {ECO:0000269	PubMed:23141292}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 21, Presinaptik, 617239, Otozomal Resesif; CMS21 (Konjenital Miyastenik Sendrom) (SLC18A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 21, presynaptic, 617239, Autosomal recessive; CMS21 (Congenital myasthenic syndrome) (SLC18A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC18A3	SLC18A3, VACHT, CMS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 21, Presinaptik, 617239, Otozomal Resesif; CMS21 (Konjenital Miyastenik Sendrom) (Prenatal) (SLC18A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 21, presynaptic, 617239, Autosomal recessive; CMS21 (Congenital myasthenic syndrome) (SLC18A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC18A3	SLC18A3, VACHT, CMS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 22, 616224; CMS22 (Konjenital Miyastenik Sendrom) (PREPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 22, 616224; CMS22 (Congenital myasthenic syndrome) (PREPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PREPL	PREPL, KIAA0436, CMS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 22, 616224; CMS22 (Konjenital Miyastenik Sendrom) (Prenatal) (PREPL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 22, 616224; CMS22 (Congenital myasthenic syndrome) (PREPL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PREPL	PREPL, KIAA0436, CMS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 2A, Yavaş Kanal, 616313, Otozomal Dominant; CMS2A (Konjenital Miyastenik Sendrom) (CHRNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 2A, slow-channel, 616313, Autosomal dominant; CMS2A (Congenital myasthenic syndrome) (CHRNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNB1	CHRNB1, ACHRB, SCCMS, CMS2A, CMS2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 2A, slow-channel (CMS2A) [MIM:616313]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS2A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:8651643, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 2A, Yavaş Kanal, 616313, Otozomal Dominant; CMS2A (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 2A, slow-channel, 616313, Autosomal dominant; CMS2A (Congenital myasthenic syndrome) (CHRNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNB1	CHRNB1, ACHRB, SCCMS, CMS2A, CMS2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 2A, slow-channel (CMS2A) [MIM:616313]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS2A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:8651643, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 2C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616314, Otozomal Resesif; CMS2A (Konjenital Miyastenik Sendrom) (CHRNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314, Autosomal recessive; CMS2A (Congenital myasthenic syndrome) (CHRNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNB1	CHRNB1, ACHRB, SCCMS, CMS2A, CMS2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 2A, slow-channel (CMS2A) [MIM:616313]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS2A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:8651643, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 2C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616314, Otozomal Resesif; CMS2A (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 2C, associated with acetylcholine receptor deficiency, 616314, Autosomal recessive; CMS2A (Congenital myasthenic syndrome) (CHRNB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNB1	CHRNB1, ACHRB, SCCMS, CMS2A, CMS2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 2A, slow-channel (CMS2A) [MIM:616313]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS2A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:8651643, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 3A, Yavaş Kanal, 616321, Otozomal Dominant; CMS3A (Konjenital Miyastenik Sendrom) (CHRND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 3A, slow-channel, 616321, Autosomal dominant; CMS3A (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 3A, Yavaş Kanal, 616321, Otozomal Dominant; CMS3A (Konjenital Miyastenik Sendrom) (Prenatal) (CHRND geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 3A, slow-channel, 616321, Autosomal dominant; CMS3A (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 3B, Hızlı Kanal, 616322, Otozomal Resesif; CMS3B (Konjenital Miyastenik Sendrom) (CHRND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 3B, fast-channel, 616322, Autosomal recessive; CMS3B (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 3B, Hızlı Kanal, 616322, Otozomal Resesif; CMS3B (Konjenital Miyastenik Sendrom) (Prenatal) (CHRND geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 3B, fast-channel, 616322, Autosomal recessive; CMS3B (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 3C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616323, Otozomal Resesif; CMS3C (Konjenital Miyastenik Sendrom) (CHRND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323, Autosomal recessive; CMS3C (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Miyastenik Sendrom, Konjenital, 3C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 616323, Otozomal Resesif; CMS3C (Konjenital Miyastenik Sendrom) (Prenatal) (CHRND geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 3C, associated with acetylcholine receptor deficiency, 616323, Autosomal recessive; CMS3C (Congenital myasthenic syndrome) (CHRND gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRND	CHRND, ACHRD, SCCMS, CMS3A, CMS3B, CMS3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple pterygium syndrome, lethal type (LMPS) [MIM:253290]: Multiple pterygia are found infrequently in children with arthrogryposis and in fetuses with fetal akinesia syndrome. In lethal multiple pterygium syndrome there is intrauterine growth retardation, multiple pterygia, and flexion contractures causing severe arthrogryposis and fetal akinesia. Subcutaneous edema can be severe, causing fetal hydrops with cystic hygroma and lung hypoplasia. Oligohydramnios and facial anomalies are frequent. {ECO:0000269	PubMed:18252226}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4A, Yavaş Kanal, 605809, Otozomal Resesif, Otozomal Dominant; CMS4A (Konjenital Miyastenik Sendrom) (CHRNE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 4A, slow-channel, 605809, Autosomal recessive, Autosomal dominant; CMS4A (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4A, Yavaş Kanal, 605809, Otozomal Resesif, Otozomal Dominant; CMS4A (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 4A, slow-channel, 605809, Autosomal recessive, Autosomal dominant; CMS4A (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4B Hızlı Kanal, 616324, Otozomal Resesif; CMS4B (Konjenital Miyastenik Sendrom) (CHRNE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 4B, fast-channel, 616324, Autosomal recessive; CMS4B (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4B Hızlı Kanal, 616324, Otozomal Resesif; CMS4B (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 4B, fast-channel, 616324, Autosomal recessive; CMS4B (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 608931, Otozomal Resesif; CMS4C (Konjenital Miyastenik Sendrom) (CHRNE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931, Autosomal recessive; CMS4C (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 4C, Asetilkolin Reseptörü Eksikliği İle İlişkili, 608931, Otozomal Resesif; CMS4C (Konjenital Miyastenik Sendrom) (Prenatal) (CHRNE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 4C, associated with acetylcholine receptor deficiency, 608931, Autosomal recessive; CMS4C (Congenital myasthenic syndrome) (CHRNE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRNE	CHRNE, SCCMS, CMS4A, CMS4B, CMS4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 4A, slow-channel (CMS4A) [MIM:605809]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS4A is a slow-channel myasthenic syndrome. It is caused by kinetic abnormalities of the AChR, resulting in prolonged AChR channel opening episodes, prolonged endplate currents, and depolarization block. This is associated with calcium overload, which may contribute to subsequent degeneration of the endplate and postsynaptic membrane. {ECO:0000269|PubMed:12141316, ECO:0000269|PubMed:7531341, ECO:0000269|PubMed:7538206, ECO:0000269|PubMed:8872460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 5, 603034, Otozomal Resesif; CMS5 (Konjenital Miyastenik Sendrom) (COLQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 5, 603034, Autosomal recessive; CMS5 (Congenital myasthenic syndrome) (COLQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COLQ	COLQ, EAD, CMS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 5 (CMS5) [MIM:603034]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS5 inheritance is autosomal recessive. {ECO:0000269|PubMed:10665486, ECO:0000269|PubMed:11865139, ECO:0000269|PubMed:24938146, ECO:0000269|PubMed:9758617}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Miyastenik Sendrom, Konjenital, 5, 603034, Otozomal Resesif; CMS5 (Konjenital Miyastenik Sendromi (Prenatal) (COLQ gen) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 5, 603034, Autosomal recessive; CMS5 (Congenital myasthenic syndrome) (COLQ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COLQ	COLQ, EAD, CMS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 5 (CMS5) [MIM:603034]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS5 inheritance is autosomal recessive. {ECO:0000269|PubMed:10665486, ECO:0000269|PubMed:11865139, ECO:0000269|PubMed:24938146, ECO:0000269|PubMed:9758617}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 6, Presinaptik, 254210, Otozomal Resesif; CMS6 (Konjenital Miyastenik Sendromi) (CHAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 6, presynaptic, 254210, Autosomal recessive; CMS6 (Congenital myasthenic syndrome) (CHAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHAT	CHAT, CMS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 6, presynaptic (CMS6) [MIM:254210]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS6 affected individuals have myasthenic symptoms since birth or early infancy, negative tests for anti-AChR antibodies, and abrupt episodic crises with increased weakness, bulbar paralysis, and apnea precipitated by undue exertion, fever, or excitement. CMS6 inheritance is autosomal recessive. {ECO:0000269|PubMed:11172068, ECO:0000269|PubMed:12756141}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 6, Presinaptik, 254210, Otozomal Resesif; CMS6 (Konjenital Miyastenik Sendromi) (Prenatal) (CHAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 6, presynaptic, 254210, Autosomal recessive; CMS6 (Congenital myasthenic syndrome) (CHAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHAT	CHAT, CMS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 6, presynaptic (CMS6) [MIM:254210]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness affecting the axial and limb muscles (with hypotonia in early-onset forms), the ocular muscles (leading to ptosis and ophthalmoplegia), and the facial and bulbar musculature (affecting sucking and swallowing, and leading to dysphonia). The symptoms fluctuate and worsen with physical effort. CMS6 affected individuals have myasthenic symptoms since birth or early infancy, negative tests for anti-AChR antibodies, and abrupt episodic crises with increased weakness, bulbar paralysis, and apnea precipitated by undue exertion, fever, or excitement. CMS6 inheritance is autosomal recessive. {ECO:0000269|PubMed:11172068, ECO:0000269|PubMed:12756141}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 7, Presinaptik, 616040, Otozomal Dominant; CMS7 (Konjenital Miyastenik Sendromi) (SYT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 7, presynaptic, 616040, Autosomal dominant; CMS7 (Congenital myasthenic syndrome) (SYT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYT2	SYT2, CMS7, MYSPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 7, presynaptic (CMS7) [MIM:616040]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. CMS7 is an autosomal dominant, presynaptic disorder resembling Lambert-Eaton myasthenic syndrome. Affected individuals have a variable degree of proximal and distal limb weakness, muscle fatigue that improves with rest, mild gait difficulties, and reduced or absent deep tendon reflexes.. {ECO:0000269|PubMed:25192047}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 7, Presinaptik, 616040, Otozomal Dominant; CMS7 (Konjenital Miyastenik Sendromi) (Prenatal) (SYT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 7, presynaptic, 616040, Autosomal dominant; CMS7 (Congenital myasthenic syndrome) (SYT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYT2	SYT2, CMS7, MYSPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 7, presynaptic (CMS7) [MIM:616040]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. CMS7 is an autosomal dominant, presynaptic disorder resembling Lambert-Eaton myasthenic syndrome. Affected individuals have a variable degree of proximal and distal limb weakness, muscle fatigue that improves with rest, mild gait difficulties, and reduced or absent deep tendon reflexes. {ECO:0000269|PubMed:25192047}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 8, Pre ve Post- Sinaptik Defekli, 615120, Otozomal Resesif; CMS8 (Konjenital Miyastenik Sendromi) (AGRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120, Autosomal recessive; CMS8 (Congenital myasthenic syndrome) (AGRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGRN	AGRN, CMS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myasthenic syndrome, congenital, 8 (CMS8) [MIM:615120]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. CMS8 is an autosomal recessive disease characterized by prominent defects of both the pre- and postsynaptic regions. Affected individuals have onset of muscle weakness in early childhood; the severity of the weakness and muscles affected is variable. {ECO:0000269|PubMed:19631309, ECO:0000269|PubMed:22205389, ECO:0000269|PubMed:24951643}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 8, Pre ve Post- Sinaptik Defekli, 615120, Otozomal Resesif; CMS8 (Konjenital Miyastenik Sendromi) (Prenatal) (AGRN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myasthenic syndrome, congenital, 8, with pre- and postsynaptic defects, 615120, Autosomal recessive; CMS8 (Congenital myasthenic syndrome) (AGRN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGRN	AGRN, CMS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myasthenic syndrome, congenital, 8 (CMS8) [MIM:615120]: A form of congenital myasthenic syndrome, a group of disorders characterized by failure of neuromuscular transmission, including pre-synaptic, synaptic, and post-synaptic disorders that are not of autoimmune origin. Clinical features are easy fatigability and muscle weakness. CMS8 is an autosomal recessive disease characterized by prominent defects of both the pre- and postsynaptic regions. Affected individuals have onset of muscle weakness in early childhood; the severity of the weakness and muscles affected is variable. {ECO:0000269|PubMed:19631309, ECO:0000269|PubMed:22205389, ECO:0000269|PubMed:24951643}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyastenik Sendrom, Konjenital, 9, Asetilkolin Reseptörü Eksikliği İle İlişkili 616325, Otozomal Resesif; CMS9 (Konjenital Miyastenik Sendromi) (MUSK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myasthenic syndrome, congenital, 9, associated with acetylcholine receptor deficiency, 616325, Autosomal recessive; CMS9 (Congenital myasthenic syndrome) (MUSK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUSK	MUSK, CMS9, FADS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fetal akinesia deformation sequence (FADS) [MIM:208150]: A clinically and genetically heterogeneous group of disorders with congenital malformations related to impaired fetal movement. Clinical features include fetal akinesia, intrauterine growth retardation, polyhydramnios, arthrogryposis, pulmonary hypoplasia, craniofacial abnormalities, and cryptorchidism. {ECO:0000269|PubMed:25537362, ECO:0000269|PubMed:25612909}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
MYB (FISH)	MYB (FISH)	6q23.3	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Mycobacterium Tuberculosis, Karşı Koruma, 607948 (Tüberküloz) (MC3R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, protection against, 607948 (Tuberculosis) (MC3R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC3R	MC3R, BMIQ9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mycobacterium Tuberculosis, Karşı Koruma, 607948 (Tüberküloz) (IRGM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, protection against, 607948 (Tuberculosis) (IRGM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRGM	IRGM, LRG47, IFI1, IBD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 19 (IBD19) [MIM:612278]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:17554261, ECO:0000269|PubMed:19174780, ECO:0000269|PubMed:21278745}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Mycobacterium Tuberculosis, Erteklsiyona Karşı Hassasiyet, 607948 (Tüberküloz) (SLC11A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, susceptibility to infection by, 607948 (Tuberculosis) (SLC11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC11A1	NRAMP1, NRAMP, SLC11A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mycobacterium Tuberculosis, Yatkınlık, 607948 (TLR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, susceptibility to, 607948 (TLR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR2	TLR2, TIL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mycobacterium Tuberculosis, Yatkınlık, 607948 (Tüberküloz) (SP110 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, susceptibility to, 607948 (Tuberculosis) (SP110 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SP110	SP110, IFI41, IFI75, VODI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic venocclusive disease with immunodeficiency (VODI) [MIM:235550]: Autosomal recessive primary immunodeficiency associated with hepatic vascular occlusion and fibrosis. The immunodeficiency is characterized by severe hypogammaglobulinemia, combined T and B-cell immunodeficiency, absent lymph node germinal centers, and absent tissue plasma cells. {ECO:0000269|PubMed:16648851}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Mycobacterium Tuberculosis, Yatkınlık, 607948 (Tüberküloz) (CD209 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, susceptibility to, 607948 (Tuberculosis) (CD209 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD209	CD209, CDSIGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mycobacterium Tuberculosis, Yatkınlık, 607948 (Tüberküloz) (CCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Mycobacterium tuberculosis, susceptibility to, 607948 (Tuberculosis) (CCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL2	CCL2, SCYA2, MCP1, MCAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyelodisplazi ve Lösemi Sendromu + Monozomi 7, 252270, Otozomal Resesif (200)	Myelodysplasia and leukemia syndrome with monosomy 7, 252270, Autosomal recessive (440)	.	MLSM7, DEL7q, C7DELq	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Miyelodisplastik Sendrom (ACSL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome (ACSL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACSL6	ACSL6, FACL6, ACS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving ACSL6 may be a cause of myelodysplastic syndrome with basophilia. Translocation t(5;12)(q31;p13) with ETV6. {ECO:0000269|PubMed:10923161,	EDTA Blood Sample

Miyelodisplastik Sendrom, Prelösemik (IRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome, preleukemic (IRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF1	IRF1, MAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gastric cancer (GASC) [MIM:613659]: A malignant disease which starts in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. The term gastric cancer or gastric carcinoma refers to adenocarcinoma of the stomach that accounts for most of all gastric malignant tumors. Two main histologic types are recognized, diffuse type and intestinal type carcinomas. Diffuse tumors are poorly differentiated infiltrating lesions, resulting in thickening of the stomach. In contrast, intestinal tumors are usually exophytic, often ulcerating, and associated with intestinal metaplasia of the stomach, most often observed in sporadic disease. {ECO:0000269|PubMed:10395927, ECO:0000269|PubMed:9679752}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyelodisplastik Sendrom, Somatik, 614286 (Miyelodisplastik Sendromi) (TET2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome, somatic, 614286 (Myelodysplastic syndrome) (TET2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TET2	TET2, KIAA1546, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Miyelodisplastik Sendrom, Somatik, 614286 (Miyelodisplastik Sendrom) (SF3B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome, somatic, 614286 (Myelodysplastic syndrome) (SF3B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SF3B1	SF3B1, SF3B155, SAP155, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Miyelodisplastik Sendrom, Somatik, 614286 (Miyelodisplastik Sendrom) (ASXL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome, somatic, 614286 (Myelodysplastic syndrome) (ASXL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASXL1	ASXL1, KIAA0978, BOPS, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Miyelodisplastik Sendrom, Yatkınlık, 614286, MDS (Miyelodisplastik Sendrom) (GATA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelodysplastic syndrome, susceptibility to, 614286; MDS (Myelodysplastic syndrome) (GATA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA2	GATA2, DCML, MONOMAC, IMD21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphedema, primary, with myelodysplasia (LMPM) [MIM:614038]: A chronic disabling condition characterized by swelling of the extremities due to altered lymphatic flow, associated with myelodysplasia. Patients with lymphedema suffer from recurrent local infections, and physical impairment. {ECO:0000269|PubMed:21892158}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyelofibrozis, Miyeloid Metaplazi, Somatik, 254450 (Miyeloid Metaplazi- Miyelofibrozis) (MPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelofibrosis with myeloid metaplasia, somatic, 254450 (Myelofibrosis with myeloid metaplasia) (MPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPL	MPL, TPOR, MPLV, THCYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Myelofibrosis with myeloid metaplasia (MMM) [MIM:254450]: A chronic myeloproliferative disorder characterized by replacement of the bone marrow by fibrous tissue, extramedullary hematopoiesis, anemia, leukoerythroblastosis and hepatosplenomegaly. {ECO:0000269|PubMed:16834459, ECO:0000269|PubMed:16868251}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA blood sample / EDTA bone marrow
Miyelofibrozis, Somatik, 254450 (Miyeloid Metaplazi- Miyelofibrozis) (SH2B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelofibrosis, somatic, 254450 (Myelofibrosis with myeloid metaplasia) (SH2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH2B3	SH2B3, LNK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Miyelofibrozis, Somatik, 254450 (Miyeloid Metaplazi- Miyelofibrozis) V617F Mutasyonu ve (Ekzon 12-14) (JAK2 geni) (Dizi Analizi) (Postnatal)	Myelofibrosis, somatic, 254450 (Myelofibrosis with myeloid metaplasia) V617F Mutation and (Exon 12-14) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	JAK2, THCYT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Myelofibrosis (MYELOF) [MIM:254450]: A disorder characterized by replacement of the bone marrow by fibrous tissue, occurring in association with a myeloproliferative disorder. Clinical manifestations may include anemia, pallor, splenomegaly, hypermetabolic state, petechiae, ecchymosis, bleeding, lymphadenopathy, hepatomegaly, portal hypertension. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA blood sample / EDTA bone marrow
Miyelofibrozis, Somatik, 254450 (Miyeloid Metaplazi- Miyelofibrozis) (CALR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelofibrosis, somatic, 254450 (Myelofibrosis with myeloid metaplasia) (CALR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALR	CALR, SSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Miyeloid Lösemi, Akut (IRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelogenous leukemia, acute (IRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF1	IRF1, MAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gastric cancer (GASC) [MIM:613659]: A malignant disease which starts in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. The term gastric cancer or gastric carcinoma refers to adenocarcinoma of the stomach that accounts for most of all gastric malignant tumors. Two main histologic types are recognized, diffuse type and intestinal type carcinomas. Diffuse tumors are poorly differentiated infiltrating lesions, resulting in thickening of the stomach. In contrast, intestinal tumors are usually exophytic, often ulcerating, and associated with intestinal metaplasia of the stomach, most often observed in sporadic disease. {ECO:0000269|PubMed:10395927, ECO:0000269|PubMed:9679752}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyeloid Lösemi, Akut (ACSL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelogenous leukemia, acute (ACSL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACSL6	ACSL6, FACL6, ACS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving ACSL6 may be a cause of myelodysplastic syndrome with basophilia. Translocation t(5;12)(q31;p13) with ETV6. {ECO:0000269|PubMed:10903516}.	EDTA Blood Sample
Miyeloid Lösemi, Akut, M4 / M4Eo Alt Tipi Somatik, 601626 (Sınıflandırılmamış Akut Miyeloid Lösemi) (CBFB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myeloid leukemia, acute, M4/M4Eo subtype, somatic, 601626 (Unclassified acute myeloid leukemia) (CBFB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBFB	CBFB, PEBP2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving CBFB is associated with acute myeloid leukemia of M4EO subtype. Pericentric inversion inv(16)(p13.q22). The inversion produces a fusion protein that consists of the 165 N-terminal residues of CBF-beta (PEPB2) with the tail region of MYH11. {ECO:0000269|PubMed:8351518}.	EDTA blood sample / EDTA bone marrow
Miyelokatekxis, İzole (CXCR4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myelokathexis, isolated (CXCR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CXCR4	CXCR4, D2S201E, NPY3R, WHIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: WHIM syndrome (WHIMS) [MIM:193670]: Immunodeficiency disease characterized by neutropenia, hypogammaglobulinemia and extensive human papillomavirus (HPV) infection. Despite the peripheral neutropenia, bone marrow aspirates from affected individuals contain abundant mature myeloid cells, a condition termed myelokathexis. {ECO:0000269|PubMed:12692554}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyeloperoksidaz Eksikliği, 254600, Otozomal Resesif; MPOD (Miyeloperoksidaz Eksikliği) (MPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myeloperoxidase deficiency, 254600, Autosomal recessive; MPOD (Myeloperoxidase deficiency) (MPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPO	MPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloperoxidase deficiency (MPOD) [MIM:254600]: A disorder characterized by decreased myeloperoxidase activity in neutrophils and monocytes that results in disseminated candidiasis. {ECO:0000269|PubMed:7904599, ECO:0000269|PubMed:8142659, ECO:0000269|PubMed:9354683, ECO:0000269|PubMed:9637725}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyeloproliferatif Bozukluk-Eozinofili, 131440, Otozomal Dominant (Sınıflandırılmamış Kronik Miyeloproliferatif Hastalık) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myeloproliferative disorder with eosinophilia, 131440, Autosomal dominant (Unclassified chronic myeloproliferative disease) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloproliferative disorder chronic with eosinophilia (MPE) [MIM:131440]: A hematologic disorder characterized by malignant eosinophils proliferation. Note=The gene represented in this entry may be involved in disease pathogenesis. Chromosomal aberrations involving PDGFRB have been found in many instances of chronic myeloproliferative disorder with eosinophilia. Translocation t(5;12) with ETV6 on chromosome 12 creating an PDGFRB-ETV6 fusion protein (PubMed:12181402). Translocation t(5;15)(q33;q22) with TPS3BP1 creating a PDGFRB-TPS3BP1 fusion protein (PubMed:15492236). Translocation t(1;5)(q23;q33) that forms a PDE4DIP-PDGFRB fusion protein (PubMed:12907457). Translocation t(5;9)(q33-34;q23) with CEP85L that fuses the 5'-end of CEP85L (isoform 4) to the 3'-end of PDGFRB (PubMed:21938754). {ECO:0000269|PubMed:12181402, ECO:0000269|PubMed:12907457, ECO:0000269|PubMed:15492236, ECO:0000269|PubMed:21938754}.	EDTA Blood Sample
Miyeloproliferatif Hastalık, 605392 (FGFR1OP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myeloproliferative disorder, 605392 (FGFR1OP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1OP	FGFR1OP, FOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
(Miyeloproliferatif Neoplaziler, Ailesel, Yatkınlık), 616604, Otozomal Dominant (2200)	Myeloproliferative neoplasms, familial, susceptibility to, 616604, Autosomal dominant (440)	.	DUP14q32, C14DUPq32	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Miyeloproliferatif / Lenfoproliferatif Neoplazmalar, Ailesel (Çok Tip), Yatkınlık, 616871, Otozomal Dominant; MPLPF (DDX41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myeloproliferative/lymphoproliferative neoplasms, familial (multiple types), susceptibility to, 616871, Autosomal dominant; MPLPF (DDX41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDX41	DDX41, ABS, MPLPF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Myhre Sendromu, 139210, Otozomal Dominant (Myhre Sendromu) (SMAD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myhre syndrome, 139210, Autosomal dominant (Myhre syndrome) (SMAD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Myhre Sendromu, 139210, Otozomal Dominant (Myhre Sendromu) (MLPA)	Myhre syndrome, 139210, Autosomal dominant (Myhre syndrome) (MLPA)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Myhre Sendromu, 139210, Otozomal Dominant (Myhre Sendromu) (Prenatal) (SMAD4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myhre syndrome, 139210, Autosomal dominant (Myhre syndrome) (SMAD4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Myhre Sendromu, 139210, Otozomal Dominant (Myhre Sendromu) (Prenatal) (MLPA)	Myhre syndrome, 139210, Autosomal dominant (Myhre syndrome) (Prenatal) (MLPA)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyokard İnfarctüsü, Yatkınlık, 608446 (PSMA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarcation, susceptibility to, 608446 (PSMA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSMA6	PSMA6, PROS27, P27K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Miyokard Enfarktüsü, Azalmış Yatkınlık, 608446 (F7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, decreased susceptibility to, 608446 (F7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F7	F7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor VII deficiency (FA7D) [MIM:227500]: A hemorrhagic disease with variable presentation. The clinical picture can be very severe, with the early occurrence of intracerebral hemorrhages or repeated hemarthroses, or, in contrast, moderate with cutaneous-mucosal hemorrhages (epistaxis, menorrhagia) or hemorrhages provoked by a surgical intervention. Finally, numerous subjects are completely asymptomatic despite very low factor VII levels. {ECO:0000269|PubMed:10682079, ECO:0000269|PubMed:11091194, ECO:0000269|PubMed:11129332, ECO:0000269|PubMed:12472587, ECO:0000269|PubMed:14717781, ECO:0000269|PubMed:1634227, ECO:0000269|PubMed:18976247, ECO:0000269|PubMed:19432927, ECO:0000269|PubMed:19751712, ECO:0000269|PubMed:2070047, ECO:0000269|PubMed:21206266, ECO:0000269|PubMed:21372693, ECO:0000269|PubMed:7974346, ECO:0000269|PubMed:7981691, ECO:0000269|PubMed:8043443, ECO:0000269|PubMed:8204979, ECO:0000269|PubMed:8364544, ECO:0000269|PubMed:8652821, ECO:0000269|PubMed:8844208, ECO:0000269|PubMed:8883260, ECO:0000269|PubMed:8940045, ECO:0000269|PubMed:9414278, ECO:0000269|PubMed:9452082, ECO:0000269|PubMed:9576180}. Note=The disease is caused	EDTA Blood Sample
Miyokard Enfarktüsü, Korunmaya Karşı, 608446 (F13A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, protection against, 608446 (F13A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F13A1	F13A1, F13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Factor XIII subunit A deficiency (FA13AD) [MIM:613225]: An autosomal recessive hematologic disorder characterized by a life-long bleeding tendency, impaired wound healing and spontaneous abortion in affected women. {ECO:0000269|PubMed:1353995, ECO:0000269|PubMed:24286209, ECO:0000269|PubMed:24329762, ECO:0000269|PubMed:24889649}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269|PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (GCLC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (GCLC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCLC	GCLC, GLCLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency (HAGGSD) [MIM:230450]: A disease characterized by hemolytic anemia, glutathione deficiency, myopathy, late-onset spinocerebellar degeneration, and peripheral neuropathy. {ECO:0000269|PubMed:10515893, ECO:0000269|PubMed:10733484, ECO:0000269|PubMed:12663448}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (TNFSF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (TNFSF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFSF4	TNFSF4, GP34, OX40L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:18059267}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The upstream region of TNFSF4 contains a single risk haplotype for SLE, which is correlated with increased expression of both cell-surface TNFSF4 and TNFSF4 transcripts. Increased levels of TNFSF4 are thought to augment T-cell-APC interaction and the functional consequences of T-cell activation, thereby destabilizing peripheral tolerance.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (OLR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (OLR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OLR1	OLR1, LOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Independent association genetic studies have implicated OLR1 gene variants in myocardial infarction susceptibility.	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (MIAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (MIAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIAT	MIAT, C22orf35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (LTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (LTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTA	LTA, TNFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet;arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). {ECO:0000269|PubMed:12746914}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (LRP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (LRP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP8	LRP8, APOER2, MCI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myocardial infarction 1 (MCI1) [MIM:608446]: A condition defined by the irreversible necrosis of heart muscle secondary to prolonged ischemia. {ECO:0000269|PubMed:17847002}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (LGALS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (LGALS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LGALS2	LGALS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (GCLM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (GCLM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GCLM	GCLM, GLCLR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (ESR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (ESR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESR1	ESR1, ESR, ESTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Estrogen resistance (ESTRR) [MIM:615363]: A disorder characterized by partial or complete resistance to estrogens, in the presence of elevated estrogen serum levels. Clinical features include absence of the pubertal growth spurt, delayed bone maturation, unfused epiphyses, reduced bone mineral density, osteoporosis, continued growth into adulthood and very tall adult stature. Glucose intolerance, hyperinsulinemia and lipid abnormalities may also be present. {ECO:0000269|PubMed:23841731, ECO:0000269|PubMed:8961262}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokard Enfarktüsü, Yatkınlık, 608446 (Glanzmann Trombastenisi) (ITGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myocardial infarction, susceptibility to, 608446 (Glanzmann thrombasthenia) (ITGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB3	ITGB3, GP3A, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (BDPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. {ECO:0000269|PubMed:18065693}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyoklonik Epilepsi, İnfantil, Alesel, 605021, Otozomal Resesif (Alesel İnfantil Miyoklonik Epilepsi) (TBC1D24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoclonic epilepsy, infantile, familial, 605021, Autosomal recessive (Familial infantile myoclonic epilepsy) (TBC1D24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D24	TBC1D24, KIAA1171, FIME, EIEE16, DOORS, DFNB86, DFNA65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, onychodystrophy, osteodystrophy, mental retardation, and seizures syndrome (DOORS) [MIM:220500]: A syndrome characterized by sensorineural deafness, mental retardation, hypoplastic or absent nails, small or absent distal phalanges of hands and feet. Additional features include coarse facies, a large nose with wide nasal bridge, bulbous tip and anteverted nares, a long prominent philtrum and downturned corners of the mouth. Progressive neurological manifestations include seizures from infancy, optic atrophy, and peripheral polyneuropathy. {ECO:0000269|PubMed:24291220}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoklonik Epilepsi, Jüvenil, Yatkınlık, 1, 254770, Otozomal Dominant; EJM (Jüvenil Miyoklonik Epilepsi) (EFHC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoclonic epilepsy, juvenile, susceptibility to, 1, 254770, Autosomal dominant; EJM (Juvenile myoclonic epilepsy) (EFHC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EFHC1	EFHC1, FLJ10466, EJM1, JAE, EJA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Juvenile absence epilepsy 1 (JAE1) [MIM:607631]: A subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures. {ECO:0000305|PubMed:17159113}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoklonik Epilepsi, Jüvenil, Yatkınlık, 1, 254770, Otozomal Dominant; EJM (Jüvenil Miyoklonik Epilepsi) (Prenatal) (EFHC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myoclonic epilepsy, juvenile, susceptibility to, 1, 254770, Autosomal dominant; EJM (Juvenile myoclonic epilepsy) (EFHC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EFHC1	EFHC1, FLJ10466, EJM1, JAE, EJA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Juvenile absence epilepsy 1 (JAE1) [MIM:607631]: A subtype of idiopathic generalized epilepsy characterized by onset occurring around puberty, absence seizures, generalized tonic-clonic seizures (GTCS), GTCS on awakening, and myoclonic seizures. {ECO:0000305|PubMed:17159113}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyoklonik-Atonik Epilepsi, 616421, Otozomal Dominant; MAE (Miyoklonik-Astastik Epilepsi) (SLC6A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoclonic-atonic epilepsy, 616421, Autosomal dominant; MAE (Myoclonic-astastic epilepsy) (SLC6A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A1	SLC6A1, GABATR, MAE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myoclonic-atonic epilepsy (MAE) [MIM:616421]: A form of epilepsy characterized by myoclonic-atonic and absence seizures, appearing in early childhood. Patients have delayed development before the onset of seizures and show varying degrees of intellectual disability following seizure onset. {ECO:0000269	PubMed:25865495}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyoklonus, Ailesel Kortikal, 614937, Otozomal Dominant; FCM (Ailesel Kortikal Miyoklonus) (NOL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoclonus, familial cortical, 614937, Autosomal dominant; FCM (Familial cortical myoclonus) (NOL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOL3	NOL3, NOP, MYC, ARC, FCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myoclonus, familial cortical (FCM) [MIM:614937]: An autosomal dominant neurologic condition characterized by adult onset of cortical myoclonus manifest as involuntary jerks or movements affecting the face and limbs. Affected individuals can also experience falls without seizure activity or loss of consciousness. {ECO:0000269	PubMed:22926851}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyoklonus, Dirençli, Neonatal, 617235, Otozomal Dominant; NEIMY (Otozomal Dominant Spastik Paraplejі Tip 10) (KIF5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoclonus, intractable, neonatal, 617235, Autosomal dominant; NEIMY (Autosomal dominant spastic paraplegia type 10) (KIF5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF5A	KIF5A, NKHC, SPG10, NEIMY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 10, autosomal dominant (SPG10) [MIM:604187]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:12355402, ECO:0000269|PubMed:15452312, ECO:0000269|PubMed:16476820, ECO:0000269|PubMed:16489470, ECO:0000269|PubMed:18245137, ECO:0000269|PubMed:18853458, ECO:0000269|PubMed:21107874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyofibromatoz, İnfantil 2, 615293, Otozomal Dominant (İnfantil Miyofibromatozis) (NOTCH3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myofibromatosis, infantile 2, 615293, Autosomal dominant (infantile myofibromatosis) (NOTCH3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOTCH3	NOTCH3, CADASIL1, CASIL, IMF2, LMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, autosomal dominant (CADASIL) [MIM:125310]: A cerebrovascular disease characterized by multiple subcortical infarcts, pseudobulbar palsy, dementia, and the presence of granular deposits in small cerebral arteries producing ischemic stroke. {ECO:0000269|PubMed:10227618, ECO:0000269|PubMed:10371548, ECO:0000269|PubMed:10802807, ECO:0000269|PubMed:10854111, ECO:0000269|PubMed:11058919, ECO:0000269|PubMed:11102981, ECO:0000269|PubMed:11593313, ECO:0000269|PubMed:11755616, ECO:0000269|PubMed:11810186, ECO:0000269|PubMed:12136071, ECO:0000269|PubMed:12146805, ECO:0000269|PubMed:12589106, ECO:0000269|PubMed:12810003, ECO:0000269|PubMed:15229130, ECO:0000269|PubMed:15300888, ECO:0000269|PubMed:15364702, ECO:0000269|PubMed:15378071, ECO:0000269|PubMed:15818833, ECO:0000269|PubMed:16009764, ECO:0000269|PubMed:24000151, ECO:0000269|PubMed:9388399}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyofibromatozis, İnfantil, 1, 228550, Otozomal Dominant; IMF1 (İnfantil Miyofibromatozis) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myofibromatosis, infantile, 1, 228550, Autosomal dominant; IMF1 (infantile myofibromatosis) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloproliferative disorder chronic with eosinophilia (MPE) [MIM:131440]: A hematologic disorder characterized by malignant eosinophils proliferation. Note=The gene represented in this entry may be involved in disease pathogenesis. Chromosomal aberrations involving PDGFRB have been found in many instances of chronic myeloproliferative disorder with eosinophilia. Translocation t(5;12) with ETV6 on chromosome 12 creating an PDGFRB-ETV6 fusion protein (PubMed:12181402). Translocation t(5;15)(q33;q22) with TPS3BP1 creating a PDGFRB-TPS3BP1 fusion protein (PubMed:15492236). Translocation t(1;5)(q23;q33) that forms a PDE4DIP-PDGFRB fusion protein (PubMed:12907457). Translocation t(5;9)(q33-34;q23) with CEP85L that fuses the 5'-end of CEP85L (isoform 4) to the 3' end of PDGFRB (PubMed:21938754). {ECO:0000269|PubMed:12181402, ECO:0000269|PubMed:12907457, ECO:0000269|PubMed:15492236, ECO:0000269|PubMed:21938754}.	EDTA Blood Sample
Miyoglobinüri, Akut Tekrarlayan, Otozomal Resesif, 268200, Otozomal Resesif (Genetik Tekrarlayan Miyoglobinüri) (LPIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myoglobinuria, acute recurrent, autosomal recessive, 268200, Autosomal recessive (Genetic recurrent myoglobinuria) (LPIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPIN1	LPIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myoglobinuria, acute recurrent, autosomal recessive (ARARM) [MIM:268200]: Recurrent myoglobinuria is characterized by recurrent attacks of rhabdomyolysis (necrosis or disintegration of skeletal muscle) associated with muscle pain and weakness and followed by excretion of myoglobin in the urine. Renal failure may occasionally occur. {ECO:0000269	PubMed:18817903}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyokimi, 121200, Otozomal Dominant; BFNS1 (Benign Ailesel Neonatal Epilepsi) (KCNQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myokymia, 121200, Autosomal dominant; BFNS1 (Benign familial neonatal epilepsy) (KCNQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ2	KCNQ2, EBN1, EIEE7, BFNS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seizures, benign familial neonatal 1 (BFNS1) [MIM:121200]: A disorder characterized by clusters of seizures occurring in the first days of life. Most patients have spontaneous remission by 12 months of age and show normal psychomotor development. Some rare cases manifest an atypical severe phenotype associated with epileptic encephalopathy and psychomotor retardation. The disorder is distinguished from benign familial infantile seizures by an earlier age at onset. In some patients, neonatal convulsions are followed later in life by myokymia, a benign condition characterized by spontaneous involuntary contractions of skeletal muscles fiber groups that can be observed as vermiform movement of the overlying skin. Electromyography typically shows continuous motor unit activity with spontaneous oligo- and multiplet-discharges of high intraburst frequency (myokymic discharges). Some patients may have isolated myokymia. {ECO:0000269|PubMed:11175290, ECO:0000269|PubMed:11572947, ECO:0000269|PubMed:14534157, ECO:0000269|PubMed:15249611, ECO:0000269|PubMed:9425895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Myoadenilat Deaminaz Eksikliğine Bağlı Miyopati, 615511, Otozomal Resesif; MMDD (Adenozin Monofosfat Deaminaz Eksikliği) (AMPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy due to myoadenylate deaminase deficiency, 615511, Autosomal recessive; MMDD (Adenosine monophosphate deaminase deficiency) (AMPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMPD1	AMPD1, MMDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Myoadenilat Deaminaz Eksikliğine Bağlı Miyopati, 615511, Otozomal Resesif; MMDD (Adenozin Monofosfat Deaminaz Eksikliği) (Prenatal) (AMPD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy due to myoadenylate deaminase deficiency, 615511, Autosomal recessive; MMDD (Adenosine monophosphate deaminase deficiency) (AMPD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMPD1	AMPD1, MMDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Ekstrapiramidal Bulgular, 615673, Otozomal Resesif; MPXPS (Proksimal Miyopati-Ekstrapiramidal Bulgular) (MICU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy with extrapyramidal signs, 615673, Autosomal recessive; MPXPS (Proximal myopathy with extrapyramidal signs) (MICU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MICU1	MICU1, CBARA1, MPXPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy with extrapyramidal signs (MPXPS) [MIM:615673]: An autosomal recessive disorder characterized by early-onset proximal muscle weakness with a static course and moderately to grossly elevated serum creatine kinase levels accompanied by learning difficulties. Most patients develop subtle extrapyramidal motor signs that progress to a debilitating disorder of involuntary movement with variable features, including chorea, tremor, dystonic posturing and orofacial dyskinesia. Additional variable features include ataxia, microcephaly, ophthalmoplegia, ptosis, optic atrophy and axonal peripheral neuropathy. {ECO:0000269|PubMed:24336167}. Note=The disease is caused by mutations affecting the gene represented in this entry. The complex phenotype is due to alterations in mitochondrial calcium signaling characterized by increased mitochondrial Ca(2+) load (PubMed:24336167). {ECO:0000269|PubMed:24336167};.	EDTA Blood Sample
Miyopati, Ekstrapiramidal Bulgular, 615673, Otozomal Resesif; MPXPS (Proksimal Miyopati-Ekstrapiramidal Bulgular) (Prenatal) (MICU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy with extrapyramidal signs, 615673, Autosomal recessive; MPXPS (Proximal myopathy with extrapyramidal signs) (MICU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MICU1	MICU1, CBARA1, MPXPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy with extrapyramidal signs (MPXPS) [MIM:615673]: An autosomal recessive disorder characterized by early-onset proximal muscle weakness with a static course and moderately to grossly elevated serum creatine kinase levels accompanied by learning difficulties. Most patients develop subtle extrapyramidal motor signs that progress to a debilitating disorder of involuntary movement with variable features, including chorea, tremor, dystonic posturing and orofacial dyskinesia. Additional variable features include ataxia, microcephaly, ophthalmoplegia, ptosis, optic atrophy and axonal peripheral neuropathy. {ECO:0000269|PubMed:24336167}. Note=The disease is caused by mutations affecting the gene represented in this entry. The complex phenotype is due to alterations in mitochondrial calcium signaling characterized by increased mitochondrial Ca(2+) load (PubMed:24336167). {ECO:0000269|PubMed:24336167};.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Laktik Asidoz, Kalıtsal, 255125, Otozomal Resesif; HML (Kalıtsal Miyopati-Laktik Asidoz, ISCU Eksikliğine Bağlı) (ISCU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy with lactic acidosis, hereditary, 255125, Autosomal recessive; HML (Hereditary myopathy with lactic acidosis due to ISCU deficiency) (ISCU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ISCU	ISCU, HML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Laktik Asidoz, Kalıtsal, 255125, Otozomal Resesif; HML (Kalıtsal Miyopati-Laktik Asidoz, ISCU Eksikliğine Bağlı) (Prenatal) (ISCU geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy with lactic acidosis, hereditary, 255125, Autosomal recessive; HML (Hereditary myopathy with lactic acidosis due to ISCU deficiency) (ISCU gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ISCU	ISCU, HML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyopati, Aktin, Konjenital, Çekirdeklerle, 161800, Otozomal Resesif, Otozomal Dominant (Ağır Konjenital Nemalin Miyopati) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, actin, congenital, with cores, 161800, Autosomal recessive, Autosomal dominant (Severe congenital nemaline myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 3 [NEM3] [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. {ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Aktin, Konjenital, Çekirdeklerle, 161800, Otozomal Resesif, Otozomal Dominant (Ağır Konjenital Nemalin Miyopati) (Prenatal) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, actin, congenital, with cores, 161800, Autosomal recessive, Autosomal dominant (Severe congenital nemaline myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 3 [NEM3] [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. {ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Arefleksi, Solunum Zorluğu, Disfaji, Erken Başlangıçlı, 614399, Otozomal Resesif, EMARDD (Erken Başlangıçlı Miyopati-Arefleksi-Solunum Distresi-Disfaji Sendromu) (MEGF10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399, Autosomal recessive; EMARDD (Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome) (MEGF10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEGF10	MEGF10, KIAA1780, EMARDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, early-onset, areflexia, respiratory distress, and dysphagia (EMARDD) [MIM:614399]: An autosomal recessive congenital myopathy characterized by onset at birth, or early in infancy, of respiratory distress caused by diaphragmatic weakness. Additional features are dysphagia resulting in poor feeding, failure to thrive, poor head control, facial weakness, cleft palate, contractures and scoliosis. Affected individuals become ventilator-dependent, and most require feeding by gastrostomy. The disorder results in severe muscle weakness and most patients never achieve walking. Death from respiratory failure in childhood occurs in about half of patients. Muscle biopsies from affected individuals show myopathic changes, replacement of myofibers with fatty tissue, small and incompletely fused muscle fibers, and variation in fiber size. Short regions of sarcomeric disorganization with few or no mitochondria (minicores) have been observed in some cases. {ECO:0000269|PubMed:22101682, ECO:0000269|PubMed:22371254}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Arefleksi, Solunum Zorluğu, Disfaji, Erken Başlangıçlı, 614399, Otozomal Resesif, EMARDD (Erken Başlangıçlı Miyopati-Arefleksi-Solunum Distresi-Disfaji Sendromu) (Prenatal) (MEGF10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, areflexia, respiratory distress, and dysphagia, early-onset, 614399, Autosomal recessive; EMARDD (Early-onset myopathy-areflexia-respiratory distress-dysphagia syndrome) (MEGF10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MEGF10	MEGF10, KIAA1780, EMARDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, early-onset, areflexia, respiratory distress, and dysphagia (EMARDD) [MIM:614399]: An autosomal recessive congenital myopathy characterized by onset at birth, or early in infancy, of respiratory distress caused by diaphragmatic weakness. Additional features are dysphagia resulting in poor feeding, failure to thrive, poor head control, facial weakness, cleft palate, contractures and scoliosis. Affected individuals become ventilator-dependent, and most require feeding by gastrostomy. The disorder results in severe muscle weakness and most patients never achieve walking. Death from respiratory failure in childhood occurs in about half of patients. Muscle biopsies from affected individuals show myopathic changes, replacement of myofibers with fatty tissue, small and incompletely fused muscle fibers, and variation in fiber size. Short regions of sarcomeric disorganization with few or no mitochondria (minicores) have been observed in some cases. {ECO:0000269|PubMed:22101682, ECO:0000269|PubMed:22371254}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Sentronükleer, 160150, Otozomal Dominant; CNM1 (Otozomal Dominant Santronükleer Miyopati) (DNM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, centronuclear, 160150, Autosomal dominant; CNM1 (Autosomal dominant centronuclear myopathy) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, LCCS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, centronuclear, 1 (CNM1) [MIM:160150]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:16227997, ECO:0000269|PubMed:17825552, ECO:0000269|PubMed:17932957, ECO:0000269|PubMed:19122038, ECO:0000269|PubMed:19623537, ECO:0000269|PubMed:19932619, ECO:0000269|PubMed:19932620, ECO:0000269|PubMed:20227276, ECO:0000269|PubMed:22396310}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Sentronükleer, 160150, Otozomal Dominant; CNM1 (Otozomal Dominant Santronükleer Miyopati) (Prenatal) (DNM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, centronuclear, 160150, Autosomal dominant; CNM1 (Autosomal dominant centronuclear myopathy) (DNM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNM2	DNM2, CMTDIB, CMTDI1, CMT2M, LCCS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, centronuclear, 1 (CNM1) [MIM:160150]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:16227997, ECO:0000269|PubMed:17825552, ECO:0000269|PubMed:17932957, ECO:0000269|PubMed:19122038, ECO:0000269|PubMed:19623537, ECO:0000269|PubMed:19932619, ECO:0000269|PubMed:19932620, ECO:0000269|PubMed:20227276, ECO:0000269|PubMed:22396310}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Santronükleer, 3, 614408, Otozomal Dominant; CNM3 (Otozomal Dominant Santronükleer Miyopati) (MYF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, centronuclear, 3, 614408, Autosomal dominant; CNM3 (Autosomal dominant centronuclear myopathy) (MYF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYF6	MYF6, CNM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Santronükleer, 3, 614408, Otozomal Dominant; CNM3 (Otozomal Dominant Santronükleer Miyopati) (Prenatal) (MYF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, centronuclear, 3, 614408, Autosomal dominant; CNM3 (Autosomal dominant centronuclear myopathy) (MYF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYF6	MYF6, CNM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Santronükleer, 4, 614807, Otozomal Dominant; CNM4 (Konjenital Miyopati-İç Çekirdek ve Altipik Çekirdekli) (CCDC78 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, centronuclear, 4, 614807, Autosomal dominant; CNM4 (Congenital myopathy with internal nuclei and atypical cores) (CCDC78 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC78	CCDC78, C16orf25, CNM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Santronükleer, 4, 614807, Otozomal Dominant; CNM4 (Konjenital Miyopati-İç Çekirdek ve Altipik Çekirdekli) (Prenatal) (CCDC78 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, centronuclear, 4, 614807, Autosomal dominant; CNM4 (Congenital myopathy with internal nuclei and atypical cores) (CCDC78 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC78	CCDC78, C16orf25, CNM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Sentronükleer, Otozomal Resesif, 255200, Otozomal Resesif (Otozomal Resesif Santronükleer Miyopati) (BIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, centronuclear, autosomal recessive, 255200, Autosomal recessive (Autosomal recessive centronuclear myopathy) (BIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BIN1	BIN1, AMPHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, centronuclear, 2 (CNM2) [MIM:255200]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:17676042}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Sentronükleer, Otozomal Resesif, 255200, Otozomal Resesif (Otozomal Resesif Santronükleer Miyopati) (Prenatal) (BIN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, centronuclear, autosomal recessive, 255200, Autosomal recessive (Autosomal recessive centronuclear myopathy) (BIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BIN1	BIN1, AMPHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, centronuclear, 2 (CNM2) [MIM:255200]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:17676042}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Konjenital, Compton-North, 612540, Otozomal Resesif; MYPCN (Konjenital Letal Miyopati, Compton-North Tipi) (CNTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, congenital, Compton-North, 612540, Autosomal recessive; MYPCN (Congenital lethal myopathy, Compton-North type) (CNTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTN1	CNTN1, MYPCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Konjenital, Compton-North, 612540, Otozomal Resesif; MYPCN (Konjenital Letal Miyopati, Compton-North Tipi) (Prenatal) (CNTN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, congenital, Compton-North, 612540, Autosomal recessive; MYPCN (Congenital lethal myopathy, Compton-North type) (CNTN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNTN1	CNTN1, MYPCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyopati, Konjenital, Lif Tipi Orantısızlık 1, 255310, Otozomal Resesif, Otozomal Dominant (Konjenital Fiber Type Disproportion Miyopati (Konjenital Lif Tipi Orantısızlık Miyopatisi)) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, congenital, with fiber-type disproportion 1, 255310, Autosomal recessive, Autosomal dominant (Congenital fiber-type disproportion myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. (ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Konjenital, Lif Tipi Orantısızlık 1, 255310, Otozomal Resesif, Otozomal Dominant (Konjenital Fiber Type Disproportion Miyopati (Konjenital Lif Tipi Orantısızlık Miyopatisi)) (Prenatal) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, congenital, with fiber-type disproportion 1, 255310, Autosomal recessive, Autosomal dominant (Congenital fiber-type disproportion myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. (ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Konjenital, Lif Tipi Orantısızlık, 255310, Otozomal Resesif, Otozomal Dominant (Konjenital Lif Tipi Orantısızlık Miyopatisi) (TPM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, congenital, with fiber-type disproportion, 255310, Autosomal recessive, Autosomal dominant (Congenital fiber-type disproportion myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, congenital, with fiber-type disproportion (CFTD) [MIM:255310]: A genetically heterogeneous disorder in which there is relative hypotrophy of type 1 muscle fibers compared to type 2 fibers on skeletal muscle biopsy. However, these findings are not specific and can be found in many different myopathic and neuropathic conditions. (ECO:0000269|PubMed:18300303, ECO:0000269|PubMed:19953533, ECO:0000269|PubMed:20951040, ECO:0000269|PubMed:24692096). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Konjenital, Lif Tipi Orantısızlık, 255310, Otozomal Resesif, Otozomal Dominant (Konjenital Lif Tipi Orantısızlık Miyopatisi) (Prenatal) (TPM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, congenital, with fiber-type disproportion, 255310, Autosomal recessive, Autosomal dominant (Congenital fiber-type disproportion myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, congenital, with fiber-type disproportion (CFTD) [MIM:255310]: A genetically heterogeneous disorder in which there is relative hypotrophy of type 1 muscle fibers compared to type 2 fibers on skeletal muscle biopsy. However, these findings are not specific and can be found in many different myopathic and neuropathic conditions. (ECO:0000269|PubMed:18300303, ECO:0000269|PubMed:19953533, ECO:0000269|PubMed:20951040, ECO:0000269|PubMed:24692096). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Konjenital, Lif Tipi Orantısızlık, 255310, Otozomal Resesif, Otozomal Dominant; CFTD (Konjenital Fiber Type Disproportion Miyopati) (Konjenital Lif Tipi Orantısızlık Miyopatisi) (SELENON geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, congenital, with fiber-type disproportion, 255310, Autosomal recessive, Autosomal dominant; CFTD (Congenital fiber-type disproportion myopathy) (SELENON gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SELENON	SELENON, SEPN1, SELN, RSMD1, CFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Konjenital, Lif Tipi Orantısızlık, 255310, Otozomal Resesif, Otozomal Dominant; CFTD (Konjenital Fiber Type Disproportion Miyopati) (Konjenital Lif Tipi Orantısızlık Miyopatisi) (Prenatal) (SELENON geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, congenital, with fiber-type disproportion, 255310, Autosomal recessive, Autosomal dominant; CFTD (Congenital fiber-type disproportion myopathy) (SELENON gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SELENON	SELENON, SEPN1, SELN, RSMD1, CFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Distal, 4, 614065, Otozomal Dominant; MPD4 (Flaminopati) (FLNC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, distal, 4, 614065, Autosomal dominant; MPD4 (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, myofibrillar, 5 (MFM5) [MIM:609524]: A neuromuscular disorder, usually with an adult onset, characterized by focal myofibrillar destruction, pathological cytoplasmic protein aggregations, and clinical features of a limb-girdle myopathy. (ECO:0000269	PubMed:15929027). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Distal, 4, 614065, Otozomal Dominant; MPD4 (Flaminopati) (Prenatal) (FLNC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, distal, 4, 614065, Autosomal dominant; MPD4 (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, myofibrillar, 5 (MFM5) [MIM:609524]: A neuromuscular disorder, usually with an adult onset, characterized by focal myofibrillar destruction, pathological cytoplasmic protein aggregations, and clinical features of a limb-girdle myopathy. (ECO:0000269	PubMed:15929027). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Distal, 5, 617030, Otozomal Resesif; MPD5 (ADSSL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, distal, 5, 617030, Autosomal recessive; MPD5 (ADSSL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADSSL1	ADSL1, MPD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Distal, 5, 617030, Otozomal Resesif; MPD5 (Prenatal) (ADSSL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, distal, 5, 617030, Autosomal recessive; MPD5 (ADSSL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADSSL1	ADSL1, MPD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati,Distal Tateyama Tipi, 614321, Otozomal Dominant; MPDT (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, distal, Tateyama type, 614321, Autosomal dominant; MPDT (Rippling muscle disease) (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati,Distal Tateyama Tipi, 614321, Otozomal Dominant; MPDT (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (MLPA)	Myopathy, distal, Tateyama type, 614321, Autosomal dominant; MPDT (Rippling muscle disease) (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati,Distal Tateyama Tipi, 614321, Otozomal Dominant; MPDT (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (Prenatal) (CAV3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, distal, Tateyama type, 614321, Autosomal dominant; MPDT (Rippling muscle disease) (CAV3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati,Distal Tateyama Tipi, 614321, Otozomal Dominant; MPDT (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (Prenatal) (MLPA)	Myopathy, distal, Tateyama type, 614321, Autosomal dominant; MPDT (Rippling muscle disease) (Prenatal) (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. (ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Distal, Anterior Tibial Başlangıçlı, 606768, Otozomal Resesif; DMAT (Distal Miyopati - Anterior Tibial Başlangıçlı) (DYSF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, distal, with anterior tibial onset, 606768, Autosomal recessive; DMAT (Distal myopathy with anterior tibial onset) (DYSF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYSF	DYSF, LGMD2B, MMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18053459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Distal, Anterior Tibial Başlangıçlı, 606768, Otozomal Resesif; DMAT (Distal Miyopati - Anterior Tibial Başlangıçlı) (MLPA)	Myopathy, distal, with anterior tibial onset, 606768, Autosomal recessive; DMAT (Distal myopathy with anterior tibial onset) (MLPA)	DYSF	DYSF, LGMD2B, MMD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18053459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Miyopati, Distal, Anterior Tibial Başlangıçlı, 606768, Otozomal Resesif; DMAT (Distal Miyopati - Anterior Tibial Başlangıçlı) (Prenatal) (DYSF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, distal, with anterior tibial onset, 606768, Autosomal recessive; DMAT (Distal myopathy with anterior tibial onset) (DYSF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYSF	DYSF, LGMD2B, MMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Distal, Anterior Tibial Başlangıçlı, 606768, Otozomal Resesif; DMAT (Distal Miyopati - Anterior Tibial Başlangıçlı) (Prenatal) (MLPA)	Myopathy, distal, with anterior tibial onset, 606768, Autosomal recessive; DMAT (Distal myopathy with anterior tibial onset) (Prenatal) (MLPA)	DYSF	DYSF, LGMD2B, MMD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Limb-girdle muscular dystrophy 2B (LGMD2B) [MIM:253601]: An autosomal recessive degenerative myopathy characterized by weakness and atrophy starting in the proximal pelvi/femoral muscles, with onset in the late teens or later, massive elevation of serum creatine kinase levels and slow progression. Scapular muscle involvement is minor and not present at onset. Upper limb girdle involvement follows some years after the onset in lower limbs. (ECO:0000269|PubMed:10196377, ECO:0000269|PubMed:11134403, ECO:0000269|PubMed:14678801, ECO:0000269|PubMed:15469449, ECO:0000269|PubMed:16010686, ECO:0000269|PubMed:16100712, ECO:0000269|PubMed:16705711, ECO:0000269|PubMed:16996541, ECO:0000269|PubMed:17287450, ECO:0000269|PubMed:18306167, ECO:0000269|PubMed:18853459, ECO:0000269|PubMed:9731526). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Müsküler Distrofi, Distal, Çerçeveli Vakuoller, 617158, Otozomal Dominant; DMRV (GNE Miyopati (Nonaka Miyopati)) (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, distal, with rimmed vacuoles, 617158, Autosomal dominant; DMRV (GNE myopathy) (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 3 (PDB3) [MIM:167250]: A disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. (ECO:0000269|PubMed:11992264, ECO:0000269|PubMed:12374763, ECO:0000269|PubMed:14584883, ECO:0000269|PubMed:15125799, ECO:0000269|PubMed:15146436, ECO:0000269|PubMed:15176995, ECO:0000269|PubMed:15207768). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Müsküler Distrofi, Distal, Çerçeveli Vakuoller, 617158, Otozomal Dominant; DMRV (GNE Miyopati (Nonaka Miyopati)) (Prenatal) (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, distal, with rimmed vacuoles, 617158, Autosomal dominant; DMRV (GNE myopathy) (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paget disease of bone 3 (PDB3) [MIM:167250]: A disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. (ECO:0000269|PubMed:11992264, ECO:0000269|PubMed:12374763, ECO:0000269|PubMed:14584883, ECO:0000269|PubMed:15125799, ECO:0000269|PubMed:15146436, ECO:0000269|PubMed:15176995, ECO:0000269|PubMed:15207768). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Erken Başlangıçlı, Letal Kardiyomiyopati, 611705, Otozomal Resesif; EOMFC (Erken Başlangıçlı Miyopati-Letal Kardiyomiyopati) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, early-onset, with fatal cardiomyopathy, 611705, Autosomal recessive; EOMFC (Early-onset myopathy with fatal cardiomyopathy) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. (ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Erken Başlangıçlı, Letal Kardiyomiyopati, 611705, Otozomal Resesif; EOMFC (Erken Başlangıçlı Miyopati-Letal Kardiyomiyopati) (Prenatal) (TTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, early-onset, with fatal cardiomyopathy, 611705, Autosomal recessive; EOMFC (Early-onset myopathy with fatal cardiomyopathy) (TTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. (ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, İzole Mitokondriyal, Otozomal Dominant, 616209, Otozomal Dominant; IMMD (Otozomal Dominant Mitokondriyal Miyopati-Egzersiz İntoleransı) (CHCHD10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, isolated mitochondrial, autosomal dominant, 616209, Autosomal dominant; IMMD (Autosomal dominant mitochondrial myopathy with exercise intolerance) (CHCHD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHCHD10	CHCHD10, FTDALS2, SMAJ, IMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, Jokela type (SMAJ) [MIM:615048]: An autosomal dominant, slowly progressive, lower motor neuron disease. SMAJ is characterized by adult-onset of muscle cramps and fasciculations affecting the proximal and distal muscles of the upper and lower limbs. The disorder results in weakness and mild muscle atrophy later in life. (ECO:0000269|PubMed:25428574). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, İzole Mitokondriyal, Otozomal Dominant, 616209, Otozomal Dominant; IMMD (Otozomal Dominant Mitokondriyal Miyopati-Egzersiz İntoleransı) (Prenatal) (CHCHD10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, isolated mitochondrial, autosomal dominant, 616209, Autosomal dominant; IMMD (Autosomal dominant mitochondrial myopathy with exercise intolerance) (CHCHD10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHCHD10	CHCHD10, FTDALS2, SMAJ, IMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, Jokela type (SMAJ) [MIM:615048]: An autosomal dominant, slowly progressive, lower motor neuron disease. SMAJ is characterized by adult-onset of muscle cramps and fasciculations affecting the proximal and distal muscles of the upper and lower limbs. The disorder results in weakness and mild muscle atrophy later in life. (ECO:0000269|PubMed:25428574). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Laktik Asidoz ve Sideroblastik Anemi 1, 600462, Otozomal Resesif; MLASA1 (Mitokondriyal Miyopati ve Sideroblastik Anemi) (PUS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462, Autosomal recessive; MLASA1 (Mitochondrial myopathy and sideroblastic anemia) (PUS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PUS1	PUS1, MLASA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Laktik Asidoz ve Sideroblastik Anemi 1, 600462, Otozomal Resesif; MLASA1 (Mitokondriyal Miyopati ve Sideroblastik Anemi) (Prenatal) (PUS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, lactic acidosis, and sideroblastic anemia 1, 600462, Autosomal recessive; MLASA1 (Mitochondrial myopathy and sideroblastic anemia) (PUS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PUS1	PUS1, MLASA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Laktik Asidoz ve Sideroblastik Anemi 2, 613561, Otozomal Resesif; MLASA2 (Mitokondriyal Miyopati ve Sideroblastik Anemi) (YARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561, Autosomal recessive; MLASA2 (Mitochondrial myopathy and sideroblastic anemia) (YARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YARS2	YARS2, TYRRS, MLASA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy with lactic acidosis and sideroblastic anemia 2 (MLASA2) [MIM:613561]: A rare oxidative phosphorylation disorder specific to skeletal muscle and bone marrow. Affected individuals manifest sideroblastic anemia, progressive lethargy, muscle weakness, and exercise intolerance associated with persistent lactic acidemia. (ECO:0000269|PubMed:20598274, ECO:0000269|PubMed:22504945). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Laktik Asidoz ve Sideroblastik Anemi 2, 613561, Otozomal Resesif; MLASA2 (Mitokondriyal Miyopati ve Sideroblastik Anemi) (Prenatal) (YARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, lactic acidosis, and sideroblastic anemia 2, 613561, Autosomal recessive; MLASA2 (Mitochondrial myopathy and sideroblastic anemia) (YARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	YARS2	YARS2, TYRRS, MLASA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy with lactic acidosis and sideroblastic anemia 2 (MLASA2) [MIM:613561]: A rare oxidative phosphorylation disorder specific to skeletal muscle and bone marrow. Affected individuals manifest sideroblastic anemia, progressive lethargy, muscle weakness, and exercise intolerance associated with persistent lactic acidemia. (ECO:0000269|PubMed:20598274, ECO:0000269|PubMed:22504945). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Mitokondriyal Progresif, Konjenital Katarakt, İşitme Kaybı ve Gelişim Geriliği, 613076 (Konjenital Katarakt-Progresif Kas Hipotonisi-İşitme Kaybı-Gelişimsel Gecikme Sendromu) (GFER geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076 (Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome) (GFER gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFER	GFER, ERV1, ALR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Mitokondriyal Progresif, Konjenital Katarakt, İşitme Kaybı ve Gelişim Geriliği, 613076 (Konjenital Katarakt-Progresif Kas Hipotonisi-İşitme Kaybı-Gelişimsel Gecikme Sendromu) (Prenatal) (GFER geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, mitochondrial progressive, with congenital cataract, hearing loss, and developmental delay, 613076 (Congenital cataract-progressive muscular hypotonia-hearing loss-developmental delay syndrome) (GFER gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFER	GFER, ERV1, ALR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 1, 601419, Otozomal Resesif, Otozomal Dominant; MFM1 (Desminopati) (DES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, myofibrillar, 1, 601419, Autosomal recessive, Autosomal dominant; MFM1 (Desminopathy) (DES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DES	DES, CMD1I, MFM1, SCPNK, ARVD7, ARVC7, LGMD2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurogenic scapulo peroneal syndrome Kaeser type (Kaeser syndrome) [MIM:181400]: Autosomal dominant disorder with a peculiar scapulo peroneal distribution of weakness and atrophy. A large clinical variability is observed ranging from scapulo peroneal, limb girdle and distal phenotypes with variable cardiac or respiratory involvement. Facial weakness, dysphagia and gynaecomastia are frequent additional symptoms. Affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Histological and immunohistochemical examination of muscle biopsy specimens reveal a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin. (ECO:0000269|PubMed:17439987, ECO:0000269|PubMed:25393488). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Miyopati, Miyofibriller, 1, 601419, Otozomal Resesif, Otozomal Dominant; MFM1 (Desminopati) (Prenatal) (DES geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 1, 601419, Autosomal recessive, Autosomal dominant; MFM1 (Desminopathy) (DES gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DES	DES, CMD1I, MFM1, SCPNK, ARVD7, ARVC7, LGMD2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurogenic scapuloperoneal syndrome Kaeser type (Kaeser syndrome) [MIM:161400]: Autosomal dominant disorder with a peculiar scapuloperoneal distribution of weakness and atrophy. A large clinical variability is observed ranging from scapuloperoneal, limb grindle and distal phenotypes with variable cardiac or respiratory involvement. Facial weakness, dysphagia and gynaecomastia are frequent additional symptoms. Affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Histological and immunohistochemical examination of muscle biopsy specimens reveal a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin. {ECO:0000269|PubMed:17439987, ECO:0000269|PubMed:25394388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 2, 608810, Otozomal Dominant; MFM2 (Early-Orset Lamellar Cataract) (CRYAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 2, 608810, Autosomal dominant; MFM2 (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1II, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, myofibrillar, 2 (MFM2) [MIM:608810]: A neuromuscular disorder that results in weakness of the proximal and distal limb muscles, weakness of the neck, velopharynx and trunk muscles, hypertrophic cardiomyopathy, and cataract in a subset of patients. {ECO:0000269|PubMed:14681890, ECO:0000269|PubMed:21920752, ECO:0000269|PubMed:9731540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyofibriller, 2, 608810, Otozomal Dominant; MFM2 (Early-Onset Lamellar Cataract) (Prenatal) (CRYAB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 2, 608810, Autosomal dominant; MFM2 (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1II, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, myofibrillar, 2 (MFM2) [MIM:608810]: A neuromuscular disorder that results in weakness of the proximal and distal limb muscles, weakness of the neck, velopharynx and trunk muscles, hypertrophic cardiomyopathy, and cataract in a subset of patients. {ECO:0000269|PubMed:14681890, ECO:0000269|PubMed:21920752, ECO:0000269|PubMed:9731540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 3, 609200, Otozomal Dominant; MFM3 (Distal Miyotilinopati) (MYOT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 3, 609200, Autosomal dominant; MFM3 (Distal myotilinopathy) (MYOT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYOT	MYOT, TTOD, MFM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Miyofibriller, 3, 609200, Otozomal Dominant; MFM3 (Distal Miyotilinopati) (MLPA)	Miyopathy, myofibrillar, 3, 609200, Autosomal dominant; MFM3 (Distal myotilinopathy) (MLPA)	MYOT	MYOT, TTOD, MFM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Miyofibriller, 3, 609200, Otozomal Dominant; MFM3 (Distal Miyotilinopati) (Prenatal) (MYOT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 3, 609200, Autosomal dominant; MFM3 (Distal myotilinopathy) (MYOT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYOT	MYOT, TTOD, MFM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 3, 609200, Otozomal Dominant; MFM3 (Distal Miyotilinopati) (Prenatal) (MLPA)	Miyopathy, myofibrillar, 3, 609200, Autosomal dominant; MFM3 (Distal myotilinopathy) (Prenatal) (MLPA)	MYOT	MYOT, TTOD, MFM3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. {ECO:0000269	PubMed:16380616}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 4, 609452, Otozomal Dominant; MFM4 (Geç Başlangıçlı Distal Miyopati, Markesbery-Griggs Tipi) (LDB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 4, 609452, Autosomal dominant; MFM4 (Late-onset distal myopathy, Markesbery-Griggs type) (LDB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LDB3	LDB3, ZASP, CYPHER, KIAA01613, MFM4, CMD1C, CMH24, LVNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Left ventricular non-compaction 3 (LVNC3) [MIM:601493]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyofibriller, 4, 609452, Otozomal Dominant; MFM4 (Geç Başlangıçlı Distal Miyopati, Markesbery-Griggs Tipi) (Prenatal) (LDB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 4, 609452, Autosomal dominant; MFM4 (Late-onset distal myopathy, Markesbery-Griggs type) (LDB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LDB3	LDB3, ZASP, CYPHER, KIAA01613, MFM4, CMD1C, CMH24, LVNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Left ventricular non-compaction 3 (LVNC3) [MIM:601493]: A disease due to an arrest of myocardial morphogenesis. It is characterized by a hypertrophic left ventricle with deep trabeculations and with poor systolic function, with or without associated left ventricular dilation. In some cases, it is associated with other congenital heart anomalies. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 5, 609524, Otozomal Dominant; MFM5 (Flaminopati) (FLNC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 5, 609524, Autosomal dominant; MFM5 (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, myofibrillar, 5 (MFM5) [MIM:609524]: A neuromuscular disorder, usually with an adult onset, characterized by focal myofibrillar destruction, pathological cytoplasmic protein aggregations, and clinical features of a limb-girdle myopathy. {ECO:0000269	PubMed:15929027}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyofibriller, 5, 609524, Otozomal Dominant; MFM5 (Flaminopati) (Prenatal) (FLNC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 5, 609524, Autosomal dominant; MFM5 (Filaminopathy) (FLNC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNC	FLNC, FLN2, ABPA, ABPL, MFM5, MPD4, CMH26, RCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, myofibrillar, 5 (MFM5) [MIM:609524]: A neuromuscular disorder, usually with an adult onset, characterized by focal myofibrillar destruction, pathological cytoplasmic protein aggregations, and clinical features of a limb-girdle myopathy. {ECO:0000269	PubMed:15929027}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 6, 612954, Otozomal Dominant; MFM6 (Müsküler Distrofi, Selcen Tipi) (BAG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 6, 612954, Autosomal dominant; MFM6 (Muscular dystrophy, Selcen type) (BAG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAG3	BAG3, MFM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, myofibrillar, 6 (MFM6) [MIM:612954]: A neuromuscular disorder that results in early-onset, severe, progressive, diffuse muscle weakness associated with cardiomyopathy, severe respiratory insufficiency during adolescence, and a rigid spine in some patients. At ultrastructural level, muscle fibers display structural alterations consisting of replacement of the normal myofibrillar markings by small, dense granules, or larger hyaline masses, or amorphous material. {ECO:0000269|PubMed:19085932, ECO:0000269|PubMed:21361913}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyofibriller, 6, 612954, Otozomal Dominant; MFM6 (Müsküler Distrofi, Selcen Tipi) (Prenatal) (BAG3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 6, 612954, Autosomal dominant; MFM6 (Muscular dystrophy, Selcen type) (BAG3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BAG3	BAG3, MFM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, myofibrillar, 6 (MFM6) [MIM:612954]: A neuromuscular disorder that results in early-onset, severe, progressive, diffuse muscle weakness associated with cardiomyopathy, severe respiratory insufficiency during adolescence, and a rigid spine in some patients. At ultrastructural level, muscle fibers display structural alterations consisting of replacement of the normal myofibrillar markings by small, dense granules, or larger hyaline masses, or amorphous material. {ECO:0000269|PubMed:19085932, ECO:0000269|PubMed:21361913}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 7, 617114, Otozomal Resesif, MFM7 (KY geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 7, 617114, Autosomal recessive, MFM7 (KY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KY	KY, MFM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Miyofibriller, 7, 617114, Otozomal Resesif, MFM7 (Prenatal) (KY geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 7, 617114, Autosomal recessive, MFM7 (KY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KY	KY, MFM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, 8, 617258, Otozomal Resesif, MFM8 (PYROXD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, 8, 617258, Autosomal recessive, MFM8 (PYROXD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PYROXD1	PYROXD1, MFM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopati, Miyofibriller, 8, 617258, Otozomal Resesif, MFM8 (Prenatal) (PYROXD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, 8, 617258, Autosomal recessive, MFM8 (PYROXD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PYROXD1	PYROXD1, MFM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyofibriller, Letal İnfantil Hipertofi, Alfa-B Kristalin İlişkili, 613869, Otozomal Resesif (Early-Onset Lamellar Cataract) (CRYAB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869, Autosomal recessive (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1II, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, myofibrillar, 2 (MFM2) [MIM:608810]: A neuromuscular disorder that results in weakness of the proximal and distal limb muscles, weakness of the neck, velopharynx and trunk muscles, hypertrophic cardiomyopathy, and cataract in a subset of patients. {ECO:0000269|PubMed:14681890, ECO:0000269|PubMed:21920752, ECO:0000269|PubMed:9731540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyofibriller, Letal İnfantil Hipertofi, Alfa-B Kristalin İlişkili, 613869, Otozomal Resesif (Early-Onset Lamellar Cataract) (Prenatal) (CRYAB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myofibrillar, fatal infantile hypertrophy, alpha-B crystallin-related, 613869, Autosomal recessive (Early-onset lamellar cataract) (CRYAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRYAB	CRYAB, CRYA2, CTPP2, CMD1II, CTRCT16, MFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, myofibrillar, 2 (MFM2) [MIM:608810]: A neuromuscular disorder that results in weakness of the proximal and distal limb muscles, weakness of the neck, velopharynx and trunk muscles, hypertrophic cardiomyopathy, and cataract in a subset of patients. {ECO:0000269|PubMed:14681890, ECO:0000269|PubMed:21920752, ECO:0000269|PubMed:9731540}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Miyosin Depo, Otozomal Dominant, 608358, Otozomal Dominant; MSMA (Hyalen Body Miyopati) (MYH7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myosin storage, autosomal dominant, 608358, Autosomal dominant; MSMA (Hyaline body myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548577}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyosin Depo, Otozomal Dominant, 608358, Otozomal Dominant; MSMA (Hyalen Body Miyopati) (Prenatal) (MYH7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myosin storage, autosomal dominant, 608358, Autosomal dominant; MSMA (Hyaline body myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548577}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyopati, Miyosin Depo, Otozomal Resesif, 255160, Otozomal Resesif; MSMB (Hyalen Body Miyopati) (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Miyopathy, myosin storage, autosomal recessive, 255160, Autosomal recessive; MSMB (Hyaline body myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. (ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Miyosin Depo, Otozomal Resesif, 255160, Otozomal Resesif; MSMB (Hyalen Body Miyopati) (Prenatal) (MYH7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Miyopathy, myosin storage, autosomal recessive, 255160, Autosomal recessive; MSMB (Hyaline body myopathy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. (ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Proksimal, Erken Solunum Kası Tutulumu, 603689 (Kalıtsal Proksimal Miyopati -Erken Solunum Yetmezliği) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, proximal, with early respiratory muscle involvement, 603689 (Hereditary proximal myopathy with early respiratory failure) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. (ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Proksimal, Erken Solunum Kası Tutulumu, 603689 (Kalıtsal Proksimal Miyopati -Erken Solunum Yetmezliği) (Prenatal) (TTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, proximal, with early respiratory muscle involvement, 603689 (Hereditary proximal myopathy with early respiratory failure) (TTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. (ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Skapulohumeroperoneal, 616852, Otozomal Dominant, SHPM (Progressive Scapulohumeroperoneal Distal Myopathy) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, scapulohumeroperoneal, 616852, Autosomal dominant; SHPM (Progressive scapulohumeroperoneal distal myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. (ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Skapulohumeroperoneal, 616852, Otozomal Dominant, SHPM (Progressive Scapulohumeroperoneal Distal Myopathy) (Prenatal) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, scapulohumeroperoneal, 616852, Autosomal dominant; SHPM (Progressive scapulohumeroperoneal distal myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. (ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Sferoid Cisimcik, 182920, Otozomal Dominant (Sferoid Cisimcikli Miyopatisi) (MYOT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, spheroid body, 182920, Autosomal dominant (Spheroid body myopathy) (MYOT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYOT	MYOT, TTOD, MFM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. (ECO:0000269|PubMed:16380616). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Sferoid Cisimcik, 182920, Otozomal Dominant (Sferoid Cisimcikli Miyopatisi) (MLPA)	Myopathy, spheroid body, 182920, Autosomal dominant (Spheroid body myopathy) (MLPA)	MYOT	MYOT, TTOD, MFM3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. (ECO:0000269|PubMed:16380616). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Sferoid Cisimcik, 182920, Otozomal Dominant (Sferoid Cisimcikli Miyopatisi) (Prenatal) (MYOT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, spheroid body, 182920, Autosomal dominant (Spheroid body myopathy) (MYOT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYOT	MYOT, TTOD, MFM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. (ECO:0000269|PubMed:16380616). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Sferoid Cisimcik, 182920, Otozomal Dominant (Sferoid Cisimcikli Miyopatisi) (Prenatal) (MLPA)	Myopathy, spheroid body, 182920, Autosomal dominant (Spheroid body myopathy) (Prenatal) (MLPA)	MYOT	MYOT, TTOD, MFM3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spheroid body myopathy (SBM) [MIM:182920]: Autosomal dominant form of myofibrillar myopathy (MFM), characterized by slowly progressing proximal muscle weakness and dysarthric nasal speech. There is no evidence of cardiomyopathy. Muscle biopsy shows spheroid bodies within the type I muscle fibers. (ECO:0000269|PubMed:16380616). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Tübüler Agregat, 1, 160565, Otozomal Dominant; TAM1 (Tübüler Agregat Miyopati) (STIM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, tubular aggregate, 1, 160565, Autosomal dominant; TAM1 (Tubular aggregate myopathy) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. (ECO:0000269|PubMed:23332920). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Tübüler Agregat, 1, 160565, Otozomal Dominant; TAM1 (Tübüler Agregat Miyopati) (Prenatal) (STIM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, tubular aggregate, 1, 160565, Autosomal dominant; TAM1 (Tubular aggregate myopathy) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. (ECO:0000269|PubMed:23332920). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Tübüler Agregat, 2, 615883, Otozomal Dominant; TAM2 (Tübüler Agregat Miyopati) (ORAI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, tubular aggregate, 2, 615883, Autosomal dominant; TAM2 (Tubular aggregate myopathy) (ORAI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ORAI1	ORAI1, TMEM142A, CRACM1, IMD9, TAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 9 (IMD9) [MIM:612782]: An immune disorder characterized by recurrent infections, impaired activation and proliferative response of T-cells, decreased T-cell production of cytokines, and normal lymphocytes counts and serum immunoglobulin levels. In surviving patients ectodermal dysplasia with anhidrosis and non-progressive myopathy may be observed. (ECO:0000269|PubMed:16147976, ECO:0000269|PubMed:16582901). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, Tübüler Agregat, 2, 615883, Otozomal Dominant; TAM2 (Tübüler Agregat Miyopati) (Prenatal) (ORAI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, tubular aggregate, 2, 615883, Autosomal dominant; TAM2 (Tubular aggregate myopathy) (ORAI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ORAI1	ORAI1, TMEM142A, CRACM1, IMD9, TAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Immunodeficiency 9 (IMD9) [MIM:612782]: An immune disorder characterized by recurrent infections, impaired activation and proliferative response of T-cells, decreased T-cell production of cytokines, and normal lymphocytes counts and serum immunoglobulin levels. In surviving patients ectodermal dysplasia with anhidrosis and non-progressive myopathy may be observed. (ECO:0000269|PubMed:16147976, ECO:0000269|PubMed:16582901). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, Vakúoler, CASQ1 Agregatları, 616231, Otozomal Dominant; VMCQA (Miyopati, Calsequestrine Bağlı ve SERCA1 Protein Aşırı Yüklenmesi) (CASQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, vacuolar, with CASQ1 aggregates, 616231, Autosomal dominant; VMCQA (Myopathy due to calsequestrin and SERCA1 protein overload) (CASQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASQ1	CASQ1, VMCQA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, vacuolar, with CASQ1 aggregates (VMCQA) [MIM:616231]: An autosomal dominant mild muscle disorder characterized by adult onset of muscle cramping and weakness as well as increased levels of serum creatine kinase. The disorder is not progressive, and some patients may be asymptomatic. (ECO:0000269|PubMed:25116801). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyopati, Vakúoler, CASQ1 Agregatları, 616231, Otozomal Dominant; VMCQA (Miyopati, Calsequestrine Bağlı ve SERCA1 Protein Aşırı Yüklenmesi) (Prenatal) (CASQ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, vacuolar, with CASQ1 aggregates, 616231, Autosomal dominant; VMCQA (Myopathy due to calsequestrin and SERCA1 protein overload) (CASQ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CASQ1	CASQ1, VMCQA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, vacuolar, with CASQ1 aggregates (VMCQA) [MIM:616231]: An autosomal dominant mild muscle disorder characterized by adult onset of muscle cramping and weakness as well as increased levels of serum creatine kinase. The disorder is not progressive, and some patients may be asymptomatic. (ECO:0000269|PubMed:25116801). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miyopati, Aşırı Otofaji İle, X'e Bağlı Miyopati, 310440, X'e Bağlı Resesif; MEAX (X'e Bağlı Miyopati-Aşırı Otofajili) (VMA21 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, X-linked, with excessive autophagy, 310440, X-linked recessive; MEAX (X-linked myopathy with excessive autophagy) (VMA21 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VMA21	VMA21, XMEA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, X-linked, with excessive autophagy (MEAX) [MIM:310440]: A muscle disorder characterized by childhood early onset of a slowly progressive proximal limb muscle weakness (especially in legs) and elevation of serum creatine kinase, without evidence of cardiac, respiratory or central nervous system involvement. Histopathological analysis reveals diseased muscle fibers that are not necrotic, but show abnormal autophagic vacuolation as a manifestation of excessive autophagic activity in skeletal muscle cells. {ECO:0000269|PubMed:19379691, ECO:0000269|PubMed:23315026, ECO:0000269|PubMed:24488655, ECO:0000269|PubMed:25683699}. Note=The disease is caused by mutations affecting the gene represented in this entry. VMA21 deficiency results in an increase of lysosomal pH from 4.7 to 5.2, which reduces lysosomal degradative ability and blocks autophagy. This reduces cellular free amino acids, which upregulates the mTOR pathway and mTOR-dependent macroautophagy, resulting in proliferation of large and ineffective autolysosomes that engulf sections of cytoplasm, merge together, and vacuolate the cell. {ECO:0000269|PubMed:23315026}.	EDTA Blood Sample
Miyopati, Aşırı Otofaji İle, X'e Bağlı Miyopati, 310440, X'e Bağlı Resesif; MEAX (X'e Bağlı Miyopati-Aşırı Otofajili) (Prenatal) (VMA21 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, X-linked, with excessive autophagy, 310440, X-linked recessive; MEAX (X-linked myopathy with excessive autophagy) (VMA21 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VMA21	VMA21, XMEA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, X-linked, with excessive autophagy (MEAX) [MIM:310440]: A muscle disorder characterized by childhood early onset of a slowly progressive proximal limb muscle weakness (especially in legs) and elevation of serum creatine kinase, without evidence of cardiac, respiratory or central nervous system involvement. Histopathological analysis reveals diseased muscle fibers that are not necrotic, but show abnormal autophagic vacuolation as a manifestation of excessive autophagic activity in skeletal muscle cells. {ECO:0000269|PubMed:19379691, ECO:0000269|PubMed:23315026, ECO:0000269|PubMed:24488655, ECO:0000269|PubMed:25683699}. Note=The disease is caused by mutations affecting the gene represented in this entry. VMA21 deficiency results in an increase of lysosomal pH from 4.7 to 5.2, which reduces lysosomal degradative ability and blocks autophagy. This reduces cellular free amino acids, which upregulates the mTOR pathway and mTOR-dependent macroautophagy, resulting in proliferation of large and ineffective autolysosomes that engulf sections of cytoplasm, merge together, and vacuolate the cell. {ECO:0000269|PubMed:23315026}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopati, X'e Bağlı, Postural Müsküler Atrofi, 300696, X'e Bağlı Resesif; XMPMA (X'e Bağlı Miyopati, Postüral Müsküler Atrofi) (FHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopathy, X-linked, with postural muscle atrophy, 300696, X-linked recessive; XMPMA (X-linked myopathy with postural muscle atrophy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopati, X'e Bağlı, Postural Müsküler Atrofi, 300696, X'e Bağlı Resesif; XMPMA (X'e Bağlı Miyopati, Postüral Müsküler Atrofi) (Prenatal) (FHL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myopathy, X-linked, with postural muscle atrophy, 300696, X-linked recessive; XMPMA (X-linked myopathy with postural muscle atrophy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyopi 21, Otozomal Dominant, 614167, Otozomal Dominant; MYP21 (ZNF644 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 21, autosomal dominant, 614167, Autosomal dominant; MYP21 (ZNF644 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF644	ZNF644, MYP21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Miyopi 22 Otozomal Dominant, 615420, Otozomal Dominant; MYP22 (PRIMPOL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 22, autosomal dominant, 615420, Autosomal dominant; MYP22 (PRIMPOL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRIMPOL	CCDC111, MYP22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopia 22, autosomal dominant (MYP22) [MIM:615420]: A refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269	PubMed:23579484}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Miyopi 23, Otozomal Resesif, 615431, Otozomal Resesif; MYP23 (Nadir İzole Miyopi) (LRPAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 23, autosomal recessive, 615431, Autosomal recessive; MYP23 (Rare isolated myopia) (LRPAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRPAP1	LRPAP1, A2MRAP, MYP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopia 23, autosomal recessive (MYP23) [MIM:615431]: A refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269	PubMed:23830514}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyopi 24, Otozomal Dominant, 615946, Otozomal Dominant; MYP24 (SLC39A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 24, autosomal dominant, 615946, Autosomal dominant; MYP24 (SLC39A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC39A5	SLC39A5, MYP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopia 24, autosomal dominant (MYP24) [MIM:615946]: A refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269|PubMed:24891338, ECO:0000269|PubMed:25525168}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Miyopi 25, Otozomal Dominant, 617238, Otozomal Dominant; MYP25 (P4HA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 25, autosomal dominant, 617238, Autosomal dominant; MYP25 (P4HA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P4HA2	P4HA2, MYP25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=P4HA2 mutations may be a cause of myopia, a refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269	PubMed:25741866}.	EDTA Blood Sample
Miyopi 6, 608908, Otozomal Dominant; MYP6 (Nadir İzole Miyopi) (SCO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia 6, 608908, Autosomal dominant; MYP6 (Rare isolated myopia) (SCO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCO2	SCO2, CEMCOX1, MYP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopia 6 (MYP6) [MIM:608908]: A refractive error of the eye, in which parallel rays from a distant object come to focus in front of the retina, vision being better for near objects than for far. {ECO:0000269|PubMed:23643385, ECO:0000269|PubMed:25525168}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Miyop, 614292, Otozomal Resesif, Katarakt ve Vitreoretinal Dejenerasyonu İle, Yüksek; MCVD (Nadir İzole Miyopi) (P3H2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myopia, high, with cataract and vitreoretinal degeneration, 614292, Autosomal recessive; MCVD (Rare isolated myopia) (P3H2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P3H2	P3H2, LEPREL1, MCVD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Miyoskleroz, Konjenital, 255600, Otozomal Resesif (Miyosklerozis) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myosclerosis, congenital, 255600, Autosomal recessive (Myosclerosis) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. {ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Miyoskleroz, Konjenital, 255600, Otozomal Resesif (Miyosklerozis) (Prenatal) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myosclerosis, congenital, 255600, Autosomal recessive (Myosclerosis) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. {ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia Kongenita Atipik, Asetazolamid Duyarlı, 608390, Otozomal Dominant (Potasyum İle Uyarılan Miyotoni) (SCN4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myotonia congenita, atypical, acetazolamide-responsive, 608390, Autosomal dominant (Potassium-aggravated myotonia) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Myotonia Kongenita Atipik, Asetazolamid Duyarlı, 608390, Otozomal Dominant (Potasyum İle Uyarılan Miyotoni) (MLPA)	Myotonia congenita, atypical, acetazolamide-responsive, 608390, Autosomal dominant (Potassium-aggravated myotonia) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Konjenital Miyotoni, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Konjenital Miyotoni, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (MLPA)	MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE (Thomsen and Becker disease) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Konjenital Miyotoni, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (Prenatal) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Miyotoni, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (Prenatal) (MLPA)	MYOTONIA CONGENITA, AUTOSOMAL RECESSIVE (Thomsen and Becker disease) (Prenatal) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia Congenita, Dominant, 160800, Otozomal Dominant (Thomsen ve Becker Hastalığı, Otozomal Dominant) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myotonia congenita, dominant, 160800, Autosomal dominant (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Myotonia Congenita, Dominant, 160800, Otozomal Dominant (Thomsen ve Becker Hastalığı, Otozomal Dominant) (MLPA)	Myotonia congenita, dominant, 160800, Autosomal dominant (Thomsen and Becker disease) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Myotonia Congenita, Dominant, 160800, Otozomal Dominant (Thomsen ve Becker Hastalığı, Otozomal Dominant) (Prenatal) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myotonia congenita, dominant, 160800, Autosomal dominant (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia Congenita, Dominant, 160800, Otozomal Dominant (Thomsen ve Becker Hastalığı, Otozomal Dominant) (Prenatal) (MLPA)	Myotonia congenita, dominant, 160800, Autosomal dominant (Thomsen and Becker disease) (Prenatal) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia Congenita, Resesif, 255700, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myotonia congenita, recessive, 255700, Autosomal recessive (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Myotonia Congenita, Resesif, 255700, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (MLPA)	Myotonia congenita, recessive, 255700, Autosomal recessive (Thomsen and Becker disease) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Myotonia Congenita, Resesif, 255700, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (Prenatal) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myotonia congenita, recessive, 255700, Autosomal recessive (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia Congenita, Resesif, 255700, Otozomal Resesif (Thomsen ve Becker Hastalığı, Otozomal Resesif) (Prenatal) (MLPA)	Myotonia congenita, recessive, 255700, Autosomal recessive (Thomsen and Becker disease) (Prenatal) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonia kongenita, Thomsen hastalığı, Becker hastalığı, Andersen-Tawil sendromu paneli (CLCN1, KCNJ2) (MLPA)	Myotonia kongenita, Thomsen's disease, Becker's disease, Andersen-Tawil syndrome panel (CLCN1, KCNJ2) (MLPA)	CLCN1, KCNJ2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Myotonia kongenita, Thomsen hastalığı, Becker hastalığı, Andersen-Tawil sendromu paneli (CLCN1, KCNJ2) (MLPA) (Prenatal)	Myotonia kongenita, Thomsen's disease, Becker's disease, Andersen-Tawil syndrome panel (CLCN1, KCNJ2) (MLPA) (Prenatal)	CLCN1, KCNJ2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyotoni Levior, Resesif (Thomsen ve Becker Hastalığı, Otozomal Dominant) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myotonia levior, recessive (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyotoni Levior, Resesif (Thomsen ve Becker Hastalığı, Otozomal Dominant) (MLPA)	Myotonia levior, recessive (Thomsen and Becker disease) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Miyotoni Levior, Resesif (Thomsen ve Becker Hastalığı, Otozomal Dominant) (Prenatal) (CLCN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myotonia levior, recessive (Thomsen and Becker disease) (CLCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN1	CLCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miyotoni Levior, Resesif (Thomsen ve Becker Hastalığı, Otozomal Dominant) (Prenatal) (MLPA)	Myotonia levior, recessive (Thomsen and Becker disease) (Prenatal) (MLPA)	CLCN1	CLCN1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myotonia congenita, autosomal recessive (MCAR) [MIM:255700]: A non-dystrophic skeletal muscle disorder characterized by muscle stiffness and an inability of the muscle to relax after voluntary contraction. Most patients have symptom onset in the legs, which later progresses to the arms, neck, and facial muscles. Many patients show marked hypertrophy of the lower limb muscles. The autosomal recessive form (Becker disease) is more severe than the autosomal dominant one (Thomsen disease). {ECO:0000269|PubMed:10215406, ECO:0000269|PubMed:1379744, ECO:0000269|PubMed:7874130, ECO:0000269|PubMed:7951242, ECO:0000269|PubMed:7981681, ECO:0000269|PubMed:8571958}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Myotonik Distrofi 1, 160900, Otozomal Dominant; DM1 (Steinert Myotonik Distrofi) (Tekrar Sayısı-)	Myotonic dystrophy 1, 160900, Autosomal dominant; DM1 (Steinert myotonic dystrophy)(Repeat Analysis)	DMPK	DMPK, DM, DMK	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)	*	20	90	.	.	60	Postnatal	DISEASE: Dystrophia myotonica 1 (DM1) [MIM:160900]: A muscular disorder characterized by myotonia, muscle wasting in the distal extremities, cataract, hypogonadism, defective endocrine functions, male baldness and cardiac arrhythmias. {ECO:0000269|PubMed:1302022, ECO:0000269|PubMed:1310900, ECO:0000269|PubMed:1546326, ECO:0000269|PubMed:19514047}. Note=The disease is caused by mutations affecting the gene represented in this entry. The causative mutation is a CTG expansion in the 3'- UTR of the DMPK gene. A length exceeding 50 CTG repeats is pathogenic, while normal individuals have 5 to 37 repeats. Intermediate alleles with 35-49 triplets are not disease-causing but show instability in intergenerational transmissions. Disease severity varies with the number of repeats: mildly affected persons have 50 to 150 repeats, patients with classic DM have 100 to 1,000 repeats, and those with congenital onset can have more than 2,000 repeats. {ECO:0000269|PubMed:1310900, ECO:0000269|PubMed:19514047}.;	EDTA Blood Sample
Myotübüler Miyopati, X'e Bağlı, 310400, X'e Bağlı, CNMX (X'e Bağlı Santronükleer Miyopati) (MTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myotubular myopathy, X-linked, 310400, X-linked recessive; CNMX (X-linked centronuclear myopathy) (MTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTM1	MTM1, MTMX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, centronuclear, X-linked (CNMX) [MIM:310400]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:10063835, ECO:0000269|PubMed:10466421, ECO:0000269|PubMed:10502779, ECO:0000269|PubMed:11793470, ECO:0000269|PubMed:12031625, ECO:0000269|PubMed:12522554, ECO:0000269|PubMed:12859411, ECO:0000269|PubMed:17005396, ECO:0000269|PubMed:19129059, ECO:0000269|PubMed:9285787, ECO:0000269|PubMed:9305655, ECO:0000269|PubMed:9829274}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Myotübüler Miyopati, X'e Bağlı, 310400, X'e Bağlı, CNMX (X'e Bağlı Santronükleer Miyopati) (Prenatal) (MTM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myotubular myopathy, X-linked, 310400, X-linked recessive; CNMX (X-linked centronuclear myopathy) (MTM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTM1	MTM1, MTMX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, centronuclear, X-linked (CNMX) [MIM:310400]: A congenital muscle disorder characterized by progressive muscular weakness and wasting involving mainly limb girdle, trunk, and neck muscles. It may also affect distal muscles. Weakness may be present during childhood or adolescence or may not become evident until the third decade of life. Ptosis is a frequent clinical feature. The most prominent histopathologic features include high frequency of centrally located nuclei in muscle fibers not secondary to regeneration, radial arrangement of sarcoplasmic strands around the central nuclei, and predominance and hypotrophy of type 1 fibers. {ECO:0000269|PubMed:10063835, ECO:0000269|PubMed:10466421, ECO:0000269|PubMed:10502779, ECO:0000269|PubMed:11793470, ECO:0000269|PubMed:12031625, ECO:0000269|PubMed:12522554, ECO:0000269|PubMed:12859411, ECO:0000269|PubMed:17005396, ECO:0000269|PubMed:19129059, ECO:0000269|PubMed:9285787, ECO:0000269|PubMed:9305655, ECO:0000269|PubMed:9829274}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Miksoid Liposarkom, 613488 (Liposarkom) (DDIT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myxoid liposarcoma, 613488 (Liposarcoma) (DDIT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDIT3	DDIT3, GADD153, CHOP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myxoid liposarcoma (MXLIPO) [MIM:613488]: A soft tissue tumor that tends to occur in the limbs (especially the thigh) of patients ranging in age from 35 to 55 years. It is defined by the presence of a hypocellular spindle cell proliferation set in a myxoid background, often with mucin pooling. Lipoblasts tend to be small and often monovacuolated and to cluster around vessels or at the periphery of the lesion. {ECO:0000269|PubMed:7503811}. Note=The gene represented in this entry may be involved in disease pathogenesis. A chromosomal aberration involving DDIT3 has been found in a patient with malignant myxoid liposarcoma. Translocation t(12;16)(q13;p11) with FUS (PubMed:7503811). {ECO:0000269|PubMed:7503811}.	EDTA Blood Sample
Miksoid Liposarkom, 613488 (Liposarkom) (Prenatal) (DDIT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Myxoid liposarcoma, 613488 (Liposarcoma) (DDIT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDIT3	DDIT3, GADD153, CHOP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myxoid liposarcoma (MXLIPO) [MIM:613488]: A soft tissue tumor that tends to occur in the limbs (especially the thigh) of patients ranging in age from 35 to 55 years. It is defined by the presence of a hypocellular spindle cell proliferation set in a myxoid background, often with mucin pooling. Lipoblasts tend to be small and often monovacuolated and to cluster around vessels or at the periphery of the lesion. {ECO:0000269|PubMed:7503811}. Note=The gene represented in this entry may be involved in disease pathogenesis. A chromosomal aberration involving DDIT3 has been found in a patient with malignant myxoid liposarcoma. Translocation t(12;16)(q13;p11) with FUS (PubMed:7503811). {ECO:0000269|PubMed:7503811}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Miksoma, İntrakardiyak, 255960, Otozomal Dominant (Ailesel Atriyal Miksoma) (PRKAR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Myxoma, intracardiac, 255960, Autosomal dominant (Familial atrial myxoma) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrodysostosis 1, with or without hormone resistance (ACRDYS1) [MIM:101800]: A form of skeletal dysplasia characterized by short stature, severe brachydactyly, facial dysostosis, and nasal hypoplasia. Affected individuals often have advanced bone age and obesity. Laboratory studies show resistance to multiple hormones, including parathyroid, thyrotropin, calcitonin, growth hormone-releasing hormone, and gonadotropin. However, not all patients show endocrine abnormalities. {ECO:0000269|PubMed:21651393, ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:22723333, ECO:0000269|PubMed:23043190, ECO:0000269|PubMed:23425300}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
N-Asetilaspartat Eksikliği, 614063, Otozomal Resesif; NACED (NAT8L geni) (Tüm Gen Dizi Analizi) (Postnatal)	N-acetylaspartate deficiency, 614063, Autosomal recessive; NACED (NAT8L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAT8L	NAT8L, CML3, NACED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: N-acetylaspartate deficiency (NACED) [MIM:614063]: A metabolic disorder resulting in truncal ataxia, marked developmental delay, seizures, and secondary microcephaly. {ECO:0000269|PubMed:19807691}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
N-Asetilaspartat Eksikliği, 614063, Otozomal Resesif; NACED (Prenatal) (NAT8L geni) (Tüm Gen Dizi Analizi) (Prenatal)	N-acetylaspartate deficiency, 614063, Autosomal recessive; NACED (NAT8L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAT8L	NAT8L, CML3, NACED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: N-acetylaspartate deficiency (NACED) [MIM:614063]: A metabolic disorder resulting in truncal ataxia, marked developmental delay, seizures, and secondary microcephaly. {ECO:0000269|PubMed:19807691}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
N-Asetilglutamat Sentaz Eksikliği, 237310, Otozomal Resesif; NAGSD (Hiperamonemi, N-Asetilglutamat Sentaz Eksikliğine Bağlı) (NAGS geni) (Tüm Gen Dizi Analizi) (Postnatal)	N-acetylglutamate synthase deficiency, 237310, Autosomal recessive; NAGSD (Hyperammonemia due to N-acetylglutamate synthase deficiency) (NAGS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGS	NAGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: N-acetylglutamate synthase deficiency (NAGSD) [MIM:237310]: Rare autosomal recessively inherited metabolic disorder leading to severe neonatal or late-onset hyperammonemia without increased excretion of orotic acid. Clinical symptoms are somnolence, tachypnea, feeding difficulties, a severe neurologic presentation characterized by uncontrollable movements, developmental delay, visual impairment, failure to thrive and hyperammonemia precipitated by the introduction of high-protein diet or febrile illness. {ECO:0000269|PubMed:12754705, ECO:0000269|PubMed:15878741}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
N-Asetilglutamat Sentaz Eksikliği, 237310, Otozomal Resesif; NAGSD (Hiperamonemi, N-Asetilglutamat Sentaz Eksikliğine Bağlı) (Prenatal) (NAGS geni) (Tüm Gen Dizi Analizi) (Prenatal)	N-acetylglutamate synthase deficiency, 237310, Autosomal recessive; NAGSD (Hyperammonemia due to N-acetylglutamate synthase deficiency) (NAGS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAGS	NAGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: N-acetylglutamate synthase deficiency (NAGSD) [MIM:237310]: Rare autosomal recessively inherited metabolic disorder leading to severe neonatal or late-onset hyperammonemia without increased excretion of orotic acid. Clinical symptoms are somnolence, tachypnea, feeding difficulties, a severe neurologic presentation characterized by uncontrollable movements, developmental delay, visual impairment, failure to thrive and hyperammonemia precipitated by the introduction of high-protein diet or febrile illness. {ECO:0000269|PubMed:12754705, ECO:0000269|PubMed:15878741}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nablus Maskesi Benzeri Yüz Sendromu, 608156, Otozomal Resesif, Sporadik Vakalar (8q22.1 Mikrodelesyon Sendromu) (2200)	Nablus mask-like facial syndrome, 608156, Autosomal recessive, Isolated cases (8q22.1 microdeletion syndrome) (440)	.	NMLFS, DEL8q22.1, CBDELq22.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Naegeli-Franceschetti-Jadassohn Sendromu, 161000, Otozomal Dominant; NFJS (Naegeli-Franceschetti-Jadassohn Sendromu) (KRT14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Naegeli-Franceschetti-Jadassohn syndrome, 161000, Autosomal dominant; NFJS (Naegeli-Franceschetti-Jadassohn syndrome) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Naegeli-Franceschetti-Jadassohn Sendromu, 161000, Otozomal Dominant; NFJS (Naegeli-Franceschetti-Jadassohn Sendromu) (MLPA)	Naegeli-Franceschetti-Jadassohn syndrome, 161000, Autosomal dominant; NFJS (Naegeli-Franceschetti-Jadassohn syndrome) (MLPA)	KRT14	KRT14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Naegeli-Franceschetti-Jadassohn Sendromu, 161000, Otozomal Dominant; NFJS (Naegeli-Franceschetti-Jadassohn Sendromu) (Prenatal) (KRT14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Naegeli-Franceschetti-Jadassohn syndrome, 161000, Autosomal dominant; NFJS (Naegeli-Franceschetti-Jadassohn syndrome) (KRT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRT14	KRT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Naegeli-Franceschetti-Jadassohn Sendromu, 161000, Otozomal Dominant; NFJS (Naegeli-Franceschetti-Jadassohn Sendromu) (Prenatal) (MLPA)	Naegeli-Franceschetti-Jadassohn syndrome, 161000, Autosomal dominant; NFJS (Naegeli-Franceschetti-Jadassohn syndrome) (Prenatal) (MLPA)	KRT14	KRT14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dermatopathia pigmentosa reticularis (DPR) [MIM:125595]: A rare ectodermal dysplasia characterized by lifelong persistent reticulate hyperpigmentation, non-cicatricial alopecia, and nail dystrophy. Variable features include adermatoglyphia, hypohidrosis or hyperhidrosis, and palmoplantar hyperkeratosis. {ECO:0000269|PubMed:16960809}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tırnak Bozukluğu, Sendromik Olmayan Konjenital, 10, Kıskaç Şeklinde Tırnak, 614157, Otozomal Resesif; NDNC10 (Otozomal Resesif Tırnak Displazisi) (FZD6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nail disorder, nonsyndromic congenital, 10, (claw-shaped nails), 614157, Autosomal recessive; NDNC10 (Autosomal recessive nail dysplasia) (FZD6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FZD6	FZD6, NDNC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nail disorder, non-syndromic congenital, 10 (NDNC10) [MIM:614157]: A nail disorder characterized by a variable degree of onychauxis (thick nails), hyponychia, and onycholysis of all nails, with claw-shaped fingernails in some individuals. No other anomalies of ectodermal tissues, including hair, teeth, sweat glands, or skin, are noted, and individuals with dysplastic nails have normal hearing and normal psychomotor development. {ECO:0000269|PubMed:21666003}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tırnak Bozukluğu, Sendromik Olmayan Konjenital, 3, (Lökonişi), 151600, Otozomal Resesif, Otozomal Dominant; NDNC3 (Lökonişia Totalis) (PLCD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600, Autosomal recessive, Autosomal dominant; NDNC3 (Leukonychia totalis) (PLCD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCD1	PLCD1, NDNC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nail disorder, non-syndromic congenital, 3 (NDNC3) [MIM:151600]: A nail disorder characterized by a white appearance of the nail plate (true leukonychia), the nail bed (pseudoleukonychia), or neither (apparent leukonychia). Leukonychia may involve all of the nail (leukonychia totalis) or only part of the nail (leukonychia partialis), or can appear as one or more transverse bands (leukonychia striata) or white spots (leukonychia punctata). {ECO:0000269|PubMed:21665001}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tırnak Patella Sendromu, 161200, Otozomal Dominant; NPS (Tırnak Patella Sendromu) (LMX1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nail-patella syndrome, 161200, Autosomal dominant; NPS (Nail-patella syndrome) (LMX1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMX1B	LMX1B, NPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tırnak Patella Sendromu, 161200, Otozomal Dominant; NPS (Tırnak Patella Sendromu) (MLPA)	Nail-patella syndrome, 161200, Autosomal dominant; NPS (Nail-patella syndrome) (MLPA)	LMX1B	LMX1B, NPS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Tırnak Patella Sendromu, 161200, Otozomal Dominant; NPS (Tırnak Patella Sendromu) (Prenatal) (LMX1B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nail-patella syndrome, 161200, Autosomal dominant; NPS (Nail-patella syndrome) (LMX1B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMX1B	LMX1B, NPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tırnak Patella Sendromu, 161200, Otozomal Dominant; NPS (Tırnak Patella Sendromu) (Prenatal) (MLPA)	Nail-patella syndrome, 161200, Autosomal dominant; NPS (Nail-patella syndrome) (Prenatal) (MLPA)	LMX1B	LMX1B, NPS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nance-Horan Sendromu, 302350, X'e Bağlı Dominant; NHS (Nance-Horan Sendromu) (NHS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nance-Horan syndrome, 302350, X-linked dominant; NHS (Nance-Horan syndrome) (NHS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NHS	NHS, CXN, CTRCT40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 40 (CTRCT40) [MIM:302200]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT40 manifests as a congenital nuclear opacity with severe visual impairment in affected males. Heterozygous females have suture cataracts and only slight reduction in vision. In some cases, cataract is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. {ECO:0000269|PubMed:19414485}. Note=The disease is caused by mutations affecting the gene represented in this entry. Caused by copy number variations predicted to result in altered transcriptional regulation of the NHS gene: a 0.8 Mb segmental duplication-triplication encompassing the NHS, SCML1 and RAI2 genes, and an 4.8 kb intragenic deletion in NHS intron 1.;	EDTA Blood Sample

Nance-Horan Sendromu, 302350, X'e Bağlı Dominant; NHS (Nance-Horan Sendromu) (Prenatal) (NHS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nance-Horan syndrome, 302350, X-linked dominant; NHS (Nance-Horan syndrome) (NHS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NHS	NHS, CXN, CTRCT40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cataract 40 (CTRCT40) [MIM:302200]: An opacification of the crystalline lens of the eye that frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. CTRCT40 manifests as a congenital nuclear opacity with severe visual impairment in affected males. Heterozygous females have suture cataracts and only slight reduction in vision. In some cases, cataract is associated with microcornea without any other systemic anomaly or dysmorphism. Microcornea is defined by a corneal diameter inferior to 10 mm in both meridians in an otherwise normal eye. {ECO:0000269	PubMed:19414485}. Note=The disease is caused by mutations affecting the gene represented in this entry. Caused by copy number variations predicted to result in altered transcriptional regulation of the NHS gene: a 0.8 Mb segmental duplication-triplication encompassing the NHS, SCML1 and RAI2 genes, and an 4.8 kb intragenic deletion in NHS intron 1.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nanoftalmus 2, 609549; NNO2 (Nanoftalmi 4) (MFRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nanophthalmos 2, 609549; NNO2 (Nanophthalmia) (MFRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MFRP	MFRP, MCOPS, NNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nanophthalmos 2 (NNO2) [MIM:609549]: Rare autosomal recessive disorder of eye development characterized by extreme hyperopia and small functional eyes. {ECO:0000269	PubMed:15976030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nanoftalmus 4, 615972, Otozomal Dominant; NNO4 (Nanoftalmi 4) (TMEM98 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nanophthalmos 4, 615972, Autosomal dominant; NNO4 (Nanophthalmia) (TMEM98 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM98	TMEM98, NNO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Narkolepsi 1, 161400, Otozomal Dominant; NRCLP1 (Narkolepsi-Katapleksi) (HCRT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Narcolepsy 1, 161400, Autosomal dominant; NRCLP1 (Narcolepsy-cataplexy) (HCRT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCRT	HCRT, OX, NRCLP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Narcolepsy 1 (NRCLP1) [MIM:161400]: Neurological disabling sleep disorder, characterized by excessive daytime sleepiness, sleep fragmentation, symptoms of abnormal rapid-eye- movement (REM) sleep, cataplexy, hypnagogic hallucinations, and sleep paralysis. Cataplexy is a sudden loss of muscle tone triggered by emotions, which is the most valuable clinical feature used to diagnose narcolepsy. Human narcolepsy is primarily a sporadically occurring disorder but familial clustering has been observed. {ECO:0000269	PubMed:10973318}. Note=The disease is caused by mutations affecting the gene represented in this entry. Human narcolepsy is associated with a deficient orexin system. Orexins are absent and/or greatly diminished in the brain and cerebrospinal fluid (CSF) of most narcoleptic patients.;	EDTA Blood Sample
Narkolepsi 7, 614250, Otozomal Dominant; NRCLP7 (Narkolepsi-Katapleksi) (MOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Narcolepsy 7, 614250, Autosomal dominant; NRCLP7 (Narcolepsy-cataplexy) (MOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MOG	MOG, NRCLP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nazofarenks Karsinomu, 607107 (Nazofarenks Karsinomu (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nasopharyngeal carcinoma, 607107 (Nasopharyngeal carcinoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nazofarenks Karsinomu, 607107 (Nazofarenks Karsinomu) (MLPA)	Nasopharyngeal carcinoma, 607107 (Nasopharyngeal carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nazofarenks Karsinomu, 607107 (Nazofarenks Karsinomu) (MLPA)	Nasopharyngeal carcinoma, 607107 (Nasopharyngeal carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nazofaringeal Karsinoma, Yatkınlık, 3, 617075, Otozomal Dominant; NPCA3 (Nazofarenks Karsinomu) (MST1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nasopharyngeal carcinoma, susceptibility to, 3, 617075, Autosomal dominant; NPCA3 (Nasopharyngeal carcinoma) (MST1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MST1R	MST1R, RON, NPCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Native American Miyopati, 255995, Otozomal Resesif; NAM (Amerikan Yerlilerinin Miyopatisi) (STAC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Native American myopathy, 255995, Autosomal recessive; NAM (Native American myopathy) (STAC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAC3	STAC3, NAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Native American myopathy (NAM) [MIM:255995]: A disease characterized by congenital weakness and arthrogryposis, cleft palate, ptosis, short stature, kyphoscoliosis, talipes deformities, and susceptibility to malignant hyperthermia provoked by anesthesia. {ECO:0000269	PubMed:23736855}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Doğal Letal Hücre (Natural Killer Cell) ve Glukokortikoid Eksikliği, DNA Tamir Defekti, 609981, Otozomal Resesif; NKGCD (Primer İmmün Yetmezlik, NK Hücre Eksikliği ve Adrenal Yetmezlik) (MCM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981, Autosomal recessive; NKGCD (Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency) (MCM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCM4	MCM4, NKGCD, NKCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Natural killer cell and glucocorticoid deficiency with DNA repair defect (NKGCD) [MIM:609981]: An autosomal recessive disorder characterized by severe intra- and extrauterine growth retardation, microcephaly, decreased numbers of natural killer cells, and recurrent viral infections, most often affecting the respiratory tract and leading to respiratory failure. Affected individuals also have adrenal insufficiency requiring corticosteroid replacement therapy and may have an increased susceptibility to cancer. {ECO:0000269|PubMed:22354167, ECO:0000269|PubMed:22354170, ECO:0000269|PubMed:22493342}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Doğal Letal Hücre (Natural Killer Cell) ve Glukokortikoid Eksikliği, DNA Tamir Defekti, 609981, Otozomal Resesif; NKGCD (Primer İmmün Yetmezlik, NK Hücre Eksikliği ve Adrenal Yetmezlik) (Prenatal) (MCM4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Natural killer cell and glucocorticoid deficiency with DNA repair defect, 609981, Autosomal recessive; NKGCD (Primary immunodeficiency with natural-killer cell deficiency and adrenal insufficiency) (MCM4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MCM4	MCM4, NKGCD, NKCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Natural killer cell and glucocorticoid deficiency with DNA repair defect (NKGCD) [MIM:609981]: An autosomal recessive disorder characterized by severe intra- and extrauterine growth retardation, microcephaly, decreased numbers of natural killer cells, and recurrent viral infections, most often affecting the respiratory tract and leading to respiratory failure. Affected individuals also have adrenal insufficiency requiring corticosteroid replacement therapy and may have an increased susceptibility to cancer. {ECO:0000269|PubMed:22354167, ECO:0000269|PubMed:22354170, ECO:0000269|PubMed:22493342}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Naxos Hastalığı, 601214, Otozomal Resesif; NXD (Naxos Hastalığı) (JUP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Naxos disease, 601214, Autosomal recessive; NXD (Naxos disease) (JUP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JUP	JUP, DP3, PDGB, ARVD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Naxos disease (NXD) [MIM:601214]: An autosomal recessive disorder characterized by the association of diffuse non- epidermolytic palmoplantar keratoderma with woolly hair and cardiac abnormalities such as dilated cardiomyopathy and arrhythmogenic right ventricular dysplasia. {ECO:0000269	PubMed:10902626}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Naxos Hastalığı, 601214, Otozomal Resesif; NXD (Naxos Hastalığı) (Prenatal) (JUP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Naxos disease, 601214, Autosomal recessive; NXD (Naxos disease) (JUP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JUP	JUP, DP3, PDGB, ARVD12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Naxos disease (NXD) [MIM:601214]: An autosomal recessive disorder characterized by the association of diffuse non- epidermolytic palmoplantar keratoderma with woolly hair and cardiac abnormalities such as dilated cardiomyopathy and arrhythmogenic right ventricular dysplasia. {ECO:0000269	PubMed:10902626}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 1, Otozomal Dominant Veya Resesif, 609284, Otozomal Resesif, Otozomal Dominant; NEM1 (Intermediate Nemalin Miyopati) (TPM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 1, autosomal dominant or recessive, 609284, Autosomal recessive, Autosomal dominant; NEM1 (Intermediate nemaline myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, congenital, with fiber-type disproportion (CFTD) [MIM:255310]: A genetically heterogeneous disorder in which there is relative hypotrophy of type 1 muscle fibers compared to type 2 fibers on skeletal muscle biopsy. However, these findings are not specific and can be found in many different myopathic and neuropathic conditions. {ECO:0000269|PubMed:18300303, ECO:0000269|PubMed:19953533, ECO:0000269|PubMed:20951040, ECO:0000269|PubMed:24692096}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nemalin Miyopati 1, Otozomal Dominant Veya Resesif, 609284, Otozomal Resesif, Otozomal Dominant; NEM1 (Intermediate Nemalin Miyopati) (Prenatal) (TPM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 1, autosomal dominant or recessive, 609284, Autosomal recessive, Autosomal dominant; NEM1 (Intermediate nemaline myopathy) (TPM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM3	TPM3, NEM1, CFTD, CAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, congenital, with fiber-type disproportion (CFTD) [MIM:255310]: A genetically heterogeneous disorder in which there is relative hypotrophy of type 1 muscle fibers compared to type 2 fibers on skeletal muscle biopsy. However, these findings are not specific and can be found in many different myopathic and neuropathic conditions. {ECO:0000269|PubMed:18300303, ECO:0000269|PubMed:19953533, ECO:0000269|PubMed:20951040, ECO:0000269|PubMed:24692096}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 10, 616165, Otozomal Resesif; NEM10 (Ağır Konjenital Nemalin Miyopati) (LMOD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 10, 616165, Autosomal recessive; NEM10 (Severe congenital nemaline myopathy) (LMOD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMOD3	LMOD3, NEM10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 10 (NEM10) [MIM:616165]: An autosomal recessive severe form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. NEM10 is characterized by early-onset generalized muscle weakness and hypotonia with respiratory insufficiency and feeding difficulties. Additional features include arthrogryposis or congenital contractures, ophthalmoplegia, a history of prematurity, reduced fetal movements, and polyhydramnios. Most patients die of respiratory failure in early infancy. {ECO:0000269	PubMed:25250574}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nemalin Miyopati 10, 616165, Otozomal Resesif; NEM10 (Ağır Konjenital Nemalin Miyopati) (Prenatal) (LMOD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 10, 616165, Autosomal recessive; NEM10 (Severe congenital nemaline myopathy) (LMOD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMOD3	LMOD3, NEM10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 10 (NEM10) [MIM:616165]: An autosomal recessive severe form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. NEM10 is characterized by early-onset generalized muscle weakness and hypotonia with respiratory insufficiency and feeding difficulties. Additional features include arthrogryposis or congenital contractures, ophthalmoplegia, a history of prematurity, reduced fetal movements, and polyhydramnios. Most patients die of respiratory failure in early infancy. {ECO:0000269	PubMed:25250574}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nemalin Miyopati 11, Otozomal Resesif, 617336, Otozomal Resesif; NEM11 (Allesel İzole Dilate Kardiyomiyopati) (MYPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 11, autosomal recessive, 617336, Autosomal recessive; NEM11 (Familial isolated dilated cardiomyopathy) (MYPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYPN	MYPN, CMD1DD, CMH22, RCM4, NEM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 22 (CMH22) [MIM:615248]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269	PubMed:22286171}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nemalin Miyopati 11, Otozomal Resesif, 617336, Otozomal Resesif; NEM11 (Allesel İzole Dilate Kardiyomiyopati) (Prenatal) (MYPN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 11, autosomal recessive, 617336, Autosomal recessive; NEM11 (Familial isolated dilated cardiomyopathy) (MYPN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYPN	MYPN, CMD1DD, CMH22, RCM4, NEM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiomyopathy, familial hypertrophic 22 (CMH22) [MIM:615248]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269	PubMed:22286171}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 2, Otozomal Resesif, 256030, Otozomal Resesif; NEM2 (Ağır Konjenital Nemalin Miyopati) (NEB geni) (Tüm Gen Dizi Analizi-(ex82-105 hariç) (Prenatal)	Nemaline myopathy 2, autosomal recessive, 256030, Autosomal recessive; NEM2 (Severe congenital nemaline myopathy) (NEB gene) (Sequence Analysis-All Coding Exons (out of ex82-105)) (Prenatal)	NEB	NEB, NEM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nemalin Miyopati 2, Otozomal Resesif, 256030, Otozomal Resesif; NEM2 (Ağır Konjenital Nemalin Miyopati) (Prenatal) (NEB geni) (Tüm Gen Dizi Analizi (ex82-105 hariç) (Prenatal)	Nemaline myopathy 2, autosomal recessive, 256030, Autosomal recessive; NEM2 (Severe congenital nemaline myopathy) (NEB gene) (Sequence Analysis-All Coding Exons (out of ex82-105)) (Prenatal)	NEB	NEB, NEM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 3, Otozomal Dominant veya Resesif, 161800, Otozomal Resesif, Otozomal Dominant; NEM3 (Ağır Konjenital Nemalin Miyopati) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 3, autosomal dominant or recessive, 161800, Autosomal recessive, Autosomal dominant; NEM3 (Severe congenital nemaline myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. {ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nemalin Miyopati 3, Otozomal Dominant veya Resesif, 161800, Otozomal Resesif, Otozomal Dominant; NEM3 (Ağır Konjenital Nemalin Miyopati) (Prenatal) (ACTA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 3, autosomal dominant or recessive, 161800, Autosomal recessive, Autosomal dominant; NEM3 (Severe congenital nemaline myopathy) (ACTA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACTA1	ACTA1, ASMA, NEM3, CFTD1, SHPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 3 (NEM3) [MIM:161800]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. {ECO:0000269|PubMed:10508519, ECO:0000269|PubMed:11166164, ECO:0000269|PubMed:11333380, ECO:0000269|PubMed:15198992, ECO:0000269|PubMed:15236405, ECO:0000269|PubMed:15336687, ECO:0000269|PubMed:15520409, ECO:0000269|PubMed:16427282, ECO:0000269|PubMed:16945537}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 4, Otozomal Dominant, 609285, Otozomal Dominant; NEM4 (Tipik Nemalin Miyopati) (TPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 4, autosomal dominant, 609285, Autosomal dominant; NEM4 (Typical nemaline myopathy) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 1A (DA1A) [MIM:108120]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269|PubMed:12592607, ECO:0000269|PubMed:24692096}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nemalin Miyopati 4, Otozomal Dominant, 609285, Otozomal Dominant; NEM4 (Tipik Nemalin Miyopati) (Prenatal) (TPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 4, autosomal dominant, 609285, Autosomal dominant; NEM4 (Typical nemaline myopathy) (TPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPM2	TPM2, TMSB, AMCD1, DA1, DA2B, NEM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Arthrogryposis, distal, 1A (DA1A) [MIM:108120]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. Distal arthrogryposis type 1 is characterized largely by camptodactyly and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common. The shoulders and hips are less frequently affected. {ECO:0000269|PubMed:12592607, ECO:0000269|PubMed:24692096}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 5, Amish Tipi, 605355, Otozomal Resesif; NEM5 (Amish Nemaline Miyopati) (TNNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 5, Amish type, 605355, Autosomal recessive; NEM5 (Amish nemaline myopathy) (TNNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNNT1	TNNT1, ANM, NEM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 5 (NEM5) [MIM:605355]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. Nemaline myopathy type 5 is a severe and progressive form common among Old Order Amish. Affected infants display tremors with hypotonia and mild contractures of the shoulders and hips. Proximal contractures progressively weaken and a pectus carinatum deformity develops before children die of respiratory insufficiency, usually in the second year. {ECO:0000269|PubMed:10952871}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nemalin Miyopati 5, Amish Tipi, 605355, Otozomal Resesif; NEM5 (Amish Nemaline Miyopati) (Prenatal) (TNNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 5, Amish type, 605355, Autosomal recessive; NEM5 (Amish nemaline myopathy) (TNNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNNT1	TNNT1, ANM, NEM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 5 (NEM5) [MIM:605355]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. Nemaline myopathy type 5 is a severe and progressive form common among Old Order Amish. Affected infants display tremors with hypotonia and mild contractures of the shoulders and hips. Proximal contractures progressively weaken and a pectus carinatum deformity develops before children die of respiratory insufficiency, usually in the second year. {ECO:0000269|PubMed:10952871}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 6, Otozomal Dominant, 609273, Otozomal Dominant; NEM6 (Çocukluk Çağı Başlangıçlı Nemalin Miyopati) (KBTBD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 6, autosomal dominant, 609273, Autosomal dominant; NEM6 (Childhood-onset nemaline myopathy) (KBTBD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KBTBD13	KBTBD13, NEM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 6 (NEM6) [MIM:609273]: A form of nemaline myopathy characterized by childhood onset of slowly progressive proximal muscle weakness, exercise intolerance, and slow movements with stiff muscles. Patients are unable to run or correct themselves from falling over. {ECO:0000269|PubMed:21109227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nemalin Miyopati 6, Otozomal Dominant, 609273, Otozomal Dominant; NEM6 (Çocukluk Çağı Başlangıçlı Nemalin Miyopati) (Prenatal) (KBTBD13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 6, autosomal dominant, 609273, Autosomal dominant; NEM6 (Childhood-onset nemaline myopathy) (KBTBD13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KBTBD13	KBTBD13, NEM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 6 (NEM6) [MIM:609273]: A form of nemaline myopathy characterized by childhood onset of slowly progressive proximal muscle weakness, exercise intolerance, and slow movements with stiff muscles. Patients are unable to run or correct themselves from falling over. {ECO:0000269|PubMed:21109227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 7, Otozomal Resesif, 610687, Otozomal Resesif; NEM7 (Tipik Nemalin Miyopati) (CFL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 7, autosomal recessive, 610687, Autosomal recessive; NEM7 (Typical nemaline myopathy) (CFL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFL2	CFL2, NEM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 7 (NEM7) [MIM:610687]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. Nemaline myopathy type 7 presents at birth with hypotonia and generalized weakness. Major motor milestones are delayed, but independent ambulation is achieved. {ECO:0000269|PubMed:17160903}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nemalin Miyopati 7, Otozomal Resesif, 610687, Otozomal Resesif; NEM7 (Tipik Nemalin Miyopati) (Prenatal) (CFL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 7, autosomal recessive, 610687, Autosomal recessive; NEM7 (Typical nemaline myopathy) (CFL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFL2	CFL2, NEM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 7 (NEM7) [MIM:610687]: A form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. Nemaline myopathy type 7 presents at birth with hypotonia and generalized weakness. Major motor milestones are delayed, but independent ambulation is achieved. {ECO:0000269|PubMed:17160903}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 8, Otozomal Resesif, 615348; NEM8 (Ağır Konjenital Nemalin Miyopati) (KLHL40 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 8, autosomal recessive, 615348; NEM8 (Severe congenital nemaline myopathy) (KLHL40 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL40	KLHL40, SYRP, KBTBD5, NEM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nemalin Miyopati 8, Otozomal Resesif, 615348; NEM8 (Ağır Konjenital Nemalin Miyopati) (Prenatal) (KLHL40 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 8, autosomal recessive, 615348; NEM8 (Severe congenital nemaline myopathy) (KLHL40 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLHL40	KLHL40, SYRP, KBTBD5, NEM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nemalin Miyopati 9, 615731, Otozomal Resesif; NEM9 (Ağır Konjenital Nemalin Miyopati) (KLHL41 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nemaline myopathy 9, 615731, Autosomal recessive; NEM9 (Severe congenital nemaline myopathy) (KLHL41 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL41	KLHL41, KBTBD10, SARCOSIN, NEM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nemaline myopathy 9 (NEM9) [MIM:615731]: An autosomal recessive form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. NEM9 phenotype is highly variable, ranging from death in infancy due to lack of antigravity movements, to slowly progressive distal muscle weakness with preserved ambulation later in childhood. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Nemalin Miyopati 9, 615731, Otozomal Resesif; NEM9 (Ağır Konjenital Nemalin Miyopati) (Prenatal) (KLHL41 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nemaline myopathy 9, 615731, Autosomal recessive; NEM9 (Severe congenital nemaline myopathy) (KLHL41 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KLHL41	KLHL41, KBTBD10, SARCOSIN, NEM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nemaline myopathy 9 (NEM9) [MIM:615731]: An autosomal recessive form of nemaline myopathy. Nemaline myopathies are muscular disorders characterized by muscle weakness of varying severity and onset, and abnormal thread-like or rod-shaped structures in muscle fibers on histologic examination. NEM9 phenotype is highly variable, ranging from death in infancy due to lack of antigravity movements, to slowly progressive distal muscle weakness with preserved ambulation later in childhood. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrojenik Sendrom-Uygunsuz Antidiürezis, 300539, X'e Bağlı Resesif; NSIAD (Uygunsuz Antidiüreisse Bağlı Nefrojenik Sendrom) (AVPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrogenic syndrome of inappropriate antidiuresis, 300539, X-linked recessive; NSIAD (Nephrogenic syndrome of inappropriate antidiuresis) (AVPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AVPR2	AVPR2, DIR, DI1, ADHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes insipidus, nephrogenic, X-linked (XNDI) [MIM:304800]: A disorder caused by the inability of the renal collecting ducts to absorb water in response to arginine vasopressin. Characterized by excessive water drinking (polydipsia), excessive urine excretion (polyuria), persistent hypotonic urine, and hypokalemia. {ECO:0000269|PubMed:10694923, ECO:0000269|PubMed:10770218, ECO:0000269|PubMed:11232028, ECO:0000269|PubMed:11916004, ECO:0000269|PubMed:1303257, ECO:0000269|PubMed:1303271, ECO:0000269|PubMed:1356229, ECO:0000269|PubMed:16845277, ECO:0000269|PubMed:7560098, ECO:0000269|PubMed:7833930, ECO:0000269|PubMed:7984150, ECO:0000269|PubMed:7987330, ECO:0000269|PubMed:7999078, ECO:0000269|PubMed:8037205, ECO:0000269|PubMed:8045948, ECO:0000269|PubMed:8078903, ECO:0000269|PubMed:8267567, ECO:0000269|PubMed:8479490, ECO:0000269|PubMed:8514744, ECO:0000269|PubMed:9402087, ECO:0000269|PubMed:9452109, ECO:0000269|PubMed:9711877}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrolitiazis, Kalsiyum Oksalat, 167030, Otozomal Resesif; CACN (SLC26A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrolithiasis, calcium oxalate, 167030, Autosomal recessive; CACN (SLC26A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A1	SLC26A1, SAT1, CACN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nefrolitiyazis, Tip 1, 310468, X'e Bağlı; XRN (Dent Hastalığı) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrolithiasis, type I, 310468, X-linked recessive; XRN (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. {ECO:0000269|PubMed:15086899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8559248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrolitiazis, Ürik Asit, Yatkınlık, 605990 (ZNF365 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrolithiasis, uric acid, susceptibility to, 605990 (ZNF365 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF365	ZNF365, UAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Uric acid nephrolithiasis (UAN) [MIM:605990]: A form of nephrolithiasis, a common multifactorial disease characterized by stones formation in the kidney and urinary tract. Nephrolithiasis is due to supersaturation of the urine by stone-forming constituents, including calcium, oxalate and uric acid. Crystals or foreign bodies can act as nidi, upon which ions from the supersaturated urine form microscopic crystalline structures. Uric acid nephrolithiasis occurs when the urine becomes overly concentrated with uric acid and accounts for 20% of all stones. {ECO:0000269|PubMed:12740763}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrolitiazis / Osteoporoz, Hipofosfatemik, 1, 612286, Otozomal Dominant; NPHLOP1 (Dominant Hipofosfatemi-Nefrolitiazis ve Osteoporoz) (SLC34A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrolithiasis/osteoporosis, hypophosphatemic, 1, 612286, Autosomal dominant; NPHLOP1 (Dominant hypophosphatemia with nephrolithiasis or osteoporosis) (SLC34A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC34A1	SLC34A1, SLC17A2, NPT2, NPHLOP1, FRTS2, HCINF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fanconi renotubular syndrome 2 (FRTS2) [MIM:613388]: A disease due to a generalized dysfunction of the proximal kidney tubule resulting in decreased solute and water reabsorption. Patients have polydipsia and polyuria with phosphaturia, glycosuria and aminoaciduria. They may develop hypophosphatemic rickets or osteomalacia, acidosis and a tendency toward dehydration. Some eventually develop renal insufficiency. {ECO:0000269|PubMed:20335586}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrolitiazis / Osteoporoz, Hipofosfatemik, 2, 612287, Otozomal Dominant; NPHLOP2 (Dominant Hipofosfatemi-Nefrolitiazis ve Osteoporoz) (SLC9A3R1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrolithiasis/osteoporosis, hypophosphatemic, 2, 612287, Autosomal dominant; NPHLOP2 (Dominant hypophosphatemia with nephrolithiasis or osteoporosis) (SLC9A3R1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC9A3R1	SLC9A3R1, EBP50, NHERF1, NPHLOP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis/osteoporosis, hypophosphatemic, 2 (NPHLOP2) [MIM:612287]: A disease characterized by decreased renal phosphate absorption, renal phosphate wasting, hypophosphatemia, hyperphosphaturia, hypercalciuria, nephrolithiasis and osteoporosis. {ECO:0000269|PubMed:18784102, ECO:0000269|PubMed:27506049}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefronoftizis 1, Jüvenil, 256100, Otozomal Resesif; NPHP1 (Nefronoftizis) (NPHP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 1, juvenile, 256100, Autosomal recessive; NPHP1 (Nephronophthisis) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269|PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 1, Jüvenil, 256100, Otozomal Resesif; NPHP1 (Nefronoftizis) (MLPA)	Nephronophthisis 1, juvenile, 256100, Autosomal recessive; NPHP1 (Nephronophthisis) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269|PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 1, Jüvenil, 256100, Otozomal Resesif; NPHP1 (Nefronoftizis) (Prenatal) (NPHP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 1, juvenile, 256100, Autosomal recessive; NPHP1 (Nephronophthisis) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269|PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 1, Jüvenil, 256100, Otozomal Resesif; NPHP1 (Nefronoftizis) (Prenatal) (MLPA)	Nephronophthisis 1, juvenile, 256100, Autosomal recessive; NPHP1 (Nephronophthisis) (Prenatal) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269|PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 11, 613550, Otozomal Resesif; NPHP11 (Senior-Boichis Sendromu) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 11, 613550, Autosomal recessive; NPHP11 (Senior-Boichis syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:18327255}. Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	EDTA Blood Sample

Nefronoftizis 11, 613550. Otozomal Resesif; NPHP11 (Senior-Boichis Sendromu) (Prenatal) (TMEM67 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 11, 613550, Autosomal recessive; NPHP11 (Senior-Boichis syndrome) (TMEM67 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM67	TMEM67, MKS3, JBTS6, NPHP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:18327255). Note=The gene represented in this entry may act as a disease modifier. TMEM67 variations may influence the expression of Bardet-Biedl syndrome in patients who have causative mutations in other genes. Heterozygosity for a complex mutation in the TMEM67 gene coding for a protein with 2 in cis changes, and homozygosity for a truncating mutation of the CEP290 gene has been found in a patient with Bardet-Biedl syndrome 14.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 12, 613820, Otozomal Resesif, Otozomal Dominant; NPHP12 (Nefronoftizis) (TTC21B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 12, 613820, Autosomal recessive, Autosomal dominant; NPHP12 (Nephronophthisis) (TTC21B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC21B	TTC21B, THM1, NPHP12, SRTD4, ATD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:21258341). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 12, 613820, Otozomal Resesif, Otozomal Dominant; NPHP12 (Nefronoftizis) (Prenatal) (TTC21B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 12, 613820, Autosomal recessive, Autosomal dominant; NPHP12 (Nephronophthisis) (TTC21B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTC21B	TTC21B, THM1, NPHP12, SRTD4, ATD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:21258341). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 13, 614377, Otozomal Resesif; NPHP13 (Nefronoftizis) (WDR19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 13, 614377, Autosomal recessive; NPHP13 (Nephronophthisis) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269	PubMed:22019273). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 13, 614377, Otozomal Resesif; NPHP13 (Nefronoftizis) (Prenatal) (WDR19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 13, 614377, Autosomal recessive; NPHP13 (Nephronophthisis) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269	PubMed:22019273). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 14, 614844, Otozomal Resesif, Otozomal Dominant (ZNF423 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 14, 614844, Autosomal recessive, Autosomal dominant (ZNF423 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF423	ZNF423, ZFP423, OAZ, KIAA0760, NPHP14, JBTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 14 (NPHP14) [MIM:614844]: An autosomal recessive disorder manifesting as infantile-onset kidney disease, cerebellar vermis hypoplasia, and situs inversus. Nephronophthisis is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. (ECO:0000269	PubMed:22863007). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 14, 614844, Otozomal Resesif, Otozomal Dominant (Prenatal) (ZNF423 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 14, 614844, Autosomal recessive, Autosomal dominant (ZNF423 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNF423	ZNF423, ZFP423, OAZ, KIAA0760, NPHP14, JBTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 14 (NPHP14) [MIM:614844]: An autosomal recessive disorder manifesting as infantile-onset kidney disease, cerebellar vermis hypoplasia, and situs inversus. Nephronophthisis is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. (ECO:0000269	PubMed:22863007). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 15, 614845, Otozomal Resesif (Senior-Loken Sendromu) (CEP164 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 15, 614845, Autosomal recessive; NPHP15 (Senior-Loken syndrome) (CEP164 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP164	CEP164, KIAA1052	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 15 (NPHP15) [MIM:614845]: An autosomal recessive disorder characterized by the association of nephronophthisis with Leber congenital amaurosis and retinal degeneration, often resulting in blindness during childhood. Additional features include seizures, cerebellar vermis hypoplasia, facial dysmorphism, bronchiectasis and liver failure. Nephronophthisis is a chronic tubulo-interstitial nephritis that progresses to end-stage renal failure. (ECO:0000269	PubMed:22863007). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Nefronoftizis 15, 614845, Otozomal Resesif (Senior-Loken Sendromu) (Prenatal) (CEP164 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 15, 614845, Autosomal recessive; NPHP15 (Senior-Loken syndrome) (CEP164 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP164	CEP164, KIAA1052	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 15 (NPHP15) [MIM:614845]: An autosomal recessive disorder characterized by the association of nephronophthisis with Leber congenital amaurosis and retinal degeneration, often resulting in blindness during childhood. Additional features include seizures, cerebellar vermis hypoplasia, facial dysmorphism, bronchiectasis and liver failure. Nephronophthisis is a chronic tubulo-interstitial nephritis that progresses to end-stage renal failure. (ECO:0000269	PubMed:22863007). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 16, 615382, Otozomal Resesif; NPHP16 (Nefronoftizis) (ANKS6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 16, 615382, Autosomal recessive; NPHP16 (Nephronophthisis) (ANKS6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKS6	ANKS6, PKDR1, NPHP16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 16 (NPHP16) [MIM:615382]: A form of nephronophthisis, a chronic tubulo-interstitial nephritis that progresses to end-stage renal failure. Some patients have cystic kidneys of normal size and no extrarenal manifestations, whereas others have enlarged renal size and severe extrarenal defects, including hypertrophic obstructive cardiomyopathy, aortic stenosis, pulmonary stenosis, patent ductus arteriosus, situs inversus, and periportal liver fibrosis. (ECO:0000269	PubMed:23792029). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Nefronoftizis 16, 615382, Otozomal Resesif; NPHP16 (Nefronoftizis) (Prenatal) (ANKS6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 16, 615382, Autosomal recessive; NPHP16 (Nephronophthisis) (ANKS6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANKS6	ANKS6, PKDR1, NPHP16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 16 (NPHP16) [MIM:615382]: A form of nephronophthisis, a chronic tubulo-interstitial nephritis that progresses to end-stage renal failure. Some patients have cystic kidneys of normal size and no extrarenal manifestations, whereas others have enlarged renal size and severe extrarenal defects, including hypertrophic obstructive cardiomyopathy, aortic stenosis, pulmonary stenosis, patent ductus arteriosus, situs inversus, and periportal liver fibrosis. (ECO:0000269	PubMed:23792029). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 18, 615862, Otozomal Resesif; NPHP18 (Nefronoftizis) (CEP83 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 18, 615862, Autosomal recessive; NPHP18 (Nephronophthisis) (CEP83 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP83	CEP83, CCDC41, NPHP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 18 (NPHP18) [MIM:615862]: An autosomal recessive disorder characterized by chronic tubulointerstitial nephritis resulting in end-stage renal disease in early childhood. Extrarenal manifestations, including intellectual disability or liver changes, may occur in some patients. (ECO:0000269	PubMed:24882706). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Nefronoftizis 18, 615862, Otozomal Resesif; NPHP18 (Nefronoftizis) (Prenatal) (CEP83 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 18, 615862, Autosomal recessive; NPHP18 (Nephronophthisis) (CEP83 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP83	CEP83, CCDC41, NPHP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 18 (NPHP18) [MIM:615862]: An autosomal recessive disorder characterized by chronic tubulointerstitial nephritis resulting in end-stage renal disease in early childhood. Extrarenal manifestations, including intellectual disability or liver changes, may occur in some patients. (ECO:0000269	PubMed:24882706). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 19, 616217, Otozomal Resesif; NPHP19 (Senior-Boichis Sendromu) (DCDC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 19, 616217, Autosomal recessive; NPHP19 (Senior-Boichis syndrome) (DCDC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCDC2	DCDC2, RU2, KIAA1154, NPHP19, DFNB66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 66 (DFNB66) [MIM:610212]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. (ECO:0000269	PubMed:25601850). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Nefronoftizis 19, 616217, Otozomal Resesif; NPHP19 (Senior-Boichis Sendromu) (Prenatal) (DCDC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 19, 616217, Autosomal recessive; NPHP19 (Senior-Boichis syndrome) (DCDC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCDC2	DCDC2, RU2, KIAA1154, NPHP19, DFNB66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Deafness, autosomal recessive, 66 (DFNB66) [MIM:610212]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. (ECO:0000269	PubMed:25601850). Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nefronoftizis 2, İnfantil, 602088, Otozomal Resesif; NPHP2 (Nefronoftizis) (INVS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 2, infantile, 602088, Autosomal recessive; NPHP2 (Nephronophthisis) (INVS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INVS	INVS, INV, NPHP2, NPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 2 (NPHP2) [MIM:602088]: An autosomal recessive disorder resulting in end-stage renal disease. It is characterized by early onset and rapid progression. Phenotypic manifestations include enlarged kidneys, chronic tubulo- interstitial nephritis, anemia, hyperkalemic metabolic acidosis. Some patients also display situs inversus. Pathologically, it differs from later-onset nephronophthisis by the absence of medullary cysts and thickened tubular basement membranes, and by the presence of cortical microcysts. [ECO:0000269	PubMed:12872123]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefronoftizis 2, İnfantil, 602088, Otozomal Resesif; NPHP2 (Nefronoftizis) (Prenatal) (INVS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 2, infantile, 602088, Autosomal recessive; NPHP2 (Nephronophthisis) (INVS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INVS	INVS, INV, NPHP2, NPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 2 (NPHP2) [MIM:602088]: An autosomal recessive disorder resulting in end-stage renal disease. It is characterized by early onset and rapid progression. Phenotypic manifestations include enlarged kidneys, chronic tubulo- interstitial nephritis, anemia, hyperkalemic metabolic acidosis. Some patients also display situs inversus. Pathologically, it differs from later-onset nephronophthisis by the absence of medullary cysts and thickened tubular basement membranes, and by the presence of cortical microcysts. [ECO:0000269	PubMed:12872123]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 20, 617271, Otozomal Resesif; NPHP20 (MAPKBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 20, 617271, Autosomal recessive; NPHP20 (MAPKBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPKBP1	MAPKBP1, JNKBP1, NPHP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nefronoftizis 20, 617271, Otozomal Resesif; NPHP20 (Prenatal) (MAPKBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 20, 617271, Autosomal recessive; NPHP20 (MAPKBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAPKBP1	MAPKBP1, JNKBP1, NPHP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 3, 604387, Otozomal Resesif; NPHP3 (Nefronoftizis) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 3, 604387, Autosomal recessive; NPHP3 (Nephronophthisis) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:18371931]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefronoftizis 3, 604387, Otozomal Resesif; NPHP3 (Nefronoftizis) (Prenatal) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 3, 604387, Autosomal recessive; NPHP3 (Nephronophthisis) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:18371931]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 4, 606966, Otozomal Resesif; NPHP4 (Nefronoftizis) (NPHP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 4, 606966, Autosomal recessive; NPHP4 (Nephronophthisis) (NPHP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP4	NPHP4, SLSN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Senior-Loken syndrome 4 (SLSN4) [MIM:606996]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. [ECO:0000269|PubMed:12244321, ECO:0000269|PubMed:15776426]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefronoftizis 4, 606966, Otozomal Resesif; NPHP4 (Nefronoftizis) (Prenatal) (NPHP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 4, 606966, Autosomal recessive; NPHP4 (Nephronophthisis) (NPHP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP4	NPHP4, SLSN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Senior-Loken syndrome 4 (SLSN4) [MIM:606996]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. [ECO:0000269|PubMed:12244321, ECO:0000269|PubMed:15776426]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 7, 611498; NPHP7 (Nefronoftizis) (GLIS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 7, 611498; NPHP7 (Nephronophthisis) (GLIS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLIS2	GLIS2, NPHP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 7 (NPHP7) [MIM:611498]: An autosomal recessive disorder resulting in end-stage renal disease during childhood or adolescence. It is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. [ECO:0000269	PubMed:17618285]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefronoftizis 7, 611498; NPHP7 (Nefronoftizis) (Prenatal) (GLIS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 7, 611498; NPHP7 (Nephronophthisis) (GLIS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLIS2	GLIS2, NPHP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 7 (NPHP7) [MIM:611498]: An autosomal recessive disorder resulting in end-stage renal disease during childhood or adolescence. It is a progressive tubulo-interstitial kidney disorder histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. [ECO:0000269	PubMed:17618285]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis 9, 613824; NPHP9 (Nefronoftizis) (NEK8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis 9, 613824; NPHP9 (Nephronophthisis) (NEK8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK8	NEK8, JCK, NPHP9, RHPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal-hepatic-pancreatic dysplasia 2 (RHPD2) [MIM:615415]: A form of renal-hepatic-pancreatic dysplasia, a disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:23418306]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefronoftizis 9, 613824; NPHP9 (Nefronoftizis) (Prenatal) (NEK8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis 9, 613824; NPHP9 (Nephronophthisis) (NEK8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEK8	NEK8, JCK, NPHP9, RHPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Renal-hepatic-pancreatic dysplasia 2 (RHPD2) [MIM:615415]: A form of renal-hepatic-pancreatic dysplasia, a disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. [ECO:0000269	PubMed:23418306]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefronoftizis Benzeri Nefropati 1, 613159, Otozomal Resesif; NPHPL1 (Nefronoftizis) (XPNPEP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephronophthisis-like nephropathy 1, 613159, Autosomal recessive; NPHPL1 (Nephronophthisis) (XPNPEP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XPNPEP3	XPNPEP3, APP3, NPHPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nefronoftizis Benzeri Nefropati 1, 613159, Otozomal Resesif; NPHPL1 (Nefronoftizis) (Prenatal) (XPNPEP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephronophthisis-like nephropathy 1, 613159, Autosomal recessive; NPHPL1 (Nephronophthisis) (XPNPEP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XPNPEP3	XPNPEP3, APP3, NPHPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefropati CFHR5 Eksikliğine Bağlı, 614809, Otozomal Dominant Nedeniyle (C3-Glomerulonefrit) (CFHR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephropathy due to CFHR5 deficiency, 614809, Autosomal dominant (C3 glomerulonephritis) (CFHR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFHR5	CFHR5, CFHL5, FHR5, CFHR5D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: CFHR5 deficiency (CFHR5D) [MIM:614809]: A progressive disease characterized by glomerulonephritis, hematuria, renal failure, end-stage renal disease, subendothelial and mesangial glomerular C3 deposits, mesangial matrix expansion, increased glomerular cellularity, and segmental capillary wall thickening. Hematuria may become apparent after respiratory infections. [ECO:0000269|PubMed:20800271, ECO:0000269|PubMed:22503529]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefropati CFHR5 Eksikliğine Bağlı, 614809, Otozomal Dominant Nedeniyle (C3-Glomerulonefrit) (Prenatal) (CFHR5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephropathy due to CFHR5 deficiency, 614809, Autosomal dominant (C3 glomerulonephritis) (CFHR5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFHR5	CFHR5, CFHL5, FHR5, CFHR5D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: CFHR5 deficiency (CFHR5D) [MIM:614809]: A progressive disease characterized by glomerulonephritis, hematuria, renal failure, end-stage renal disease, subendothelial and mesangial glomerular C3 deposits, mesangial matrix expansion, increased glomerular cellularity, and segmental capillary wall thickening. Hematuria may become apparent after respiratory infections. [ECO:0000269|PubMed:20800271, ECO:0000269|PubMed:22503529]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pretibial Epidermolizis Bullosa, Nefropati ve Sağırlık, 609057 (Nefrotik Sendrom-Sağırlık-Pretibiyal Epidermolizis Bülloza Sendromu) (CD151 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome) (CD151 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD151	CD151, PETA3, SFA1, MER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephropathy with pretibial epidermolysis bullosa and deafness (NPEBD) [MIM:609057]: A disorder characterized by the association of hereditary nephritis, epidermolysis bullosa, deafness, and beta-thalassemia minor. [ECO:0000269	PubMed:15265795]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pretibial Epidermolizis Bullosa, Nefropati ve Sağırlık, 609057 (Nefrotik Sendrom-Sağırlık-Pretibiyal Epidermolizis Bülloza Sendromu) (Prenatal) (CD151 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (Nephrotic syndrome-deafness-pretibial epidermolysis bullosa syndrome) (CD151 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD151	CD151, PETA3, SFA1, MER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephropathy with pretibial epidermolysis bullosa and deafness (NPEBD) [MIM:609057]: A disorder characterized by the association of hereditary nephritis, epidermolysis bullosa, deafness, and beta-thalassemia minor. [ECO:0000269	PubMed:15265795]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 1, 256300; NPHS1 (Konjenital Nefrotik Sendrom, Fin Tipi) (NPHS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 1, 256300, Autosomal recessive; NPHS1 (Congenital nephrotic syndrome, Finnish type) (NPHS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHS1	NPHS1, NPHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 1 (NPHS1) [MIM:256300]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. ECO:0000269|PubMed:10652016, ECO:0000269|PubMed:11317351, ECO:0000269|PubMed:17290294, ECO:0000269|PubMed:18503012, ECO:0000269|PubMed:18614772, ECO:0000269|PubMed:20172850, ECO:0000269|PubMed:22009864, ECO:0000269|PubMed:22565185, ECO:0000269|PubMed:9660941, ECO:0000269|PubMed:9915943]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Nefrotik Sendrom, Otozomal Resesif Tip 1, 256300; NPHS1 (Konjenital Nefrotik Sendrom, Fin Tipi) (Prenatal) (NPHS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 1, 256300, Autosomal recessive; NPHS1 (Congenital nephrotic syndrome, Finnish type) (NPHS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHS1	NPHS1, NPHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 1 (NPHS1) [MIM:256300]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. {ECO:0000269|PubMed:10652016, ECO:0000269|PubMed:11317351, ECO:0000269|PubMed:17290394, ECO:0000269|PubMed:18503012, ECO:0000269|PubMed:18614772, ECO:0000269|PubMed:20172850, ECO:0000269|PubMed:22009864, ECO:0000269|PubMed:22565185, ECO:0000269|PubMed:9660941, ECO:0000269|PubMed:9915943}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 10, 615861; NPHS10 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (EMP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 10, 615861, Autosomal recessive; NPHS10 (Familial idiopathic steroid-resistant nephrotic syndrome) (EMP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EMP2	EMP2, NPHS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 10 (NPHS10) [MIM:615861]: A form of nephrotic syndrome, a renal disease clinically characterized by focal segmental glomerulosclerosis, progressive renal failure, severe proteinuria, hypoalbuminemia, hyperlipidemia and edema. NPHS10 is a steroid-sensitive form characterized by onset in childhood and remission without end-stage kidney disease. {ECO:0000269|PubMed:24814193}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 10, 615861; NPHS10 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (EMP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 10, 615861, Autosomal recessive; NPHS10 (Familial idiopathic steroid-resistant nephrotic syndrome) (EMP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EMP2	EMP2, NPHS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 10 (NPHS10) [MIM:615861]: A form of nephrotic syndrome, a renal disease clinically characterized by focal segmental glomerulosclerosis, progressive renal failure, severe proteinuria, hypoalbuminemia, hyperlipidemia and edema. NPHS10 is a steroid-sensitive form characterized by onset in childhood and remission without end-stage kidney disease. {ECO:0000269|PubMed:24814193}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 11, 616730; NPHS11 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (NUP107 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 11, 616730, Autosomal recessive; NPHS11 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP107 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP107	NUP107, NUP84, NPHS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 11, 616730; NPHS11 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (NUP107 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 11, 616730, Autosomal recessive; NPHS11 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP107 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUP107	NUP107, NUP84, NPHS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 12, 616892; NPHS12 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (NUP93 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 12, 616892, Autosomal recessive; NPHS12 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP93 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP93	NUP93, NIC96, KIAA0095, NPHS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 12, 616892; NPHS12 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (NUP93 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 12, 616892, Autosomal recessive; NPHS12 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP93 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUP93	NUP93, NIC96, KIAA0095, NPHS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Tip 13, 616893; NPHS13 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (NUP205 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 13, 616893; NPHS13 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP205 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP205	NUP205, C7orf14, KIAA0225, NPHS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nefrotik Sendrom, Tip 13, 616893; NPHS13 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (NUP205 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 13, 616893; NPHS13 (Familial idiopathic steroid-resistant nephrotic syndrome) (NUP205 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUP205	NUP205, C7orf14, KIAA0225, NPHS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 2, 600995; NPHS2 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (NPHS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 2, 600995, Autosomal recessive; NPHS2 (Familial idiopathic steroid-resistant nephrotic syndrome) (NPHS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHS2	PDCN, NPHS2, SRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 2 (NPHS2) [MIM:600995]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. The disorder is resistant to steroid treatment and progresses to end-stage renal failure in the first or second decades. Some patients show later onset of the disorder. {ECO:0000269|PubMed:10742096, ECO:0000269|PubMed:15253708, ECO:0000269|PubMed:17899208, ECO:0000269|PubMed:20947785, ECO:0000269|PubMed:22565185, ECO:0000269|PubMed:22578956, ECO:0000269|PubMed:23242530, ECO:0000269|PubMed:23913389, ECO:0000269|PubMed:24072147, ECO:0000269|PubMed:24227627}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 2, 600995; NPHS2 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (NPHS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 2, 600995, Autosomal recessive; NPHS2 (Familial idiopathic steroid-resistant nephrotic syndrome) (NPHS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHS2	PDCN, NPHS2, SRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 2 (NPHS2) [MIM:600995]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. The disorder is resistant to steroid treatment and progresses to end-stage renal failure in the first or second decades. Some patients show later onset of the disorder. {ECO:0000269|PubMed:10742096, ECO:0000269|PubMed:15253708, ECO:0000269|PubMed:17899208, ECO:0000269|PubMed:20947785, ECO:0000269|PubMed:22565185, ECO:0000269|PubMed:22578956, ECO:0000269|PubMed:23242530, ECO:0000269|PubMed:23913389, ECO:0000269|PubMed:24072147, ECO:0000269|PubMed:24227627}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 3, 610725; NPHS3 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (PLCE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 3, 610725, Autosomal recessive; NPHS3 (Familial idiopathic steroid-resistant nephrotic syndrome) (PLCE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCE1	PLCE1, KIAA1516, NPHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 3 (NPHS3) [MIM:610725]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. Most patients with NPHS3 show diffuse mesangial sclerosis on renal biopsy, which is a pathologic entity characterized by mesangial matrix expansion with no mesangial hypercellularity, hypertrophy of the podocytes, vacuolized podocytes, thickened basement membranes, and diminished patency of the capillary lumen. {ECO:0000269|PubMed:17086182}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 3, 610725; NPHS3 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (PLCE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 3, 610725, Autosomal recessive; NPHS3 (Familial idiopathic steroid-resistant nephrotic syndrome) (PLCE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLCE1	PLCE1, KIAA1516, NPHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 3 (NPHS3) [MIM:610725]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. Most patients with NPHS3 show diffuse mesangial sclerosis on renal biopsy, which is a pathologic entity characterized by mesangial matrix expansion with no mesangial hypercellularity, hypertrophy of the podocytes, vacuolized podocytes, thickened basement membranes, and diminished patency of the capillary lumen. {ECO:0000269|PubMed:17086182}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Dominant Tip 4, 256370; NPHS4 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (WT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 4, 256370, Autosomal dominant; NPHS4 (Familial idiopathic steroid-resistant nephrotic syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269|PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Dominant Tip 4, 256370; NPHS4 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (MLPA)	Nephrotic syndrome, type 4, 256370, Autosomal dominant; NPHS4 (Familial idiopathic steroid-resistant nephrotic syndrome) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269|PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Dominant Tip 4, 256370; NPHS4 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (WT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 4, 256370, Autosomal dominant; NPHS4 (Familial idiopathic steroid-resistant nephrotic syndrome) (WT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269|PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nefrotik Sendrom, Otozomal Dominant Tip 4, 256370; NPHS4 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (MLPA)	Nephrotic syndrome, type 4, 256370, Autosomal dominant; NPHS4 (Familial idiopathic steroid-resistant nephrotic syndrome) (Prenatal) (MLPA)	WT1	WT1, NPHS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Frasier syndrome (FS) [MIM:136680]: Characterized by a slowly progressing nephropathy leading to renal failure in adolescence or early adulthood, male pseudohermaphroditism, and no Wilms tumor. As for histological findings of the kidneys, focal glomerular sclerosis is often observed. There is phenotypic overlap with Denys-Drash syndrome. Inheritance is autosomal dominant. {ECO:0000269|PubMed:10571943}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Tip 5, ± Oküler Anomaliler, 614199; NPHS5 (LAMB2 İlişkili İnfantil Başlangıçlı Nefrotik Sendrom) (LAMB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199; NPHS5 (LAMB2-related infantile-onset nephrotic syndrome) (LAMB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB2	LAMB2, LAMS, NPHS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pierson syndrome (PIERS5) [MIM:609049]: Characterized by nephrotic syndrome with neonatal onset, diffuse mesangial sclerosis and eye abnormalities with microcoria as the leading clinical feature. Death usually occurs within the first weeks of life. Disease severity depends on the mutation type: nontruncating LAMB2 mutations may display variable phenotypes ranging from a milder variant of Pierson syndrome to isolated congenital nephrotic syndrome. {ECO:0000269|PubMed:15367484, ECO:0000269|PubMed:16912710}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrotik Sendrom, Tip 5, ± Oküler Anomaliler, 614199; NPHS5 (LAMB2 İlişkili İnfantil Başlangıçlı Nefrotik Sendrom) (Prenatal) (LAMB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 5, with or without ocular abnormalities, 614199; NPHS5 (LAMB2-related infantile-onset nephrotic syndrome) (LAMB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMB2	LAMB2, LAMS, NPHS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pierson syndrome (PIERS5) [MIM:609049]: Characterized by nephrotic syndrome with neonatal onset, diffuse mesangial sclerosis and eye abnormalities with microcoria as the leading clinical feature. Death usually occurs within the first weeks of life. Disease severity depends on the mutation type: nontruncating LAMB2 mutations may display variable phenotypes ranging from a milder variant of Pierson syndrome to isolated congenital nephrotic syndrome. {ECO:0000269|PubMed:15367484, ECO:0000269|PubMed:16912710}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Tip 6, 614196, Otozomal Resesif; NPHS6 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (PTPRO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 6, 614196, Autosomal recessive; NPHS6 (Familial idiopathic steroid-resistant nephrotic syndrome) (PTPRO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRO	PTPRO, GLEPP1, NPHS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 6 (NPHS6) [MIM:614196]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. {ECO:0000269|PubMed:21722858}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrotik Sendrom, Tip 6, 614196, Otozomal Resesif; NPHS6 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (PTPRO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 6, 614196, Autosomal recessive; NPHS6 (Familial idiopathic steroid-resistant nephrotic syndrome) (PTPRO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPRO	PTPRO, GLEPP1, NPHS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 6 (NPHS6) [MIM:614196]: A form of nephrotic syndrome, a renal disease clinically characterized by severe proteinuria, resulting in complications such as hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show non-specific histologic changes such as focal segmental glomerulosclerosis and diffuse mesangial proliferation. Some affected individuals have an inherited steroid-resistant form and progress to end-stage renal failure. {ECO:0000269|PubMed:21722858}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Tip 7, 615008; NPHS7 (Primer Membranoproliferatif Glomerulonefrit) (DGKE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 7, 615008, Autosomal recessive; NPHS7 (Primary membranoproliferative glomerulonephritis) (DGKE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGKE	DGKE, NPHS7, AHUS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 7 (AHUS7) [MIM:615008]: An atypical form of hemolytic uremic syndrome characterized by acute onset in the first year of life of microangiopathic hemolytic anemia, thrombocytopenia, and renal failure. After the acute episode, most patients develop chronic renal insufficiency. Unlike other genetic forms of aHUS, AHUS7 is not related to abnormal activation of the complement system. {ECO:0000269|PubMed:23542698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 7, 615008; NPHS7 (Primer Membranoproliferatif Glomerulonefrit) (Prenatal) (DGKE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 7, 615008, Autosomal recessive; NPHS7 (Primary membranoproliferative glomerulonephritis) (DGKE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGKE	DGKE, NPHS7, AHUS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hemolytic uremic syndrome atypical 7 (AHUS7) [MIM:615008]: An atypical form of hemolytic uremic syndrome characterized by acute onset in the first year of life of microangiopathic hemolytic anemia, thrombocytopenia, and renal failure. After the acute episode, most patients develop chronic renal insufficiency. Unlike other genetic forms of aHUS, AHUS7 is not related to abnormal activation of the complement system. {ECO:0000269|PubMed:23542698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 8, 615244; NPHS8 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (ARHGDIA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 8, 615244, Autosomal recessive; NPHS8 (Familial idiopathic steroid-resistant nephrotic syndrome) (ARHGDIA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGDIA	ARHGDIA, GDIA1, NPHS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrotic syndrome 8 (NPHS8) [MIM:615244]: A form of nephrotic syndrome, a renal disease clinically characterized by progressive renal failure, severe proteinuria, hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show diffuse mesangial sclerosis, with small glomeruli, hypercellularity, increased extracellular matrix, and contracted/collapsed glomerular tufts surrounded by immature or abnormal podocytes. {ECO:0000269|PubMed:23434736}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 8, 615244; NPHS8 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (ARHGDIA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 8, 615244, Autosomal recessive; NPHS8 (Familial idiopathic steroid-resistant nephrotic syndrome) (ARHGDIA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARHGDIA	ARHGDIA, GDIA1, NPHS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephrotic syndrome 8 (NPHS8) [MIM:615244]: A form of nephrotic syndrome, a renal disease clinically characterized by progressive renal failure, severe proteinuria, hypoalbuminemia, hyperlipidemia and edema. Kidney biopsies show diffuse mesangial sclerosis, with small glomeruli, hypercellularity, increased extracellular matrix, and contracted/collapsed glomerular tufts surrounded by immature or abnormal podocytes. {ECO:0000269|PubMed:23434736}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 9, 615573; NPHS9 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (COQ8B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nephrotic syndrome, type 9, 615573, Autosomal recessive; NPHS9 (Familial idiopathic steroid-resistant nephrotic syndrome) (COQ8B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COQ8B	COQ8B, ADCK4, NPHS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nefrotik Sendrom, Otozomal Resesif Tip 9, 615573; NPHS9 (Ailesel İdyopatik Steroide Dirençli Nefrotik Sendrom) (Prenatal) (COQ8B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nephrotic syndrome, type 9, 615573, Autosomal recessive; NPHS9 (Familial idiopathic steroid-resistant nephrotic syndrome) (COQ8B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COQ8B	COQ8B, ADCK4, NPHS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nestor-Guillermo Progeria Sendromu, 614008, Otozomal Resesif; NGPS (Nestor-Guillermo Progeria Sendromu) (BANF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nestor-Guillermo progeria syndrome, 614008, Autosomal recessive; NGPS (Nestor-Guillermo progeria syndrome) (BANF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BANF1	BANF1, BAF, NGPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nestor-Guillermo progeria syndrome (NGPS) [MIM:614008]: An atypical progeroid syndrome characterized by normal development in the first years of life, later followed by the emergence of generalized lipoatrophy, severe osteoporosis, and marked osteolysis. The atrophic facial subcutaneous fat pad and the marked osteolysis of the maxilla and mandible result in a typical pseudosenile facial appearance with micrognathia, prominent subcutaneous venous patterning, a convex nasal ridge, and proptosis. Cognitive development is completely normal. Patients do not have cardiovascular dysfunction, atherosclerosis, or metabolic anomalies. {ECO:0000269|PubMed:21549337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nestor-Guillermo Progeria Sendromu, 614008, Otozomal Resesif; NGPS (Nestor-Guillermo Progeria Sendromu) (Prenatal) (BANF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nestor-Guillermo progeria syndrome, 614008, Autosomal recessive; NGPS (Nestor-Guillermo progeria syndrome) (BANF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BANF1	BANF1, BAF, NGPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nestor-Guillermo progeria syndrome (NGPS) [MIM:614008]: An atypical progeroid syndrome characterized by normal development in the first years of life, later followed by the emergence of generalized lipoatrophy, severe osteoporosis, and marked osteolysis. The atrophic facial subcutaneous fat pad and the marked osteolysis of the maxilla and mandible result in a typical pseudosenile facial appearance with micrognathia, prominent subcutaneous venous patterning, a convex nasal ridge, and proptosis. Cognitive development is completely normal. Patients do not have cardiovascular dysfunction, atherosclerosis, or metabolic anomalies. {ECO:0000269|PubMed:21549337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Netherton Sendromu, 256500, Otozomal Resesif; NETH (Netherton Sendromu) (SPINK5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Netherton syndrome, 256500, Autosomal recessive; NETH (Netherton syndrome) (SPINK5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINK5	SPINK5, LEKTI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Netherton syndrome (NETH) [MIM:256500]: An autosomal recessive congenital ichthyosis associated with hair shaft abnormalities and anomalies of the immune system. Typical features are ichthyosis linearis circumflexa, ichthyosiform erythroderma, trichorrhexis invaginata (bamboo hair), atopic dermatitis, and hayfever. High postnatal mortality is due to failure to thrive, infections and hypernatremic dehydration. {ECO:0000269|PubMed:10835624}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Netherton Sendromu, 256500, Otozomal Resesif; NETH (Netherton Sendromu) (Prenatal) (SPINK5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Netherton syndrome, 256500, Autosomal recessive; NETH (Netherton syndrome) (SPINK5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPINK5	SPINK5, LEKTI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Netherton syndrome (NETH) [MIM:256500]: An autosomal recessive congenital ichthyosis associated with hair shaft abnormalities and anomalies of the immune system. Typical features are ichthyosis linearis circumflexa, ichthyosiform erythroderma, trichorrhexis invaginata (bamboo hair), atopic dermatitis, and hayfever. High postnatal mortality is due to failure to thrive, infections and hypernatremic dehydration. {ECO:0000269|PubMed:10835624}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Neu-Laxova Sendromu 1, 256520, Otozomal Resesif; NLS1 (Neu-Laxova Sendromu) (PHGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neu-Laxova syndrome 1, 256520, Autosomal recessive; NLS1 (Neu-Laxova syndrome) (PHGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHGDH	PHGDH, NLS1, PHGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neu-Laxova syndrome 1 (NLS1) [MIM:256520]: A lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies including lissencephaly, cerebellar hypoplasia and/or abnormalgenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269|PubMed:24636451}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Neu-Laxova Sendromu 1, 256520, Otozomal Resesif; NLS1 (Neu-Laxova Sendromu) (Prenatal) (PHGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neu-Laxova syndrome 1, 256520, Autosomal recessive; NLS1 (Neu-Laxova syndrome) (PHGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHGDH	PHGDH, NLS1, PHGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neu-Laxova syndrome 1 (NLS1) [MIM:256520]: A lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies including lissencephaly, cerebellar hypoplasia and/or abnormalgenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269|PubMed:24636451}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Neu-Laxova Sendromu 2, 616038, Otozomal Resesif; NLS2 (Neu-Laxova Sendromu) (PSAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neu-Laxova syndrome 2, 616038, Autosomal recessive; NLS2 (Neu-Laxova syndrome) (PSAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAT1	PSAT1, PSAT, EPIP, PSATD, NLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neu-Laxova syndrome 2 (NLS2) [MIM:616038]: A form of Neu- Laxova syndrome, a lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies. These include lissencephaly, cerebellar hypoplasia and/or abnormalagenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269	PubMed:25152457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Neu-Laxova Sendromu 2, 616038, Otozomal Resesif; NLS2 (Neu-Laxova Sendromu) (Prenatal) (PSAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neu-Laxova syndrome 2, 616038, Autosomal recessive; NLS2 (Neu-Laxova syndrome) (PSAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAT1	PSAT1, PSAT, EPIP, PSATD, NLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neu-Laxova syndrome 2 (NLS2) [MIM:616038]: A form of Neu- Laxova syndrome, a lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies. These include lissencephaly, cerebellar hypoplasia and/or abnormalagenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269	PubMed:25152457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöral Tüp Defekti, 182940, Otozomal Dominant (Nöral Tüp Kapanma Defekti) (VANGL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, 182940, Autosomal dominant (Neural tube closure defect) (VANGL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VANGL2	VANGL2, LTAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neural tube defects (NTD) [MIM:182940]: Congenital malformations of the central nervous system and adjacent structures related to defective neural tube closure during the first trimester of pregnancy. Failure of neural tube closure can occur at any level of the embryonic axis. Common NTD forms include anencephaly, myelomeningocele and spina bifida, which result from the failure of fusion in the cranial and spinal region of the neural tube. NTDs have a multifactorial etiology encompassing both genetic and environmental components. {ECO:0000269	PubMed:20558380}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöral Tüp Defekti, 182940, Otozomal Dominant (Nöral Tüp Kapanma Defekti) (FUZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, 182940, Autosomal dominant (Neural tube closure defect) (FUZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUZ	FUZ, NTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöral Tüp Defekti, Folat Duyarlı, Yatkınlık, 601634, Otozomal Resesif (Nöral Tüp Kapanma Defekti) (MTRR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, folate-sensitive, susceptibility to, 601634, Autosomal recessive (Neural tube closure defect) (MTRR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTRR	MTRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neural tube defects, folate-sensitive (NTDFS) [MIM:601634]: The most common NTDs are open spina bifida (myelomeningocele) and anencephaly. {ECO:0000269|PubMed:10444342, ECO:0000269|PubMed:12375236}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Nöral Tüp Defekti, Folat Duyarlı, Yatkınlık, 601634, Otozomal Resesif (Nöral Tüp Kapanma Defekti) (MTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, folate-sensitive, susceptibility to, 601634, Autosomal recessive (Neural tube closure defect) (MTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTR	MTR, HMAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Homocystinuria-megaloblastic anemia, cblG complementation type (HMAG) [MIM:250940]: An autosomal recessive inborn error of metabolism resulting from defects in the cobalamin-dependent pathway that converts homocysteine to methionine. It causes delayed psychomotor development, megaloblastic anemia, homocystinuria, and hypomethioninemia. {ECO:0000269|PubMed:8968736, ECO:0000269|PubMed:8968737}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöral Tüp Defekti, Yatkınlık, 182940, Otozomal Dominant (Nöral Tüp Kapanma Defekti) (VANGL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, susceptibility to, 182940, Autosomal dominant (Neural tube closure defect) (VANGL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VANGL1	VANGL1, STBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sacral defect with anterior meningocele (SDAM) [MIM:600145]: Form of caudal dysgenesis. It is present at birth and becomes symptomatic later in life, usually because of obstructive labor in females, chronic constipation, or meningitis. Inheritance is autosomal dominant. {ECO:0000269	PubMed:17409324}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöral Tüp Defekti, Yatkınlık, 182940, Otozomal Dominant (Nöral Tüp Kapanma Defekti) (T geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, susceptibility to, 182940, Autosomal dominant (Neural tube closure defect) (T gene) (Sequence Analysis-All Coding Exons) (Postnatal)	T	T, TFT, SAVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chordoma (CHDM) [MIM:215400]: Rare, clinically malignant tumors derived from notochordal remnants. They occur along the length of the spinal axis, predominantly in the sphenooccipital, vertebral and sacrococcygeal regions. They are characterized by slow growth, local destruction of bone, extension into adjacent soft tissues and rarely, distant metastatic spread. {ECO:0000269	PubMed:19801981}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to development of chordomas is due to a T gene duplication.	EDTA Blood Sample
Nöral Tüp Defekti, Yatkınlık, 601634, Otozomal Resesif; NTDFS (Nöral Tüp Kapanma Defekti) (MTHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neural tube defects, susceptibility to, 601634, Autosomal recessive; NTDFS (Neural tube closure defect) (MTHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9791030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöral Tüp Defekti, Yatkınlık NTD (Nöral Tüp Kapanma Defekti) (VANGL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEURAL TUBE DEFECTS, SUSCEPTIBILITY TO; NTD (Neural tube closure defect) (VANGL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VANGL2	VANGL2, LTAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neural tube defects (NTD) [MIM:182940]: Congenital malformations of the central nervous system and adjacent structures related to defective neural tube closure during the first trimester of pregnancy. Failure of neural tube closure can occur at any level of the embryonic axis. Common NTD forms include anencephaly, myelomeningocele and spina bifida, which result from the failure of fusion in the cranial and spinal region of the neural tube. NTDs have a multifactorial etiology encompassing both genetic and environmental components. {ECO:0000269	PubMed:20558380}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöraminidaz Eksikliği (Sialidoz Tip 1) (NEU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEURAMINIDASE DEFICIENCY (Sialidosis type 1) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insiduous visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. {ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöraminidaz Eksikliği (Sialidoz Tip 1) (Prenatal) (NEU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	NEURAMINIDASE DEFICIENCY (Sialidosis type 1) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insiduous visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. {ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöroblastoma (TP73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroblastoma (TP73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP73	TP73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nöroblastom (Miks 1) (1p36, 3p22/3p21.3, 11q23) (MLPA)	Neuroblastoma (Mix 1) (1p36, 3p22 / 3p21.3, 11q23) (MLPA)	1p36, 3p22/3p21.3, 11q23	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Nöroblastom (Miks 1) (1p36, 3p22/3p21.3, 11q23) (MLPA)/ Nöroblastom (Miks 2) (2p24.1/MYCN, 2q33, 17p13/TP53, 17q) (MLPA)/ Nöroblastom (Miks 3) (Kromozom 4, 7, 9, 12, 14) (MLPA)	Neuroblastoma (Mix 1) (1p36, 3p22 / 3p21.3, 11q23) (MLPA) / Neuroblastoma (Mix 2) (2p24.1 / MYCN, 2q33, 17p13 / Chromosome 4, 7, 9, 12, 14) (MLPA)	1p36, 3p22/3p21.3, 11q23/ 2p24.1/MYCN, 2q33, 17p13/TP53, 17q/ Kromozom 4, 7, 9, 12, 14/	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Nöroblastom (Miks 2) (2p24.1/MYCN, 2q33, 17p13/TP53, 17q) (MLPA)	Neuroblastoma (Mix 2) (2p24.1 / MYCN, 2q33, 17p13 / TP53, 17p) (MLPA)	2p24.1/MYCN, 2q33, 17p13/TP53, 17p	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Nöroblastoma, N - MYC (MYCN) Amplifikasyonu (2p24.3-2q11.2) (FISH)	Neuroblastoma, N - MYC (MYCN) Amplification (2p24.3-2q11.2) (FISH)	2p24.3	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal		Tumor tissue (Fresh or etanol fixed) /Parafinized tissue samole
Nöroblastoma-Hirschsprung Hastalığı, 613013 (Nöroblastom) (PHOX2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroblastoma with Hirschsprung disease, 613013 (Neuroblastoma) (PHOX2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHOX2B	PMX2B, NBPHOX, PHOX2B, NBLST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuroblastoma 2 (NBLST2) [MIM:613013]: A common neoplasm of early childhood arising from embryonic cells that form the primitive neural crest and give rise to the adrenal medulla and the sympathetic nervous system. {ECO:0000305	PubMed:15024693}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Nöroblastoma-Hirschsprung Hastalığı, 613013 (Nöroblastom) (Prenatal) (PHOX2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuroblastoma with Hirschsprung disease, 613013 (Neuroblastoma) (PHOX2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHOX2B	PMX2B, NBPHOX, PHOX2B, NBLST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuroblastoma 2 (NBLST2) [MIM:613013]: A common neoplasm of early childhood arising from embryonic cells that form the primitive neural crest and give rise to the adrenal medulla and the sympathetic nervous system. {ECO:0000305	PubMed:15024693}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nöroblastoma, 256700, Otozomal Dominant, Sporadik Vakalar (Nöroblastom) (NME1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroblastoma, 256700, Autosomal dominant, Isolated cases (Neuroblastoma) (NME1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NME1	NME1, NM23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöroblastoma, Yatkınlık 1, 256700, Otozomal Dominant, Sporadik Vakalar (Nöroblastom) (KIF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroblastoma, susceptibility to, 1, 256700, Autosomal dominant, Isolated cases (Neuroblastoma) (KIF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1B	KIF1B, CMT2A, CMT2A1, NBLST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pheochromocytoma (PCC) [MIM:171300]: A catecholamine- producing tumor of chromaffin tissue of the adrenal medulla or sympathetic paraganglia. The cardinal symptom, reflecting the increased secretion of epinephrine and norepinephrine, is hypertension, which may be persistent or intermittent. {ECO:0000269	PubMed:18334619}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöroblastoma, Yatkınlık, 2, 613013; NBLST2 (Nöroblastom) (PHOX2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroblastoma, susceptibility to, 2, 613013; NBLST2 (Neuroblastoma) (PHOX2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHOX2B	PMX2B, NBPHOX, PHOX2B, NBLST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuroblastoma 2 (NBLST2) [MIM:613013]: A common neoplasm of early childhood arising from embryonic cells that form the primitive neural crest and give rise to the adrenal medulla and the sympathetic nervous system. {ECO:0000305	PubMed:15024693}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöroblastoma, Yatkınlık, 3, 613014; NBLST3 (Nöroblastom) (Exon21 - 29) (ALK geni) (Postnatal)	Neuroblastoma, susceptibility to, 3, 613014; NBLST3 (Neuroblastoma)(Exons 21-29 - Sequence analysis (ALK gene) (Postnatal)	ALK	ALK, NBLST3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuroblastoma 3 (NBLST3) [MIM:613014]: A common neoplasm of early childhood arising from embryonic cells that form the primitive neural crest and give rise to the adrenal medulla and the sympathetic nervous system. {ECO:0000269|PubMed:18724359, ECO:0000269|PubMed:18923523, ECO:0000269|PubMed:18923525}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Nörokütan Melanoz, Somatik, 249400 (Nörokütan Melanositozis) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurocutaneous melanosis, somatic, 249400 (Neurocutaneous melanocytosis) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood. both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Serebral Folat Taşıma Eksikliğine Bağlı Nörodejenerasyon, 613068, Otozomal Resesif (Nörodejeneratif Sendrom, Serebral Folat Taşıma Eksikliğine Bağlı) (FOLR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration due to cerebral folate transport deficiency, 613068, Autosomal recessive (Neurodegenerative syndrome due to cerebral folate transport deficiency) (FOLR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOLR1	FOLR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration due to cerebral folate transport deficiency (NCFTD) [MIM:613068]: A neurodegenerative disorder resulting from brain-specific folate deficiency early in life. Onset is apparent in late infancy with severe developmental regression, movement disturbances, epilepsy and leukodystrophy. {ECO:0000269	PubMed:19732866}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Serebral Folat Taşıma Eksikliğine Bağlı Nörodejenerasyon, 613068, Otozomal Resesif (Nörodejeneratif Sendrom, Serebral Folat Taşıma Eksikliğine Bağlı) (Prenatal) (FOLR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration due to cerebral folate transport deficiency, 613068, Autosomal recessive (Neurodegenerative syndrome due to cerebral folate transport deficiency) (FOLR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOLR1	FOLR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration due to cerebral folate transport deficiency (NCFTD) [MIM:613068]: A neurodegenerative disorder resulting from brain-specific folate deficiency early in life. Onset is apparent in late infancy with severe developmental regression, movement disturbances, epilepsy and leukodystrophy. {ECO:0000269	PubMed:19732866}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon-Ataksi, Distoni ve Görme Bozukluğu, Çocukluk Dönem Başlangıçlı, 617145, Otozomal Resesif; NADGP (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145, Autosomal recessive; NADGP (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 3 (PDB3) [MIM:167250]: A disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269|PubMed:11992264, ECO:0000269|PubMed:12374763, ECO:0000269|PubMed:14584883, ECO:0000269|PubMed:15125799, ECO:0000269|PubMed:15146436, ECO:0000269|PubMed:15176995, ECO:0000269|PubMed:15207768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörodejenerasyon-Ataksi, Distoni ve Görme Bozukluğu, Çocukluk Dönem Başlangıçlı, 617145, Otozomal Resesif; NADGP (Prenatal) (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with ataxia, dystonia, and gaze palsy, childhood-onset, 617145, Autosomal recessive; NADGP (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paget disease of bone 3 (PDB3) [MIM:167250]: A disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269|PubMed:11992264, ECO:0000269|PubMed:12374763, ECO:0000269|PubMed:14584883, ECO:0000269|PubMed:15125799, ECO:0000269|PubMed:15146436, ECO:0000269|PubMed:15176995, ECO:0000269|PubMed:15207768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 1, 234200, Otozomal Resesif; NBIA1 (Pantotenat Kinaz İle İlişkili Nörodejenerasyon) (PANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 1, 234200, Autosomal recessive; NBIA1 (Pantothenate kinase-associated neurodegeneration) (PANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PANK2	PANK2, NBIA1, PKAN, HARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269	PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 1, 234200, Otozomal Resesif; NBIA1 (Pantotenat Kinaz İle İlişkili Nörodejenerasyon) (MLPA)	Neurodegeneration with brain iron accumulation 1, 234200, Autosomal recessive; NBIA1 (Pantothenate kinase-associated neurodegeneration) (MLPA)	PANK2	PANK2, NBIA1, PKAN, HARP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269	PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 1, 234200, Otozomal Resesif; NBIA1 (Pantotenat Kinaz İle İlişkili Nörodejenerasyon) (Prenatal) (PANK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 1, 234200, Autosomal recessive; NBIA1 (Pantothenate kinase-associated neurodegeneration) (PANK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PANK2	PANK2, NBIA1, PKAN, HARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269	PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 1, 234200, Otozomal Resesif; NBIA1 (Pantotenat Kinaz İle İlişkili Nörodejenerasyon) (Prenatal) (MLPA)	Neurodegeneration with brain iron accumulation 1, 234200, Autosomal recessive; NBIA1 (Pantothenate kinase-associated neurodegeneration) (Prenatal) (MLPA)	PANK2	PANK2, NBIA1, PKAN, HARP	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hypoprebetalipoproteinemia, acanthocytosis, retinitis pigmentosa, and pallidal degeneration (HARP) [MIM:607236]: Rare syndrome with many clinical similarities to PKAN. {ECO:0000269	PubMed:12058097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 2B, 610217, Otozomal Resesif; NBIA2B (İnfantil Nöroaksonal Distrofi) (PLA2G6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 2B, 610217, Autosomal recessive; NBIA2B (Infantile neuroaxonal dystrophy) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 2B, 610217, Otozomal Resesif; NBIA2B (İnfantil Nöroaksonal Distrofi) (MLPA)	Neurodegeneration with brain iron accumulation 2B, 610217, Autosomal recessive; NBIA2B (Infantile neuroaxonal dystrophy) (MLPA)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 2B, 610217, Otozomal Resesif; NBIA2B (İnfantil Nöroaksonal Distrofi) (PLA2G6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 2B, 610217, Autosomal recessive; NBIA2B (Infantile neuroaxonal dystrophy) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 2B, 610217, Otozomal Resesif; NBIA2B (İnfantil Nöroaksonal Distrofi) (Prenatal) (MLPA)	Neurodegeneration with brain iron accumulation 2B, 610217, Autosomal recessive; NBIA2B (Infantile neuroaxonal dystrophy) (Prenatal) (MLPA)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. {ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 3, 606159, Otozomal Dominant; NBIA3 (Nöroferritinopati) (FTL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 3, 606159, Autosomal dominant; NBIA3 (Neuroferritinopathy) (FTL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTL	FTL, NBIA3, LFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. {ECO:0000269	PubMed:19176363}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 3, 606159, Otozomal Dominant; NBIA3 (Nöroferritinopati) (Prenatal) (FTL geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 3, 606159, Autosomal dominant; NBIA3 (Neuroferritinopathy) (FTL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FTL	FTL, NBIA3, LFTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary hyperferritinemia-cataract syndrome (HHCS) [MIM:600886]: An autosomal dominant disease characterized by elevated level of ferritin in serum and tissues, and early-onset bilateral cataract. {ECO:0000269	PubMed:19176363}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nörodejenerasyon, Beyin Demir Birikmesi 4, 614298, Otozomal Resesif; NBIA4 (Mitokondriyal Membran Proteini İle İlişkili Nörodejenerasyon) (C19orf12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 4, 614298, Autosomal recessive; NBIA4 (Mitochondrial membrane protein-associated neurodegeneration) (C19orf12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C19orf12	C19orf12, NBIA4, SPG43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 4 (NBIA4) [MIM:614298]: A neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. NBIA4 results in speech difficulty, extrapyramidal signs, oromandibular and generalized dystonia, and parkinsonism. Most patients have progressive involvement of the corticospinal tract, with spasticity, hyperreflexia, and extensor plantar responses. {ECO:0000269|PubMed:21981780, ECO:0000269|PubMed:22508347, ECO:0000269|PubMed:22584950, ECO:0000269|PubMed:23269600, ECO:0000269|PubMed:23521069, ECO:0000269|PubMed:23857908}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 4, 614298, Otozomal Resesif; NBIA4 (Mitokondriyal Membran Proteini İle İlişkili Nörodejenerasyon) (Prenatal) (C19orf12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 4, 614298, Autosomal recessive; NBIA4 (Mitochondrial membrane protein-associated neurodegeneration) (C19orf12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C19orf12	C19orf12, NBIA4, SPG43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurodegeneration with brain iron accumulation 4 (NBIA4) [MIM:614298]: A neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. NBIA4 results in speech difficulty, extrapyramidal signs, oromandibular and generalized dystonia, and parkinsonism. Most patients have progressive involvement of the corticospinal tract, with spasticity, hyperreflexia, and extensor plantar responses. {ECO:0000269|PubMed:21981780, ECO:0000269|PubMed:22508347, ECO:0000269|PubMed:22584950, ECO:0000269|PubMed:23269600, ECO:0000269|PubMed:23521069, ECO:0000269|PubMed:23857908}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 5, 300894, X'e Bağlı Dominant; NBIA5 (Beta-Propeller Protein İle İlişkili Nörodejenerasyon) (WDR45 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 5, 300894, X-linked dominant; NBIA5 (Beta-propeller protein-associated neurodegeneration) (WDR45 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR45	WDR45, WIPI4, NBIA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 5, 300894, X'e Bağlı Dominant; NBIA5 (Beta-Propeller Protein İle İlişkili Nörodejenerasyon) (Prenatal) (WDR45 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 5, 300894, X-linked dominant; NBIA5 (Beta-propeller protein-associated neurodegeneration) (WDR45 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR45	WDR45, WIPI4, NBIA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 6, 615643, Otozomal Resesif (Coasy Proteini İle İlişkili Nörodejenerasyon) (COASY geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodegeneration with brain iron accumulation 6, 615643, Autosomal recessive; NBIA6 (COASY protein-associated neurodegeneration) (COASY gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COASY	COASY, NBIA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nörodejenerasyon, Beyin Demir Birikmesi 6, 615643, Otozomal Resesif (Coasy Proteini İle İlişkili Nörodejenerasyon) (Prenatal) (COASY geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodegeneration with brain iron accumulation 6, 615643, Autosomal recessive; NBIA6 (COASY protein-associated neurodegeneration) (COASY gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COASY	COASY, NBIA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörogelişimsel Bozukluk, Hipotoni, Nöbetler ve Konuşması Olmayan 617268, Otozomal Dominant; NDHSA (HECW2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268, Autosomal dominant; NDHSA (HECW2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HECW2	HECW2, NEDL2, KIAA1301, NDHSAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nörogelişimsel Bozukluk, Hipotoni, Nöbetler ve Konuşması Olmayan 617268, Otozomal Dominant; NDHSA (Prenatal) (HECW2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodevelopmental disorder with hypotonia, seizures, and absent language, 617268, Autosomal dominant; NDHSA (HECW2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HECW2	HECW2, NEDL2, KIAA1301, NDHSAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörogelişimsel Bozukluk, ± Beyin, Göz Veya Kalp Anomalileri, 616975, Otozomal Dominant; NEDIBEH (RERE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975, Autosomal dominant; NEDIBEH (RERE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RERE	RERE, NEDBEH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving RERE is found in the neuroblastoma cell line NGP. Translocation t(1;15)(p36.2;q24).-	EDTA Blood Sample
Nörogelişimsel Bozukluk, ± Beyin, Göz Veya Kalp Anomalileri, 616975, Otozomal Dominant; NEDIBEH (Prenatal) (RERE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurodevelopmental disorder with or without anomalies of the brain, eye, or heart, 616975, Autosomal dominant; NEDIBEH (RERE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RERE	RERE, NEDBEH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving RERE is found in the neuroblastoma cell line NGP. Translocation t(1;15)(p36.2;q24).-	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöroepitelyoma, 612219 (Nöroepitelyoma) (EWSR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuroepithelioma, 612219 (Neuroepithelioma) (EWSR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EWSR1	EWSR1, EWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angiomatoid fibrous histiocytoma (AFH) [MIM:612160]: A distinct variant of malignant fibrous histiocytoma that typically occurs in children and adolescents and is manifest by nodular subcutaneous growth. Characteristic microscopic features include lobulated sheets of histiocyte-like cells intimately associated with areas of hemorrhage and cystic pseudovascular spaces, as well as a striking cuffing of inflammatory cells, mimicking a lymph node metastasis. {ECO:0000269|PubMed:15884099, ECO:0000269|PubMed:17724745}. Note=The gene represented in this entry is involved in disease pathogenesis. Chromosomal aberrations involving EWSR1 are found in patients with angiomatoid fibrous histiocytoma. Translocation t(12;22)(q13;q12) with ATF1 generates a chimeric EWSR1/ATF1 protein (PubMed:15884099). Translocation t(2;22)(q33;q12) with CREB1 generates a EWSR1/CREB1 fusion gene that is most common genetic abnormality in this tumor type (PubMed:17724745). {ECO:0000269|PubMed:15884099, ECO:0000269|PubMed:17724745}.	EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (NF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (NF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (MLPA)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (MLPA)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (Prenatal) (NF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (NF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (Prenatal) (MLPA)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis-Noonan Sendromu, 601321, Otozomal Dominant; NFNS (Nörofibromatoz-Noonan Sendromu) (Prenatal) (MLPA)	Neurofibromatosis-Noonan syndrome, 601321, Autosomal dominant; NFNS (Neurofibromatosis-Noonan syndrome) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Allesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (NF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (NF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Allesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (MLPA)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Allesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (MLPA)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Allesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (Prenatal) (NF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (NF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nörofibromatozis, Ailesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (Prenatal) (MLPA)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Ailesel Spinal, 162210, Otozomal Dominant (Nörofibromatozis Tip 1) (Prenatal) (MLPA)	Neurofibromatosis, familial spinal, 162210, Autosomal dominant (Neurofibromatosis type 1) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (NF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (NF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (MLPA)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (MLPA)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (Prenatal) (NF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (NF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (Prenatal) (MLPA)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Tip 1, 162200, Otozomal Dominant; NF1 (Nörofibromatozis Tip 1) (Prenatal) (MLPA)	Neurofibromatosis, type 1, 162200, Autosomal dominant; NF1 (Neurofibromatosis type 1) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The gene represented in this entry may be involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Tip 2, 101000, Otozomal Dominant (Nörofibromatozis Tip 2) (NF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurofibromatosis, type 2, 101000, Autosomal dominant; NF2 (Neurofibromatosis type 2) (NF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF2	NF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörofibromatozis, Tip 2, 101000, Otozomal Dominant (Nörofibromatozis Tip 2) (NF2) (MLPA)	Neurofibromatosis, type 2, 101000, Autosomal dominant; NF2 (Neurofibromatosis type 2) (MLPA)	NF2	NF2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nörofibromatozis, Tip 2, 101000, Otozomal Dominant (Nörofibromatozis Tip 2) (Prenatal) (NF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neurofibromatosis, type 2, 101000, Autosomal dominant; NF2 (Neurofibromatosis type 2) (NF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NF2	NF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibromatozis, Tip 2, 101000, Otozomal Dominant (Nörofibromatozis Tip 2) (Prenatal) (MLPA)	Neurofibromatosis, type 2, 101000, Autosomal dominant; NF2 (Neurofibromatosis type 2) (Prenatal) (MLPA)	NF2	NF2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nörofibrosarkom (MXI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neurofibrosarcoma (MXI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MXI1	MXI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nörometabolik bozukluklar paneli (ASPA, MLC1, MLYCD, D2HGDH, L2HGDH) (MLPA)	Neurometabolic disorders (ASPA, MLC1, MLYCD, D2HGDH, L2HGDH) (MLPA)	ASPA, MLC1, MLYCD, D2HGDH, L2HGDH	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Nörometabolik bozukluklar paneli (ASPA, MLC1, MLYCD, D2HGDH, L2HGDH) (MLPA) (Prenatal)	Neurometabolic disorders (ASPA, MLC1, MLYCD, D2HGDH, L2HGDH) (MLPA) (Prenatal)	ASPA, MLC1, MLYCD, D2HGDH, L2HGDH	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Nöromüsküler Hastalık, Konjenital, Homojen Tip 1 Lif, 117000, Otozomal Resesif, Otozomal Dominant (RYR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000, Autosomal recessive, Autosomal dominant (RYR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095	EDTA Blood Sample
Nöromüsküler Hastalık, Konjenital, Homojen Tip 1 Lif, 117000, Otozomal Resesif, Otozomal Dominant (Prenatal) (RYR1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Neuromuscular disease, congenital, with uniform type 1 fiber, 117000, Autosomal recessive, Autosomal dominant (RYR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RYR1	RYR1, MHS, CCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Central core disease of muscle (CCD) [MIM:117000]: Autosomal dominant congenital myopathy, but a severe autosomal recessive form also exists. Both clinical and histological variability is observed. Affected individuals typically display hypotonia and proximal muscle weakness in infancy, leading to the delay of motor milestones. The clinical course of the disorder is usually slow or nonprogressive in adulthood, and the severity of the symptoms may vary from normal to significant muscle weakness. Microscopic examination of CCD-affected skeletal muscle reveals a predominance of type I fibers containing amorphous-looking areas (cores) that do not stain with oxidative and phosphorylase histochemical techniques. {ECO:0000269|PubMed:10051009, ECO:0000269|PubMed:10097181, ECO:0000269|PubMed:11113224, ECO:0000269|PubMed:11709545, ECO:0000269|PubMed:11741831, ECO:0000269|PubMed:12112081, ECO:0000269|PubMed:12136074, ECO:0000269|PubMed:12565913, ECO:0000269|PubMed:12566385, ECO:0000269|PubMed:12937085, ECO:0000269|PubMed:14670767, ECO:0000269|PubMed:14985404, ECO:0000269|PubMed:17204054, ECO:0000269|PubMed:17226826, ECO:0000269|PubMed:18253926, ECO:0000269|PubMed:18312400, ECO:0000269|PubMed:20142353, ECO:0000269|PubMed:21674524, ECO:0000269|PubMed:23558838, ECO:0000269|PubMed:24561095	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöromiyotoni ve Aksonal Nöropati, Otozomal Resesif, 137200, Otozomal Resesif; NMAN (Otozomal Resesif Aksonal Nöropati-Nöromiyotoni) (HINT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200, Autosomal recessive; NMAN (Autosomal recessive axonal neuropathy with neuromyotonia) (HINT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HINT1	HINT1, PRKCNH1, NMAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöromiyotoni ve Aksonal Nöropati, Otozomal Resesif, 137200, Otozomal Resesif; NMAN (Otozomal Resesif Aksonal Nöropati-Nöromiyotoni) (Prenatal) (HINT1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Neuromyotonia and axonal neuropathy, autosomal recessive, 137200, Autosomal recessive; NMAN (Autosomal recessive axonal neuropathy with neuromyotonia) (HINT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HINT1	HINT1, PRKCNH1, NMAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Neuronopathy, Distal Kalıtsal Motor, Tip 2C, 613376, Otozomal Dominant; HMN2C (Distal Kalıtsal Motor Nöropati Tip 2) (HSPB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuronopathy, distal hereditary motor, type IIC, 613376, Autosomal dominant; HMN2C (Distal hereditary motor neuropathy type 2) (HSPB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPB3	HSPB3, HSPL27, HMN2C, DHMN2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 2C (HMN2C) [MIM:613376]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269|PubMed:20142617}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Neuronopathy, Distal Kalıtsal Motor, Tip 2C, 613376, Otozomal Dominant; HMN2C (Distal Kalıtsal Motor Nöropati Tip 2) (Prenatal) (HSPB3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuronopathy, distal hereditary motor, type IIC, 613376, Autosomal dominant; HMN2C (Distal hereditary motor neuropathy type 2) (HSPB3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPB3	HSPB3, HSPL27, HMN2C, DHMN2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuronopathy, distal hereditary motor, 2C (HMN2C) [MIM:613376]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269|PubMed:20142617}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöronopati, Distal Kalıtsal Motor, Tip 2D, 615575, Otozomal Dominant; HMN2D (Distal Kalıtsal Motor Nöropati Tip 2) (FBXO38 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuronopathy, distal hereditary motor, type IID, 615575, Autosomal dominant; HMN2D (Distal hereditary motor neuropathy type 2) (FBXO38 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBXO38	FBXO38, FBX38, MOKA, HMN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 2D (HMN2D) [MIM:615575]: A disorder characterized by onset of slowly progressive distal lower limb weakness and atrophy between the second and fourth decades of life. Weakness usually begins in the calf muscles and later involves more proximal muscles. The severity is variable, and some patients have difficulty walking or running. Most also have upper limb involvement, particularly of the triceps and intrinsic hand muscles. Some patients may lose independent ambulation later in the disease course. Sensory impairment is typically not present, and cognition and bulbar function are normal. {ECO:0000269|PubMed:24207122}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Neuronopathy, Distal Kalıtsal Motor Tipi VB, 614751, Otozomal Dominant; HMN5B (Distal Kalıtsal Motor Nöropati Tipi 5) (REEP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuronopathy, distal hereditary motor, type VB, 614751, Autosomal dominant; HMN5B (Distal hereditary motor neuropathy type 5) (REEP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REEP1	REEP1, C2ORF23, SPG31, HMN5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 5B (HMN5B) [MIM:614751]: A disorder characterized by distal muscular atrophy mainly affecting the upper extremities, in contrast to other distal motor neuronopathies. These constitute a heterogeneous group of neuromuscular diseases caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN5B is characterized by onset in the first or second decade of distal muscle weakness and atrophy, primarily affecting the intrinsic hand muscles, but also affecting the lower legs, resulting in abnormal gait and pes cavus. {ECO:0000269|PubMed:22703882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Neuronopathy, Distal Kalıtsal Motor Tipi VB, 614751, Otozomal Dominant; HMN5B (Distal Kalıtsal Motor Nöropati Tipi 5) (Prenatal) (REEP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuronopathy, distal hereditary motor, type VB, 614751, Autosomal dominant; HMN5B (Distal hereditary motor neuropathy type 5) (REEP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	REEP1	REEP1, C2ORF23, SPG31, HMN5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuronopathy, distal hereditary motor, 5B (HMN5B) [MIM:614751]: A disorder characterized by distal muscular atrophy mainly affecting the upper extremities, in contrast to other distal motor neuronopathies. These constitute a heterogeneous group of neuromuscular diseases caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN5B is characterized by onset in the first or second decade of distal muscle weakness and atrophy, primarily affecting the intrinsic hand muscles, but also affecting the lower legs, resulting in abnormal gait and pes cavus. {ECO:0000269|PubMed:22703882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöronopati, Distal Kalıtsal Motor, Tip 6, 604320, Otozomal Resesif; DSMA1 (Spinal Müsküler Atrofi-Solunum Zorluğu Tip 1) (IGHMBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuronopathy, distal hereditary motor, type VI, 604320, Autosomal recessive; DSMA1 (Spinal muscular atrophy with respiratory distress type 1) (IGHMBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGHMBP2	IGHMBP2, SMUBP2, CATF1, SMARD1, HMN6, CMT2S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2S (CMT2S) [MIM:616155]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. {ECO:0000269|PubMed:25439726}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Neuronopathy, Distal Kalıtsal Motor Tipi VIIA, 158580, Otozomal Dominant; HMN7A (Distal Kalıtsal Motor Nöropati Tip 7) (SLC5A7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuronopathy, distal hereditary motor, type VIIA, 158580, Autosomal dominant; HMN7A (Distal hereditary motor neuropathy type 7) (SLC5A7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC5A7	SLC5A7, CHT1, HMN7A, CMS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 7A (HMN7A) [MIM:158580]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN7A is characterized by onset in the second decade of progressive distal muscle wasting and weakness affecting the upper and lower limbs and resulting in walking difficulties and hand grip. There is significant muscle atrophy of the hands and lower limbs. The disorder is associated with vocal cord paresis due to involvement of the tenth cranial nerve. {ECO:0000269	PubMed:23141292}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Neuronopathy, Distal Kalıtsal Motor Tipi VIIA, 158580, Otozomal Dominant; HMN7A (Distal Kalıtsal Motor Nöropati Tip 7) (Prenatal) (SLC5A7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuronopathy, distal hereditary motor, type VIIA, 158580, Autosomal dominant; HMN7A (Distal hereditary motor neuropathy type 7) (SLC5A7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC5A7	SLC5A7, CHT1, HMN7A, CMS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neuronopathy, distal hereditary motor, 7A (HMN7A) [MIM:158580]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN7A is characterized by onset in the second decade of progressive distal muscle wasting and weakness affecting the upper and lower limbs and resulting in walking difficulties and hand grip. There is significant muscle atrophy of the hands and lower limbs. The disorder is associated with vocal cord paresis due to involvement of the tenth cranial nerve. {ECO:0000269	PubMed:23141292}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöropati, Distal Kalıtsal Motor, Tip 8; Hmn8 (Otozomal Dominant Konjenital Benign Spinal Musküler Atrofi) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE VIII; HMN8 (Autosomal dominant congenital benign spinal muscular atrophy) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachydmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269	PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 1, 605253, Otozomal Resesif, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 4E) (EGR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, congenital hypomyelinating, 1, 605253, Autosomal recessive, Autosomal dominant (Charcot-Marie-Tooth disease type 4E) (EGR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EGR2	EGR2, KROX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 1, 605253, Otozomal Resesif, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 4E) (MLPA)	Neuropathy, congenital hypomyelinating, 1, 605253, Autosomal recessive, Autosomal dominant (Charcot-Marie-Tooth disease type 4E) (MLPA)	EGR2	EGR2, KROX20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 1, 605253, Otozomal Resesif, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 4E) (Prenatal) (EGR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuropathy, congenital hypomyelinating, 1, 605253, Autosomal recessive, Autosomal dominant (Charcot-Marie-Tooth disease type 4E) (EGR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EGR2	EGR2, KROX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 1, 605253, Otozomal Resesif, Otozomal Dominant (Charcot-Marie-Tooth Hastalığı Tip 4E) (Prenatal) (MLPA)	Neuropathy, congenital hypomyelinating, 1, 605253, Autosomal recessive, Autosomal dominant (Charcot-Marie-Tooth disease type 4E) (Prenatal) (MLPA)	EGR2	EGR2, KROX20	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dejerine-Sottas syndrome (DSS) [MIM:145900]: A severe degenerating neuropathy of the demyelinating Charcot-Marie-Tooth disease category, with onset by age 2 years. Characterized by motor and sensory neuropathy with very slow nerve conduction velocities, increased cerebrospinal fluid protein concentrations, hypertrophic nerve changes, delayed age of walking as well as areflexia. There are both autosomal dominant and autosomal recessive forms of Dejerine-Sottas syndrome. {ECO:0000269	PubMed:10371530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 605253, Otozomal Resesif, Otozomal Dominant; CHN (Charcot-Marie-Tooth Hastalığı Tip 4E) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, congenital hypomyelinating, 605253, Autosomal recessive, Autosomal dominant; CHN (Charcot-Marie-Tooth disease type 4E) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adie pupil (ADEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269	PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 605253, Otozomal Resesif, Otozomal Dominant; CHN (Charcot-Marie-Tooth Hastalığı Tip 4E) (MLPA)	Neuropathy, congenital hypomyelinating, 605253, Autosomal recessive, Autosomal dominant; CHN (Charcot-Marie-Tooth disease type 4E) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adie pupil (ADEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269	PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 605253, Otozomal Resesif, Otozomal Dominant; CHN (Charcot-Marie-Tooth Hastalığı Tip 4E) (Prenatal) (MPZ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neuropathy, congenital hypomyelinating, 605253, Autosomal recessive, Autosomal dominant; CHN (Charcot-Marie-Tooth disease type 4E) (MPZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adie pupil (ADEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269	PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöropati, Konjenital Hipomiyelinizan, 605253, Otozomal Resesif, Otozomal Dominant; CHN (Charcot-Marie-Tooth Hastalığı Tip 4E) (Prenatal) (MLPA)	Neuropathy, congenital hypomyelinating, 605253, Autosomal recessive, Autosomal dominant; CHN (Charcot-Marie-Tooth disease type 4E) (Prenatal) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adie pupil (ADEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269	PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöropati, Distal Kalıtsal Motor, Tip 2A, 158590, Otozomal Dominant; HMN2A (Distal Kalıtsal Motor Nöropati Tip 2) (HSPB8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, distal hereditary motor, type IIA, 158590, Autosomal dominant; HMN2A (Distal hereditary motor neuropathy type 2) (HSPB8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPB8	HSPB8, HL1, E2IG1, DHMN2, CMT2L, HMN2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 2A (HMN2A) [MIM:158590]: A neuromuscular disorder. Distal hereditary motor neuronopathies constitute a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:15122253}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Distal Kalıtsal Motor, Tip 2B, 608634, Otozomal Dominant; HMN2B (Distal Kalıtsal Motor Nöropati Tip 2) (HSPB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, distal hereditary motor, type IIB, 608634, Autosomal dominant; HMN2B (Distal hereditary motor neuropathy type 2) (HSPB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPB1	HSPB1, HSP27, CMT2F, HMN2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2F (CMT2F) [MIM:606595]: A dominant axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. Nerve conduction velocities are normal or slightly reduced. Onset of Charcot-Marie-Tooth disease type 2F is between 15 and 25 years with muscle weakness and atrophy usually beginning in feet and legs (peroneal distribution). Upper limb involvement occurs later. {ECO:0000269|PubMed:15122254, ECO:0000269|PubMed:22206013}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Distal Kalıtsal Motor, Tip 5A, 600794, Otozomal Dominant (Distal Kalıtsal Motor Nöropati Tipi 5) (BSCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, distal hereditary motor, type VA, 600794, Autosomal dominant (Distal hereditary motor neuropathy type 5) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 17, autosomal dominant (SPG17) [MIM:270685]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG17 is characterized by prominent amyotrophy of the hand muscles, the presence of mild to severe pyramidal tract signs and spastic paraplegia. SPG17 is a motor neuron disease overlapping with distal spinal muscular atrophy type 5. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Nöropati, Distal Kalıtsal Motor, Tip 5A, 600794, Otozomal Dominant; HMN5A (Distal Kalıtsal Motor Nöropati Tipi 5) (GARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, distal hereditary motor, type VA, 600794, Autosomal dominant; HMN5A (Distal hereditary motor neuropathy type 5) (GARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GARS	GARS, SMAD1, CMT2D, HMN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 5A (HMN5A) [MIM:600794]: A disorder characterized by distal muscular atrophy mainly affecting the upper extremities, in contrast to other distal motor neuronopathies. These constitute a heterogeneous group of neuromuscular diseases caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269|PubMed:12690580, ECO:0000269|PubMed:24627186}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Distal Kalıtsal Motor, Tip 7B, 607641, Otozomal Dominant; HMN7B (Distal Kalıtsal Motor Nöropati Tip 7) (DCTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, distal hereditary motor, type VIIB, 607641, Autosomal dominant; HMN7B (Distal hereditary motor neuropathy type 7) (DCTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCTN1	DCTN1, HMN7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis (ALS) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:15326253, ECO:0000269|PubMed:16240349}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal Motor ve Duyu, Russe Tipi, 605285, Otozomal Resesif; HMSNR (Charcot-Marie-Tooth Hastalığı Tip 4G) (HK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary motor and sensory, Russe type, 605285, Autosomal recessive; HMSNR (Charcot-Marie-Tooth disease type 4G) (HK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HK1	HK1, HKD, HMSNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hexokinase deficiency (HK deficiency) [MIM:235700]: Rare autosomal recessive disease with nonspherocytic hemolytic anemia as the predominant clinical feature. {ECO:0000269|PubMed:12393545, ECO:0000269|PubMed:7655856}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal Motor ve Duyu, Tip 6B, 616505, Otozomal Resesif; HMSN6B (Kalıtsal Motor ve Duyusal Nöropati Tip 6) (SLC25A46 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary motor and sensory, type VIB, 616505, Autosomal recessive; HMSN6B (Hereditary motor and sensory neuropathy type 6) (SLC25A46 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A46	SLC25A46, HMSN6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 1A, 162400, Otozomal Dominant; HSAN1A (Kalıtsal Duysal ve Otonom Nöropati, Tip 1) (SPTLC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type IA, 162400, Autosomal dominant; HSAN1A (Hereditary sensory and autonomic neuropathy type 1) (SPTLC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTLC1	SPTLC1, LBC1, SPT1, HSN1, HSAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 1A (HSAN1A) [MIM:162400]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by prominent sensory abnormalities with a variable degree of motor and autonomic dysfunction. The neurological phenotype is often complicated by severe infections, osteomyelitis, and amputations. HSAN1A is an autosomal dominant axonal form with onset in the second or third decades. Initial symptoms are loss of pain, touch, heat, and cold sensation over the feet, followed by distal muscle wasting and weakness. Loss of pain sensation leads to chronic skin ulcers and distal amputations. {ECO:0000269|PubMed:11242114, ECO:0000269|PubMed:19651702, ECO:0000269|PubMed:21618344, ECO:0000269|PubMed:22302274, ECO:0000269|PubMed:24247255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik Tip 1C, 613640, Otozomal Dominant; HSAN1C (Kalıtsal Duysal ve Otonom Nöropati, Tip 2) (SPTLC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type IC, 613640, Autosomal dominant; HSAN1C (Hereditary sensory and autonomic neuropathy type 1) (SPTLC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTLC2	SPTLC2, KIAA0526, SPT2, LCB2, HSN1C, NSAN1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 2, 201300, Otozomal Resesif; HSAN2A (Alesel Duysal ve Otonomik Nöropati Tip 2) (WNK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type II, 201300, Autosomal recessive; HSAN2A (Hereditary sensory and autonomic neuropathy type 2) (WNK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNK1	WNK1, PRKWNK1, KDP, PHA2C, HSAN2, HSN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 2A (HSAN2A) [MIM:201300]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN2A is an autosomal recessive disorder characterized by impairment of pain, temperature and touch sensation, onset of symptoms in infancy or early childhood, occurrence of distal extremity pathologies (paronychia, whitlows, ulcers, and Charcot joints), frequent amputations, sensory loss that affects all modalities of sensation (lower and upper limbs and perhaps the trunk as well), absence or diminution of tendon reflexes (usually in all limbs), minimal autonomic dysfunction, absence of sensory nerve action potentials, and virtual absence of myelinated fibers with decreased numbers of unmyelinated fibers in sural nerves. {ECO:0000269|PubMed:15060842, ECO:0000269|PubMed:15911806, ECO:0000269|PubMed:18521183}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 2B, 613115, Otozomal Resesif; HSAN2B (Alesel Duysal ve Otonomik Nöropati Tip 4) (RETREG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type IIB, 613115, Autosomal recessive; HSAN2B (Hereditary sensory and autonomic neuropathy type 2) (RETREG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RETREG1	FAM134B, HSAN2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonom, Tip 3; Hsan3 (Alesel Disotonomi) (ELP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE III; HSAN3 (Familial dysautonomia) (ELP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELP1	IKBKAP, IKAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, 5 Tipi, 608654, Otozomal Resesif; HSAN5 (Kalıtsal Duysal ve Otonom Nöropati Tip 5) (NGF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type V, 608654, Autosomal recessive; HSAN5 (Hereditary sensory and autonomic neuropathy type 5) (NGF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NGF	NGF, NGFB, HSAN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 5 (HSAN5) [MIM:608654]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN5 patients manifest loss of pain perception and impaired temperature sensitivity, ulcers, and in some cases self- mutilation. The autonomic involvement is variable. {ECO:0000269|PubMed:14976160, ECO:0000269|PubMed:22302274}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 6, 614653, Otozomal Resesif; HSAN6 (Kalıtsal Duysal ve Otonom Nöropati Tip 6) (DST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type VI, 614653, Autosomal recessive; HSAN6 (Hereditary sensory and autonomic neuropathy type 6) (DST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DST	DST, BPAG1, DMH, D6S1101, HSAN6, EBSB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 6 (HSAN6) [MIM:614653]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN6 is a severe autosomal recessive disorder characterized by neonatal hypotonia, respiratory and feeding difficulties, lack of psychomotor development, and autonomic abnormalities including labile cardiovascular function, lack of corneal reflexes leading to corneal scarring, areflexia, and absent axonal flare response after intradermal histamine injection. {ECO:0000269|PubMed:22522446}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 7, 615548, Otozomal Dominant; HSAN7 (Kalıtsal Duysal ve Otonom Nöropati Tip 7) (SCN11A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type VII, 615548, Autosomal dominant; HSAN7 (Hereditary sensory and autonomic neuropathy type 7) (SCN11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN11A	SCN11A, HSAN7, FEPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 7 (HSAN7) [MIM:615548]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN7 is characterized by congenital inability to experience pain resulting in self-mutilations, slow-healing wounds, and multiple painless fractures, mild muscle weakness, delayed motor development, slightly reduced motor and sensory nerve conduction velocities, hyperhidrosis and gastrointestinal dysfunction. {ECO:0000269|PubMed:24039648}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal Duysal ve Otonomik, Tip 8, 616488, Otozomal Resesif; HSAN8 (Konjenital Ağrı Duyarsızlığı-Hipohidroz Sendromu) (PRDM12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory and autonomic, type VIII, 616488, Autosomal recessive; HSAN8 (Congenital insensitivity to pain-hypohidrosis syndrome) (PRDM12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM12	PRDM12, HSAN8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 8 (HSAN8) [MIM:616488]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN8 patients manifest congenital insensitivity to pain resulting in ulceration to the fingers, tongue, lips, and other distal appendages. Some patients may also have decreased sweating and tear production. {ECO:0000269|PubMed:26005867}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal Duysal Tip 1D, 613708, Otozomal Dominant; HSN1D (Kalıtsal Duysal ve Otonom Nöropati, Tip 3) (ATL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory, type ID, 613708, Autosomal dominant; HSN1D (Hereditary sensory and autonomic neuropathy type 1) (ATL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATL1	ATL1, SPG3A, HSN1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 1D (HSN1D) [MIM:613708]: A disease characterized by adult-onset distal axonal sensory neuropathy leading to mutilating ulcerations as well as hyporeflexia. Some patients may show features suggesting upper neuron involvement. {ECO:0000269|PubMed:21194679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal Duysal Tip 1E, 614116, Otozomal Dominant; HSN1E (Kalıtsal Duysal Nöropati-Sağırlık-Demans Sendromu) (DNMT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory, type IE, 614116, Autosomal dominant; HSN1E (Hereditary sensory neuropathy-deafness-dementia syndrome) (DNMT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNMT1	DNMT1, MCMT, HSN1E, ADCADN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant (ADCADN) [MIM:604121]: An autosomal dominant neurologic disorder characterized by adult onset of progressive cerebellar ataxia, narcolepsy, cataplexy, sensorineural deafness, and dementia. More variable features include optic atrophy, sensory neuropathy, psychosis, and depression. {ECO:0000269|PubMed:22328086}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal Duysal Tip 615632, Otozomal Dominant 1F; HSN1F (Kalıtsal Duysal ve Otonom Nöropati, Tip 4) (ATL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory, type IF, 615632, Autosomal dominant; HSN1F (Hereditary sensory and autonomic neuropathy type 1) (ATL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATL3	ATL3, HSN1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Nöropati, Kalıtsal Duysal Tip 2C, 614213, Otozomal Resesif; HSN2C (Alesel Duysal ve Otonomik Nöropati Tip 5) (KIF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory, type IIC, 614213, Autosomal recessive; HSN2C (Hereditary sensory and autonomic neuropathy type 2) (KIF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1A	KIF1A, ATSV, UNC104, SPG30, HSN2C, MRD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 2C (HSN2C) [MIM:614213]: A neurodegenerative disorder characterized by onset in the first decade of progressive distal sensory loss leading to ulceration and amputation of the fingers and toes. Affected individuals also develop distal muscle weakness, primarily affecting the lower limbs. {ECO:0000269|PubMed:21820098}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Spastik Paraplejj Kalıtsal Duysal, 256840, Otozomal Resesif (Kalıtsal Duysal ve Otonom Nöropati-Spastik Paraplejj) (CCT5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary sensory, with spastic paraplegia, 256840, Autosomal recessive (Hereditary sensory and autonomic neuropathy with spastic paraplegia) (CCT5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCT5	CCT5, KIAA0098, CCTE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, with spastic paraplegia, autosomal recessive (HSNSP) [MIM:256840]: A disease characterized by spastic paraplegia and progressive distal sensory neuropathy leading to mutilating ulcerations of the upper and lower limbs. {ECO:0000269|PubMed:16399879}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nöropati, Kalıtsal, Basınca Duyarlı Paralizi; Hnpp (Alesel Basınca Duyarlı Nöropati (Kalıtsal Nöropati With Liability To Pressure Palsies-HNPP)) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP (Hereditary neuropathy with liability to pressure palsies) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal, Basınca Duyarlı Paralizi; Hnpp (Alesel Basınca Duyarlı Nöropati (Kalıtsal Nöropati With Liability To Pressure Palsies-HNPP)) (MLPA)	NEUROPATHY, HEREDITARY, WITH LIABILITY TO PRESSURE PALSIES; HNPP (Hereditary neuropathy with liability to pressure palsies) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Kalıtsal, ± Yaşla İlişkili Makülа Dejenerasyonu, 608895, Otozomal Dominant; HNARMD (Kalıtsal Sensorimotor Nöropati-Hiperelastik Deri) (FBLN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, hereditary, with or without age-related macular degeneration, 608895, Autosomal dominant; HNARMD (Hereditary sensorimotor neuropathy with hyperelastic skin) (FBLN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBLN5	FBLN5, ARMD3, ADCL2, ARCL1A, HNARMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 1A (ARCL1A) [MIM:219100]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. The clinical spectrum of autosomal recessive cutis laxa is highly heterogeneous with respect to organ involvement and severity. Type I autosomal recessive cutis laxa is a specific, life-threatening disorder with organ involvement, lung atelectasis and emphysema, diverticula of the gastrointestinal and genitourinary systems, and vascular anomalies. Associated cranial anomalies, late closure of the fontanel, joint laxity, hip dislocation, and inguinal hernia have been observed but are uncommon. {ECO:0000269|PubMed:12189163, ECO:0000269|PubMed:16652333, ECO:0000269|PubMed:16691202, ECO:0000269|PubMed:17035250, ECO:0000269|PubMed:18185537}. Note=The disease is caused by mutations affecting the gene represented in this entry. Mutations affecting this gene can modify the phenotype of diseases caused by ELN mutations. {ECO:0000269|PubMed:19194475}.	EDTA Blood Sample
Nöropati, İnflamatuvar Demiyelinizan, 139393,Otozomal Dominant (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, inflammatory demyelinating, 139393, Autosomal dominant (Acute inflammatory demyelinating polyradiculoneuropathy) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, İnflamatuvar Demiyelinizan, 139393,Otozomal Dominant (Akut Enflamatuvar Demiyelinizan Poliradikülonöropati) (MLPA)	Neuropathy, inflammatory demyelinating, 139393, Autosomal dominant (Acute inflammatory demyelinating polyradiculoneuropathy) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Paraneoplastik Duysal (ELAVL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, paraneoplastic sensory (ELAVL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELAVL4	ELAVL4, HUD, PNEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Nöropati, Tekrarlayan, Basınç Paralizisi, 162500, Otozomal Dominant (Aileselbasınca Duyarlı Nöropati (Kalıtsal Nöropati With Liability To Pressure Palsies-HNPP)) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neuropathy, recurrent, with pressure palsies, 162500, Autosomal dominant (Hereditary neuropathy with liability to pressure palsies) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöropati, Tekrarlayan, Basınç Paralizisi, 162500, Otozomal Dominant (Aileselbasınca Duyarlı Nöropati (Kalıtsal Nöropati With Liability To Pressure Palsies-HNPP)) (MLPA)	Neuropathy, recurrent, with pressure palsies, 162500, Autosomal dominant (Hereditary neuropathy with liability to pressure palsies) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nötral Lipid Depo Hastalığı, Miyopati, 610717, Otozomal Resesif; NLSDM (Nötral Lipid Depo Miyopatisi) (PNPLA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutral lipid storage disease with myopathy, 610717, Autosomal recessive; NLSDM (Neutral lipid storage myopathy) (PNPLA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA2	PNPLA2, TTS2, ATGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutral lipid storage disease with myopathy (NLSDM) [MIM:610717]: Neutral lipid storage disorder (NLSD) with myopathy but without ichthyosis. NLSDs are characterized by the presence of triglyceride-containing cytoplasmic droplets in leukocytes and in other tissues, including bone marrow, skin, and muscle. Individuals with NLSDM did not show obesity, in spite of a defect in triglyceride degradation in fibroblasts and in marked triglyceride storage in liver, muscles, and other visceral cells. {ECO:0000269|PubMed:17187067}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötral Lipid Depo Hastalığı, Miyopati, 610717, Otozomal Resesif; NLSDM (Nötral Lipid Depo Miyopatisi) (Prenatal) (PNPLA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutral lipid storage disease with myopathy, 610717, Autosomal recessive; NLSDM (Neutral lipid storage myopathy) (PNPLA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPLA2	PNPLA2, TTS2, ATGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutral lipid storage disease with myopathy (NLSDM) [MIM:610717]: Neutral lipid storage disorder (NLSD) with myopathy but without ichthyosis. NLSDs are characterized by the presence of triglyceride-containing cytoplasmic droplets in leukocytes and in other tissues, including bone marrow, skin, and muscle. Individuals with NLSDM did not show obesity, in spite of a defect in triglyceride degradation in fibroblasts and in marked triglyceride storage in liver, muscles, and other visceral cells. {ECO:0000269|PubMed:17187067}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Alloimmün Neonatal (FCGR3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, alloimmune neonatal (FCGR3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR3B	FCGR3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nötropeni, Alloimmün Neonatal (Prenatal) (FCGR3B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, alloimmune neonatal (FCGR3B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FCGR3B	FCGR3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Siklik, 162800, Otozomal Dominant (Siklik Nötropeni) (ELANE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, cyclic, 162800, Autosomal dominant (Cyclic neutropenia) (ELANE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELANE	ELANE, ELA2, SCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital 1, autosomal dominant (SCN1) [MIM:202700]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269|PubMed:11001877, ECO:0000269|PubMed:11675333, ECO:0000269|PubMed:12091371, ECO:0000269|PubMed:14962902, ECO:0000269|PubMed:17436313, ECO:0000269|PubMed:18946670, ECO:0000269|PubMed:19036076, ECO:0000269|PubMed:19415009, ECO:0000269|PubMed:19927291, ECO:0000269|PubMed:20220065, ECO:0000269|PubMed:20803142, ECO:0000269|PubMed:21425445, ECO:0000269|PubMed:23463630}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, Siklik, 162800, Otozomal Dominant (Siklik Nötropeni (Prenatal) (ELANE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, cyclic, 162800, Autosomal dominant (Cyclic neutropenia) (ELANE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELANE	ELANE, ELA2, SCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutropenia, severe congenital 1, autosomal dominant (SCN1) [MIM:202700]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269|PubMed:11001877, ECO:0000269|PubMed:11675333, ECO:0000269|PubMed:12091371, ECO:0000269|PubMed:14962902, ECO:0000269|PubMed:17436313, ECO:0000269|PubMed:18946670, ECO:0000269|PubMed:19036076, ECO:0000269|PubMed:19415009, ECO:0000269|PubMed:19927291, ECO:0000269|PubMed:20220065, ECO:0000269|PubMed:20803142, ECO:0000269|PubMed:21425445, ECO:0000269|PubMed:23463630}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, İmmün Olmayan Kronik İdyopatik, Yetişkin, 607847, Otozomal Dominant (Yetişkin İdyopatik Nötropeni) (GFI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, nonimmune chronic idiopathic, of adults, 607847, Autosomal dominant (Adult idiopathic neutropenia) (GFI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFI1	GFI1, ZNF163, SCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant nonimmune chronic idiopathic neutropenia of adults (NI-CINA) [MIM:607847]: Relatively mild form of neutropenia diagnosed in adults, but predisposing to leukemia in a subset of patients. {ECO:0000269|PubMed:12778173}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, İmmün Olmayan Kronik İdyopatik, Yetişkin, 607847, Otozomal Dominant (Yetişkin İdyopatik Nötropeni) (Prenatal) (GFI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, nonimmune chronic idiopathic, of adults, 607847, Autosomal dominant (Adult idiopathic neutropenia) (GFI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFI1	GFI1, ZNF163, SCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dominant nonimmune chronic idiopathic neutropenia of adults (NI-CINA) [MIM:607847]: Relatively mild form of neutropenia diagnosed in adults, but predisposing to leukemia in a subset of patients. {ECO:0000269|PubMed:12778173}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital 1, Otozomal Dominant, 202700, Otozomal Dominant; SCN1 (Otozomal Dominant Ağır Konjenital Nötropeni) (ELANE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital 1, autosomal dominant, 202700, Autosomal dominant; SCN1 (Autosomal dominant severe congenital neutropenia) (ELANE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELANE	ELANE, ELA2, SCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital 1, autosomal dominant (SCN1) [MIM:202700]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269|PubMed:11001877, ECO:0000269|PubMed:11675333, ECO:0000269|PubMed:12091371, ECO:0000269|PubMed:14962902, ECO:0000269|PubMed:17436313, ECO:0000269|PubMed:18946670, ECO:0000269|PubMed:19036076, ECO:0000269|PubMed:19415009, ECO:0000269|PubMed:19927291, ECO:0000269|PubMed:20220065, ECO:0000269|PubMed:20803142, ECO:0000269|PubMed:21425445, ECO:0000269|PubMed:23463630}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Nötropeni, Ağır Konjenital 1, Otozomal Dominant, 202700, Otozomal Dominant; SCN1 (Otozomal Dominant Ağır Konjenital Nötropeni) (Prenatal) (ELANE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital 1, autosomal dominant, 202700, Autosomal dominant; SCN1 (Autosomal dominant severe congenital neutropenia) (ELANE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ELANE	ELANE, ELA2, SCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutropenia, severe congenital 1, autosomal dominant (SCN1) [MIM:202700]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269|PubMed:11001877, ECO:0000269|PubMed:11675333, ECO:0000269|PubMed:12091371, ECO:0000269|PubMed:14962902, ECO:0000269|PubMed:17436313, ECO:0000269|PubMed:18946670, ECO:0000269|PubMed:19036076, ECO:0000269|PubMed:19415009, ECO:0000269|PubMed:19927291, ECO:0000269|PubMed:20220065, ECO:0000269|PubMed:20803142, ECO:0000269|PubMed:21425445, ECO:0000269|PubMed:23463630}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital 2, Otozomal Dominant 613107, Otozomal Dominant; SCN2 (Otozomal Dominant Ağır Konjenital Nötropeni) (GFI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital 2, autosomal dominant, 613107, Autosomal dominant; SCN2 (Autosomal dominant severe congenital neutropenia) (GFI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GFI1	GFI1, ZNF163, SCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant nonimmune chronic idiopathic neutropenia of adults (NI-CINA) [MIM:607847]: Relatively mild form of neutropenia diagnosed in adults, but predisposing to leukemia in a subset of patients. {ECO:0000269	PubMed:12778173}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, Ağır Konjenital 2, Otozomal Dominant 613107, Otozomal Dominant; SCN2 (Otozomal Dominant Ağır Konjenital Nötropeni) (Prenatal) (GFI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital 2, autosomal dominant, 613107, Autosomal dominant; SCN2 (Autosomal dominant severe congenital neutropenia) (GFI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GFI1	GFI1, ZNF163, SCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dominant nonimmune chronic idiopathic neutropenia of adults (NI-CINA) [MIM:607847]: Relatively mild form of neutropenia diagnosed in adults, but predisposing to leukemia in a subset of patients. {ECO:0000269	PubMed:12778173}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital 3, Otozomal Resesif, 610738, Otozomal Resesif; SCN3 (Kostmann Sendromu) (HAX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital 3, autosomal recessive, 610738, Autosomal recessive; SCN3 (Kostmann syndrome) (HAX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HAX1	HAX1, SCN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital 3, autosomal recessive (SCN3) [MIM:610738]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. Some patients affected by severe congenital neutropenia type 3 have neurological manifestations such as psychomotor retardation and seizures. {ECO:0000269|PubMed:17187068, ECO:0000269|PubMed:18337561, ECO:0000269|PubMed:19796188, ECO:0000269|PubMed:20220065}. Note=The disease is caused by mutations affecting the gene represented in this entry. The clinical phenotype due to HAX1 deficiency appears to depend on the localization of the mutations and their influence on the transcript variants. Mutations affecting exclusively isoform 1 are associated with isolated congenital neutropenia, whereas mutations affecting both isoform 1 and isoform 5 are associated with additional neurologic symptoms (PubMed:18337561). {ECO:0000269|PubMed:18337561}.;	EDTA Blood Sample
Nötropeni, Ağır Konjenital 3, Otozomal Resesif, 610738, Otozomal Resesif; SCN3 (Kostmann Sendromu) (Prenatal) (HAX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital 3, autosomal recessive, 610738, Autosomal recessive; SCN3 (Kostmann syndrome) (HAX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HAX1	HAX1, SCN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutropenia, severe congenital 3, autosomal recessive (SCN3) [MIM:610738]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. Some patients affected by severe congenital neutropenia type 3 have neurological manifestations such as psychomotor retardation and seizures. {ECO:0000269|PubMed:17187068, ECO:0000269|PubMed:18337561, ECO:0000269|PubMed:19796188, ECO:0000269|PubMed:20220065}. Note=The disease is caused by mutations affecting the gene represented in this entry. The clinical phenotype due to HAX1 deficiency appears to depend on the localization of the mutations and their influence on the transcript variants. Mutations affecting exclusively isoform 1 are associated with isolated congenital neutropenia, whereas mutations affecting both isoform 1 and isoform 5 are associated with additional neurologic symptoms (PubMed:18337561). {ECO:0000269|PubMed:18337561}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital 4, Otozomal Resesif, 612541, Otozomal Resesif (Otozomal Resesif Ağır Konjenital Nötropeni, G6PC3 Eksikliğine Bağlı) (G6PC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital 4, autosomal recessive, 612541, Autosomal recessive (Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency) (G6PC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PC3	G6PC3, UGRP, SCN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dursun syndrome (DURSS) [MIM:612541]: A disease characterized by pulmonary arterial hypertension, cardiac abnormalities including secundum-type atrial septal defect, intermittent neutropenia, lymphopenia, monocytosis and anemia. {ECO:0000269	PubMed:20799326}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, Ağır Konjenital 4, Otozomal Resesif, 612541, Otozomal Resesif (Otozomal Resesif Ağır Konjenital Nötropeni, G6PC3 Eksikliğine Bağlı) (Prenatal) (G6PC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital 4, autosomal recessive, 612541, Autosomal recessive (Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency) (G6PC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	G6PC3	G6PC3, UGRP, SCN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dursun syndrome (DURSS) [MIM:612541]: A disease characterized by pulmonary arterial hypertension, cardiac abnormalities including secundum-type atrial septal defect, intermittent neutropenia, lymphopenia, monocytosis and anemia. {ECO:0000269	PubMed:20799326}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital, 5, Otozomal Resesif, 615285, Otozomal Resesif; SCN5 (Konjenital Nötropeni-Miyelofibroz-Nefromegali Sendromu) (VPS45 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital, 5, autosomal recessive, 615285, Autosomal recessive; SCN5 (Congenital neutropenia-myelofibrosis-nephromegaly syndrome) (VPS45 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS45	VPS45A, VPS45, SCN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nötropeni, Ağır Konjenital, 5, Otozomal Resesif, 615285, Otozomal Resesif; SCN5 (Konjenital Nötropeni-Miyelofibroz-Nefromegali Sendromu) (Prenatal) (VPS45 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital, 5, autosomal recessive, 615285, Autosomal recessive; SCN5 (Congenital neutropenia-myelofibrosis-nephromegaly syndrome) (VPS45 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS45	VPS45A, VPS45, SCN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital, 6, Otozomal Resesif, 616022, Otozomal Resesif; SCN6 (Otozomal Resesif Ağır Konjenital Nötropeni, JAGN1 Eksikliğine Bağlı) (JAGN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital, 6, autosomal recessive, 616022, Autosomal recessive; SCN6 (Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency) (JAGN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAGN1	JAGN1, SCN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Nötropeni, Ağır Konjenital, 6, Otozomal Resesif, 616022, Otozomal Resesif; SCN6 (Otozomal Resesif Ağır Konjenital Nötropeni, JAGN1 Eksikliğine Bağlı) (Prenatal) (JAGN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital, 6, autosomal recessive, 616022, Autosomal recessive; SCN6 (Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency) (JAGN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JAGN1	JAGN1, SCN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital, 7, Otozomal Resesif, 617014, Otozomal Resesif; SCN7 (Otozomal Resesif Ağır Konjenital Nötropeni, CSF3R Eksikliğine Bağlı) (CSF3R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital, 7, autosomal recessive, 617014, Autosomal recessive; SCN7 (Autosomal recessive severe congenital neutropenia due to CSF3R deficiency) (CSF3R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSF3R	CSF3R, GCSFR, SCN7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary neutrophilia (NEUTROPHILIA) [MIM:162830]: A form of lifelong, persistent neutrophilia, a condition characterized by an increase in the number of neutrophils in the blood. {ECO:0000269	PubMed:19620628}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, Ağır Konjenital, 7, Otozomal Resesif, 617014, Otozomal Resesif; SCN7 (Otozomal Resesif Ağır Konjenital Nötropeni, CSF3R Eksikliğine Bağlı) (Prenatal) (CSF3R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital, 7, autosomal recessive, 617014, Autosomal recessive; SCN7 (Autosomal recessive severe congenital neutropenia due to CSF3R deficiency) (CSF3R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSF3R	CSF3R, GCSFR, SCN7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hereditary neutrophilia (NEUTROPHILIA) [MIM:162830]: A form of lifelong, persistent neutrophilia, a condition characterized by an increase in the number of neutrophils in the blood. {ECO:0000269	PubMed:19620628}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötropeni, Ağır Konjenital, X'e Bağlı, 300299, X'e Bağlı Recessive; SCNX (X'e Bağlı Şiddetli Konjenital Nötropeni) (WAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutropenia, severe congenital, X-linked, 300299, X-linked recessive; SCNX (X-linked severe congenital neutropenia) (WAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital, X-linked (XLN) [MIM:300299]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269	PubMed:11242115}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötropeni, Ağır Konjenital, X'e Bağlı, 300299, X'e Bağlı Recessive; SCNX (X'e Bağlı Şiddetli Konjenital Nötropeni) (Prenatal) (WAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutropenia, severe congenital, X-linked, 300299, X-linked recessive; SCNX (X-linked severe congenital neutropenia) (WAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutropenia, severe congenital, X-linked (XLN) [MIM:300299]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. {ECO:0000269	PubMed:11242115}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nötrofil İmmün Yetmezlik Sendromu, 608203 (Nötrofil İmmun Yetmezlik Sendromu) (RAC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Neutrophil immunodeficiency syndrome, 608203 (Neutrophil immunodeficiency syndrome) (RAC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAC2	RAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutrophil immunodeficiency syndrome (NEUID) [MIM:608203]: An immunodeficiency syndrome due to defective neutrophils. Affected individuals present with leukocytosis, neutrophilia, severe recurrent bacterial infections and poor wound healing. {ECO:0000269	PubMed:10758162}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nötrofil İmmün Yetmezlik Sendromu, 608203 (Nötrofil İmmun Yetmezlik Sendromu) (Prenatal) (RAC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Neutrophil immunodeficiency syndrome, 608203 (Neutrophil immunodeficiency syndrome) (RAC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAC2	RAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neutrophil immunodeficiency syndrome (NEUID) [MIM:608203]: An immunodeficiency syndrome due to defective neutrophils. Affected individuals present with leukocytosis, neutrophilia, severe recurrent bacterial infections and poor wound healing. {ECO:0000269	PubMed:10758162}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nevus Komedonikus, Somatik, 617025; NC (Nevus Komedonikus Sendromu) (NEK9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nevus comedonicus, somatic, 617025, NC (Nevus comedonicus syndrome) (NEK9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK9	NEK9, NERCC1, LCCS10, APUG, NC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Nevus Sebaseus veya Yünsü Saç Nevus, Somatik, 162900 (Yünsü Saç Nevus) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nevus sebaceous or woolly hair nevus, somatic, 162900 (Woolly hair nevus) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Nevüs, Epidermal (Yünsü Saç Nevus) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	NEVUS, EPIDERMAL (Woolly hair nevus) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Nevüs, Epidermal (Yünsü Saç Nevus) (MLPA)	NEVUS, EPIDERMAL (Woolly hair nevus) (MLPA)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neurometanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nevüs, Epidermal, Somatik, 162900 (PIK3CA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nevus, epidermal, somatic, 162900 (PIK3CA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3CA	PIK3CA, CLOVE, MCAP, MCM, MCMTC, CWSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Newfoundland Rod-Kon Distrofisi, 607476 (Fundus Albipunctatus) (RLBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Newfoundland rod-cone dystrophy, 607476 (Fundus albipunctatus) (RLBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RLBP1	RLBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. {ECO:0000269|PubMed:10102299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nicolaides-Baraitser Sendromu, 601358, Otozomal Dominant; NCBRS (Entellektüel Yetersizlik-Seyrek Saç-Brakidaktili Sendromu (Nicolaides-Baraitser Sendromu) (SMARCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicolaides-Baraitser syndrome, 601358, Autosomal dominant; NCBRS (Intellectual disability-sparse hair-brachydactyly syndrome) (SMARCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCA2	SMARCA2, SNF2L2, NCBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schizophrenia (SCZD) [MIM:181500]: A complex, multifactorial psychotic disorder or group of disorders characterized by disturbances in the form and content of thought (e.g. delusions, hallucinations), in mood (e.g. inappropriate affect), in sense of self and relationship to the external world (e.g. loss of ego boundaries, withdrawal), and in behavior (e.g. bizarre or apparently purposeless behavior). Although it affects emotions, it is distinguished from mood disorders in which such disturbances are primary. Similarly, there may be mild impairment of cognitive function, and it is distinguished from the dementias in which disturbed cognitive function is considered primary. Some patients manifest schizophrenic as well as bipolar disorder symptoms and are often given the diagnosis of schizoaffective disorder. {ECO:0000269|PubMed:19363039}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Nicolaides-Baraitser Sendromu, 601358, Otozomal Dominant; NCBRS (Entellektüel Yetersizlik-Seyrek Saç-Brakidaktili Sendromu (Nicolaides-Baraitser Sendromu) (Prenatal) (SMARCA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nicolaides-Baraitser syndrome, 601358, Autosomal dominant; NCBRS (Intellectual disability-sparse hair-brachydactyly syndrome) (SMARCA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCA2	SMARCA2, SNF2L2, NCBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schizophrenia (SCZD) [MIM:181500]: A complex, multifactorial psychotic disorder or group of disorders characterized by disturbances in the form and content of thought (e.g. delusions, hallucinations), in mood (e.g. inappropriate affect), in sense of self and relationship to the external world (e.g. loss of ego boundaries, withdrawal), and in behavior (e.g. bizarre or apparently purposeless behavior). Although it affects emotions, it is distinguished from mood disorders in which such disturbances are primary. Similarly, there may be mild impairment of cognitive function, and it is distinguished from the dementias in which disturbed cognitive function is considered primary. Some patients manifest schizophrenic as well as bipolar disorder symptoms and are often given the diagnosis of schizoaffective disorder. {ECO:0000269|PubMed:19363039}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nikotin Bağımlılığı, Koruma, 188890 (CYP2A6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine addiction, protection from, 188890 (CYP2A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nikotin Bağımlılığı, Koruma, 188890 (MLPA)	Nicotine addiction, protection from, 188890 (MLPA)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Nikotin Bağımlılığı, Yatkınlık, 188890 (CHRNA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine addiction, susceptibility to, 188890 (CHRNA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA4	CHRNA4, ENFL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epilepsy, nocturnal frontal lobe, 1 (ENFL1) [MIM:600513]: An autosomal dominant focal epilepsy characterized by nocturnal seizures with hyperkinetic automatisms and poorly organized stereotyped movements. {ECO:0000269|PubMed:10563623, ECO:0000269|PubMed:14623738, ECO:0000269|PubMed:7550350}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nikotin Bağımlılığı, Karşı Koruma, 188890 (SLC6A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine dependence, protection against, 188890 (SLC6A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A3	SLC6A3, DAT1, PKDYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nikotin Bağımlılığı, Karşı Koruma, 188890 (GABBR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine dependence, protection against, 188890 (GABBR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABBR2	GPRS1, GABBR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nikotin Bağımlılığı, Yatkınlık, 188890 (GABBR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine dependence, susceptibility to, 188890 (GABBR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GABBR2	GPRS1, GABBR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nikotin Bağımlılığı, Yatkınlık, 612052 (CHRNA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nicotine dependence, susceptibility to, 612052 (CHRNA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA5	CHRNA5, LNCR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Niemann-Pick hastalığı tip C paneli (NPC1, NPC2) (MLPA)	Niemann-Pick disease type C panel (NPC1, NPC2) (MLPA)	NPC1, NPC2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Niemann-Pick hastalığı tip C paneli (NPC1, NPC2) (MLPA) (Prenatal)	Niemann-Pick disease type C panel (NPC1, NPC2) (MLPA) (Prenatal)	NPC1, NPC2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, Tip A, 257200, Otozomal Resesif (Niemann-Pick Hastalığı Tip A) (SMPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Niemann-Pick disease, type A, 257200, Autosomal recessive (Niemann-Pick disease type A) (SMPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMPD1	SMPD1, NPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Niemann-Pick disease A (NPDA) [MIM:257200]: An early- onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B. {ECO:0000269|PubMed:12556236, ECO:0000269|PubMed:1391960, ECO:0000269|PubMed:15221801, ECO:0000269|PubMed:15877209, ECO:0000269|PubMed:1618760, ECO:0000269|PubMed:1718266, ECO:0000269|PubMed:19405096, ECO:0000269|PubMed:2023926, ECO:0000269|PubMed:20386867, ECO:0000269|PubMed:8680412, ECO:0000269|PubMed:8693491, ECO:0000269|PubMed:9266408}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Niemann-Pick Hastalığı, Tip A, 257200, Otozomal Resesif (Niemann-Pick Hastalığı Tip A) (Prenatal) (SMPD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Niemann-Pick disease, type A, 257200, Autosomal recessive (Niemann-Pick disease type A) (SMPD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMPD1	SMPD1, NPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease A (NPDA) [MIM:257200]: An early- onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B. {ECO:0000269|PubMed:12556236, ECO:0000269|PubMed:1391960, ECO:0000269|PubMed:15221801, ECO:0000269|PubMed:15877209, ECO:0000269|PubMed:1618760, ECO:0000269|PubMed:1718266, ECO:0000269|PubMed:19405096, ECO:0000269|PubMed:2023926, ECO:0000269|PubMed:20386867, ECO:0000269|PubMed:8680412, ECO:0000269|PubMed:8693491, ECO:0000269|PubMed:9266408}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, Tip B, 607616, Otozomal Resesif (Niemann-Pick Hastalığı Tip B) (SMPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Niemann-Pick disease, type B, 607616, Autosomal recessive (Niemann-Pick disease type B) (SMPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMPD1	SMPD1, NPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Niemann-Pick disease A (NPDA) [MIM:257200]: An early- onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B. {ECO:0000269|PubMed:12556236, ECO:0000269|PubMed:1391960, ECO:0000269|PubMed:15221801, ECO:0000269|PubMed:15877209, ECO:0000269|PubMed:1618760, ECO:0000269|PubMed:1718266, ECO:0000269|PubMed:19405096, ECO:0000269|PubMed:2023926, ECO:0000269|PubMed:20386867, ECO:0000269|PubMed:8680412, ECO:0000269|PubMed:8693491, ECO:0000269|PubMed:9266408}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Niemann-Pick Hastalığı, Tip B, 607616, Otozomal Resesif (Niemann-Pick Hastalığı Tip B) (Prenatal) (SMPD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Niemann-Pick disease, type B, 607616, Autosomal recessive (Niemann-Pick disease type B) (SMPD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMPD1	SMPD1, NPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease A (NPDA) [MIM:257200]: An early- onset lysosomal storage disorder caused by failure to hydrolyze sphingomyelin to ceramide. It results in the accumulation of sphingomyelin and other metabolically related lipids in reticuloendothelial and other cell types throughout the body, leading to cell death. Niemann-Pick disease type A is a primarily neurodegenerative disorder characterized by onset within the first year of life, mental retardation, digestive disorders, failure to thrive, major hepatosplenomegaly, and severe neurologic symptoms. The severe neurological disorders and pulmonary infections lead to an early death, often around the age of four. Clinical features are variable. A phenotypic continuum exists between type A (basic neurovisceral) and type B (purely visceral) forms of Niemann-Pick disease, and the intermediate types encompass a cluster of variants combining clinical features of both types A and B. (ECO:0000269|PubMed:12556236, ECO:0000269|PubMed:1391960, ECO:0000269|PubMed:15221801, ECO:0000269|PubMed:15877209, ECO:0000269|PubMed:1618760, ECO:0000269|PubMed:1718266, ECO:0000269|PubMed:19405096, ECO:0000269|PubMed:2023926, ECO:0000269|PubMed:20386887, ECO:0000269|PubMed:8680412, ECO:0000269|PubMed:8693491, ECO:0000269|PubMed:9266408). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, C1, 257220, Otozomal Resesif; NPC1 (Niemann-Pick Hastalığı Tip C) (NPC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Niemann-Pick disease, type C1, 257220, Autosomal recessive; NPC1 (Niemann-Pick disease type C) (NPC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPC1	NPC1, NPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. (ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849, ECO:0000269|PubMed:9634529). Note=The disease is caused	EDTA Blood Sample
Niemann-Pick Hastalığı, C1, 257220, Otozomal Resesif; NPC1 (Niemann-Pick Hastalığı Tip C) (MLPA)	Niemann-Pick disease, type C1, 257220, Autosomal recessive; NPC1 (Niemann-Pick disease type C) (MLPA)	NPC1	NPC1, NPC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	30	Postnatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. (ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849, ECO:0000269|PubMed:9634529). Note=The disease is caused	EDTA Blood Sample
Niemann-Pick Hastalığı, C1, 257220, Otozomal Resesif; NPC1 (Niemann-Pick Hastalığı Tip C) (Prenatal) (NPC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Niemann-Pick disease, type C1, 257220, Autosomal recessive; NPC1 (Niemann-Pick disease type C) (NPC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPC1	NPC1, NPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. (ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849, ECO:0000269|PubMed:9634529). Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, C1, 257220, Otozomal Resesif; NPC1 (Niemann-Pick Hastalığı Tip C) (Prenatal) (MLPA)	Niemann-Pick disease, type C1, 257220, Autosomal recessive; NPC1 (Niemann-Pick disease type C) (Prenatal) (MLPA)	NPC1	NPC1, NPC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. (ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849, ECO:0000269|PubMed:9634529). Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, C2 Tipi, 607625, Otozomal Resesif; NPC2 (Niemann-Pick Hastalığı Tip C) (NPC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Niemann-pick disease, type C2, 607625, Autosomal recessive; NPC2 (Niemann-Pick disease type C) (NPC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPC2	NPC2, HE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	60	Postnatal	DISEASE: Niemann-Pick disease C2 (NPC2) [MIM:607625]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C2 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. (ECO:0000269|PubMed:11125141, ECO:0000269|PubMed:11567215, ECO:0000269|PubMed:12447927, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15937921, ECO:0000269|PubMed:16126423). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Niemann-Pick Hastalığı, C2 Tipi, 607625, Otozomal Resesif; NPC2 (Niemann-Pick Hastalığı Tip C) (MLPA)	Niemann-pick disease, type C2, 607625, Autosomal recessive; NPC2 (Niemann-Pick disease type C) (MLPA)	NPC2	NPC2, HE1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Niemann-Pick disease C2 (NPC2) [MIM:607625]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C2 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. {ECO:0000269|PubMed:11125141, ECO:0000269|PubMed:11567215, ECO:0000269|PubMed:12447927, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15937921, ECO:0000269|PubMed:16126423}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Niemann-Pick Hastalığı, C2 Tipi, 607625, Otozomal Resesif; NPC2 (Niemann-Pick Hastalığı Tip C) (Prenatal) (NPC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Niemann-pick disease, type C2, 607625, Autosomal recessive; NPC2 (Niemann-Pick disease type C) (NPC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPC2	NPC2, HE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C2 (NPC2) [MIM:607625]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C2 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. {ECO:0000269|PubMed:11125141, ECO:0000269|PubMed:11567215, ECO:0000269|PubMed:12447927, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15937921, ECO:0000269|PubMed:16126423}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, C2 Tipi, 607625, Otozomal Resesif; NPC2 (Niemann-Pick Hastalığı Tip C) (Prenatal) (MLPA)	Niemann-pick disease, type C2, 607625, Autosomal recessive; NPC2 (Niemann-Pick disease type C) (Prenatal) (MLPA)	NPC2	NPC2, HE1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C2 (NPC2) [MIM:607625]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C2 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. {ECO:0000269|PubMed:11125141, ECO:0000269|PubMed:11567215, ECO:0000269|PubMed:12447927, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15937921, ECO:0000269|PubMed:16126423}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Niemann-Pick Hastalığı, Tip D, 257220, Otozomal Resesif (Niemann-Pick Hastalığı Tip C) (NPC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Niemann-Pick disease, type D, 257220, Autosomal recessive (Niemann-Pick disease type C) (NPC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPC1	NPC1, NPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. {ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849,	EDTA Blood Sample
Niemann-Pick Hastalığı, Tip D, 257220, Otozomal Resesif (Niemann-Pick Hastalığı Tip C) (MLPA)	Niemann-Pick disease, type D, 257220, Autosomal recessive (Niemann-Pick disease type C) (MLPA)	NPC1	NPC1, NPC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. {ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849,	EDTA Blood Sample
Niemann-Pick Hastalığı, Tip D, 257220, Otozomal Resesif (Niemann-Pick Hastalığı Tip C) (Prenatal) (NPC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Niemann-Pick disease, type D, 257220, Autosomal recessive (Niemann-Pick disease type C) (NPC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPC1	NPC1, NPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. {ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Niemann-Pick Hastalığı, Tip D, 257220, Otozomal Resesif (Niemann-Pick Hastalığı Tip C) (Prenatal) (MLPA)	Niemann-Pick disease, type D, 257220, Autosomal recessive (Niemann-Pick disease type C) (Prenatal) (MLPA)	NPC1	NPC1, NPC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Niemann-Pick disease C1 (NPC1) [MIM:257220]: A lysosomal storage disorder that affects the viscera and the central nervous system. It is due to defective intracellular processing and transport of low-density lipoprotein derived cholesterol. It causes accumulation of cholesterol in lysosomes, with delayed induction of cholesterol homeostatic reactions. Niemann-Pick disease type C1 has a highly variable clinical phenotype. Clinical features include variable hepatosplenomegaly and severe progressive neurological dysfunction such as ataxia, dystonia and dementia. The age of onset can vary from infancy to late adulthood. An allelic variant of Niemann-Pick disease type C1 is found in people with Nova Scotia ancestry. Patients with the Nova Scotian clinical variant are less severely affected. (ECO:0000269|PubMed:10480349, ECO:0000269|PubMed:10521290, ECO:0000269|PubMed:10521297, ECO:0000269|PubMed:11182931, ECO:0000269|PubMed:11333381, ECO:0000269|PubMed:11349231, ECO:0000269|PubMed:11479732, ECO:0000269|PubMed:11545687, ECO:0000269|PubMed:11754101, ECO:0000269|PubMed:12401890, ECO:0000269|PubMed:12408188, ECO:0000269|PubMed:12554680, ECO:0000269|PubMed:12955717, ECO:0000269|PubMed:15774455, ECO:0000269|PubMed:16098014, ECO:0000269|PubMed:16126423, ECO:0000269|PubMed:16802107, ECO:0000269|PubMed:9211849, ECO:0000269|PubMed:9634529). Note=The disease is caused	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), 1A, X'e Bağlı, 310500, X'e Bağlı; CSNB1A (Konjenital Statisyoner Gece Körlüğü) (NYX gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1A, X-linked, 310500, X-linked recessive; CSNB1A (Congenital stationary night blindness) (NYX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NYX	NYX, CSNB1A, NBM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), Resesif, 1B, Otozomal, 257270, Otozomal Resesif; CSNB1B (Konjenital Statisyoner Gece Körlüğü) (GRM6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1B, autosomal recessive, 257270, Autosomal recessive; CSNB1B (Congenital stationary night blindness) (GRM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRM6	GRM6, MGLUR6, CSNB1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), 1C, Otozomal Resesif, 613216; CSNB1C (Konjenital Statisyoner Gece Körlüğü) (TRPM1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1C, autosomal recessive, 613216; CSNB1C (Congenital stationary night blindness) (TRPM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPM1	TRPM1, MLSN1, CSNB1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, 1C (CSNB1C) [MIM:613216]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269|PubMed:19878917, ECO:0000269|PubMed:19896109, ECO:0000269|PubMed:19896113). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), 1D, Otozomal Resesif, 613830, Otozomal Resesif; CSNB1D (Konjenital Statisyoner Gece Körlüğü) (SLC24A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1D, autosomal recessive, 613830, Autosomal recessive; CSNB1D (Congenital stationary night blindness) (SLC24A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC24A1	SLC24A1, NCKX1, CSNB1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, 1D (CSNB1D) [MIM:613830]: An autosomal recessive form of congenital stationary night blindness, a non-progressive retinal disorder characterized by impaired night vision. CSNB1D is characterized by a Riggs type of electroretinogram (proportionally reduced a- and b-waves). Patients have visual acuity within the normal range and no symptoms of myopia and/or nystagmus. (ECO:0000269	PubMed:20850105). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), 1E, Otozomal Resesif, 614565, Otozomal Resesif; CSNB1E (Konjenital Statisyoner Gece Körlüğü) (GPR179 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1E, autosomal recessive, 614565, Autosomal recessive; CSNB1E (Congenital stationary night blindness) (GPR179 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR179	GPR179, GPR15BL, GPR158L1, CSNB1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Tam), Resesif, 1F, Otozomal, 615058, Otozomal Resesif; CSNB1F (Konjenital Statisyoner Gece Körlüğü) (LRIT3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (complete), 1F, autosomal recessive, 615058, Autosomal recessive; CSNB1F (Congenital stationary night blindness) (LRIT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRIT3	LRIT3, FIGLER4, CSNB1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, 1F (CSNB1F) [MIM:615058]: An autosomal recessive form of congenital stationary night blindness, a non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269	PubMed:23246293). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner (Parsiyel), 2A, X'e Bağlı 300071, X'e Bağlı; CSNB2A (Konjenital Statisyoner Gece Körlüğü) (CACNA1F gen) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary (incomplete), 2A, X-linked, 300071, X-linked; CSNB2A (Congenital stationary night blindness) (CACNA1F gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1F	CACNA1F, CSNB2, CORDX3, CSNB2A, AIED, OA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, 2A (CSNB2A) [MIM:300071]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269|PubMed:11281458, ECO:0000269|PubMed:12111638, ECO:0000269|PubMed:12187427, ECO:0000269|PubMed:15897456, ECO:0000269|PubMed:22194652, ECO:0000269|PubMed:9662399, ECO:0000269|PubMed:9662400). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner, Otozomal Dominant 1, 610445, CSNBAD1 (Konjenital Statisyoner Gece Körlüğü) (RHO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary, autosomal dominant 1, 610445, CSNBAD1 (Congenital stationary night blindness) (RHO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHO	RHO, RP4, OPN2, CSNBAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 1 (CSNBAD1) [MIM:610445]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269|PubMed:7846071, ECO:0000269|PubMed:8358437, ECO:0000269|PubMed:9888392). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner, Otozomal Dominant 2, 163500, Otozomal Dominant; CSNBAD2 (Konjenital Statisyoner Gece Körlüğü) (PDE6B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary, autosomal dominant 2, 163500, Autosomal dominant; CSNBAD2 (Congenital stationary night blindness) (PDE6B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6B	PDE6B, PDEB, RP40, CSNBAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 2 (CSNBAD2) [MIM:163500]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269	PubMed:8075643). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner, Otozomal Dominant 3, 610444, Otozomal Dominant; CSNBAD3 (Konjenital Statisyoner Gece Körlüğü) (GNAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary, autosomal dominant 3, 610444, Autosomal dominant; CSNBAD3 (Congenital stationary night blindness) (GNAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAT1	GNAT1, CSNBAD3, CSNB1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 3 (CSNBAD3) [MIM:610444]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269|PubMed:17584859, ECO:0000269|PubMed:8673138). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner, Tip 1 G, 616389, Otozomal Resesif; CSNB1G (Konjenital Statisyoner Gece Körlüğü) (GNAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary, type 1G, 616389, Autosomal recessive; CSNB1G (Congenital stationary night blindness) (GNAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAT1	GNAT1, CSNBAD3, CSNB1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 3 (CSNBAD3) [MIM:610444]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. (ECO:0000269|PubMed:17584859, ECO:0000269|PubMed:8673138). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Gece Körlüğü, Konjenital Statisyoner, Tip 1 H, 617024, Otozomal Resesif; CSNB1H (GNB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Night blindness, congenital stationary, type 1H, 617024, Autosomal recessive; CSNB1H (GNB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNB3	GNB3, CSNB1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nijmegen Kırık Sendromu Benzeri Hastalık, 613078; NBSLD (Nijmegen Kırık Sendromu Gibi Bozukluk) (RAD50 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nijmegen breakage syndrome-like disorder, 613078; NBSLD (Nijmegen breakage syndrome-like disorder) (RAD50 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAD50	RAD50, NBSLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nijmegen breakage syndrome-like disorder (NBSLD) [MIM:613078]: A disorder similar to Nijmegen breakage syndrome and characterized by chromosomal instability, radiation sensitivity, microcephaly, growth retardation, short stature and bird-like face. Immunodeficiency is absent. (ECO:0000269	PubMed:19409520). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nijmegen Kırık Sendromu Benzeri Hastalık, 613078; NBSLD (Nijmegen Kırık Sendromu Gibi Bozukluk) (Prenatal) (RAD50 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nijmegen breakage syndrome-like disorder, 613078; NBSLD (Nijmegen breakage syndrome-like disorder) (RAD50 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAD50	RAD50, NBSLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nijmegen breakage syndrome-like disorder (NBSLD) [MIM:613078]: A disorder similar to Nijmegen breakage syndrome and characterized by chromosomal instability, radiation sensitivity, microcephaly, growth retardation, short stature and bird-like face. Immunodeficiency is absent. (ECO:0000269	PubMed:19409520). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nijmegen Kırık Sendromu, 251260, Otozomal Resesif (Nijmegen Kırık Sendromu) (NBN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nijmegen breakage syndrome, 251260, Autosomal recessive; NBS (Nijmegen breakage syndrome) (NBN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBN	NBN, NBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269	PubMed:14684699). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Nijmegen Kırık Sendromu, 251260, Otozomal Resesif (Nijmegen Kırık Sendromu) (Prenatal) (NBN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Nijmegen breakage syndrome, 251260, Autosomal recessive; NBS (Nijmegen breakage syndrome) (NBN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NBN	NBN, NBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. (ECO:0000269	PubMed:14684699). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nonaka Miyopati, 605820, Otozomal Resesif; NM (GNE Miyopati (Nonaka Miyopati)) (GNE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nonaka myopathy, 605820, Autosomal recessive; NM (GNE myopathy) (GNE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNE	GNE, GLCNE, IBM2, DMRV, NM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sialuria (SIALURIA) [MIM:269921]: In sialuria, free sialic acid accumulates in the cytoplasm and gram quantities of neuraminic acid are secreted in the urine. The metabolic defect involves lack of feedback inhibition of UDP-GlcNAc 2-epimerase by CMP-NeuSAc, resulting in constitutive overproduction of free NeuSAc. Clinical features include variable degrees of developmental delay, coarse facial features and hepatomegaly. Sialuria inheritance is autosomal dominant. (ECO:0000269|PubMed:10330343, ECO:0000269|PubMed:10356312, ECO:0000269|PubMed:2808337). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Nonarteritik Anterior İskemik Optik Nöropati, Yatkınlık, 258660, Otozomal Resesif (Bernard-Soulier Sendromu) (GP1BA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nonarteritic anterior ischemic optic neuropathy, susceptibility to, 258660, Autosomal recessive (Bernard-Soulier syndrome) (GP1BA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BA	GP1BA, BSS, BDPLT1, VWDP, BDPLT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bernard-Soulier syndrome A2, autosomal dominant (BSSA2) [MIM:153670]: A coagulation disorder characterized by mild to moderate bleeding tendency, thrombocytopenia, and an increased mean platelet volume. Some individuals have no symptoms. Mild bleeding tendencies manifest as epistaxis, gingival bleeding, menorrhagia, easy bruising, or prolonged bleeding after dental surgery. {ECO:0000269	PubMed:11272377}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Küçük Hücreli Akciğer Kanseri, Tirozin Kinaz İnhibitörü Yanıt, 211980, Otozomal Resesif (Ekzon 18 - 19 - 20 - 21) (EGFR geni) (Dizi Analizi) (Postnatal)	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980, Autosomal recessive (EGFR gene) (Sequence Analysis) (Postnatal)	EGFR	EGFR, NSBD2	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lung cancer (LNCR) [MIM:211980]: A common malignancy affecting tissues of the lung. The most common form of lung cancer is non-small cell lung cancer (NSCLC) that can be divided into 3 major histologic subtypes: squamous cell carcinoma, adenocarcinoma, and large cell lung cancer. NSCLC is often diagnosed at an advanced stage and has a poor prognosis. {ECO:0000269|PubMed:15118125, ECO:0000269|PubMed:16533793, ECO:0000269|PubMed:16672372}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Küçük Hücreli Akciğer Kanseri, Somatik (BRAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nonsmall cell lung cancer, somatic (BRAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Küçük Hücreli Akciğer Kanseri, Somatik, 211980 (IRF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nonsmall cell lung cancer, somatic, 211980 (IRF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF1	IRF1, MAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Gastric cancer (GASC) [MIM:613659]: A malignant disease which starts in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. The term gastric cancer or gastric carcinoma refers to adenocarcinoma of the stomach that accounts for most of all gastric malignant tumors. Two main histologic types are recognized, diffuse type and intestinal type carcinomas. Diffuse tumors are poorly differentiated infiltrating lesions, resulting in thickening of the stomach. In contrast, intestinal tumors are usually exophytic, often ulcerating, and associated with intestinal metaplasia of the stomach, most often observed in sporadic disease. {ECO:0000269|PubMed:10395927, ECO:0000269|PubMed:9679752}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Küçük Hücreli Dışı Akciğer Kanseri, Yatkınlık, 211980, Otozomal Resesif (Ekzon 18 - 19 - 20 - 21) (EGFR geni) (Dizi Analizi) (Postnatal)	Nonsmall cell lung cancer, susceptibility to, 211980, Autosomal recessive (EGFR gene) (Sequence Analysis) (Postnatal)	EGFR	EGFR, NSBD2	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lung cancer (LNCR) [MIM:211980]: A common malignancy affecting tissues of the lung. The most common form of lung cancer is non-small cell lung cancer (NSCLC) that can be divided into 3 major histologic subtypes: squamous cell carcinoma, adenocarcinoma, and large cell lung cancer. NSCLC is often diagnosed at an advanced stage and has a poor prognosis. {ECO:0000269|PubMed:15118125, ECO:0000269|PubMed:16533793, ECO:0000269|PubMed:16672372}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Noonan Sendromu 1, 163950, Otozomal Dominant; NS1 (Noonan Sendromu) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 1, 163950, Autosomal dominant; NS1 (Noonan syndrome) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. {ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 1, 163950, Otozomal Dominant; NS1 (Noonan Sendromu) (Prenatal) (PTPN11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 1, 163950, Autosomal dominant; NS1 (Noonan syndrome) (PTPN11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPN11	PTPN11, PTP2C, SHP2, NS1, JMML, METCDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: LEOPARD syndrome 1 (LPRD1) [MIM:151100]: A disorder characterized by lentigines, electrocardiographic conduction abnormalities, ocular hypertelorism, pulmonic stenosis, abnormalities of genitalia, retardation of growth, and sensorineural deafness. {ECO:0000269|PubMed:12058348, ECO:0000269|PubMed:14961557, ECO:0000269|PubMed:15121796, ECO:0000269|PubMed:15389709, ECO:0000269|PubMed:15520399, ECO:0000269|PubMed:15690106, ECO:0000269|PubMed:16679933, ECO:0000269|PubMed:24891296}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 10, 616564, Otozomal Dominant; NS10 (Noonan Sendromu) (LZTR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 10, 616564, Autosomal dominant; NS10 (Noonan syndrome) (LZTR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LZTR1	LZTR1, SWNTS2, NS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schwannomatosis 2 (SWNTS2) [MIM:615670]: A cancer predisposition syndrome in which patients develop multiple non- vestibular schwannomas, benign neoplasms that arise from Schwann cells of the cranial, peripheral, and autonomic nerves. {ECO:0000269	PubMed:24362817}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 10, 616564, Otozomal Dominant; NS10 (Noonan Sendromu) (Prenatal) (LZTR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 10, 616564, Autosomal dominant; NS10 (Noonan syndrome) (LZTR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LZTR1	LZTR1, SWNTS2, NS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schwannomatosis 2 (SWNTS2) [MIM:615670]: A cancer predisposition syndrome in which patients develop multiple non- vestibular schwannomas, benign neoplasms that arise from Schwann cells of the cranial, peripheral, and autonomic nerves. {ECO:0000269	PubMed:24362817}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 3, 609942; NS3 (Noonan Sendromu) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 3, 609942; NS3 (Noonan syndrome) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 3, 609942; NS3 (Noonan Sendromu) (MLPA)	Noonan syndrome 3, 609942; NS3 (Noonan syndrome) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 3, 609942; NS3 (Noonan Sendromu) (Prenatal) (KRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 3, 609942; NS3 (Noonan syndrome) (KRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 3, 609942; NS3 (Noonan Sendromu) (Prenatal) (MLPA)	Noonan syndrome 3, 609942; NS3 (Noonan syndrome) (Prenatal) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 4, 610733, Otozomal Dominant; NS4 (Noonan Sendromu) (SOS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 4, 610733, Autosomal dominant; NS4 (Noonan syndrome) (SOS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOS1	SOS1, GINGF, GF1, HGF, NS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome 4 (NS4) [MIM:610733]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyeslant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. Some patients with NS4 have polyarticular villonodular synovitis. {ECO:0000269|PubMed:17143282, ECO:0000269|PubMed:17143285, ECO:0000269|PubMed:19020799, ECO:0000269|PubMed:19438935, ECO:0000269|PubMed:19953625, ECO:0000269|PubMed:20673819, ECO:0000269|PubMed:20683980, ECO:0000269|PubMed:21387466}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Noonan Sendromu 4, 610733, Otozomal Dominant; NS4 (Noonan Sendromu) (Prenatal) (SOS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 4, 610733, Autosomal dominant; NS4 (Noonan syndrome) (SOS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOS1	SOS1, GINGF, GF1, HGF, NS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Noonan syndrome 4 (NS4) [MIM:610733]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyestant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. Some patients with NS4 have polyarticular villonodular synovitis. {ECO:0000269|PubMed:17143282, ECO:0000269|PubMed:17143285, ECO:0000269|PubMed:19020799, ECO:0000269|PubMed:19438935, ECO:0000269|PubMed:19953625, ECO:0000269|PubMed:20673819, ECO:0000269|PubMed:20683980, ECO:0000269|PubMed:21387466}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 5, 611553; NS5 (Noonan Sendromu) (RAF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 5, 611553; NS5 (Noonan syndrome) (RAF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAF1	RAF1, CRAF, NS5, CMD1NN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome 5 (NS5) [MIM:611553]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyestant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. {ECO:0000269|PubMed:17603482, ECO:0000269|PubMed:17603483, ECO:0000269|PubMed:20683980}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 5, 611553; NS5 (Noonan Sendromu) (Prenatal) (RAF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 5, 611553; NS5 (Noonan syndrome) (RAF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAF1	RAF1, CRAF, NS5, CMD1NN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Noonan syndrome 5 (NS5) [MIM:611553]: A form of Noonan syndrome, a disease characterized by short stature, facial dysmorphic features such as hypertelorism, a downward eyestant and low-set posteriorly rotated ears, and a high incidence of congenital heart defects and hypertrophic cardiomyopathy. Other features can include a short neck with webbing or redundancy of skin, deafness, motor delay, variable intellectual deficits, multiple skeletal defects, cryptorchidism, and bleeding diathesis. Individuals with Noonan syndrome are at risk of juvenile myelomonocytic leukemia, a myeloproliferative disorder characterized by excessive production of myelomonocytic cells. {ECO:0000269|PubMed:17603482, ECO:0000269|PubMed:17603483, ECO:0000269|PubMed:20683980}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 6, 613224, Otozomal Dominant; NS6 (Noonan Sendromu) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 6, 613224, Autosomal dominant; NS6 (Noonan syndrome) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 6, 613224, Otozomal Dominant; NS6 (Noonan Sendromu) (MLPA)	Noonan syndrome 6, 613224, Autosomal dominant; NS6 (Noonan syndrome) (MLPA)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 6, 613224, Otozomal Dominant; NS6 (Noonan Sendromu) (Prenatal) (NRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 6, 613224, Autosomal dominant; NS6 (Noonan syndrome) (NRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 6, 613224, Otozomal Dominant; NS6 (Noonan Sendromu) (Prenatal) (MLPA)	Noonan syndrome 6, 613224, Autosomal dominant; NS6 (Noonan syndrome) (Prenatal) (MLPA)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 7, 613706, Otozomal Dominant; NS7 (Noonan Sendromu) (BRAF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 7, 613706, Autosomal dominant; NS7 (Noonan syndrome) (BRAF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Noonan Sendromu 7, 613706, Otozomal Dominant; NS7 (Noonan Sendromu) (Prenatal) (BRAF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 7, 613706, Autosomal dominant; NS7 (Noonan syndrome) (BRAF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRAF	BRAF, NS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:12198537, ECO:0000269|PubMed:23263490, ECO:0000269|PubMed:24455489}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 8, 615355, Otozomal Dominant; NS8 (Noonan Sendromu) (RIT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 8, 615355, Autosomal dominant; NS8 (Noonan syndrome) (RIT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIT1	RIT1, RIT, ROC1, NS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Noonan Sendromu 8, 615355, Otozomal Dominant; NS8 (Noonan Sendromu) (Prenatal) (RIT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 8, 615355, Autosomal dominant; NS8 (Noonan syndrome) (RIT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RIT1	RIT1, RIT, ROC1, NS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu 9, 616559, Otozomal Dominant; NS9 (Noonan Sendromu) (SOS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome 9, 616559, Autosomal dominant; NS9 (Noonan syndrome) (SOS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOS2	SOS2, NS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Noonan Sendromu 9, 616559, Otozomal Dominant; NS9 (Noonan Sendromu) (Prenatal) (SOS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome 9, 616559, Autosomal dominant; NS9 (Noonan syndrome) (SOS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOS2	SOS2, NS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Sendromu Benzeri Hastalık, ± Juvenil Miyelomonositik Lösemi, 613563, Otozomal Dominant; NSLL (Noonan Sendromu Benzeri Bozukluk-Juvenil Miyelomonositik Lösemi) (CBL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563, Autosomal dominant; NSLL (Noonan syndrome-like disorder with juvenile myelomonocytic leukemia) (CBL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBL	CBL, CBL2, NSLL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL) [MIM:613563]: A syndrome characterized by a phenotype reminiscent of Noonan syndrome. Clinical features are highly variable, including facial dysmorphism, short neck, developmental delay, hyperextensible joints and thorax abnormalities with widely spaced nipples. The facial features consist of triangular face with hypertelorism, large low-set ears, ptosis, and flat nasal bridge. Some patients manifest cardiac defects. Some have an increased risk for certain malignancies, particularly juvenile myelomonocytic leukemia. {ECO:0000269	PubMed:20619386}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Noonan Sendromu Benzeri Hastalık, ± Jüvenil Miyelomonositik Lösemi, 613563, Otozomal Dominant; NSLL (Noonan Sendromu Benzeri Bozukluk-Jüvenil Miyelomonositik Lösemi) (Prenatal) (CBL gen) (Tüm Gen Dizi Analizi) (Prenatal)	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563, Autosomal dominant; NSLL (Noonan syndrome-like disorder with juvenile myelomonocytic leukemia) (CBL gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CBL	CBL, CBL2, NSLL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia (NSLL) [MIM:613563]: A syndrome characterized by a phenotype reminiscent of Noonan syndrome. Clinical features are highly variable, including facial dysmorphism, short neck, developmental delay, hyperextensible joints and thorax abnormalities with widely spaced nipples. The facial features consist of triangular face with hypertelorism, large low-set ears, ptosis, and flat nasal bridge. Some patients manifest cardiac defects. Some have an increased risk for certain malignancies, particularly juvenile myelomonocytic leukemia. {ECO:0000269	PubMed:20619386}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Noonan Benzeri Sendrom, Gevşek Anajen Saç, 607721, Otozomal Dominant; NSLH (Noonan Sendromu Benzeri Bozukluk-Gevşek Anajen Saç) (SHOC2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Noonan-like syndrome with loose anagen hair, 607721, Autosomal dominant; NSLH (Noonan syndrome-like disorder with loose anagen hair) (SHOC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHOC2	SHOC2, SIAA0862, SOC2, SUR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Noonan syndrome-like disorder with loose anagen hair (NSLH) [MIM:607721]: A syndrome characterized by Noonan dysmorphic features such as macrocephaly, high forehead, hypertelorism, palpebral ptosis, low-set and posteriorly rotated ears, short and webbed neck, pectus anomalies, in association with pluckable, sparse, thin and slow-growing hair. {ECO:0000269|PubMed:19684605, ECO:0000269|PubMed:25137548}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
NOR Poliaglütinasyon Sendromu, 111400 (A4GALT geni) (Tüm Gen Dizi Analizi) (Postnatal)	NOR polyagglutination syndrome, 111400 (A4GALT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	A4GALT	A4GALT, P1PK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Norrie Hastalığı, 310600, X'e Bağlı Resesif; ND (Norrie Hastalığı) (NDP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Norrie disease, 310600, X-linked recessive; ND (Norrie disease) (NDP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDP	NDP, ND, EVR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitreoretinopathy, exudative 2 (EVR2) [MIM:305390]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. {ECO:0000269|PubMed:16163268, ECO:0000269|PubMed:16970763, ECO:0000269|PubMed:17296899, ECO:0000269|PubMed:17325173, ECO:0000269|PubMed:8252044, ECO:0000269|PubMed:8946107, ECO:0000269|PubMed:9143917}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Norrie Hastalığı, 310600, X'e Bağlı Resesif; ND (Norrie Hastalığı) (Prenatal) (NDP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Norrie disease, 310600, X-linked recessive; ND (Norrie disease) (NDP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDP	NDP, ND, EVR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Vitreoretinopathy, exudative 2 (EVR2) [MIM:305390]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. {ECO:0000269|PubMed:16163268, ECO:0000269|PubMed:16970763, ECO:0000269|PubMed:17296899, ECO:0000269|PubMed:17325173, ECO:0000269|PubMed:8252044, ECO:0000269|PubMed:8946107, ECO:0000269|PubMed:9143917}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Norwalk Virüsü Enfeksiyonu, Direnç (FUT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Norwalk virus infection, resistance to (FUT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT2	FUT2, SE, B12QTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Nistagmus 1, Konjenital, X'e Bağlı, 310700, X'e Bağlı; NYS1 (FRMD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nystagmus 1, congenital, X-linked, 310700, X-linked; NYS1 (FRMD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRMD7	FRMD7, NYS1, XIPAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nystagmus congenital X-linked 1 (NYS1) [MIM:310700]: A condition defined as conjugated, spontaneous and involuntary ocular oscillations that appear at birth or during the first three months of life. Other associated features may include mildly decreased visual acuity, strabismus, astigmatism, and occasionally head nodding. {ECO:0000269|PubMed:17013395, ECO:0000269|PubMed:17397053, ECO:0000269|PubMed:17768376, ECO:0000269|PubMed:17893669, ECO:0000269|PubMed:17962394, ECO:0000269|PubMed:18087240, ECO:0000269|PubMed:18246032, ECO:0000269|PubMed:18431453, ECO:0000269|PubMed:21303855, ECO:0000269|PubMed:21365021, ECO:0000269|PubMed:22490987, ECO:0000269|PubMed:23946638}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Nistagmus 6, Konjenital, X'e Bağlı, 300814; NYS6 (X'e Bağlı Resesif Okiler Albinizm) (GPR143 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nystagmus 6, congenital, X-linked, 300814; NYS6 (X-linked recessive ocular albinism) (GPR143 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR143	GPR143, OA1, NYS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism ocular 1 (OA1) [MIM:300500]: Form of albinism affecting only the eye. Pigment of the hair and skin is normal or only slightly diluted. Eyes may be severely affected with photophobia and reduced visual acuity. Nystagmus or strabismus are often associated. The irides and fundus are depigmented. {ECO:0000269|PubMed:11214907, ECO:0000269|PubMed:16646960, ECO:0000269|PubMed:17822861, ECO:0000269|PubMed:17960122, ECO:0000269|PubMed:18078956, ECO:0000269|PubMed:8634705, ECO:0000269|PubMed:9529334, ECO:0000269|PubMed:9887374}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nistagmus 6, Konjenital, X'e Bağlı, 300814; NYS6 (X'e Bağlı Resesif Okiler Albinizm) (MLPA)	Nystagmus 6, congenital, X-linked, 300814; NYS6 (X-linked recessive ocular albinism) (MLPA)	GPR143	GPR143, OA1, NYS6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Albinism ocular 1 (OA1) [MIM:300500]: Form of albinism affecting only the eye. Pigment of the hair and skin is normal or only slightly diluted. Eyes may be severely affected with photophobia and reduced visual acuity. Nystagmus or strabismus are often associated. The irides and fundus are depigmented. {ECO:0000269|PubMed:11214907, ECO:0000269|PubMed:16646960, ECO:0000269|PubMed:17822861, ECO:0000269|PubMed:17960122, ECO:0000269|PubMed:18078956, ECO:0000269|PubMed:8634705, ECO:0000269|PubMed:9529334, ECO:0000269|PubMed:9887374}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nistagmus, İnfantil Periyodik Değişken, X'e Bağlı, 310700, X'e Bağlı (FRMD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Nystagmus, infantile periodic alternating, X-linked, 310700, X-linked (FRMD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRMD7	FRMD7, NYS1, XIPAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nystagmus congenital X-linked 1 (NYS1) [MIM:310700]: A condition defined as conjugated, spontaneous and involuntary ocular oscillations that appear at birth or during the first three months of life. Other associated features may include mildly decreased visual acuity, strabismus, astigmatism, and occasionally head nodding. {ECO:0000269|PubMed:17013395, ECO:0000269|PubMed:17397053, ECO:0000269|PubMed:17768376, ECO:0000269|PubMed:17893669, ECO:0000269|PubMed:17962394, ECO:0000269|PubMed:18087240, ECO:0000269|PubMed:18246032, ECO:0000269|PubMed:18431453, ECO:0000269|PubMed:21303855, ECO:0000269|PubMed:21365021, ECO:0000269|PubMed:22490987, ECO:0000269|PubMed:23946638}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite paneli (LEPR, POMC, LEP, SIM1, MC3R, MC4R) (MLPA)	Obesity paneli (LEPR, POMC, LEP, SIM1, MC3R, MC4R) (MLPA)	LEPR, POMC, LEP, SIM1, MC3R, MC4R	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Obesite, Prohormon Bozukluğuna Bağlı, 600955, Sporadik Vakalar (Obezite, Prohormon Konvertaz 1 Eksikliğine Bağlı) (PCSK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity with impaired prohormone processing, 600955, Isolated cases (Obesity due to prohormone convertase I deficiency) (PCSK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCSK1	PCSK1, NEC1, PC1, PC3, BMIQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Proprotein convertase 1 deficiency (PC1 deficiency) [MIM:600955]: Characterized by obesity, hypogonadism, hypoadrenalism, reactive hypoglycemia as well as marked small-intestinal absorptive dysfunction It is due to impaired processing of prohormones. {ECO:0000269|PubMed:14617756, ECO:0000269|PubMed:17596246, ECO:0000269|PubMed:9207799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Adrenal Yetmezlik ve POMC Eksikliğine Bağlı Kızıl Saç, 609734 (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (POMC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (Obesity due to pro-opiomelanocortin deficiency) (POMC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMC	POMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. {ECO:0000269	PubMed:12165561}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Obezite, Adrenal Yetmezlik ve POMC Eksikliğine Bağlı Kızıl Saç, 609734 (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (MLPA)	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (Obesity due to pro-opiomelanocortin deficiency) (MLPA)	POMC	POMC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. {ECO:0000269	PubMed:12165561}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Obezite, İlişkl, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (SDC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, association with, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (SDC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDC3	SDC3, SYND3, SDCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Otozomal Dominant, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (MC4R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, autosomal dominant, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (MC4R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC4R	MC4R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Obezite, Otozomal Dominant, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (MLPA)	Obesity, autosomal dominant, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (MLPA)	MC4R	MC4R	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Obezite, Erken Başlangıçlı, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (POMC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, early-onset, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (POMC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMC	POMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [ECO:0000269	PubMed:12165561]. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Obezite, Erken Başlangıçlı, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (MLPA)	Obesity, early-onset, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (MLPA)	POMC	POMC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [ECO:0000269	PubMed:12165561]. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Obezite, Hiperfaji ve Gelişme Geriliği (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (AKR1C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, hyperphagia, and developmental delay (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKR1C2	AKR1C2, DDH2, DD2, HAKRD, SRXY8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Hiperfaji ve Gelişme Geriliği (46,XY Cinsel Farklılaşma Bozukluğu, Testiküler 17,20-Dezmolaz Eksikliğine Bağlı) (Prenatal) (AKR1C2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Obesity, hyperphagia, and developmental delay (46,XY disorder of sex development due to testicular 17,20-desmolase deficiency) (AKR1C2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AKR1C2	AKR1C2, DDH2, DD2, HAKRD, SRXY8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Obezite, Hiperfaji ve Gelişme Geriliği, OBHD (NTRK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, hyperphagia, and developmental delay, 613886, OBHD (NTRK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTRK2	NTRK2, TRKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity, hyperphagia, and developmental delay (OBHD) [MIM:613886]: A disorder characterized by early-onset obesity, hyperphagia, and severe developmental delay in motor function, speech, and language. [ECO:0000269	PubMed:15494731]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Geç Başlangıçlı, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (AGRP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, late-onset, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (AGRP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGRP	AGRP, ART, AGRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Hafif, Erken Başlangıçlı, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (NR0B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, mild, early-onset, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (NR0B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR0B2	NR0B2, SHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [ECO:0000269	PubMed:11136233]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Morbid, Leptin Eksikliğine Bağlı, 614962, Otozomal Resesif (Obezite, Konjenital Leptin Eksikliğine Bağlı) (LEP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, morbid, due to leptin deficiency, 614962, Autosomal recessive (Obesity due to congenital leptin deficiency) (LEP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEP	LEP, OB, LEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leptin deficiency (LEPD) [MIM:614962]: A rare disease characterized by low levels of serum leptin, severe hyperphagia and intractable obesity from an early age. [ECO:0000269	PubMed:9500540]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Morbid, Leptin Eksikliğine Bağlı, 614962, Otozomal Resesif (Obezite, Konjenital Leptin Eksikliğine Bağlı) (MLPA)	Obesity, morbid, due to leptin deficiency, 614962, Autosomal recessive (Obesity due to congenital leptin deficiency) (MLPA)	LEP	LEP, OB, LEPD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leptin deficiency (LEPD) [MIM:614962]: A rare disease characterized by low levels of serum leptin, severe hyperphagia and intractable obesity from an early age. [ECO:0000269	PubMed:9500540]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Morbid, Leptin Reseptörü Eksikliğine Bağlı, 614963 (Obezite, Leptin Reseptör Geni Eksikliğine Bağlı) (LEPR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, morbid, due to leptin receptor deficiency, 614963 (Obesity due to leptin receptor gene deficiency) (LEPR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEPR	LEPR, OBR, LEPRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leptin receptor deficiency (LEPRD) [MIM:614963]: A rare disease characterized by normal levels of serum leptin, hyperphagia and severe obesity from an early age. Additional features include alterations in immune function, and delayed puberty due to hypogonadotropic hypogonadism. [ECO:0000269	PubMed:9537324]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Morbid, Leptin Reseptörü Eksikliğine Bağlı, 614963 (Obezite, Leptin Reseptör Gen Eksikliğine Bağlı) (MLPA)	Obesity, morbid, due to leptin receptor deficiency, 614963 (Obesity due to leptin receptor gene deficiency) (MLPA)	LEPR	LEPR, OBR, LEPRD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leptin receptor deficiency (LEPRD) [MIM:614963]: A rare disease characterized by normal levels of serum leptin, hyperphagia and severe obesity from an early age. Additional features include alterations in immune function, and delayed puberty due to hypogonadotropic hypogonadism. [ECO:0000269	PubMed:9537324]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Şiddetli, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (SIM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, severe, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (SIM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIM1	SIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Şiddetli, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (MLPA)	Obesity, severe, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (MLPA)	SIM1	SIM1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Obezite, Şiddetli, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (PPARG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, severe, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (PPARG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARG	PPARG, PPARG1, PPARG2, CIMT1, GLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glioma 1 (GLM1) [MIM:137800]: Gliomas are benign or malignant central nervous system neoplasms derived from glial cells. They comprise astrocytomas and glioblastoma multiforme that are derived from astrocytes, oligodendrogliomas derived from oligodendrocytes and ependymomas derived from ependymocytes. {ECO:0000269	PubMed:10851250}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry. Polymorphic PPARG alleles have been found to be significantly over-represented among a cohort of American patients with sporadic glioblastoma multiforme suggesting a possible contribution to disease susceptibility.;	EDTA Blood Sample
Obezite, Ağır, ve Tip 2 Diyabet, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (UCP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, severe, and type II diabetes, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (UCP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UCP3	UCP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Şiddetli Yatkınlık, BMIQ9, 602025 (MC3R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, severe, susceptibility to, BMIQ9, 602025 (MC3R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC3R	MC3R, BMIQ9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Şiddetli Yatkınlık, BMIQ9, 602025 (MLPA)	Obesity, severe, susceptibility to, BMIQ9, 602025 (MLPA)	MC3R	MC3R, BMIQ9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (GHRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (GHRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GHRL	GHRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (ENPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (ENPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENPP1	ENPP1, PDNP1, NPPS, MES1, PCA1, ARHR2, CCLED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. [ECO:0000269	PubMed:16186408]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (CARTPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (CARTPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARTPT	CARTPT, CART	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (UCP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (UCP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UCP1	UCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (ADRB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (ADRB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB3	ADRB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöryel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (ADRB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (ADRB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADRB2	ADRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık, 607514 (FFAR4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, 607514 (FFAR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FFAR4	FFAR4, G3FAR1, GPR120, PGR4, BMIQ10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık BMIQ11 İçin, 300306 (SLC6A14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, BMIQ11, 300306 (SLC6A14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A14	SLC6A14, OBX, BMIQ11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obezite, Yatkınlık BMIQ12 İçin, 612362 (PCSK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, BMIQ12, 612362 (PCSK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCSK1	PCSK1, NEC1, PC1, PC3, BMIQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Proprotein convertase 1 deficiency (PC1 deficiency) [MIM:600955]: Characterized by obesity, hypogonadism, hypoadrenalism, reactive hypoglycemia as well as marked small-intestinal absorptive dysfunction It is due to impaired processing of prohormones. {ECO:0000269|PubMed:14617756, ECO:0000269|PubMed:17595246, ECO:0000269|PubMed:9207799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Yatkınlık, BMIQ14, 612460, Otozomal Resesif (FTO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, BMIQ14, 612460, Autosomal recessive (FTO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FTO	FTO, GDFD, BMIQ14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [ECO:0000269|PubMed:26287746]. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A pathogenic intronic FTO variation (rs1421085) disrupts an evolutionarily conserved motif for ARID5B binding. Loss of ARID5B binding results in overexpression of two genes distal to FTO, IRX3 and IRX5. IRX3 and IRX5 overexpression shifts pre-adipocytes differentiation from brown to white fat cells, resulting in increased lipid storage and loss of mitochondrial thermogenesis. {ECO:0000269|PubMed:26287746}.;	EDTA Blood Sample
Obezite, BMIQ18, Yatkınlık, 615457, Otozomal Dominant (MRAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, BMIQ18, 615457, Autosomal dominant (MRAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MRAP2	MRAP2, C6orf117, BMIQ18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. [ECO:0000269	PubMed:23969016]. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Obezite, Yatkınlık BMIQ4 İçin, 607447 (UCP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, susceptibility to, BMIQ4, 607447 (UCP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UCP2	UCP2, BMIQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Obezite, Varyasyon, 601665, Otozomal Resesif, Otozomal Dominant, Multifaktöriyel (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (PPARGC1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obesity, variation in, 601665, Autosomal recessive, Autosomal dominant, Multifactorial (Obesity due to pro-opiomelanocortin deficiency) (PPARGC1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPARGC1B	PPARGC1B, PGC1B, PERC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obsesif-Kompulsif Bozukluk, 164230, Otozomal Dominant (SLC6A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obsessive-compulsive disorder, 164230, Autosomal dominant (SLC6A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A4	SLC6A4, HTT, OCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Obsesif-Kompulsif Bozukluk, Karşı Koruma, 164230, Otozomal Dominant (BDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obsessive-compulsive disorder, protection against, 164230, Autosomal dominant (BDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BDNF	BDNF, BULN2, ANON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. {ECO:0000269	PubMed:11840487}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Obsesif-Kompulsif Bozukluk, Yatkınlık, 164230, Otozomal Dominant; OCD (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Obsessive-compulsive disorder, susceptibility to, 164230, Autosomal dominant; OCD (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Oksipital Boynuz Sendromu, 304150, X'e Bağlı; OHS (Oksipital Boynuz Sendromu) (ATP7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Occipital horn syndrome, 304150, X-linked recessive; OHS (Occipital horn syndrome) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oksipital Boynuz Sendromu, 304150, X'e Bağlı; OHS (Oksipital Boynuz Sendromu) (MLPA)	Occipital horn syndrome, 304150, X-linked recessive; OHS (Occipital horn syndrome) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oksipital Boynuz Sendromu, 304150, X'e Bağlı; OHS (Oksipital Boynuz Sendromu) (Prenatal) (ATP7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Occipital horn syndrome, 304150, X-linked recessive; OHS (Occipital horn syndrome) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oksipital Boynuz Sendromu, 304150, X'e Bağlı; OHS (Oksipital Boynuz Sendromu) (Prenatal) (MLPA)	Occipital horn syndrome, 304150, X-linked recessive; OHS (Occipital horn syndrome) (Prenatal) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Gizli Maküler Distrofi, 613587, Otozomal Dominant; OCMD (Gizli Maküler Distrofi) (RP1L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Occult macular dystrophy, 613587, Autosomal dominant; OCMD (Occult macular dystrophy) (RP1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RP1L1	RP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Occult macular dystrophy (OCMD) [MIM:613587]: An inherited macular dystrophy characterized by progressive loss of macular function but normal ophthalmoscopic appearance. It is typically characterized by a central cone dysfunction leading to a loss of vision despite normal ophthalmoscopic appearance, normal fluorescein angiography, and normal full-field electroretinogram (ERGs), but the amplitudes of the focal macular ERGs and multifocal ERGs are significantly reduced at the central retina. {ECO:0000269|PubMed:20826268, ECO:0000269|PubMed:22605915}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oküler Albinizm, Tip 1, Nettleship-Falls Tipi, 300500, X'e Bağlı; OA1 (X'e Bağlı Resesif Oküler Albinizm) (GPR143 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ocular albinism, type I, Nettleship-Falls type, 300500, X-linked; OA1 (X-linked recessive ocular albinism) (GPR143 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR143	GPR143, OA1, NYS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Albinism ocular 1 (OA1) [MIM:300500]: Form of albinism affecting only the eye. Pigment of the hair and skin is normal or only slightly diluted. Eyes may be severely affected with photophobia and reduced visual acuity. Nystagmus or strabismus are often associated. The irides and fundus are depigmented. {ECO:0000269|PubMed:11214907, ECO:0000269|PubMed:16646960, ECO:0000269|PubMed:17822861, ECO:0000269|PubMed:17960122, ECO:0000269|PubMed:18978956, ECO:0000269|PubMed:8634705, ECO:0000269|PubMed:9529334, ECO:0000269|PubMed:9887374}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Oküler Albinizm, Tip 1, Nettleship-Falls Tipi, 300500, X'e Bağlı; OA1 (X'e Bağlı Resesif Oküler Albinizm) (MLPA)	Ocular albinism, type I, Nettleship-Falls type, 300500, X-linked; OA1 (X-linked recessive ocular albinism) (MLPA)	GPR143	GPR143, OA1, NYS6	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Albinism ocular 1 (OA1) [MIM:300500]: Form of albinism affecting only the eye. Pigment of the hair and skin is normal or only slightly diluted. Eyes may be severely affected with photophobia and reduced visual acuity. Nystagmus or strabismus are often associated. The irides and fundus are depigmented. {ECO:0000269|PubMed:11214907, ECO:0000269|PubMed:16646960, ECO:0000269|PubMed:17822861, ECO:0000269|PubMed:17960122, ECO:0000269|PubMed:18978956, ECO:0000269|PubMed:8634705, ECO:0000269|PubMed:9529334, ECO:0000269|PubMed:9887374}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Oküler malformasyonlar paneli (PAX6 11p13, SOX2 3q26, WT1, 11p13) (MLPA)	Ocular malformations, hereditary panel (PAX6 11p13, SOX2 3q26, WT1, 11p13) (MLPA)	PAX6 11p13, SOX2 3q26, WT1, 11p13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Okuloauriküler Sendrom, 612109, Otozomal Resesif; OCACS (Okuloauriküler Sendromu, Schorderet Tipi) (HMX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oculoauricular syndrome, 612109, Autosomal recessive; OCACS (Oculoauricular syndrome, Schorderet type) (HMX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMX1	HMX1, H6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculoauricular syndrome (OCACS) [MIM:612109]: A syndrome characterized by microphthalmia, microcornea, anterior segment dysgenesis, cataract, ocular coloboma, retinal pigment epithelium abnormalities, rod-cone dystrophy, and anomalies of the external ear. {ECO:0000269	PubMed:18423520}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Okülokütanöz albinizm paneli (OCA2 15q12; TYR 11q14.3) (MLPA)	Oculocutaneous albinism panel (OCA2 15q12; TYR 11q14.3) (MLPA)	OCA2 15q12; TYR 11q14.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Okülokütanöz albinizm paneli (OCA2 15q12; TYR 11q14.3) (MLPA) (Prenatal)	Oculocutaneous albinism panel (OCA2 15q12; TYR 11q14.3) (MLPA) (Prenatal)	OCA2 15q12; TYR 11q14.3	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oküdentodijital Displazi, 164200, Otozomal Dominant; ODDD (Oküdentodijital Displazi) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oculodentodigital dysplasia, 164200, Autosomal dominant; ODDD (Oculodentodigital dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. {ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Okülodentadijital Displazi, 164200, Otozomal Dominant; ODDD (Okulodentodijital Displazi) (Prenatal) (GJA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Oculodentodigital dysplasia, 164200, Autosomal dominant; ODDD (Oculodentodigital dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. (ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Okülodentodijital Displazi, Otozomal Resesif, 257850, Otozomal Resesif (Okulodentodijital Displazi) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oculodentodigital dysplasia, autosomal recessive, 257850, Autosomal recessive (Oculodentodigital dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. (ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Okülodentodijital Displazi, Otozomal Resesif, 257850, Otozomal Resesif (Okulodentodijital Displazi) (Prenatal) (GJA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Oculodentodigital dysplasia, autosomal recessive, 257850, Autosomal recessive (Oculodentodigital dysplasia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. (ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Okülofaringeal Müsküler Distrofi, 164300, Otozomal Dominant; OPMD (Okulofaringeal Musküler Distrofi) (Tekrar Sayısı)	Oculopharyngeal muscular dystrophy, 164300, Autosomal dominant; OPMD (Oculopharyngeal muscular dystrophy)(Repeat Analysis)	PABPN1	PABPN1, PABP2, PAB2	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Oculopharyngeal muscular dystrophy (OPMD) [MIM:164300]: A form of late-onset slowly progressive myopathy characterized by eyelid ptosis, dysphagia and, sometimes by other cranial and limb- muscle involvement. {ECO:0000269	PubMed:12673802}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Okülofaringeal Müsküler Distrofi, 164300, Otozomal Dominant; OPMD (Okulofaringeal Musküler Distrofi) (Prenatal) (Tekrar Sayısı)	Oculopharyngeal muscular dystrophy, 164300, Autosomal dominant; OPMD (Oculopharyngeal muscular dystrophy) (Prenatal)(Repeat Analysis)	PABPN1	PABPN1, PABP2, PAB2	Tekrar sayısı/ Repeat Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Oculopharyngeal muscular dystrophy (OPMD) [MIM:164300]: A form of late-onset slowly progressive myopathy characterized by eyelid ptosis, dysphagia and, sometimes by other cranial and limb- muscle involvement. {ECO:0000269	PubMed:12673802}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Odontohipofosfatazia, 146300, Otozomal Resesif, Otozomal Dominant (Odontohipofosfatazia) (ALPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Odontohypophosphatasia, 146300, Autosomal recessive, Autosomal dominant (Odontohypophosphatasia) (ALPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALPL	ALPL, HOPS, TNSALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatasia (HOPS) [MIM:146300]: A metabolic bone disease characterized by defective skeletal mineralization and biochemically by deficient activity of the tissue non-specific isoenzyme of alkaline phosphatase. Four forms are distinguished, depending on the age of onset: perinatal, infantile, childhood and adult type. The perinatal form is the most severe and is almost always fatal. The adult form is mild and characterized by recurrent fractures, osteomalacia, rickets, and loss of teeth. Some cases are asymptomatic, while some patients manifest dental features without skeletal manifestations (odontohypophosphatasia). {ECO:0000269|PubMed:10094560, ECO:0000269|PubMed:10332035, ECO:0000269|PubMed:10679946, ECO:0000269|PubMed:10690885, ECO:0000269|PubMed:10834525, ECO:0000269|PubMed:11438998, ECO:0000269|PubMed:11745997, ECO:0000269|PubMed:11760847, ECO:0000269|PubMed:11834095, ECO:0000269|PubMed:11855933, ECO:0000269|PubMed:11999978, ECO:0000269|PubMed:12815606, ECO:0000269|PubMed:12920074, ECO:0000269|PubMed:1408720, ECO:0000269|PubMed:15135428, ECO:0000269|PubMed:15694177, ECO:0000269|PubMed:3174660, ECO:0000269|PubMed:7833929, ECO:0000269|PubMed:8406453, ECO:0000269|PubMed:8954059, ECO:0000269|PubMed:9452105, ECO:0000269|PubMed:9747027.	EDTA Blood Sample
Odontoonikodermal Displazi, 257980, Otozomal Resesif; ODOD (Odonto-Oniko-Dermal Displazi) (WNT10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Odontoonychodermal dysplasia, 257980, Autosomal recessive; OODD (Odonto-onycho-dermal dysplasia) (WNT10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT10A	WNT10A, SSPS, STHAG4, OODD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 4 (STHAG4) [MIM:150400]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). In STHAG4, the upper lateral incisors are absent or peg-shaped. {ECO:0000269|PubMed:21484994, ECO:0000269|PubMed:22581971}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oftalmogenetik anomaliler paneli (FOXL2 TWIST1, FOXC1, FOXC2, ATR, PITX2, GPR143) (MLPA)	Oftalmogenetic anomalies panel (FOXL2, TWIST1, FOXC1, FOXC2, ATR, PITX2, GPR143) (MLPA)	FOXL2 , TWIST1, FOXC1, FOXC2, ATR, PITX2, GPR143	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ogden Sendromu, 300855, X'e Bağlı Resesif, X'e Bağlı Dominant; OGDNS (Erken Yaşlarında Görülmüş-Gelişimsel Gecikme-Kardiyak Aritmi Sendromu) (NAA10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ogden syndrome, 300855, X-linked recessive, X-linked dominant; OGDNS (Premature aging appearance-developmental delay-cardiac arrhythmia syndrome) (NAA10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAA10	NAA10, ARD1A, ARD1, TE2, NATD, OGDNS, MCOPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: N-terminal acetyltransferase deficiency (NATD) [MIM:300855]: An enzymatic deficiency resulting in postnatal growth failure with severe delays and dysmorphic features. It is clinically characterized by wrinkled forehead, prominent eyes, widely opened anterior and posterior fontanels, downsloping palpebral fissures, thickened lids, large ears, flared nares, hypoplastic alae, short columella, protruding upper lip, and micronetrognathia. There are also delayed closing of fontanels and broad great toes. Skin is characterized by redundancy or laxity with minimal subcutaneous fat, cutaneous capillary malformations, and very fine hair and eyebrows. Death results from cardiogenic shock following arrhythmia. {ECO:0000269	PubMed:21700266}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ogden Sendromu, 300855, X'e Bağlı Resesif, X'e Bağlı Dominant; OGDNS (Erken Yaşlarına Görünümü-Gelişimsel Gecikme-Kardiyak Aritmi Sendromu) (Prenatal) (NAA10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ogden syndrome, 300855, X-linked recessive, X-linked dominant; OGDNS (Premature aging appearance-developmental delay-cardiac arrhythmia syndrome) (NAA10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAA10	NAA10, ARD1A, ARD1, TE2, NATD, OGDNS, MCOPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: N-terminal acetyltransferase deficiency (NATD) [MIM:300855]: An enzymatic deficiency resulting in postnatal growth failure with severe delays and dysmorphic features. It is clinically characterized by wrinkled forehead, prominent eyes, widely opened anterior and posterior fontanels, downsloping palpebral fissures, thickened lids, large ears, flared nares, hypoplastic alae, short columella, protruding upper lip, and microretrognathia. There are also delayed closing of fontanels and broad great toes. Skin is characterized by redundancy or laxity with minimal subcutaneous fat, cutaneous capillary malformations, and very fine hair and eyebrows. Death results from cardiogenic shock following arrhythmia. {ECO:0000269	PubMed:21700266}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oguchi Hastalığı 1, 258100, Otozomal Resesif (Oguchi Hastalığı) (SAG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oguchi disease-1, 258100, Autosomal recessive (Oguchi disease) (SAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAG	SAG, RP47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 47 (RP47) [MIM:613758]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:29565049}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oguchi Hastalığı 1, 258100, Otozomal Resesif (Oguchi Hastalığı) (Prenatal) (SAG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Oguchi disease-1, 258100, Autosomal recessive (Oguchi disease) (SAG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SAG	SAG, RP47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Retinitis pigmentosa 47 (RP47) [MIM:613758]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:29565049}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oguchi Hastalığı-2, 613411 (Oguchi Hastalığı) (GRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oguchi disease-2, 613411 (Oguchi disease) (GRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRK1	GRK1, RHOK, RK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, Oguchi type 2 (CSNBO2) [MIM:613411]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. Congenital stationary night blindness Oguchi type is associated with fundus discoloration and abnormally slow dark adaptation. {ECO:0000269|PubMed:17070587, ECO:0000269|PubMed:9020843}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oguchi Hastalığı-2, 613411 (Oguchi Hastalığı) (Prenatal) (GRK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Oguchi disease-2, 613411 (Oguchi disease) (GRK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRK1	GRK1, RHOK, RK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Night blindness, congenital stationary, Oguchi type 2 (CSNBO2) [MIM:613411]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. Congenital stationary night blindness Oguchi type is associated with fundus discoloration and abnormally slow dark adaptation. {ECO:0000269|PubMed:17070587, ECO:0000269|PubMed:9020843}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ohdo Sendromu, X'e Bağlı, 300895, X'e Bağlı Resesif, OHDOX (Blefarofimozis-Mental Retardasyon Sendromu, MKB Tipi) (MED12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ohdo syndrome, X-linked, 300895, X-linked recessive, OHDOX (Blepharophimosis-intellectual disability syndrome, MKB type) (MED12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269	PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ohdo Sendromu, X'e Bağlı, 300895, X'e Bağlı Resesif, OHDOX (Blefarofimozis-Mental Retardasyon Sendromu, MKB Tipi) (Prenatal) (MED12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ohdo syndrome, X-linked, 300895, X-linked recessive, OHDOX (Blepharophimosis-intellectual disability syndrome, MKB type) (MED12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269	PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
OKT4 Epitop Eksikliği, 613949 (CD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	OKT4 epitope deficiency, 613949 (CD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD4	CD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OKT4 Epitop Eksikliği, 613949 (Prenatal) (CD4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	OKT4 epitope deficiency, 613949 (CD4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CD4	CD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Okur-Chung Nörogelişimsel Sendrom, 617062, Otozomal Dominant (CSNK2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Okur-Chung neurodevelopmental syndrome, 617062, Autosomal dominant (CSNK2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSNK2A1	CSNK2A1, CK2A1, OCNDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Okur-Chung Nörogelişimsel Sendrom, 617062, Otozomal Dominant (Prenatal) (CSNK2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Okur-Chung neurodevelopmental syndrome, 617062, Autosomal dominant (CSNK2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSNK2A1	CSNK2A1, CK2A1, OCNDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oligodonti-Kolorektal Kanser Sendromu, 608615, Otozomal Dominant; ODCRCS (Oligodonti-Kanser Yatkınlık Sendromu) (AXIN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oligodontia-colorectal cancer syndrome, 608615, Autosomal dominant; ODCRCS (Oligodontia-cancer predisposition syndrome) (AXIN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AXIN2	AXIN2, ODCRCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:11017067}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Oliver-Mcfarlane Sendromu, 275400, Otozomal Resesif; OMCS (Trikomegali-Retina Pigment Dejenerasyonu-Dwarfizm Sendromu (Oliver-Mcfarlane Sendromu)) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oliver-McFarlane syndrome, 275400, Autosomal recessive; OMCS (Trichomegaly-retina pigmentary degeneration-dwarfism syndrome) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA6	PNPLA6, NTE, SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. {ECO:0000269|PubMed:24357708, ECO:0000269|PubMed:25033069}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Oliver-Mcfarlane Sendromu, 275400, Otozomal Resesif; OMCS (Trikomegali-Retina Pigment Dejenerasyonu-Dwarfizm Sendromu (Oliver-Mcfarlane Sendromu)) (Prenatal) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Oliver-McFarlane syndrome, 275400, Autosomal recessive; OMCS (Trichomegaly-retina pigmentary degeneration-dwarfism syndrome) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPLA6	PNPLA6, NTE, SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. {ECO:0000269|PubMed:24357708, ECO:0000269|PubMed:25033069}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Olmsted Sendromu, 614594, Otozomal Dominant (Mutile Edici Palmoplantar Keratodermi - Periorifisial Keratotik Plaklar (Olmsted Sendromu)) (TRPV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Olmsted syndrome, 614594, Autosomal dominant (Mutilating palmoplantar keratoderma with periorificial keratotic plaques) (TRPV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV3	TRPV3, OLMS, FNEPPK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Palmoplantar keratoderma, non-epidermolytic, focal 2 (FNEPPK2) [MIM:614400]: A dermatological disorder characterized by non-epidermolytic, abnormal thickening of the skin on the palms and soles. Focal palmoplantar keratoderma consists of localized areas of hyperkeratosis located mainly on pressure points and sites of recurrent friction. {ECO:0000269	PubMed:25285920}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Olmsted Sendromu, 614594, Otozomal Dominant (Mutile Edici Palmoplantar Keratodermi - Periorifisial Keratotik Plaklar (Olmsted Sendromu)) (Prenatal) (TRPV3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Olmsted syndrome, 614594, Autosomal dominant (Mutilating palmoplantar keratoderma with periorificial keratotic plaques) (TRPV3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPV3	TRPV3, OLMS, FNEPPK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Palmoplantar keratoderma, non-epidermolytic, focal 2 (FNEPPK2) [MIM:614400]: A dermatological disorder characterized by non-epidermolytic, abnormal thickening of the skin on the palms and soles. Focal palmoplantar keratoderma consists of localized areas of hyperkeratosis located mainly on pressure points and sites of recurrent friction. {ECO:0000269	PubMed:25285920}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Olmsted Sendromu, X'e Bağlı, 300918, X'e Bağlı Resesif (Mutile Edici Palmoplantar Keratodermi - Periorifisial Keratotik Plaklar (Olmsted Sendromu)) (MBTPS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Olmsted syndrome, X-linked, 300918, X-linked recessive (Mutilating palmoplantar keratoderma with periorificial keratotic plaques) (MBTPS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MBTPS2	MBTPS2, S2P, IFAP, KFSDX, OLMSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Olmsted syndrome, X-linked (OLMSX) [MIM:300918]: A rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma and periorificial keratotic plaques with severe itching at all lesions. Diffuse alopecia, constriction of digits, and onychodystrophy have also been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes. {ECO:0000269	PubMed:22931912}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Olmsted Sendromu, X'e Bağlı, 300918, X'e Bağlı Resesif (Mutile Edici Palmoplantar Keratodermi - Periorifisial Keratotik Plaklar (Olmsted Sendromu)) (Prenatal) (MBTPS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Olmsted syndrome, X-linked, 300918, X-linked recessive (Mutilating palmoplantar keratoderma with periorificial keratotic plaques) (MBTPS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MBTPS2	MBTPS2, S2P, IFAP, KFSDX, OLMSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Olmsted syndrome, X-linked (OLMSX) [MIM:300918]: A rare congenital disorder characterized by bilateral mutilating palmoplantar keratoderma and periorificial keratotic plaques with severe itching at all lesions. Diffuse alopecia, constriction of digits, and onychodystrophy have also been reported. Infections and squamous cell carcinomas can arise on the keratotic areas. The digital constriction may progress to autoamputation of fingers and toes. {ECO:0000269	PubMed:22931912}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (RAG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHIDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B cells, and sparse thymic tissue on ultrasonography. {ECO:0000269	PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (RAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. (ECO:0000269	PubMed:18463379). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (DCLRE1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (DCLRE1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. (ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (MLPA)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (MLPA)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. (ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (Prenatal) (RAG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. (ECO:0000269	PubMed:18463379). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (Prenatal) (RAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. (ECO:0000269	PubMed:18463379). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (Prenatal) (DCLRE1C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (DCLRE1C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. (ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omenn Sendromu, 603554, Otozomal Resesif (Omenn Sendromu) (Prenatal) (MLPA)	Omenn syndrome, 603554, Autosomal recessive (Omenn syndrome) (Prenatal) (MLPA)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. (ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omeprazol Yavaş Metabolizma, 609535, Otozomal Resesif (Klopidogrel Direnci) (CYP2C19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Omeprazole poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (CYP2C19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C19	CYP2C, CYP2C19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Omeprazol Yavaş Metabolizma, 609535, Otozomal Resesif (Klopidogrel Direnci) (MLPA)	Omeprazole poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (MLPA)	CYP2C19	CYP2C, CYP2C19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Omodisplazi 1, 258315, Otozomal Resesif, OMOD1 (Omodisplazi) (GPC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Omodysplasia 1, 258315, Autosomal recessive; OMOD1 (Omodysplasia) (GPC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPC6	GPC6, OMIMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Omodysplasia 1 (OMOD1) [MIM:258315]: A rare autosomal recessive skeletal dysplasia characterized by facial dysmorphism and severe congenital micromelia with shortening and distal tapering of the humeri and femora, to give a club-like appearance. Typical facial features include a prominent forehead, frontal bossing, short nose with a depressed broad bridge, short columella, anteverted nostrils, long philtrum, and small chin. (ECO:0000269	PubMed:19481194). Note=The disease is caused by mutations affecting the gene represented in this entry. Point mutations leading to protein truncation, as well as larger genomic rearrangements resulting in exon deletions, have been found in family segregating omodysplasia type 1. All mutations identified in individuals affected by omodysplasia could lead to the absence of a functional protein, the mutant RNAs being suspected to be nonsense-mediated mRNA decay (NMD) targets. Even if the mRNA escapes NMD and is translated, all mutations are expected to disrupt the three-dimensional protein structure and often to abolish multiple highly conserved cysteine residues.;	EDTA Blood Sample
Omodisplazi 1, 258315, Otozomal Resesif, OMOD1 (Omodisplazi) (Prenatal) (GPC6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Omodysplasia 1, 258315, Autosomal recessive; OMOD1 (Omodysplasia) (GPC6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPC6	GPC6, OMIMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Omodysplasia 1 (OMOD1) [MIM:258315]: A rare autosomal recessive skeletal dysplasia characterized by facial dysmorphism and severe congenital micromelia with shortening and distal tapering of the humeri and femora, to give a club-like appearance. Typical facial features include a prominent forehead, frontal bossing, short nose with a depressed broad bridge, short columella, anteverted nostrils, long philtrum, and small chin. (ECO:0000269	PubMed:19481194). Note=The disease is caused by mutations affecting the gene represented in this entry. Point mutations leading to protein truncation, as well as larger genomic rearrangements resulting in exon deletions, have been found in family segregating omodysplasia type 1. All mutations identified in individuals affected by omodysplasia could lead to the absence of a functional protein, the mutant RNAs being suspected to be nonsense-mediated mRNA decay (NMD) targets. Even if the mRNA escapes NMD and is translated, all mutations are expected to disrupt the three-dimensional protein structure and often to abolish multiple highly conserved cysteine residues.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Omphalosel, 1p31.3 Duplikasyonuna Bağlı, 164750, Otozomal Dominant (Omfalosel) (2200)	Omphalocele due to duplication of 1p31.3, 164750, Autosomal dominant (Omphalocele) (440)	.	OPHLC, C1DUPp31.3, DUP1p31.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Omphalosel, 1p31.3 Duplikasyonuna Bağlı, 164750, Otozomal Dominant (Omfalosel) (Prenatal)	Omphalocele due to duplication of 1p31.3, 164750, Autosomal dominant (Omphalocele) (Prenatal)	.	OPHLC, C1DUPp31.3, DUP1p31.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Oosit Olgunlaşırma Defekti 1, 615774, Otozomal Resesif; OOMD1 (Kadın Infertilitesi, Zona Pellucida Defektine Bağlı) (ZP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Oocyte maturation defect 1, 615774, Autosomal recessive; OOMD1 (Female infertility due to zona pellucida defect) (ZP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZP1	ZP1, OOMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oocyte maturation defect (OOMD) [MIM:615774]: An infertility disorder caused by defective oocyte maturation that results in abnormal eggs lacking a zona pellucida. Affected females have normal menstrual cycles and sex hormone levels, no obstruction in the fallopian tubes or abnormalities of the uterus or adnexa. (ECO:0000269	PubMed:24670168). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Opitz GBBB Sendromu, Tip I, 300000, X'e Bağlı Resesif; GBBB1 (Opitz G / BBB Sendromu) (MID1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Opitz GBBB syndrome, type I, 300000, X-linked recessive; GBBB1 (Opitz G/BBB syndrome) (MID1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MID1	MID1, OGS1, BBBG1, FXY, OSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Opitz GBBB syndrome 1 (GBBB1) [MIM:300000]: A congenital midline malformation syndrome characterized by hypertelorism, genital-urinary defects such as hypospadias in males and splayed labia in females, cleft lip/palate, laryngotracheoesophageal abnormalities, imperforate anus, developmental delay and congenital heart defects. {ECO:0000269|PubMed:11030761, ECO:0000269|PubMed:15558842, ECO:0000269|PubMed:9354791, ECO:0000269|PubMed:9718340}. Note=The disease is caused by mutations affecting the gene represented in this entry. MID1 mutations produce proteins with a decreased affinity for microtubules.	EDTA Blood Sample
Opitz GBBB Sendromu, Tip I, 300000, X'e Bağlı Resesif; GBBB1 (Opitz G / BBB Sendromu) (Prenatal) (MID1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Opitz GBBB syndrome, type I, 300000, X-linked recessive; GBBB1 (Opitz G/BBB syndrome) (MID1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MID1	MID1, OGS1, BBBG1, FXY, OSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Opitz GBBB syndrome 1 (GBBB1) [MIM:300000]: A congenital midline malformation syndrome characterized by hypertelorism, genital-urinary defects such as hypospadias in males and splayed labia in females, cleft lip/palate, laryngotracheoesophageal abnormalities, imperforate anus, developmental delay and congenital heart defects. {ECO:0000269|PubMed:11030761, ECO:0000269|PubMed:15558842, ECO:0000269|PubMed:9354791, ECO:0000269|PubMed:9718340}. Note=The disease is caused by mutations affecting the gene represented in this entry. MID1 mutations produce proteins with a decreased affinity for microtubules.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Opitz GBBB Sendromu, Tip 2, 145410, Otozomal Dominant; GBBB2 (Opitz G / BBB Sendromu) (SPECC1L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Opitz GBBB syndrome, type II, 145410, Autosomal dominant; GBBB2 (Opitz G/BBB syndrome) (SPECC1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPECC1L	SPECC1L, KIAA0376, OBLFC1, GBBB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Opitz GBBB syndrome 2 (GBBB2) [MIM:145410]: A form of Opitz GBBB syndrome, a congenital midline malformation syndrome characterized by hypertelorism, genital-urinary defects such as hypospadias in males and splayed labia in females, cleft lip/palate, laryngotracheoesophageal abnormalities, imperforate anus, developmental delay and congenital heart defects. {ECO:0000269|PubMed:25412741}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Opitz GBBB Sendromu, Tip 2, 145410, Otozomal Dominant; GBBB2 (Opitz G / BBB Sendromu) (Prenatal) (SPECC1L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Opitz GBBB syndrome, type II, 145410, Autosomal dominant; GBBB2 (Opitz G/BBB syndrome) (SPECC1L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPECC1L	SPECC1L, KIAA0376, OBLFC1, GBBB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Opitz GBBB syndrome 2 (GBBB2) [MIM:145410]: A form of Opitz GBBB syndrome, a congenital midline malformation syndrome characterized by hypertelorism, genital-urinary defects such as hypospadias in males and splayed labia in females, cleft lip/palate, laryngotracheoesophageal abnormalities, imperforate anus, developmental delay and congenital heart defects. {ECO:0000269|PubMed:25412741}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Opitz-Kaveggia Sendromu, 305450, X'e Bağlı Resesif; OKS (FG Sendromu Tip 1) (MED12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Opitz-Kaveggia syndrome, 305450, X-linked recessive; OKS (FG syndrome type 1) (MED12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269|PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Opitz-Kaveggia Sendromu, 305450, X'e Bağlı Resesif; OKS (FG Sendromu Tip 1) (Prenatal) (MED12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Opitz-Kaveggia syndrome, 305450, X-linked recessive; OKS (FG syndrome type 1) (MED12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED12	MED12, TNRC11, TRAP230, HOPA, KIAA0192, OKS, FGS1, OHDOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ohdo syndrome, X-linked (OHDOX) [MIM:300895]: A syndrome characterized by mental retardation, feeding problems, and distinctive facial appearance with coarse facial features, severe blepharophimosis, ptosis, a bulbous nose, micrognathia and a small mouth. Dental hypoplasia and deafness can be considered as common manifestations of the syndrome. Male patients show cryptorchidism and scrotal hypoplasia. {ECO:0000269|PubMed:23395478}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Opsismodisplazi, 258480, Otozomal Resesif; OPSMD (Opsismodisplazi) (INPPL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Opsismodysplasia, 258480, Autosomal recessive; OPSMD (Opsismodysplasia) (INPPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INPPL1	INPPL1, OPSMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:12086927, ECO:0000269|PubMed:15687335}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Opsismodisplazi, 258480, Otozomal Resesif; OPSMD (Opsismodisplazi) (Prenatal) (INPPL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Opsismodysplasia, 258480, Autosomal recessive; OPSMD (Opsismodysplasia) (INPPL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	INPPL1	INPPL1, OPSMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:12086927, ECO:0000269|PubMed:15687335}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Atrofi 1, 165500, Otozomal Dominant; OPA1 (Otozomal Dominant Optik Atrofi, Klasik Form) (OPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 1, 165500, Autosomal dominant; OPA1 (Autosomal dominant optic atrophy, classic form) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Optik Atrofi 10, ± Ataksi ve Mental Retardasyon, Nöbet, 616732, Otozomal Resesif; OPA10 (Otozomal Resesif İzole Optik Atrofi) (RTN4IP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732, Autosomal recessive; OPA10 (Autosomal recessive isolated optic atrophy) (RTN4IP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTN4IP1	RTN4IP1, NIMP, OPA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Optik Atrofi 10, ± Ataksi ve Mental Retardasyon, Nöbet, 616732, Otozomal Resesif; OPA10 (Otozomal Resesif İzole Optik Atrofi) (Prenatal) (RTN4IP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Optic atrophy 10 with or without ataxia, mental retardation, and seizures, 616732, Autosomal recessive; OPA10 (Autosomal recessive isolated optic atrophy) (RTN4IP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RTN4IP1	RTN4IP1, NIMP, OPA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Atrofi 11, 617302, Otozomal Resesif; OPA11 (Otozomal Resesif İzole Optik Atrofi) (YME1L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 11, 617302, Autosomal recessive; OPA11 (Autosomal recessive isolated optic atrophy) (YME1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	YME1L1	YME1L1, YME1L, PAMP, OPA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Optik Atrofi 3-Katarakt, 165300, Otozomal Dominant; OPA3 (Otozomal Dominant Optik Atrofi ve Katarakt) (OPA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 3 with cataract, 165300, Autosomal dominant; OPA3 (Autosomal dominant optic atrophy and cataract) (OPA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA3	OPA3, MGA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Optic atrophy 3 (OPA3) [MIM:165300]: A condition that features progressive visual loss in association with optic atrophy. Atrophy of the optic disk indicates a deficiency in the number of nerve fibers which arise in the retina and converge to form the optic disk, optic nerve, optic chiasm and optic tracts. OPA3 is associated with cataract and a neurologic disorder characterized by extrapyramidal signs and ataxia. {ECO:0000269|PubMed:15342707}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Optik Atrofi 7, 612989, Otozomal Resesif; OPA7 (Otozomal Resesif Optik Atrofi, OPA7 Tipi) (TMEM126A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 7, 612989, Autosomal recessive; OPA7 (Autosomal recessive optic atrophy, OPA7 type) (TMEM126A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM126A	TMEM126A, OPA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Optik Atrofi 9, 616289, Otozomal Resesif; OPA9 (Otozomal Resesif İzole Optik Atrofi) (ACO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy 9, 616289, Autosomal recessive; OPA9 (Autosomal recessive isolated optic atrophy) (ACO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACO2	ACO2, ICRD, OPA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile cerebellar-retinal degeneration (ICRD) [MIM:614559]: A severe autosomal recessive neurodegenerative disorder characterized by onset between ages 2 and 6 months of truncal hypotonia, athetosis, seizures, and ophthalmologic abnormalities, particularly optic atrophy and retinal degeneration. Affected individuals show profound psychomotor retardation, with only some achieving rolling, sitting, or recognition of family. Brain MRI shows progressive cerebral and cerebellar degeneration. {ECO:0000269|PubMed:22405087, ECO:0000269|PubMed:25351951}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Optik Atrofi Plus Sendromu, 125250, Otozomal Dominant (Otozomal Dominant Optik Atrofi Plus Sendromu) (OPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic atrophy plus syndrome, 125250, Autosomal dominant (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Optik Atrofi ± Sağırlık, Oftalmopleji, Miyopati, Ataksi ve Nöropati (Otozomal Dominant Optik Atrofi Plus Sendromu) (OPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	OPTIC ATROPHY WITH OR WITHOUT DEAFNESS, OPHTHALMOPLEGIA, MYOPATHY, ATAXIA, AND NEUROPATHY (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Optik Atrofi + Sağırlık, Oftalmopleji, Miyopati, Ataksi ve Nöropati (Otozomal Dominant Optik Atrofi Plus Sendromu) (Prenatal) (OPA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	OPTIC ATROPHY WITH OR WITHOUT DEAFNESS, OPHTHALMOPLEGIA, MYOPATHY, ATAXIA, AND NEUROPATHY (Autosomal dominant optic atrophy plus syndrome) (OPA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OPA1	OPA1, NTG, NPG, BERHS, MTDPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dominant optic atrophy plus syndrome (DOA+) [MIM:125250]: A neurologic disorder characterized most commonly by an insidious onset of visual loss and sensorineural hearing loss in childhood with variable presentation of other clinical manifestations including progressive external ophthalmoplegia, muscle cramps, hyperreflexia, and ataxia. There appears to be a wide range of intermediate phenotypes. {ECO:0000269|PubMed:15531309, ECO:0000269|PubMed:16240368, ECO:0000269|PubMed:18065439, ECO:0000269|PubMed:18158317, ECO:0000269|PubMed:18195150, ECO:0000269|PubMed:23387428}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Disk Anomalileri, Retina ve / Veya Makula Distrofisi, 212550, Otozomal Resesif; ODRMD (Kolobomatöz Optik Disk-Maküler Atrofi-Korioretinopati Sendromu) (SIX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic disc anomalies with retinal and/or macular dystrophy, 212550, Autosomal recessive; ODRMD (Colobomatous optic disc-macular atrophy-chorioretinopathy syndrome) (SIX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX6	SIX6, ODRMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Optik Sinir Hipoplazisi ve Merkezi Sinir Sistemi Bozuklukları, 206900, Otozomal Dominant (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (SOX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900, Autosomal dominant (Anophthalmia/microphthalmia-esophageal atresia syndrome) (SOX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX2	SOX2, MCOPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Optik Sinir Hipoplazisi ve Merkezi Sinir Sistemi Bozuklukları, 206900, Otozomal Dominant (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (MLPA)	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900, Autosomal dominant (Anophthalmia/microphthalmia-esophageal atresia syndrome) (MLPA)	SOX2	SOX2, MCOPS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Optik Sinir Hipoplazisi ve Merkezi Sinir Sistemi Bozuklukları, 206900, Otozomal Dominant (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (Prenatal) (SOX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900, Autosomal dominant (Anophthalmia/microphthalmia-esophageal atresia syndrome) (SOX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX2	SOX2, MCOPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Sinir Hipoplazisi ve Merkezi Sinir Sistemi Bozuklukları, 206900, Otozomal Dominant (Anoftalmi / Mikroftalmi-Özofagus Atrezisi Sendromu) (Prenatal) (MLPA)	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900, Autosomal dominant (Anophthalmia/microphthalmia-esophageal atresia syndrome) (Prenatal) (MLPA)	SOX2	SOX2, MCOPS3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microphthalmia, syndromic, 3 (MCOPS3) [MIM:206900]: A disease characterized by the rare association of malformations including uni- or bilateral anophthalmia or microphthalmia, and esophageal atresia with tracheoesophageal fistula. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269	PubMed:12612584}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Optik Sinir Hipoplazisi, 165550, Otozomal Dominant (Izole Optik Sinir Hipoplazisi) (PAX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Optic nerve hypoplasia, 165550, Autosomal dominant (Isolated optic nerve hypoplasia) (PAX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Optik Sinir Hipoplazisi, 165550, Otozomal Dominant (Izole Optik Sinir Hipoplazisi) (MLPA)	Optic nerve hypoplasia, 165550, Autosomal dominant (Isolated optic nerve hypoplasia) (MLPA)	PAX6	PAX6, AN2, MGDA, FVH1, ASGD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Aniridia (AN) [MIM:106210]: A congenital, bilateral, panocular disorder characterized by complete absence of the iris or extreme iris hypoplasia. Aniridia is not just an isolated defect in iris development but it is associated with macular and optic nerve hypoplasia, cataract, corneal changes, nystagmus. Visual acuity is generally low but is unrelated to the degree of iris hypoplasia. Glaucoma is a secondary problem causing additional visual loss over time. {ECO:0000269|PubMed:10234503, ECO:0000269|PubMed:10737978, ECO:0000269|PubMed:11309364, ECO:0000269|PubMed:11553050, ECO:0000269|PubMed:11826019, ECO:0000269|PubMed:12552561, ECO:0000269|PubMed:12634864, ECO:0000269|PubMed:21850189, ECO:0000269|PubMed:8364574, ECO:0000269|PubMed:9147640, ECO:0000269|PubMed:9281415, ECO:0000269|PubMed:9792406, ECO:0000269|PubMed:9856761, ECO:0000269|PubMed:9931324, ECO:0000269|Ref.26, ECO:0000269|Ref.27}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Organofosfat Zehirlenmesi, Yatkınlık (PON1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Organophosphate poisoning, sensitivity to (PON1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PON1	PON1, PON, ESA, MVCD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microvascular complications of diabetes 5 (MVCD5) [MIM:612633]: Pathological conditions that develop in numerous tissues and organs as a consequence of diabetes mellitus. They include diabetic retinopathy, diabetic nephropathy leading to end-stage renal disease, and diabetic neuropathy. Diabetic retinopathy remains the major cause of new-onset blindness among diabetic adults. It is characterized by vascular permeability and increased tissue ischemia and angiogenesis. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Homozygosity for the Leu-55 allele is strongly associated with the development of retinal disease in diabetic patients.;	EDTA Blood Sample
Ornitin Transkarbamilaz Eksikliği, 311250, X'e Bağlı (Ornitin Transkarbamilaz Eksikliği) (OTC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ornithine transcarbamylase deficiency, 311250, X-linked recessive (Ornithine transcarbamylase deficiency) (OTC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTC	OTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ornithine carbamoyltransferase deficiency (OTCD) [MIM:311250]: An X-linked disorder of the urea cycle which causes a form of hyperammonemia. Mutations with no residual enzyme activity are always expressed in hemizygote males by a very severe neonatal hyperammonemic coma that generally proves to be fatal. Heterozygous females are either asymptomatic or express orotic aciduria spontaneously or after protein intake. The disorder is treatable with supplemental dietary arginine and low protein diet. The arbitrary classification of patients into the 'neonatal' group (clinical hyperammonemia in the first few days of life) and 'late' onset (clinical presentation after the neonatal period) has been used to differentiate severe from mild forms. {ECO:0000269|PubMed:10070627, ECO:0000269|PubMed:10502831, ECO:0000269|PubMed:10737985, ECO:0000269|PubMed:11793483, ECO:0000269|PubMed:1480464, ECO:0000269|PubMed:1671317, ECO:0000269|PubMed:1721894, ECO:0000269|PubMed:2347583, ECO:0000269|PubMed:2474822, ECO:0000269|PubMed:3170748, ECO:0000269|PubMed:7474905, ECO:0000269|PubMed:7951259, ECO:0000269|PubMed:8019569, ECO:0000269|PubMed:8081373, ECO:0000269|PubMed:8081398, ECO:0000269|PubMed:8099056, ECO:0000269|PubMed:8530002, ECO:0000269|PubMed:8807340, ECO:0000269|PubMed:8830175, ECO:0000269|PubMed:8956038,	EDTA Blood Sample

Ornitin Transkarbamilaz Eksikliği, 311250, X'e Bağlı (Ornitin Transkarbamilaz Eksikliği) (MLPA)	Ornithine transcarbamylase deficiency, 311250, X-linked recessive (Ornithine transcarbamylase deficiency) (MLPA)	OTC	OTC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ornithine carbamoyltransferase deficiency (OTCD) [MIM:311250]: An X-linked disorder of the urea cycle which causes a form of hyperammonemia. Mutations with no residual enzyme activity are always expressed in hemizygote males by a very severe neonatal hyperammonemic coma that generally proves to be fatal. Heterozygous females are either asymptomatic or express orotic aciduria spontaneously or after protein intake. The disorder is treatable with supplemental dietary arginine and low protein diet. The arbitrary classification of patients into the 'neonatal' group (clinical hyperammonemia in the first few days of life) and 'late' onset (clinical presentation after the neonatal period) has been used to differentiate severe from mild forms. {ECO:0000269|PubMed:10070627, ECO:0000269|PubMed:10502831, ECO:0000269|PubMed:10737985, ECO:0000269|PubMed:11793483, ECO:0000269|PubMed:1480464, ECO:0000269|PubMed:1671317, ECO:0000269|PubMed:1721894, ECO:0000269|PubMed:2347583, ECO:0000269|PubMed:2474822, ECO:0000269|PubMed:3170748, ECO:0000269|PubMed:7474905, ECO:0000269|PubMed:7951259, ECO:0000269|PubMed:8019569, ECO:0000269|PubMed:8081373, ECO:0000269|PubMed:8081398, ECO:0000269|PubMed:8099056, ECO:0000269|PubMed:8530002, ECO:0000269|PubMed:8807340, ECO:0000269|PubMed:8830175, ECO:0000269|PubMed:8956038.	EDTA Blood Sample
Ornitin Transkarbamilaz Eksikliği, 311250, X'e Bağlı (Ornitin Transkarbamilaz Eksikliği) (Prenatal) (OTC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ornithine transcarbamylase deficiency, 311250, X-linked recessive (Ornithine transcarbamylase deficiency) (OTC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OTC	OTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ornithine carbamoyltransferase deficiency (OTCD) [MIM:311250]: An X-linked disorder of the urea cycle which causes a form of hyperammonemia. Mutations with no residual enzyme activity are always expressed in hemizygote males by a very severe neonatal hyperammonemic coma that generally proves to be fatal. Heterozygous females are either asymptomatic or express orotic aciduria spontaneously or after protein intake. The disorder is treatable with supplemental dietary arginine and low protein diet. The arbitrary classification of patients into the 'neonatal' group (clinical hyperammonemia in the first few days of life) and 'late' onset (clinical presentation after the neonatal period) has been used to differentiate severe from mild forms. {ECO:0000269|PubMed:10070627, ECO:0000269|PubMed:10502831, ECO:0000269|PubMed:10737985, ECO:0000269|PubMed:11793483, ECO:0000269|PubMed:1480464, ECO:0000269|PubMed:1671317, ECO:0000269|PubMed:1721894, ECO:0000269|PubMed:2347583, ECO:0000269|PubMed:2474822, ECO:0000269|PubMed:3170748, ECO:0000269|PubMed:7474905, ECO:0000269|PubMed:7951259, ECO:0000269|PubMed:8019569, ECO:0000269|PubMed:8081373, ECO:0000269|PubMed:8081398, ECO:0000269|PubMed:8099056, ECO:0000269|PubMed:8530002, ECO:0000269|PubMed:8807340, ECO:0000269|PubMed:8830175, ECO:0000269|PubMed:8956038.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ornitin Transkarbamilaz Eksikliği, 311250, X'e Bağlı (Ornitin Transkarbamilaz Eksikliği) (Prenatal) (MLPA)	Ornithine transcarbamylase deficiency, 311250, X-linked recessive (Ornithine transcarbamylase deficiency) (Prenatal) (MLPA)	OTC	OTC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ornithine carbamoyltransferase deficiency (OTCD) [MIM:311250]: An X-linked disorder of the urea cycle which causes a form of hyperammonemia. Mutations with no residual enzyme activity are always expressed in hemizygote males by a very severe neonatal hyperammonemic coma that generally proves to be fatal. Heterozygous females are either asymptomatic or express orotic aciduria spontaneously or after protein intake. The disorder is treatable with supplemental dietary arginine and low protein diet. The arbitrary classification of patients into the 'neonatal' group (clinical hyperammonemia in the first few days of life) and 'late' onset (clinical presentation after the neonatal period) has been used to differentiate severe from mild forms. {ECO:0000269|PubMed:10070627, ECO:0000269|PubMed:10502831, ECO:0000269|PubMed:10737985, ECO:0000269|PubMed:11793483, ECO:0000269|PubMed:1480464, ECO:0000269|PubMed:1671317, ECO:0000269|PubMed:1721894, ECO:0000269|PubMed:2347583, ECO:0000269|PubMed:2474822, ECO:0000269|PubMed:3170748, ECO:0000269|PubMed:7474905, ECO:0000269|PubMed:7951259, ECO:0000269|PubMed:8019569, ECO:0000269|PubMed:8081373, ECO:0000269|PubMed:8081398, ECO:0000269|PubMed:8099056, ECO:0000269|PubMed:8530002, ECO:0000269|PubMed:8807340, ECO:0000269|PubMed:8830175, ECO:0000269|PubMed:8956038.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 10, 613705, OFC10 (Yarık Dudak / Damak) (SUMO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 10, 613705, OFC10 (Cleft lip/palate) (SUMO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUMO1	SUMO1, UBL1, SMT3, OFC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-syndromic orofacial cleft 10 (OFC10) [MIM:613705]: A birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum. {ECO:0000269|PubMed:16990542}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving SUMO1 is the cause of OFC10. Translocation t(2;8)(q33.1;p24.3). The breakpoint occurred in the SUMO1 gene and resulted in haploinsufficiency confirmed by protein assays.	EDTA Blood Sample
Orofasiyal Yarık 10, 613705, OFC10 (Yarık Dudak / Damak) (Prenatal) (SUMO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 10, 613705, OFC10 (Cleft lip/palate) (SUMO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUMO1	SUMO1, UBL1, SMT3, OFC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-syndromic orofacial cleft 10 (OFC10) [MIM:613705]: A birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum. {ECO:0000269|PubMed:16990542}. Note=The disease is caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving SUMO1 is the cause of OFC10. Translocation t(2;8)(q33.1;p24.3). The breakpoint occurred in the SUMO1 gene and resulted in haploinsufficiency confirmed by protein assays.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 11, 600625, OFC11 (Yarık Dudak / Damak) (BMP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 11, 600625, OFC11 (Cleft lip/palate) (BMP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP4	BMP4, BMP2B1, BMP2B, MCOPS6, OFC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Non-syndromic orofacial cleft 11 (OFC11) [MIM:600625]: A birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum. {ECO:0000269|PubMed:19249007}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Orofasiyal Yarık 11, 600625, OFC11 (Yarık Dudak / Damak) (Prenatal) (BMP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 11, 600625, OFC11 (Cleft lip/palate) (BMP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMP4	BMP4, BMP2B1, BMP2B, MCOPS6, OFC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Non-syndromic orofacial cleft 11 (OFC11) [MIM:600625]: A birth defect consisting of cleft lips with or without cleft palate. Cleft lips are associated with cleft palate in two-third of cases. A cleft lip can occur on one or both sides and range in severity from a simple notch in the upper lip to a complete opening in the lip extending into the floor of the nostril and involving the upper gum. {ECO:0000269|PubMed:19249007}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 15, 616788, Otozomal Dominant; OFC15 (Yarık Dudak / Damak) (DLX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 15, 616788, Autosomal dominant; OFC15 (Cleft lip/palate) (DLX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLX4	DLX4, DLX7, DLX8, OFC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Orofasiyal Yarık 15, 616788, Otozomal Dominant; OFC15 (Yarık Dudak / Damak) (Prenatal) (DLX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 15, 616788, Autosomal dominant; OFC15 (Cleft lip/palate) (DLX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLX4	DLX4, DLX7, DLX8, OFC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 5, 608874; OFC5 (Yarık Dudak / Damak) (MSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 5, 608874; OFC5 (Cleft lip/palate) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 1 (STHAG1) [MIM:106600]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). Tooth agenesis selective type 1 can be associated with orofacial cleft in some patients. {ECO:0000269|PubMed:12097313, ECO:0000269|PubMed:8696335}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyal Yarık 5, 608874; OFC5 (Yarık Dudak / Damak) (Prenatal) (MSX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 5, 608874; OFC5 (Cleft lip/palate) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tooth agenesis selective 1 (STHAG1) [MIM:106600]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). Tooth agenesis selective type 1 can be associated with orofacial cleft in some patients. {ECO:0000269|PubMed:12097313, ECO:0000269|PubMed:8696335}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Orofasiyal Yarık 6, 608864, Sporadik Vakalar; OFC6 (Yarık Dudak / Damak) (IRF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 6, 608864, Isolated cases; OFC6 (Cleft lip/palate) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. {ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyal Yarık 6, 608864, Sporadik Vakalar; OFC6 (Yarık Dudak / Damak) (Prenatal) (IRF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 6, 608864, Isolated cases; OFC6 (Cleft lip/palate) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. {ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 7, 225060, Otozomal Resesif (Yarık Dudak / Damak) (NECTIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 7, 225060, Autosomal recessive (Cleft lip/palate) (NECTIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NECTIN1	NECTIN1, PVRL1, HVEC, PVRR1, PRR1, ED4, OFC7, CLPED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia, Margarita Island type (EDMI) [MIM:225060]: An autosomal recessive form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is a syndrome characterized by the association of cleft lip/palate, ectodermal dysplasia (sparse short and dry scalp hair, sparse eyebrows and eyelashes), and partial syndactyly of the fingers and/or toes. Two thirds of the patients do not manifest oral cleft but present with abnormal teeth and nails. {ECO:0000269	PubMed:10932188}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyal Yarık 7, 225060, Otozomal Resesif (Yarık Dudak / Damak) (Prenatal) (NECTIN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 7, 225060, Autosomal recessive (Cleft lip/palate) (NECTIN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NECTIN1	NECTIN1, PVRL1, HVEC, PVRR1, PRR1, ED4, OFC7, CLPED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ectodermal dysplasia, Margarita Island type (EDMI) [MIM:225060]: An autosomal recessive form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. It is a syndrome characterized by the association of cleft lip/palate, ectodermal dysplasia (sparse short and dry scalp hair, sparse eyebrows and eyelashes), and partial syndactyly of the fingers and/or toes. Two thirds of the patients do not manifest oral cleft but present with abnormal teeth and nails. {ECO:0000269	PubMed:10932188}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyal Yarık 8, 129400, Otozomal Dominant (Yarık Dudak / Damak) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofacial cleft 8, 129400, Autosomal dominant (Cleft lip/palate) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyal Yarık 8, 129400, Otozomal Dominant (Yarık Dudak / Damak) (Prenatal) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofacial cleft 8, 129400, Autosomal dominant (Cleft lip/palate) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodijital Sendrom I, 311200, X'e Bağlı Dominant; OFD1 (Orofasiyodijital Sendrom Tip 1) (OFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome I, 311200, X-linked dominant; OFD1 (Orofaciodigital syndrome type 1) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. {ECO:0000269	PubMed:16783569}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyodijital Sendrom I, 311200, X'e Bağlı Dominant; OFD1 (Orofasiyodijital Sendrom Tip 1) (Prenatal) (OFD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome I, 311200, X-linked dominant; OFD1 (Orofaciodigital syndrome type 1) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. {ECO:0000269	PubMed:16783569}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodijital Sendrom 4, 258860, Otozomal Resesif; OFD4 (Orofasiyodijital Sendrom Tip 4) (TCTN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome IV, 258860, Autosomal recessive; OFD4 (Orofaciodigital syndrome type 4) (TCTN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCTN3	TCTN3, TECT3, C10orf61, OFD4, JBTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 4 (OFD4) [MIM:258860]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD4 patients have tongue nodules, multiple frenulae, broad flat nose, hypertelorism, and short rib polydactyly with tibial dysplasia (Majewski syntrome). The presence of severe tibial aplasia differentiates OFD4 from OFD1. Additional features of cystic dysplastic kidneys and brain malformation, including occipital encephalocele, are observed in severely affected patients. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orofasiyodijital Sendrom 4, 258860, Otozomal Resesif; OFD4 (Orofasiyodijital Sendrom Tip 4) (Prenatal) (TCTN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome IV, 258860, Autosomal recessive; OFD4 (Orofaciodigital syndrome type 4) (TCTN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCTN3	TCTN3, TECT3, C10orf61, OFD4, JBTS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 4 (OFD4) [MIM:258860]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD4 patients have tongue nodules, multiple frenulae, broad flat nose, hypertelorism, and short rib polydactyly with tibial dysplasia (Majewski syntrome). The presence of severe tibial aplasia differentiates OFD4 from OFD1. Additional features of cystic dysplastic kidneys and brain malformation, including occipital encephalocele, are observed in severely affected patients. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodigital Sendrom 5, 174300, Otozomal Resesif; OFD5 (Orofasiyodijital Sendrom Tip 5) (DDX59 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome V, 174300, Autosomal recessive; OFD5 (Orofaciodigital syndrome type 5) (DDX59 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDX59	DDX59, OFD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 5 (OFD5) [MIM:174300]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD5 patients show the core features of cleft palate, lobulated tongue, and polydactyly. Additional features include frontal bossing and intellectual disability. {ECO:0000269	PubMed:23972372}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Orofasiyodigital Sendrom 5, 174300, Otozomal Resesif; OFD5 (Orofasiyodijital Sendrom Tip 5) (Prenatal) (DDX59 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome V, 174300, Autosomal recessive; OFD5 (Orofaciodigital syndrome type 5) (DDX59 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDX59	DDX59, OFD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 5 (OFD5) [MIM:174300]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD5 patients show the core features of cleft palate, lobulated tongue, and polydactyly. Additional features include frontal bossing and intellectual disability. {ECO:0000269	PubMed:23972372}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodijital Sendrom 6 277170, Otozomal Resesif; OFD6 (Joubert Sendromu-Orofasiyodijital Defekt) (C5orf42 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome VI, 277170, Autosomal recessive; OFD6 (Joubert syndrome with orofaciodigital defect) (C5orf42 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C5orf42	C5orf42, JBTS17, OFD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 6 (OFD6) [MIM:277170]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD6 is characterized by metacarpal abnormalities with central polydactyly, cerebellar abnormalities including the molar tooth sign, tongue hamartomas, additional frenula, and upper lip notch. {ECO:0000269	PubMed:24178751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Orofasiyodijital Sendrom 6 277170, Otozomal Resesif; OFD6 (Joubert Sendromu-Orofasiyodijital Defekt) (Prenatal) (C5orf42 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome VI, 277170, Autosomal recessive; OFD6 (Joubert syndrome with orofaciodigital defect) (C5orf42 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C5orf42	C5orf42, JBTS17, OFD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 6 (OFD6) [MIM:277170]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD6 is characterized by metacarpal abnormalities with central polydactyly, cerebellar abnormalities including the molar tooth sign, tongue hamartomas, additional frenula, and upper lip notch. {ECO:0000269	PubMed:24178751}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodijital Sendrom 14, 615948, Otozomal Resesif; OFD14 (Orofasiyodijital Sendromu Tip 14) (C2CD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome XIV, 615948, Autosomal recessive; OFD14 (Orofaciodigital syndrome type 14) (C2CD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C2CD3	C2CD3, OFD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orofaciodigital syndrome 14 (OFD14) [MIM:615948]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD14 patients show severe microcephaly, cerebral malformations the molar tooth sign, and intellectual disability in addition to canonical OFDS features. {ECO:0000269	PubMed:24997988}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Orofasiyodijital Sendrom 14, 615948, Otozomal Resesif, OFD14 (Orofasiyodijital Sendromu Tip 14) (Prenatal)) (C2CD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome XIV, 615948, Autosomal recessive; OFD14 (Orofaciodigital syndrome type 14) (C2CD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C2CD3	C2CD3, OFD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orofaciodigital syndrome 14 (OFD14) [MIM:615948]: A form of orofaciodigital syndrome, a group of heterogeneous disorders characterized by malformations of the oral cavity, face and digits, and associated phenotypic abnormalities that lead to the delineation of various subtypes. OFD14 patients show severe microcephaly, cerebral malformations the molar tooth sign, and intellectual disability in addition to canonical OFDS features. {ECO:0000269	PubMed:24997988}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orofasiyodijital Sendrom 15 617127, Otozomal Resesif (Joubert Sendromu-Orofasiyodijital Defekt) (KIAA0753 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orofaciodigital syndrome XV, 617127, Autosomal recessive (Joubert syndrome with orofaciodigital defect) (KIAA0753 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA0753	KIAA0753, OFIP, OFD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Orofasiyodijital Sendrom 15 617127, Otozomal Resesif (Joubert Sendromu-Orofasiyodijital Defekt) (Prenatal) (KIAA0753 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orofaciodigital syndrome XV, 617127, Autosomal recessive (Joubert syndrome with orofaciodigital defect) (KIAA0753 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIAA0753	KIAA0753, OFIP, OFD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orolarengeal Kanser, Multipl (CDKN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orolaryngeal cancer, multiple (CDKN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN2A	CDKN2A, MTS1, P16, MLM, CMM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Li-Fraumeni syndrome (LFS) [MIM:151623]: Autosomal dominant familial cancer syndrome that in its classic form is defined by the existence of a proband affected by a sarcoma before 45 years with a first degree relative affected by any tumor before 45 years and another first degree relative with any tumor before 45 years or a sarcoma at any age. Other clinical definitions for LFS have been proposed (PubMed:8118819 and PubMed:8718514) and called Li-Fraumeni like syndrome (LFL). In these families affected relatives develop a diverse set of malignancies at unusually early ages. Four types of cancers account for 80% of tumors occurring in TP53 germline mutation carriers: breast cancers, soft tissue and bone sarcomas, brain tumors (astrocytomas) and adrenocortical carcinomas. Less frequent tumors include choroid plexus carcinoma or papilloma before the age of 15, rhabdomyosarcoma before the age of 5, leukemia, Wilms tumor, malignant phyllodes tumor, colorectal and gastric cancers. {ECO:0000269	PubMed:10484981}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orotik Asidüri, 258900, Otozomal Resesif (Kalıtsal Orotik Asitüri) (UMPS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orotic aciduria, 258900, Autosomal recessive (Hereditary orotic aciduria) (UMPS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UMPS	UMPS, OPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orotic aciduria 1 (ORAC1) [MIM:258900]: A disorder of pyrimidine metabolism resulting in megaloblastic anemia and orotic acid crystalluria that is frequently associated with some degree of physical and mental retardation. A minority of cases have additional features, particularly congenital malformations and immune deficiencies. {ECO:0000269	PubMed:9042911}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Orotik Asidüri, 258900, Otozomal Resesif (Kalıtsal Orotik Asitüri) (Prenatal) (UMPS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Orotic aciduria, 258900, Autosomal recessive (Hereditary orotic aciduria) (UMPS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UMPS	UMPS, OPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Orotic aciduria 1 (ORAC1) [MIM:258900]: A disorder of pyrimidine metabolism resulting in megaloblastic anemia and orotic acid crystalluria that is frequently associated with some degree of physical and mental retardation. A minority of cases have additional features, particularly congenital malformations and immune deficiencies. {ECO:0000269	PubMed:9042911}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ortostatik İntoleransı, 604715 (Ortostatik İntoleransı, NET Eksikliğine Bağlı) (SLC6A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Orthostatic intolerance, 604715 (Orthostatic intolerance due to NET deficiency) (SLC6A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A2	SLC6A2, NAT1, NET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Orthostatic intolerance (OI) [MIM:604715]: Syndrome characterized by lightheadedness, fatigue, altered mentation and syncope. It is associated with postural tachycardia. Plasma norepinephrine concentration is abnormally high. {ECO:0000269	PubMed:10684912}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oseözl Heteroplazi, Progresif, 166350, Otozomal Dominant; POH (Progresif Oseoz Heteroplazi) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osseous heteroplasia, progressive, 166350, Autosomal dominant; POH (Progressive osseous heteroplasia) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Oseözl Heteroplazi, Progresif, 166350, Otozomal Dominant; POH (Progresif Oseoz Heteroplazi) (MLPA)	Osseous heteroplasia, progressive, 166350, Autosomal dominant; POH (Progressive osseous heteroplasia) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoartrit Yatkınlık 1, 165720, Multifaktöryel, OS1 (FRZB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoarthritis susceptibility 1, 165720, Multifactorial; OS1 (FRZB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FRZB	FRZB, FRZB1, SRFP3, OS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoarthritis 1 (OS1) [MIM:165720]: A degenerative disease of the joints characterized by degradation of the hyaline articular cartilage and remodeling of the subchondral bone with sclerosis. Clinical symptoms include pain and joint stiffness often leading to significant disability and joint replacement. {ECO:0000269	PubMed:15210948}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteoartrit Yatkınlık 2, 140600, Otozomal Dominant; OS2 (MATN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoarthritis susceptibility 2, 140600, Autosomal dominant; OS2 (MATN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MATN3	MATN3, EDM5, HOA, OS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoarthritis 2 (OS2) [MIM:140600]: A degenerative disease of the joints characterized by degradation of the hyaline articular cartilage and remodeling of the subchondral bone with sclerosis. Clinical symptoms include pain and joint stiffness often leading to significant disability and joint replacement. In the hand, osteoarthritis can develop in the distal interphalangeal and the first carpometacarpal (base of thumb) and proximal interphalangeal joints. Patients with osteoarthritis may have one, a few, or all of these sites affected. {ECO:0000269	PubMed:12736871}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteoartrit Yatkınlık 3, 607850, Otozomal Dominant; OS3 (ASPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoarthritis susceptibility 3, 607850, Autosomal dominant; OS3 (ASPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPN	ASPN, PLAP1, OS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intervertebral disc disease (IDD) [MIM:603932]: A common musculo-skeletal disorder caused by degeneration of intervertebral disks of the lumbar spine. It results in low-back pain and unilateral leg pain. {ECO:0000269|PubMed:18304494}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to intervertebral disk disease, particularly lumbar disk degeneration, is conferred by a triplet repeat expansion polymorphism. ASPN allele having 14 aspartic acid repeats in the N-terminal region of the protein (D14), is associated with the disorder in some populations (PubMed:18304494). {ECO:0000269|PubMed:18304494}.;	EDTA Blood Sample
Hafif Kondrodisplazi İle Osteoartrit, 604864, Otozomal Dominant; OSCDP (Hafif Spondiloepifizyal Displazi-Erken Başlayan Osteoartrit, COL2A1 Mutasyonuna Bağlı) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoarthritis with mild chondrodysplasia, 604864, Autosomal dominant; OSCDP (Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hafif Kondrodisplazi İle Osteoartrit, 604864, Otozomal Dominant; OSCDP (Hafif Spondiloepifizyal Displazi-Erken Başlayan Osteoartrit, COL2A1 Mutasyonuna Bağlı) (MLPA)	Osteoarthritis with mild chondrodysplasia, 604864, Autosomal dominant; OSCDP (Mild spondyloepiphyseal dysplasia due to COL2A1 mutation with early-onset osteoarthritis) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoartrit-5, 612400 (Fibula Aplazisi-Kompleks Brakidaktili Sendromu (Du Pan Sendromu)) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoarthritis-5, 612400 (Fibular aplasia-complex brachydactyly syndrome) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:16127465, ECO:0000269|PubMed:18203755, ECO:0000269|PubMed:21976273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteokondritis Dissekans, Kısa Boy ve Erken Başlangıçlı Osteoartrit, 165800, Otozomal Dominant; OD (Ailesel Osteokondritis Dissekans) (ACAN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteochondritis dissecans, short stature, and early-onset osteoarthritis, 165800, Autosomal dominant; OD (Familial osteochondritis dissecans) (ACAN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAN	ACAN, AGC1, CSPG1, MSK16, SEDK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteochondritis dissecans short stature and early-onset osteoarthritis (OD) [MIM:165800]: A type of osteochondritis defined as a separation of cartilage and subchondral bone from the surrounding tissue, primarily affecting the knee, ankle and elbow joints. It is clinically characterized by multiple osteochondritic lesions in knees and/or hips and/or elbows, disproportionate short stature and early-onset osteoarthritis. {ECO:0000269	PubMed:20137779}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Osteokondrodisplazi, Kompleks Letal, Symoens-Barnes-Gistelinck Tipi, 616897, Otozomal Resesif, OCLSBG (Kompleks Letal Osteokondrodisplazi) (TAPT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897, Autosomal recessive; OCLSBG (Complex lethal osteochondrodysplasia) (TAPT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAPT1	TAPT1, CMVFR, OCLSBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteokondrodisplazi, Kompleks Letal, Symoens-Barnes-Gistelinck Tipi, 616897, Otozomal Resesif, OCLSBG (Kompleks Letal Osteokondrodisplazi) (Prenatal) (TAPT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897, Autosomal recessive; OCLSBG (Complex lethal osteochondrodysplasia) (TAPT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAPT1	TAPT1, CMVFR, OCLSBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteofibrous Displazi, Yatkınlık, 607278, Otozomal Dominant (MET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteofibrous dysplasia, susceptibility to, 607278, Autosomal dominant (MET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MET	MET, DFNB97, OSFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatocellular carcinoma (HCC) [MIM:114550]: A primary malignant neoplasm of epithelial liver cells. The major risk factors for HCC are chronic hepatitis B virus (HBV) infection, chronic hepatitis C virus (HCV) infection, prolonged dietary aflatoxin exposure, alcoholic cirrhosis, and cirrhosis due to other causes. {ECO:0000269	PubMed:9927037}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 1, 166200, Otozomal Dominant; OI1 (Osteogenezis İmperfekta) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type I, 166200, Autosomal dominant; OI1 (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 1, 166200, Otozomal Dominant; OI1 (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type I, 166200, Autosomal dominant; OI1 (Osteogenesis imperfecta) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 1, 166200, Otozomal Dominant; OI1 (Osteogenezis İmperfekta) (Prenatal) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type I, 166200, Autosomal dominant; OI1 (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 1, 166200, Otozomal Dominant; OI1 (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type I, 166200, Autosomal dominant; OI1 (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant (Osteogenezis İmperfekta) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant (Osteogenesis imperfecta) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant (Osteogenezis İmperfekta) (Prenatal) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant; OI2 (Osteogenezis İmperfekta) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant; OI2 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant; OI2 (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant; OI2 (Osteogenesis imperfecta) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant; OI2 (Osteogenezis İmperfekta) (Prenatal) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant; OI2 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 2, 166210, Otozomal Dominant; OI2 (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type II, 166210, Autosomal dominant; OI2 (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant (Osteogenezis İmperfekta) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant (Osteogenesis imperfecta) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269	PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant (Osteogenezis İmperfekta) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant; OI3 (Osteogenezis İmperfekta) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant; OI3 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant; OI3 (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant; OI3 (Osteogenesis imperfecta) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant; OI3 (Osteogenezis İmperfekta) (Prenatal) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant; OI3 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 3, 259420, Otozomal Dominant; OI3 (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type III, 259420, Autosomal dominant; OI3 (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant (Osteogenezis İmperfekta) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type IV, 166220, Autosomal Dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type IV, 166220, Autosomal Dominant (Osteogenesis imperfecta) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant (Osteogenezis İmperfekta) (Prenatal) (COL1A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type IV, 166220, Autosomal Dominant (Osteogenesis imperfecta) (COL1A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type IV, 166220, Autosomal Dominant (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A1	COL1A1, OI1, OI2, OI3, OI4, EDSC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Caffey disease (CAFFD) [MIM:114000]: Characterized by an infantile episode of massive subperiosteal new bone formation that typically involves the diaphyses of the long bones, mandible, and clavicles. The involved bones may also appear inflamed, with painful swelling and systemic fever often accompanying the illness. The bone changes usually begin before 5 months of age and resolve before 2 years of age. {ECO:0000269|PubMed:15864348}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant; OI4 (Osteogenezis İmperfekta) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type IV, 166220, Autosomal dominant; OI4 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant; OI4 (Osteogenezis İmperfekta) (MLPA)	Osteogenesis imperfecta, type IV, 166220, Autosomal dominant; OI4 (Osteogenesis imperfecta) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant; OI4 (Osteogenezis İmperfekta) (Prenatal) (COL1A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type IV, 166220, Autosomal dominant; OI4 (Osteogenesis imperfecta) (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 4, 166220, Otozomal Dominant; OI4 (Osteogenezis İmperfekta) (Prenatal) (MLPA)	Osteogenesis imperfecta, type IV, 166220, Autosomal dominant; OI4 (Osteogenesis imperfecta) (Prenatal) (MLPA)	COL1A2	COL1A2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hypermobility. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. {ECO:0000269|PubMed:15777455, ECO:0000269|PubMed:23947581, ECO:0000269|PubMed:36802255}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 9 259440, Otozomal Resesif; OI9 (Osteogenezis İmperfekta) (PPIB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type IX, 259440, Autosomal recessive; OI9 (Osteogenesis imperfecta) (PPIB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPIB	PPIB, CYPB, OI9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 9 (OI9) [MIM:259440]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI9 is a severe autosomal recessive form of the disorder. {ECO:0000269|PubMed:19781681, ECO:0000269|PubMed:20089953}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 9 259440, Otozomal Resesif; OI9 (Osteogenezis İmperfekta) (Prenatal) (PPIB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type IX, 259440, Autosomal recessive; OI9 (Osteogenesis imperfecta) (PPIB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPIB	PPIB, CYPB, OI9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 9 (OI9) [MIM:259440]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI9 is a severe autosomal recessive form of the disorder. {ECO:0000269|PubMed:19781681, ECO:0000269|PubMed:20089953}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Osteogenezis İmperfekta, Tip 5, 610967, Otozomal Dominant; OI5 (Osteogenezis İmperfekta) (IFITM5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type V, 610967, Autosomal dominant; OI5 (Osteogenesis imperfecta) (IFITM5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFITM5	IFITM5, OI5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 5 (OI5) [MIM:610967]: An autosomal dominant form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI5 patients manifest moderate to severe bone fragility, calcification of the forearm interosseous membrane, radial head dislocation, a subphyseal metaphyseal radiodense line, and hyperplastic callus formation. (ECO:0000269	PubMed:22863190). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 5, 610967, Otozomal Dominant; OI5 (Osteogenezis İmperfekta) (Prenatal) (IFITM5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type V, 610967, Autosomal dominant; OI5 (Osteogenesis imperfecta) (IFITM5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFITM5	IFITM5, OI5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 5 (OI5) [MIM:610967]: An autosomal dominant form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI5 patients manifest moderate to severe bone fragility, calcification of the forearm interosseous membrane, radial head dislocation, a subphyseal metaphyseal radiodense line, and hyperplastic callus formation. (ECO:0000269	PubMed:22863190). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 6, 613982, OI6 (Osteogenezis İmperfekta) (SERPINF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type VI, 613982; OI6 (Osteogenesis imperfecta) (SERPINF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINF1	SERPINF1, PEDF, OI6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 6 (OI6) [MIM:613982]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI6 is a severe, autosomal recessive form compatible with OI type III in the Sillence classification. (ECO:0000269	PubMed:21353196). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 6, 613982, OI6 (Osteogenezis İmperfekta) (Prenatal) (SERPINF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type VI, 613982; OI6 (Osteogenesis imperfecta) (SERPINF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPINF1	SERPINF1, PEDF, OI6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 6 (OI6) [MIM:613982]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI6 is a severe, autosomal recessive form compatible with OI type III in the Sillence classification. (ECO:0000269	PubMed:21353196). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 7, 610682, Otozomal Resesif; OI7 (Osteogenezis İmperfekta) (CRTAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type VII, 610682, Autosomal recessive; OI7 (Osteogenesis imperfecta) (CRTAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRTAP	CRTAP, CASP, OI7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 7, 610682, Otozomal Resesif; OI7 (Osteogenezis İmperfekta) (Prenatal) (CRTAP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type VII, 610682, Autosomal recessive; OI7 (Osteogenesis imperfecta) (CRTAP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRTAP	CRTAP, CASP, OI7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 8, 610915, Otozomal Resesif; OI8 (Osteogenezis İmperfekta) (P3H1 (LEPRE1) geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type VIII, 610915, Autosomal recessive; OI8 (Osteogenesis imperfecta) (P3H1 (LEPRE1) gene) (Sequence Analysis-All Coding Exons) (Postnatal)	P3H1	P3H1, LEPRE1, GROS1, OI8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 8 (OI8) [MIM:610915]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI8 is characterized by disproportionate short stature, severe osteoporosis, shortening of the long bones, white sclerae, a round face and a short barrel-shaped chest. (ECO:0000269|PubMed:17277775, ECO:0000269|PubMed:19088120). Note=The disease is caused by mutations affecting the gene represented in this entry. A splice site mutation leading to the absence of isoform 1 has been reported in 2 OI8 patients. Isoform 1 is the only form predicted to be located in the endoplasmic reticulum, which the appropriate location for the catalysis of collagen hydroxylation. These patients show indeed severely reduced COL1A1 hydroxylation (PubMed:19088120). {ECO:0000269	PubMed:19088120}.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 8, 610915, Otozomal Resesif; OI8 (Osteogenezis İmperfekta) (Prenatal) (P3H1 (LEPRE1) geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type VIII, 610915, Autosomal recessive; OI8 (Osteogenesis imperfecta) (P3H1 (LEPRE1) gene) (Sequence Analysis-All Coding Exons) (Prenatal)	P3H1	P3H1, LEPRE1, GROS1, OI8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 8 (OI8) [MIM:610915]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI8 is characterized by disproportionate short stature, severe osteoporosis, shortening of the long bones, white sclerae, a round face and a short barrel-shaped chest. (ECO:0000269|PubMed:17277775, ECO:0000269|PubMed:19088120). Note=The disease is caused by mutations affecting the gene represented in this entry. A splice site mutation leading to the absence of isoform 1 has been reported in 2 OI8 patients. Isoform 1 is the only form predicted to be located in the endoplasmic reticulum, which the appropriate location for the catalysis of collagen hydroxylation. These patients show indeed severely reduced COL1A1 hydroxylation (PubMed:19088120). {ECO:0000269	PubMed:19088120}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, 10, 613848, Otozomal Resesif; OI10 (Osteogenezis İmperfekta) (SERPINH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type X, 613848, Autosomal recessive; OI10 (Osteogenesis imperfecta) (SERPINH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINH1	SERPINH1, SERPINH2, PPROM, CBP2, CBP1, OI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 10 (OI10) [MIM:613848]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI10 is an autosomal recessive form characterized by multiple bone deformities and fractures, generalized osteopenia, dentinogenesis imperfecta, and blue sclerae. (ECO:0000269	PubMed:20188343). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, 10, 613848, Otozomal Resesif; OI10 (Osteogenezis İmperfekta) (Prenatal) (SERPINH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type X, 613848, Autosomal recessive; OI10 (Osteogenesis imperfecta) (SERPINH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SERPINH1	SERPINH1, SERPINH2, PPROM, CBP2, CBP1, OI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 10 (OI10) [MIM:613848]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI10 is an autosomal recessive form characterized by multiple bone deformities and fractures, generalized osteopenia, dentinogenesis imperfecta, and blue sclerae. (ECO:0000269	PubMed:20188343). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 11, 610968, Otozomal Resesif; OI11 (Osteogenezis İmperfekta) (FKBP10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XI, 610968, Autosomal recessive; OI11 (Osteogenesis imperfecta) (FKBP10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FKBP10	FKBP10, FKBP65, OI11, BRKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bruck syndrome 1 (BRKS1) [MIM:259450]: A disease characterized by generalized osteopenia, congenital joint contractures, fragile bones with onset of fractures in infancy or early childhood, short stature, severe limb deformity, progressive scoliosis, and pterygia. {ECO:0000269|PubMed:20839288, ECO:0000269|PubMed:22949511}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 11, 610968, Otozomal Resesif; OI11 (Osteogenezis İmperfekta) (Prenatal) (FKBP10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XI, 610968, Autosomal recessive; OI11 (Osteogenesis imperfecta) (FKBP10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FKBP10	FKBP10, FKBP65, OI11, BRKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bruck syndrome 1 (BRKS1) [MIM:259450]: A disease characterized by generalized osteopenia, congenital joint contractures, fragile bones with onset of fractures in infancy or early childhood, short stature, severe limb deformity, progressive scoliosis, and pterygia. {ECO:0000269|PubMed:20839288, ECO:0000269|PubMed:22949511}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 12, 613849, Otozomal Resesif; OI12 (Osteogenezis İmperfekta) (SP7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XII, 613849, Autosomal recessive; OI12 (Osteogenesis imperfecta) (SP7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SP7	SP7, OSX, OI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 12 (OI12) [MIM:613849]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI12 is an autosomal recessive form characterized by recurrent fractures, mild bone deformations, generalized osteoporosis, delayed teeth eruption, no dentinogenesis imperfecta, normal hearing, and white sclerae. {ECO:0000269	PubMed:20579626}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 12, 613849, Otozomal Resesif; OI12 (Osteogenezis İmperfekta) (Prenatal) (SP7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XII, 613849, Autosomal recessive; OI12 (Osteogenesis imperfecta) (SP7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SP7	SP7, OSX, OI12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 12 (OI12) [MIM:613849]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI12 is an autosomal recessive form characterized by recurrent fractures, mild bone deformations, generalized osteoporosis, delayed teeth eruption, no dentinogenesis imperfecta, normal hearing, and white sclerae. {ECO:0000269	PubMed:20579626}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Osteogenezis İmperfekta, Tip 13, 614856, Otozomal Resesif, OI13 (Osteogenezis Imperfekta) (BMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XIII, 614856, Autosomal recessive; OI13 (Osteogenesis imperfecta) (BMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP1	BMP1, OI13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 13 (OI13) [MIM:614856]: An autosomal recessive form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI13 is characterized by normal teeth, faint blue sclerae, severe growth deficiency, borderline osteoporosis, severe bone deformity, and recurrent fractures affecting both upper and lower limbs. (ECO:0000269|PubMed:22052668, ECO:0000269|PubMed:22482805, ECO:0000269|PubMed:25402547). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 13, 614856, Otozomal Resesif, OI13 (Osteogenezis Imperfekta) (Prenatal) (BMP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XIII, 614856, Autosomal recessive; OI13 (Osteogenesis imperfecta) (BMP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMP1	BMP1, OI13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 13 (OI13) [MIM:614856]: An autosomal recessive form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI13 is characterized by normal teeth, faint blue sclerae, severe growth deficiency, borderline osteoporosis, severe bone deformity, and recurrent fractures affecting both upper and lower limbs. (ECO:0000269|PubMed:22052668, ECO:0000269|PubMed:22482805, ECO:0000269|PubMed:25402547). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 14, 615066, OI14 (Osteogenezis Imperfekta) (TMEM38B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XIV, 615066; OI14 (Osteogenesis imperfecta) (TMEM38B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM38B	TMEM38B, TRICB, OI14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 14 (OI14) [MIM:615066]: An autosomal recessive form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI14 is characterized by variable degrees of severity of multiple fractures and osteopenia, with normal teeth, sclerae, and hearing. Fractures first occur prenatally or by age 6 years. (ECO:0000269|PubMed:23054245, ECO:0000269|PubMed:23316006). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 14, 615066, OI14 (Osteogenezis Imperfekta) (Prenatal) (TMEM38B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XIV, 615066; OI14 (Osteogenesis imperfecta) (TMEM38B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TMEM38B	TMEM38B, TRICB, OI14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteogenesis imperfecta 14 (OI14) [MIM:615066]: An autosomal recessive form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI14 is characterized by variable degrees of severity of multiple fractures and osteopenia, with normal teeth, sclerae, and hearing. Fractures first occur prenatally or by age 6 years. (ECO:0000269|PubMed:23054245, ECO:0000269|PubMed:23316006). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 15, 615220, Otozomal Resesif, OI15 (Osteogenezis Imperfekta) (WNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XV, 615220, Autosomal recessive; OI15 (Osteogenesis imperfecta) (WNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT1	WNT1, INT1, OI15, BMND16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. (ECO:0000269|PubMed:23499309, ECO:0000269|PubMed:23656646). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 15, 615220, Otozomal Resesif, OI15 (Osteogenezis Imperfekta) (Prenatal) (WNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XV, 615220, Autosomal recessive; OI15 (Osteogenesis imperfecta) (WNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT1	WNT1, INT1, OI15, BMND16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. (ECO:0000269|PubMed:23499309, ECO:0000269|PubMed:23656646). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 16, 616229, Otozomal Resesif (Osteogenezis Imperfekta) (22Q0)	Osteogenesis imperfecta, type XVI, 616229, Autosomal recessive (Osteogenesis imperfecta) (440)	.	OI16, C16DELp11.2, DEL16p11.2	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Osteogenezis İmperfekta, Tip 16, 616229, Otozomal Resesif (Osteogenezis Imperfekta) (Prenatal)	Osteogenesis imperfecta, type XVI, 616229, Autosomal recessive (Osteogenesis imperfecta) (Prenatal)	.	OI16, C16DELp11.2, DEL16p11.2	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteogenezis İmperfekta, Tip 17, 616507, Otozomal Resesif, OI17 (Osteogenezis İmperfekta) (SPARC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteogenesis imperfecta, type XVII, 616507, Autosomal recessive; OI17 (Osteogenesis imperfecta) (SPARC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPARC	SPARC, ON, OI17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteogenezis İmperfekta, Tip 17, 616507, Otozomal Resesif, OI17 (Osteogenezis İmperfekta) (Prenatal) (SPARC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteogenesis imperfecta, type XVII, 616507, Autosomal recessive; OI17 (Osteogenesis imperfecta) (SPARC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPARC	SPARC, ON, OI17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteojenik Sarkom (Osteosarkom) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	OSTEOGENIC SARCOMA (Osteosarcoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteojenik Sarkom (Osteosarkom) (MLPA)	OSTEOGENIC SARCOMA (Osteosarcoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteojenik Sarkom (Osteosarkom) (MLPA)	OSTEOGENIC SARCOMA (Osteosarcoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoglofonik Displazi, 166250, Otozomal Dominant; OGD (Osteoglofonik Displazi) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoglophonic dysplasia, 166250, Autosomal dominant; OGD (Osteoglophonic dwarfism) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7874169). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoglofonik Displazi, 166250, Otozomal Dominant; OGD (Osteoglofonik Displazi) (MLPA)	Osteoglophonic dysplasia, 166250, Autosomal dominant; OGD (Osteoglophonic dwarfism) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7874169). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoglofonik Displazi, 166250, Otozomal Dominant; OGD (Osteoglofonik Displazi) (Prenatal) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteoglophonic dysplasia, 166250, Autosomal dominant; OGD (Osteoglophonic dwarfism) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7874169). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteoglofonik Displazi, 166250, Otozomal Dominant; OGD (Osteoglofonik Displazi) (Prenatal) (MLPA)	Osteoglophonic dysplasia, 166250, Autosomal dominant; OGD (Osteoglophonic dwarfism) (Prenatal) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). (ECO:0000269	PubMed:7874169). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteoliz, Ailesel Ekspansil, 174810, Otozomal Dominant; FEO (Ailesel Ekspansil Osteoliz) (TNFRSF11A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteolysis, familial expansile, 174810, Autosomal dominant; FEO (Familial expansile osteolysis) (TNFRSF11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF11A	TNFRSF11A, RANK, ODFR, FEO, OPTB7, PDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 2, early-onset (PDB2) [MIM:602080]: A form of Paget disease, a disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. (ECO:0000269	PubMed:10615125). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteomalazi, Tümör Kaynaklı (FGF23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteomalacia, tumor-induced (FGF23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF23	FGF23, ADHR, HPDR2, PHPTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. (ECO:0000269	PubMed:11062477). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Osteomalazi, Tümör Kaynaklı (MLPA)	Osteomalacia, tumor-induced (MLPA)	FGF23	FGF23, ADHR, HPDR2, PHPTC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. {ECO:0000269	PubMed:11062477}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteopati Striata-Kranial Scleroz 300373, X'e Bağlı Dominant; OSCS (Osteopati Striata-Kranial Skleroz Sendromu) (AMER1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopathia striata with cranial sclerosis, 300373, X-linked dominant; OSCS (Osteopathia striata-cranial sclerosis syndrome) (AMER1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMER1	AMER1, FAM123B, WTX, OSCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteopati Striata-Kranial Scleroz 300373, X'e Bağlı Dominant; OSCS (Osteopati Striata-Kranial Skleroz Sendromu) (Prenatal) (AMER1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopathia striata with cranial sclerosis, 300373, X-linked dominant; OSCS (Osteopathia striata-cranial sclerosis syndrome) (AMER1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMER1	AMER1, FAM123B, WTX, OSCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Dominant 1, 607634, Otozomal Dominant; OPTA1 (Osteopetroz, Otozomal Dominant Tip 1) (LRP5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal dominant 1, 607634, Autosomal dominant; OPTA1 (Autosomal dominant osteopetrosis type 1) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteopetrozis, Otozomal Dominant 1, 607634, Otozomal Dominant; OPTA1 (Osteopetroz, Otozomal Dominant Tip 1) (Prenatal) (LRP5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal dominant 1, 607634, Autosomal dominant; OPTA1 (Autosomal dominant osteopetrosis type 1) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Dominant 2, 166600, Otozomal Dominant; OPTA2 (Albers-Schönberg Osteopetrozu) (CLCN7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal dominant 2, 166600, Autosomal dominant; OPTA2 (Albers-Schönberg osteopetrosis) (CLCN7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN7	CLCN7, CLC7, OPTA2, OPTB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal dominant 2 (OPTA2) [MIM:166600]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA2 is the most common form of osteopetrosis, occurring in adolescence or adulthood. It is characterized by sclerosis, predominantly involving the spine, the pelvis and the skull base. {ECO:0000269|PubMed:11741829, ECO:0000269|PubMed:14584882, ECO:0000269|PubMed:19953639}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteopetrozis, Otozomal Dominant 2, 166600, Otozomal Dominant; OPTA2 (Albers-Schönberg Osteopetrozu) (Prenatal) (CLCN7 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal dominant 2, 166600, Autosomal dominant; OPTA2 (Albers-Schönberg osteopetrosis) (CLCN7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN7	CLCN7, CLC7, OPTA2, OPTB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteopetrosis, autosomal dominant 2 (OPTA2) [MIM:166600]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA2 is the most common form of osteopetrosis, occurring in adolescence or adulthood. It is characterized by sclerosis, predominantly involving the spine, the pelvis and the skull base. {ECO:0000269|PubMed:11741829, ECO:0000269|PubMed:14584882, ECO:0000269|PubMed:19953639}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 1, 259700, Otozomal Resesif; OPTB1 (Otozomal Resesif Malign Osteopetroz) (TCIRG1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 1, 259700, Autosomal recessive; OPTB1 (Autosomal recessive malignant osteopetrosis) (TCIRG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCIRG1	TCIRG1, TIRC7, OC116, OPTB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 1, 259700, Otozomal Resesif; OPTB1 (Otozomal Resesif Malign Osteopetroz) (Prenatal) (TCIRG1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 1, 259700, Autosomal recessive; OPTB1 (Autosomal recessive malignant osteopetrosis) (TCIRG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCIRG1	TCIRG1, TIRC7, OC116, OPTB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 2, 259710, Otozomal Resesif; OPTB2 (Otozomal Resesif Malign Osteopetroz) (TNFSF11 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 2, 259710, Autosomal recessive; OPTB2 (Autosomal recessive malignant osteopetrosis) (TNFSF11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFSF11	TNFSF11, OPGL, TRANCE, OPTB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal recessive 2 (OPTB2) [MIM:259710]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB2 is characterized by paucity of osteoclasts, suggesting a molecular defect in osteoclast development. {ECO:0000269	PubMed:17632511}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 2, 259710, Otozomal Resesif; OPTB2 (Otozomal Resesif Malign Osteopetroz) (Prenatal) (TNFSF11 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 2, 259710, Autosomal recessive; OPTB2 (Autosomal recessive malignant osteopetrosis) (TNFSF11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFSF11	TNFSF11, OPGL, TRANCE, OPTB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteopetrosis, autosomal recessive 2 (OPTB2) [MIM:259710]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB2 is characterized by paucity of osteoclasts, suggesting a molecular defect in osteoclast development. {ECO:0000269	PubMed:17632511}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteoporozis, Renal Tübüler Asidoz, Otozomal Resesif 3, 259730, Otozomal Resesif; OPTB3 (Osteoporoz-Renal Tübüler Asidoz) (CA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730, Autosomal recessive; OPTB3 (Osteopetrosis with renal tubular acidosis) (CA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CA2	CA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal recessive 3 (OPTB3) [MIM:259730]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB3 is associated with renal tubular acidosis, cerebral calcification (marble brain disease) and in some cases with mental retardation. {ECO:0000269|PubMed:15300855, ECO:0000269|PubMed:1542674, ECO:0000269|PubMed:1928091, ECO:0000269|PubMed:8634238, ECO:0000269|PubMed:9143915}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 4, 611490; OPTB4 (Otozomal Resesif Malign Osteopetroz) (CLCN7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 4, 611490; OPTB4 (Autosomal recessive malignant osteopetrosis) (CLCN7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN7	CLCN7, CLC7, OPTA2, OPTB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal dominant 2 (OPTA2) [MIM:166600]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA2 is the most common form of osteopetrosis, occurring in adolescence or adulthood. It is characterized by sclerosis, predominantly involving the spine, the pelvis and the skull base. {ECO:0000269|PubMed:11741829, ECO:0000269|PubMed:14584882, ECO:0000269|PubMed:19953639}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 4, 611490; OPTB4 (Otozomal Resesif Malign Osteopetroz) (Prenatal) (CLCN7 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 4, 611490; OPTB4 (Autosomal recessive malignant osteopetrosis) (CLCN7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLCN7	CLCN7, CLC7, OPTA2, OPTB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteopetrosis, autosomal dominant 2 (OPTA2) [MIM:166600]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. OPTA2 is the most common form of osteopetrosis, occurring in adolescence or adulthood. It is characterized by sclerosis, predominantly involving the spine, the pelvis and the skull base. {ECO:0000269|PubMed:11741829, ECO:0000269|PubMed:14584882, ECO:0000269|PubMed:19953639}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Osteopetrozis, Otozomal Resesif 5, 259720, Otozomal Resesif; OPTB5 (Infantil Osteopetrozis-Nöroaksonal Displazi) (OSTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 5, 259720, Autosomal recessive; OPTB5 (infantile osteopetrosis with neuroaxonal dysplasia) (OSTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OSTM1	OSTM1, GL, OPTB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal recessive 5 (OPTB5) [MIM:259720]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB5 patients manifest primary central nervous system involvement in addition to the classical stigmata of severe bone sclerosis, growth failure, anemia, thrombocytopenia and visual impairment with optic atrophy. {ECO:0000269|PubMed:12627228, ECO:0000269|PubMed:16813530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 5, 259720, Otozomal Resesif; OPTB5 (Infantil Osteopetrozis-Nöroaksonal Displazi) (Prenatal) (OSTM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 5, 259720, Autosomal recessive; OPTB5 (infantile osteopetrosis with neuroaxonal dysplasia) (OSTM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OSTM1	OSTM1, GL, OPTB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteopetrosis, autosomal recessive 5 (OPTB5) [MIM:259720]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB5 patients manifest primary central nervous system involvement in addition to the classical stigmata of severe bone sclerosis, growth failure, anemia, thrombocytopenia and visual impairment with optic atrophy. {ECO:0000269|PubMed:12627228, ECO:0000269|PubMed:16813530}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 7, 612301; OPTB7 (Osteopetrozis-Hipogamaglobulinemi Sendromu) (TNFRSF11A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 7, 612301; OPTB7 (Osteopetrosis-hypogammaglobulinemia syndrome) (TNFRSF11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF11A	TNFRSF11A, RANK, ODFR, FEO, OPTB7, PDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 2, early-onset (PDB2) [MIM:602080]: A form of Paget disease, a disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269	PubMed:10615125}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 7, 612301; OPTB7 (Osteopetrozis-Hipogamaglobulinemi Sendromu) (Prenatal) (TNFRSF11A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 7, 612301; OPTB7 (Osteopetrosis-hypogammaglobulinemia syndrome) (TNFRSF11A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TNFRSF11A	TNFRSF11A, RANK, ODFR, FEO, OPTB7, PDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paget disease of bone 2, early-onset (PDB2) [MIM:602080]: A form of Paget disease, a disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269	PubMed:10615125}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 8, 615085, Otozomal Resesif; OPTB8 (Otozomal Resesif Malign Osteopetroz) (SNX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopetrosis, autosomal recessive 8, 615085, Autosomal recessive; OPTB8 (Autosomal recessive malignant osteopetrosis) (SNX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNX10	SNX10, OPTB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteopetrosis, autosomal recessive 8 (OPTB8) [MIM:615085]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB8 is clinically characterized by dense bones with no distinction between outer and inner plates, due to extensive encroachment of cortical bone into the medullary space, increased head circumference, broad open fontanelle, frontal bossing, and hepatosplenomegaly. Osteoclasts number is low and their bone resorptive capacity is impaired. {ECO:0000269|PubMed:22499339, ECO:0000269|PubMed:23123320, ECO:0000269|PubMed:23280965}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Osteopetrozis, Otozomal Resesif 8, 615085, Otozomal Resesif; OPTB8 (Otozomal Resesif Malign Osteopetroz) (Prenatal) (SNX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopetrosis, autosomal recessive 8, 615085, Autosomal recessive; OPTB8 (Autosomal recessive malignant osteopetrosis) (SNX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNX10	SNX10, OPTB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteopetrosis, autosomal recessive 8 (OPTB8) [MIM:615085]: A rare genetic disease characterized by abnormally dense bone, due to defective resorption of immature bone. Osteopetrosis occurs in two forms: a severe autosomal recessive form occurring in utero, infancy, or childhood, and a benign autosomal dominant form occurring in adolescence or adulthood. Recessive osteopetrosis commonly manifests in early infancy with macrocephaly, feeding difficulties, evolving blindness and deafness, bone marrow failure, severe anemia, and hepatosplenomegaly. Deafness and blindness are generally thought to represent effects of pressure on nerves. OPTB8 is clinically characterized by dense bones with no distinction between outer and inner plates, due to extensive encroachment of cortical bone into the medullary space, increased head circumference, broad open fontanelle, frontal bossing, and hepatosplenomegaly. Osteoclasts number is low and their bone resorptive capacity is impaired. {ECO:0000269|PubMed:22499339, ECO:0000269|PubMed:23123320, ECO:0000269|PubMed:23280965}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteopoikiloz, 166700, Otozomal Dominant (Zole Osteopoikiloz) (LEMD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteopoikilosis, 166700, Autosomal dominant (Isolated osteopoikilosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melorheostosis (MEL) [MIM:155950]: Rare mesenchymal dysplasia and one of the sclerosing bone disorders. It is caused by a developmental error, with a sclerotomal distribution, frequently involving one limb. It may be asymptomatic, but pain, stiffness with limitation of motion, leg-length discrepancy and limb deformity may occur. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Osteopoikiloz, 166700, Otozomal Dominant (Zole Osteopoikiloz) (Prenatal) (LEMD3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteopoikilosis, 166700, Autosomal dominant (Isolated osteopoikilosis) (LEMD3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LEMD3	LEMD3, MAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Melorheostosis (MEL) [MIM:155950]: Rare mesenchymal dysplasia and one of the sclerosing bone disorders. It is caused by a developmental error, with a sclerotomal distribution, frequently involving one limb. It may be asymptomatic, but pain, stiffness with limitation of motion, leg-length discrepancy and limb deformity may occur. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteoporoz-Psödoglioma Sendromu, 259770, Otozomal Resesif; OPPG (Osteoporoz-Psödoglioma Sendromu) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis-pseudoglioma syndrome, 259770, Autosomal recessive; OPPG (Osteoporosis-pseudoglioma syndrome) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoporoz-Psödoglioma Sendromu, 259770, Otozomal Resesif; OPPG (Osteoporoz-Psödoglioma Sendromu) (Prenatal) (LRP5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Osteoporosis-pseudoglioma syndrome, 259770, Autosomal recessive; OPPG (Osteoporosis-pseudoglioma syndrome) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Osteoporoz, 166710, Otozomal Dominant (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, 166710, Autosomal dominant (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269	PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Osteoporoz, Erken Başlangıçlı, Yatkınlık, Otozomal Dominant, 615221 (İdiopatik Jüvenil Osteoporoz) (WNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, early-onset, susceptibility to, autosomal dominant, 615221 (Idiopathic juvenile osteoporosis) (WNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT1	WNT1, INT1, OI15, BMND16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoporosis (OSTEOP) [MIM:166710]: A systemic skeletal disorder characterized by decreased bone mass and deterioration of bone microarchitecture without alteration in the composition of bone. The result is fragile bones and an increased risk of fractures, even after minimal trauma. Osteoporosis is a chronic condition of multifactorial etiology and is usually clinically silent until a fracture occurs. (ECO:0000269|PubMed:23499309, ECO:0000269|PubMed:23656646). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoporoz, İnvolüsyonel, 166710, Otozomal Dominant (VDR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, involutional, 166710, Autosomal dominant (VDR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VDR	VDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rickets vitamin D-dependent 2A (VDDR2A) [MIM:277440]: A disorder of vitamin D metabolism resulting in severe rickets, hypocalcemia and secondary hyperparathyroidism. Most patients have total alopecia in addition to rickets. (ECO:0000269|PubMed:1652893, ECO:0000269|PubMed:2177843, ECO:0000269|PubMed:2849209, ECO:0000269|PubMed:7828346, ECO:0000269|PubMed:8106618, ECO:0000269|PubMed:8381803, ECO:0000269|PubMed:8392085, ECO:0000269|PubMed:8675579, ECO:0000269|PubMed:8961271, ECO:0000269|PubMed:9005998). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Osteoporoz, Menopoz Sonrası, 166710, Otozomal Dominant (COL1A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, postmenopausal, 166710, Autosomal dominant (COL1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL1A2	COL1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. (ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:3680255). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoporoz, Menopoz Sonrası, 166710, Otozomal Dominant (MLPA)	Osteoporosis, postmenopausal, 166710, Autosomal dominant (MLPA)	COL1A2	COL1A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Ehlers-Danlos syndrome 7B (EDS7B) [MIM:130060]: A connective tissue disorder characterized by hyperextensible skin, atrophic cutaneous scars due to tissue fragility and joint hyperlaxity. Marked by bilateral congenital hip dislocation, hyperlaxity of the joints, and recurrent partial dislocations. (ECO:0000269|PubMed:1577745, ECO:0000269|PubMed:2394758, ECO:0000269|PubMed:3680255). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Osteoporoz, Postmenopozal, Yatkınlık, 166710, Otozomal Dominant (CALCR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, postmenopausal, susceptibility, 166710, Autosomal dominant (CALCR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALCR	CALCR, CRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Osteoporoz, Yatkınlık, 166710, Otozomal Dominant (PDLIM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteoporosis, susceptibility to, 166710, Autosomal dominant (PDLIM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDLIM4	RIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Koroid Pleksus Papilloma, 260500, Otozomal Dominant (Koroid Pleksus Karsinomu) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteosarcoma, 259500, Autosomal recessive (Choroid plexus carcinoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affectino the gene represented in this entry.	EDTA Blood Sample
Osteosarkom, Somatik, 259500 (Osteosarkom) (RB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteosarcoma, somatic, 259500 (Osteosarcoma) (RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1	RB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Osteosarkom, Somatik, 259500 (Osteosarkom) (CHEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteosarcoma, somatic, 259500 (Osteosarcoma) (CHEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal		Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Osteoskleroz, 144750, Otozomal Dominant (Otozomal Dominant Osteoskleroz, Worth Tipi) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Osteosclerosis, 144750, Autosomal dominant (Autosomal dominant osteosclerosis, Worth type) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. (ECO:0000269	PubMed:12579474). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otodental Displazi-Kromozom 11q13 Delesyon Sendromu, 166750, Otozomal Dominant (Otodental Sendromu) (C11DEL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otodental dysplasia chromosome 11q13 deletion syndrome, 166750, Autosomal dominant (Otodental syndrome) (C11DEL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	.	OTDD, DEL11q13, C11DEL@13	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Otodental Displazi-Kromozom 11q13 Delesyon Sendromu, 166750, Otozomal Dominant (Otodental Sendromu) (Prenatal)	Otodental dysplasia chromosome 11q13 deletion syndrome, 166750, Autosomal dominant (Otodental syndrome) (Prenatal)	.	OTDD, DEL11q13, C11DEL@13	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otofasikoservikal Sendrom 2, 615560, Otozomal Resesif (Otofasiyoservikal Sendrom) (PAX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otofaciocervical syndrome 2, 615560, Autosomal recessive; OTFCS2 (Otofaciocervical syndrome) (PAX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX1	PAX1, OFC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Otofaciocervical syndrome 2 (OFC2) [MIM:615560]: A disorder characterized by facial dysmorphism, cup-shaped low-set ears, preauricular fistulas, hearing loss, branchial defects, skeletal anomalies including vertebral defects, low-set clavicles, winged scapulae, sloping shoulders, and mild intellectual disability. (ECO:0000269	PubMed:23851939). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Otofasikoservikal Sendrom 2, 615560, Otozomal Resesif (Otofasiyoservikal Sendrom) (Prenatal) (PAX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Otofaciocervical syndrome 2, 615560, Autosomal recessive; OTFCS2 (Otofaciocervical syndrome) (PAX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX1	PAX1, OFC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Otofaciocervical syndrome 2 (OFC2) [MIM:615560]: A disorder characterized by facial dysmorphism, cup-shaped low-set ears, preauricular fistulas, hearing loss, branchial defects,; skeletal anomalies including vertebral defects, low-set clavicles, winged scapulae, sloping shoulders, and mild intellectual disability. (ECO:0000269	PubMed:23851939). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otofasiyoservikal Sendrom, 166780, Otozomal Dominant; OTFCS (Otofasiyoservikal Sendrom) (EYA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otofaciocervical syndrome, 166780, Autosomal dominant; OTFCS (Otofaciocervical syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. (ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otofasiyoservikal Sendrom, 166780, Otozomal Dominant; OTFCS (Otofasiyoservikal Sendrom) (Prenatal) (EYA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Otofaciocervical syndrome, 166780, Autosomal dominant; OTFCS (Otofaciocervical syndrome) (EYA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EYA1	EYA1, BOR, BOS1, OFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Branchiootorenal syndrome 1 (BOR1) [MIM:113650]: A syndrome characterized by branchial cleft fistulas or cysts, sensorineural and/or conductive hearing loss, pre-auricular pits, structural defects of the outer, middle or inner ear, and renal malformations. (ECO:0000269|PubMed:10464653, ECO:0000269|PubMed:10655545, ECO:0000269|PubMed:10991693, ECO:0000269|PubMed:11558900, ECO:0000269|PubMed:21280147, ECO:0000269|PubMed:9361030). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otopalatodijital Sendrom, Tip 1, 311300, X'e Bağlı Dominant; OPD1 (Otopalatodijital Sendrom) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otopalatodigital syndrome, type I, 311300, X-linked dominant; OPD1 (Otopalatodigital syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. (ECO:0000269	PubMed:17357080). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otopalatodijital Sendrom, Tip 1, 311300, X'e Bağlı Dominant; OPD1 (Otopalatodijital Sendromi) (MLPA)	Otopalatodigital syndrome, type I, 311300, X-linked dominant; OPD1 (Otopalatodigital syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. (ECO:0000269	PubMed:17357080). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otopalatodijital Sendrom, Tip 1, 311300, X'e Bağlı Dominant; OPD1 (Otopalatodijital Sendromi) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Otopalatodigital syndrome, type I, 311300, X-linked dominant; OPD1 (Otopalatodigital syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. (ECO:0000269	PubMed:17357080). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otopalatodijital Sendrom, Tip 1, 311300, X'e Bağlı Dominant; OPD1 (Otopalatodijital Sendromi) (Prenatal) (MLPA)	Otopalatodigital syndrome, type I, 311300, X-linked dominant; OPD1 (Otopalatodigital syndrome) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. (ECO:0000269	PubMed:17357080). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otopalatodijital Sendrom, Tip 2, 304120, X'e Bağlı Dominant; OPD2 (Otopalatodijital Sendrom) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otopalatodigital syndrome, type II, 304120, X-linked dominant; OPD2 (Otopalatodigital syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. (ECO:0000269	PubMed:17357080). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Otopalatodijital Sendrom, Tip 2, 304120, X'e Bağlı Dominant; OPD2 (Otopalatodijital Sendromi) (MLPA)	Otopalatodijital syndrome, type II, 304120, X-linked dominant; OPD2 (Otopalatodigital syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otopalatodijital Sendrom, Tip 2, 304120, X'e Bağlı Dominant; OPD2 (Otopalatodijital Sendromi) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Otopalatodigital syndrome, type II, 304120, X-linked dominant; OPD2 (Otopalatodigital syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otopalatodijital Sendrom, Tip 2, 304120, X'e Bağlı Dominant; OPD2 (Otopalatodijital Sendromi) (Prenatal) (MLPA)	Otopalatodigital syndrome, type II, 304120, X-linked dominant; OPD2 (Otopalatodigital syndrome) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Otospondilomegaepifizyal Displazi, 215150, Otozomal Resesif; OSMED (Otospondilomegaepifizyal Displazi) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Otospondylomegaepiphyseal dysplasia, 215150, Autosomal recessive; OSMED (Otospondylomegaepiphyseal dysplasia) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 3 (STL3) [MIM:184840]: An autosomal dominant non-ocular form of Stickler syndrome. Classical Stickler syndrome associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular symptoms are absent. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269	PubMed:9506662}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Otospondilomegaepifizyal Displazi, 215150, Otozomal Resesif; OSMED (Otospondilomegaepifizyal Displazi) (Prenatal) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Otospondylomegaepiphyseal dysplasia, 215150, Autosomal recessive; OSMED (Otospondylomegaepiphyseal dysplasia) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 3 (STL3) [MIM:184840]: An autosomal dominant non-ocular form of Stickler syndrome. Classical Stickler syndrome associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular symptoms are absent. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269	PubMed:9506662}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ovalositoz, SA Tipi, 166900, Otozomal Dominant; SAO (Güneydoğu Asya Ovalositozis) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovalocytosis, SA type, 166900, Autosomal dominant; SAO (Southeast Asian ovalocytosis) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Over Kanseri, Somatik (ERBB2-HER2) (FISH)	Ovarian cancer, somatic (ERBB2-HER2) (FISH)	ERBB2	ERBB2, NGL, NEU, HER2	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Over Kanseri, Somatik, 167000 (OPCML geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian cancer, somatic, 167000 (OPCML gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OPCML	OPCML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Ovarian cancer (OC) [MIM:167000]: The term ovarian cancer defines malignancies originating from ovarian tissue. Although many histologic types of ovarian tumors have been described, epithelial ovarian carcinoma is the most common form. Ovarian cancers are often asymptomatic and the recognized signs and symptoms, even of late-stage disease, are vague. Consequently, most patients are diagnosed with advanced disease. {ECO:0000269	PubMed:12919783}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Over Kanseri, Somatik, 167000 (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian cancer, somatic, 167000 (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269	PubMed:9065402}. Note=The gene represented in this entry may be involved in disease pathogenesis.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Over Kanseri, Somatik, 167000 (Ekzon 4) (AKT1 geni) (Dizi Analizi) (Postnatal)	Ovarian cancer, somatic, 167000 (Exon 4) (AKT1 gene) (Sequence Analysis) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:17611497}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Over Karsinomu (SEPT9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian carcinoma (SEPT9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEPT9	SEPT9, MSF, MSF1, NAP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Over Karsinomu (RRAS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian carcinoma (RRAS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RRAS2	RRAS2, TC21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian cancer (OC) [MIM:167000]: The term ovarian cancer defines malignancies originating from ovarian tissue. Although many histologic types of ovarian tumors have been described, epithelial ovarian carcinoma is the most common form. Ovarian cancers are often asymptomatic and the recognized signs and symptoms, even of late-stage disease, are vague. Consequently, most patients are diagnosed with advanced disease. {ECO:0000269	PubMed:8052619}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Over Karsinomu, Somatik, 167000 (CDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian carcinoma, somatic, 167000 (CDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH1	CDH1, UVO, LCAM, ECAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Hereditary diffuse gastric cancer (HDGC) [MIM:137215]: A cancer predisposition syndrome with increased susceptibility to diffuse gastric cancer. Diffuse gastric cancer is a malignant disease characterized by poorly differentiated infiltrating lesions resulting in thickening of the stomach. Malignant tumors start in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. {ECO:0000269|PubMed:10319582, ECO:0000269|PubMed:12216071}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Heterozygous CDH1 germline mutations are responsible for familial cases of diffuse gastric cancer. Somatic mutations has also been found in patients with sporadic diffuse gastric cancer and lobular breast cancer.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Ovaryan Disgenezis 1, 233300, Otozomal Resesif; ODG1 (46,XX Gonadal Disgenezis) (FSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian dysgenesis 1, 233300, Autosomal recessive; ODG1 (46,XX gonadal dysgenesis) (46,XX gonadal dysgenesis) (FSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FSHR	FSHR, ODG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 1 (ODG1) [MIM:233300]: An autosomal recessive disease characterized by primary amenorrhea, variable development of secondary sex characteristics, poorly developed streak ovaries, and high serum levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH). {ECO:0000269|PubMed:10551778, ECO:0000269|PubMed:11889179, ECO:0000269|PubMed:12571157, ECO:0000269|PubMed:12915623, ECO:0000269|PubMed:7553856, ECO:0000269|PubMed:9769327, ECO:0000269|PubMed:9851774}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ovaryan Disgenezis 2, 300510; ODG2 (46,XX Gonadal Disgenezis) (BMP15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian dysgenesis 2, 300510; ODG2 (46,XX gonadal dysgenesis) (BMP15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP15	BMP15, GDF9B, ODG2, POF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 2 (ODG2) [MIM:300510]: A disorder characterized by lack of spontaneous pubertal development, primary amenorrhea, uterine hypoplasia, and hypergonadotropic hypogonadism as a result of streak gonads. {ECO:0000269	PubMed:15136966}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ovaryan Disgenezis 3, 614324, Otozomal Resesif; ODG3 (46,XX Gonadal Disgenezis) (PSMC3IP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian dysgenesis 3, 614324, Autosomal recessive; ODG3 (46,XX gonadal dysgenesis) (PSMC3IP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSMC3IP	PSMC3IP, TBPIP, GT198, HOP2, ODG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 3 (ODG3) [MIM:614324]: A disorder characterized by lack of spontaneous pubertal development, primary amenorrhea, uterine hypoplasia, and hypergonadotropic hypogonadism as a result of streak gonads. {ECO:0000269	PubMed:21963259}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ovaryan Disgenezis 4, 616185, Otozomal Resesif; ODG4 (46,XX Ovaryan Disgenesis-Kısa Boy Sendromu) (MCM9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian dysgenesis 4, 616185, Autosomal recessive; ODG4 (46,XX ovarian dysgenesis-short stature syndrome) (MCM9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCM9	MCM9, MCMDC1, ODG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 4 (ODG4) [MIM:616185]: A disorder characterized by lack of spontaneous pubertal development, primary amenorrhea, uterine hypoplasia, and hypergonadotropic hypogonadism as a result of streak gonads. {ECO:0000269	PubMed:25480036}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ovaryan Hiperstimülasyon Sendromu, 608115, Otozomal Dominant; OHSS (Ovaryan Hiperstimülasyon Sendromu) (FSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian hyperstimulation syndrome, 608115, Autosomal dominant; OHSS (Ovarian hyperstimulation syndrome) (FSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FSHR	FSHR, ODG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 1 (ODG1) [MIM:233300]: An autosomal recessive disease characterized by primary amenorrhea, variable development of secondary sex characteristics, poorly developed streak ovaries, and high serum levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH). {ECO:0000269|PubMed:10551778, ECO:0000269|PubMed:11889179, ECO:0000269|PubMed:12571157, ECO:0000269|PubMed:12915623, ECO:0000269|PubMed:7553856, ECO:0000269|PubMed:9769327, ECO:0000269|PubMed:9851774}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

FSH Uyarısına Over Yanıtı, 276400, Otozomal Resesif (FSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovarian response to FSH stimulation, 276400, Autosomal recessive (FSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FSHR	FSHR, ODG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 1 (ODG1) [MIM:233300]: An autosomal recessive disease characterized by primary amenorrhea, variable development of secondary sex characteristics, poorly developed streak ovaries, and high serum levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH). ECO:0000269|PubMed:10051778, ECO:0000269|PubMed:11889179, ECO:0000269|PubMed:12571157, ECO:0000269|PubMed:12915623, ECO:0000269|PubMed:7553856, ECO:0000269|PubMed:9769327, ECO:0000269|PubMed:9851774). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (CACH Sendromu) (EIF2B5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (CACH syndrome) (EIF2B5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B5	EIF2B5, LVWM, CACH, CLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12325082, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:19158808, ECO:0000269|PubMed:21484434). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (CACH Sendromu) (Prenatal) (EIF2B5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (CACH syndrome) (EIF2B5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B5	EIF2B5, LVWM, CACH, CLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12325082, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:19158808, ECO:0000269|PubMed:21484434). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (Ovariolökodistrofi) (EIF2B4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (Ovarioleukodystrophy) (EIF2B4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B4	EIF2B4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11835386, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (Ovariolökodistrofi) (EIF2B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (Ovarioleukodystrophy) (EIF2B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B2	EIF2B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:21484434, ECO:0000269|PubMed:22285377, ECO:0000269|PubMed:22729508). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (Ovariolökodistrofi) (Prenatal) (EIF2B4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (Ovariolekuokodystrofi) (Prenatal) (EIF2B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B4	EIF2B4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11835386, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ovariolökodistrofi, 603896, Otozomal Resesif (Ovariolökodistrofi) (Prenatal) (EIF2B2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ovariolekuokodystrophy, 603896, Autosomal recessive (Ovariolekuokodystrofi) (Prenatal) (EIF2B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF2B2	EIF2B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy with vanishing white matter (VWM) [MIM:603896]: A leukodystrophy that occurs mainly in children. Neurological signs include progressive cerebellar ataxia, spasticity, inconstant optic atrophy and relatively preserved mental abilities. The disease is chronic-progressive with, in most individuals, additional episodes of rapid deterioration following febrile infections or minor head trauma. While childhood onset is the most common form of the disorder, some severe forms are apparent at birth. A severe, early-onset form seen among the Cree and Chippewayan populations of Quebec and Manitoba is called Cree leukoencephalopathy. Milder forms may not become evident until adolescence or adulthood. Some females with milder forms of the disease who survive to adolescence exhibit ovarian dysfunction. This variant of the disorder is called ovarioleukodystrophy. (ECO:0000269|PubMed:11704758, ECO:0000269|PubMed:12707859, ECO:0000269|PubMed:15776425, ECO:0000269|PubMed:21484434, ECO:0000269|PubMed:22285377, ECO:0000269|PubMed:22729508). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Aşırı Hidrate Kalıtsal Stomatositozis, 185000, Otozomal Dominant; OHST (Aşırı Hidrate Kalıtsal Stomatositoz) (RHAG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Overhydrated hereditary stomatocytosis, 185000, Autosomal dominant; OHST (Overhydrated hereditary stomatocytosis) (RHAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHAG	RHAG, RH50A, OHST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Aşırı Hidrate Kalıtsal Stomatositozis, 185000, Otozomal Dominant; OHST (Aşırı Hidrate Kalıtsal Stomatositoz) (Prenatal) (RHAG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Overhydrated hereditary stomatocytosis, 185000, Autosomal dominant; OHST (Overhydrated hereditary stomatocytosis) (RHAG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RHAG	RHAG, RH50A, OHST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
P16 (CDKN2A) (FISH)	P16 (CDKN2A) (FISH)	9p21.3	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
P53 Geni (TP53) (17p13.1) (FISH)	P53 Gene (TP53) (17p13.1) (FISH)	17p13.1	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu/ Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample/ Heparinized blood sample / Heparinized bone marrow
Pakonişi Konjenita 1, 167200, Otozomal Dominant; PC1 (Pakonişi Konjenita) (KRT16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pachyonychia congenita 1, 167200, Autosomal dominant; PC1 (Pachyonychia congenita) (KRT16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT16	KRT16, FNEPPK, PC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Unilateral palmoplantar verrucous nevus (UPVN) [MIM:144200]: UPVN is characterized by a localized epidermolytic hyperkeratosis in parts of the right palm and the right sole, following the lines of Blaschko. [ECO:0000269	PubMed:10844556). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Kemiğin Paget Hastalığı 2, Erken Başlangıçlı, 602080, Otozomal Dominant; PDB2 (Alkesel Ekspersil Osteoliz) (TNFRSF11A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paget disease of bone 2, early-onset, 602080, Autosomal dominant; PDB2 (Familial expansile osteolysis) (TNFRSF11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF11A	TNFRSF11A, RANK, ODFR, FEO, OPTB7, PDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 2, early-onset (PDB2) [MIM:602080]: A form of Paget disease, a disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269	PubMed:10615125}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kemiğin Paget Hastalığı 3, 167250, Otozomal Dominant; PDB3 (SQSTM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paget disease of bone 3, 167250, Autosomal dominant; PDB3 (SQSTM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SQSTM1	SQSTM1, P62, PDB3, FTDALS3, NADGP, DMRV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 3 (PDB3) [MIM:167250]: A disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. {ECO:0000269|PubMed:11992264, ECO:0000269|PubMed:12374763, ECO:0000269|PubMed:14584883, ECO:0000269|PubMed:15125799, ECO:0000269|PubMed:15146436, ECO:0000269|PubMed:15176995, ECO:0000269|PubMed:15207768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kemiğin Paget Hastalığı 5, Jüvenil Başlangıçlı, 239000, Otozomal Resesif; PDB5 (Jüvenil Paget Hastalığı) (TNFRSF11B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paget disease of bone 5, juvenile-onset, 239000, Autosomal recessive; PDB5 (Juvenile Paget disease) (TNFRSF11B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF11B	TNFRSF11B, OPG, OCIF, PDB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paget disease of bone 5, juvenile-onset (PDB5) [MIM:239000]: An autosomal recessive, juvenile-onset form of Paget disease, a disorder of bone remodeling characterized by increased bone turnover affecting one or more sites throughout the skeleton, primarily the axial skeleton. Osteoclastic overactivity followed by compensatory osteoblastic activity leads to a structurally disorganized mosaic of bone (woven bone), which is mechanically weaker, larger, less compact, more vascular, and more susceptible to fracture than normal adult lamellar bone. PDB5 clinical manifestations include short stature, progressive long bone deformities, fractures, vertebral collapse, skull enlargement, and hyperostosis with progressive deafness. {ECO:0000269	PubMed:12189164}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Kemiğin Paget Hastalığı 6, 616833, Otozomal Dominant (ZNF687 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paget disease of bone 6, 616833, Autosomal dominant (ZNF687 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF687	ZNF687, KIAA1441, PDB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
PAI - 14G/ 5G Polimorfizm (.gen) (Dizi Analizi) (Postnatal)	PAI-1 4G/5G polymorphism (.gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Pallister-Hall Sendromu, 146510, Otozomal Dominant; PHS (Pallister-Hall Sendromu) (GLI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pallister-Hall syndrome, 146510, Autosomal dominant; PHS (Pallister-Hall syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pallister-Hall Sendromu, 146510, Otozomal Dominant; PHS (Pallister-Hall Sendromu) (MLPA)	Pallister-Hall syndrome, 146510, Autosomal dominant; PHS (Pallister-Hall syndrome) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pallister-Hall Sendromu, 146510, Otozomal Dominant; PHS (Pallister-Hall Sendromu) (Prenatal) (GLI3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pallister-Hall syndrome, 146510, Autosomal dominant; PHS (Pallister-Hall syndrome) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pallister-Hall Sendromu, 146510, Otozomal Dominant; PHS (Pallister-Hall Sendromu) (Prenatal) (MLPA)	Pallister-Hall syndrome, 146510, Autosomal dominant; PHS (Pallister-Hall syndrome) (Prenatal) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Palmoplantar Karsinoma, Multiple Kendi İyileşen, 615255 (NLRP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar carcinoma, multiple self-healing, 615255 (NLRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP1	NLRP1, NALP1, KIAA0926, DEFCAP, CARD7, SLEV1, VAMAS1, MSPC, AIADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitiligo-associated multiple autoimmune disease 1 (VAMAS1) [MIM:606579]: A disorder characterized by the association of vitiligo with several autoimmune and autoinflammatory diseases including autoimmune thyroid disease, rheumatoid arthritis and systemic lupus erythematosus. {ECO:0000269	PubMed:17377159}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Hiperkeratoz ve Gerçek Hermafroditizm, 610644, Otozomal Resesif (Palmoplantar Keratoderma-XX Cinsiyet Farklılaşma Bozukluğu-Skuamöz Hücre Karsinoması Yatkınlığı Sendromu) (RSPO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar hyperkeratosis and true hermaphroditism, 610644, Autosomal recessive (Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome) (RSPO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPO1	RSPO1, FLJ40906	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratoderma, palmoplantar, with squamous cell carcinoma of skin and sex reversal (PKKSCC) [MIM:610644]: A recessive syndrome characterized by XX (female to male) SRY-independent sex reversal, palmoplantar hyperkeratosis and predisposition to squamous cell carcinoma of the skin. {ECO:0000269	PubMed:17041600}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Palmoplantar Hiperkeratozis, Derinin Skuamöz Hücreli Karsinması ve Cinsiyet Uyumsuzluğu, 610644, Otozomal Resesif (Palmoplantar Keratoderma-XX Cinsiyet Farklılaşma Bozukluğu-Skuamöz Hücre Karsinoması Yatkınlığı Sendromu) (RSPO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644, Autosomal recessive (Palmoplantar keratoderma-XX sex reversal-predisposition to squamous cell carcinoma syndrome) (RSPO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RSPO1	RSPO1, FLJ40906	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratoderma, palmoplantar, with squamous cell carcinoma of skin and sex reversal (PKKSCC) [MIM:610644]: A recessive syndrome characterized by XX (female to male) SRY-independent sex reversal, palmoplantar hyperkeratosis and predisposition to squamous cell carcinoma of the skin. {ECO:0000269	PubMed:17041600}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Palmoplantar Keratoderma ve Konjenital Alopesi 1; Ppkca1 (Otozomal Dominant Palmoplantar Keratoderma ve Konjenital Alopesi) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PALMOPLANTAR KERATODERMA AND CONGENITAL ALOPECIA 1; PPKCA1 (Autosomal dominant palmoplantar keratoderma and congenital alopecia) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. {ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:15219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Keratoderma ve Yünsü Saç, 616099, Otozomal Resesif; PPKWH (Palmoplantar Keratoderma-Yünsü Saç Sendromu) (KANK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma and woolly hair, 616099, Autosomal recessive; PPKWH (Woolly hair-palmoplantar keratoderma syndrome) (KANK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KANK2	KANK2, ANKRD25, KIAA1518, PPKWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Palmoplantar keratoderma and woolly hair (PPKWH) [MIM:616099]: A disorder characterized by abnormal thickening of the skin on the palms and soles, in association with woolly scalp hair. Affected individuals manifest a variable degree of striate palmoplantar keratoderma, generally more severe on the soles. Leukonychia is more pronounced on the fingernails than toenails. Scalp hair, body hair, eyebrows, and eyelashes are sparse. The fifth toes show variable degrees of pseudoainhum, ranging from external rotation to a deep sulcus at the digitoplantar fold, accompanied by a bulbous appearance of the distal toe. {ECO:0000269	PubMed:24671081}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Palmoplantar Keratoderma I, Çizgili, Fokal Veya Diffüz Ppks1 (Fokal Palmoplantar Keratoderma - Eldeki Keratozlar) (DSG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PALMOPLANTAR KERATODERMA I, STRIATE, FOCAL, OR DIFFUSE, PPKS1 (Focal palmoplantar keratoderma with joint keratoses) (DSG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSG1	DSG1, PPKS1, SPPK1, EPKHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Palmoplantar keratoderma 1, striate, focal, or diffuse (PPKS1) [MIM:148700]: A dermatological disorder characterized by thickening of the skin on the palms and soles, and longitudinal hyperkeratotic lesions on the palms, running the length of each finger. {ECO:0002658	PubMed:10332026}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Palmoplantar Keratoderma - Konjenital Alopesi. 121014, Otozomal Dominant (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma with congenital alopecia, 121014, autosomal dominant (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. {ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729830, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508441}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Keratoderma, Bothnian Tipi, 600231, Otozomal Dominant; PPKB (Epidermolitik Olmayan Palmoplantar Keratoderma, Bothnian Tipi) (AQP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, Bothnian type, 600231, Autosomal dominant; PPKB (non-epidermolytic palmoplantar keratoderma) (AQP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AQP5	AQP5, PPKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratoderma, palmoplantar, Bothnian type (PPKB) [MIM:600231]: A dermatological disorder characterized by diffuse non-epidermolytic hyperkeratosis of the skin of palms and soles. PPKB is frequently complicated by fungal infections. {ECO:0000269	PubMed:23830519}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Keratoderma, Epidermolitik, 144200, Otozomal Dominant (Epidermolitik Palmoplantar Keratoderma) (KRT9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, epidermolytic, 144200, Autosomal dominant (Epidermolytic palmoplantar keratoderma) (KRT9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT9	KRT9, EPPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Keratoderma, palmoplantar, epidermolytic (EPPK) [MIM:144200]: A dermatological disorder characterized by diffuse thickening of the epidermis on the entire surface of palms and soles sharply bordered with erythematous margins. Some patients may present knuckle pads, thick pads of skin appearing over the proximal phalangeal joints. {ECO:0000269|PubMed:10632938, ECO:0000269|PubMed:10844507, ECO:0000269|PubMed:12072061, ECO:0000269|PubMed:12192490, ECO:0000269|PubMed:12838553, ECO:0000269|PubMed:12926810, ECO:0000269|PubMed:14675368, ECO:0000269|PubMed:15090359, ECO:0000269|PubMed:15115518, ECO:0000269|PubMed:15605275, ECO:0000269|PubMed:16911293, ECO:0000269|PubMed:16961539, ECO:0000269|PubMed:19874353, ECO:0000269|PubMed:20964665, ECO:0000269|PubMed:21715251, ECO:0000269|PubMed:7511021, ECO:0000269|PubMed:7512862, ECO:0000269|PubMed:7516304, ECO:0000269|PubMed:7523529, ECO:0000269|PubMed:7532199, ECO:0000269|PubMed:8647270, ECO:0000269|PubMed:9204965, ECO:0000269|PubMed:9856842}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Palmoplantar Keratoderma, Epidermolitik, 144200, Otozomal Dominant; EPPK (Epidermolitik Palmoplantar Keratoderma) (KRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, epidermolytic, 144200, Autosomal dominant; EPPK (Epidermolytic palmoplantar keratoderma) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Keratoderma, Nagashima Tipi, 615598, Otozomal Resesif; PPKN (Palmoplantar Keratoderma, Nagashima Tipi) (SERPINB7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, Nagashima type, 615598, Autosomal recessive; PPKN (Palmoplantar keratoderma, Nagashima type) (SERPINB7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINB7	SERPINB7, MEGSIN, PPKN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Palmoplantar Keratoderma, Nonepidermolitik, 600962, Otozomal Dominant; NEPPK (Thost-Unna Palmoplantar Keratoderma) (KRT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, nonepidermolytic, 600962, Autosomal dominant; NEPPK (Thost-Unna palmoplantar keratoderma) (KRT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT1	KRT1, EPPK, NEPPK, EHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Epidermolytic hyperkeratosis (EHK) [MIM:113800]: An autosomal dominant skin disorder characterized by widespread blistering and an ichthyotic erythroderma at birth that persist into adulthood. Histologically there is a diffuse epidermolytic degeneration in the lower spinous layer of the epidermis. Within a few weeks from birth, erythroderma and blister formation diminish and hyperkeratoses develop. {ECO:0000269|PubMed:10232403, ECO:0000269|PubMed:10688370, ECO:0000269|PubMed:10844506, ECO:0000269|PubMed:11531804, ECO:0000269|PubMed:12406348, ECO:0000269|PubMed:1380725, ECO:0000269|PubMed:1381288, ECO:0000269|PubMed:21271994, ECO:0000269|PubMed:7507151, ECO:0000269|PubMed:7507152, ECO:0000269|PubMed:7512983, ECO:0000269|PubMed:9856846}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Palmoplantar Keratoderma, Nonepidermolitik Fokal 2, 616400, Otozomal Dominant; FNEPPK2 (Izole Fokal Non-Epidermolitik Palmoplantar Keratoderma) (TRPV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400, Autosomal dominant; FNEPPK2 (Isolated focal non-epidermolytic palmoplantar keratoderma) (TRPV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV3	TRPV3, OLMS, FNEPPK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Palmoplantar keratoderma, non-epidermolytic, focal 2 (FNEPPK2) [MIM:616400]: A dermatological disorder characterized by non-epidermolytic, abnormal thickening of the skin on the palms and soles. Focal palmoplantar keratoderma consists of localized areas of hyperkeratosis located mainly on pressure points and areas of recurrent friction. {ECO:0000269	PubMed:25285920}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Palmoplantar Keratoderma, Nonepidermolitik, Fokal veya Yaygın, 615735, Otozomal Dominant; PPKNEFD (Otozomal Dominant Fokal Epidermolitik Olmayan Palmoplantar Keratoderma-Plantar Su Toplama) (KRT6C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735, Autosomal dominant; PPKNEFD (Autosomal dominant focal non-epidermolytic palmoplantar keratoderma with plantar blistering) (KRT6C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT6C	KRT6C, PPKNEFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Palmoplantar Keratoderma, Nonepidermolitik, Fokal, 613000, Otozomal Dominant (KRT16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Palmoplantar keratoderma, nonepidermolytic, focal, 613000, Autosomal dominant (KRT16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT16	KRT16, FNEPPK, PC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Unilateral palmoplantar verrucous nevus (UPVN) [MIM:144200]: UPVN is characterized by a localized epidermolytic hyperkeratosis in parts of the right palm and the right sole, following the lines of Blaschko. {ECO:0000269	PubMed:12844556}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatik Agenezis 1, 260370, Otozomal Resesif; PAGEN1 (Parsiyel Pankreatik Agenezi) (PDX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic agenesis 1, 260370, Autosomal recessive; PAGEN1 (Partial pancreatic agenesis) (PDX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDX1	PDX1, IPF1, MODY4, PAGEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatik Agenezis 1, 260370, Otozomal Resesif; PAGEN1 (Parsiyel Pankreatik Agenezi) (Prenatal) (PDX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pancreatic agenesis 1, 260370, Autosomal recessive; PAGEN1 (Partial pancreatic agenesis) (PDX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDX1	PDX1, IPF1, MODY4, PAGEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pankreatik Agenezis 2, 615935, Otozomal Resesif; PAGEN2 (Parsiyel Pankreatik Agenezi) (PTF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic agenesis 2, 615935, Autosomal recessive; PAGEN2 (Partial pancreatic agenesis) (PTF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTF1A	PTF1A, PACA, PAGEN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatic agenesis 2 (PAGEN2) [MIM:615935]: A disease characterized by isolated hypoplasia or agenesis of the pancreas, pancreatic beta-cell failure resulting in neonatal insulin-dependent diabetes mellitus, and exocrine pancreatic insufficiency. {ECO:0000269|PubMed:24212882}. Note=The disease is caused by mutations affecting the gene represented in this entry. In some families with pancreatic agenesis, disease causing mutations affect the sequence and activity of an enhancer region of 400-bp located 25 kb downstream of PTF1A (PubMed:24212882). {ECO:0000269|PubMed:24212882}.;	EDTA Blood Sample
Pankreatik Agenezis 2, 615935, Otozomal Resesif; PAGEN2 (Parsiyel Pankreatik Agenezi) (Prenatal) (PTF1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pancreatic agenesis 2, 615935, Autosomal recessive; PAGEN2 (Partial pancreatic agenesis) (PTF1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTF1A	PTF1A, PACA, PAGEN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pancreatic agenesis 2 (PAGEN2) [MIM:615935]: A disease characterized by isolated hypoplasia or agenesis of the pancreas, pancreatic beta-cell failure resulting in neonatal insulin-dependent diabetes mellitus, and exocrine pancreatic insufficiency. {ECO:0000269|PubMed:24212882}. Note=The disease is caused by mutations affecting the gene represented in this entry. In some families with pancreatic agenesis, disease causing mutations affect the sequence and activity of an enhancer region of 400-bp located 25 kb downstream of PTF1A (PubMed:24212882). {ECO:0000269|PubMed:24212882}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Pankreatik Agenezis ve Konjenital Kalp Kusurları, 600001, Otozomal Dominant (Pankreatik Hipoplazi-Diyabet-Konjenital Kalp Hastalığı Sendromu) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic agenesis and congenital heart defects, 600001, Autosomal dominant (Pancreatic hypoplasia-diabetes-congenital heart disease syndrome) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatik Agenezis ve Konjenital Kalp Kusurları, 600001, Otozomal Dominant (Pankreatik Hipoplazi-Diyabet-Konjenital Kalp Hastalığı Sendromu) (Prenatal) (GATA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pancreatic agenesis and congenital heart defects, 600001, Autosomal dominant (Pancreatic hypoplasia-diabetes-congenital heart disease syndrome) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pankreas ve Beyincik Agenezisi, 609069, Otozomal Resesif; PACA (Kalıcı Neonatal Diyabetes Mellitus-Pankreatik ve Serebellar Agenezi Sendromu) (PTF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic and cerebellar agenesis, 609069, Autosomal recessive; PACA (Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome) (PTF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTF1A	PTF1A, PACA, PAGEN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatic agenesis 2 (PAGEN2) [MIM:615935]: A disease characterized by isolated hypoplasia or agenesis of the pancreas, pancreatic beta-cell failure resulting in neonatal insulin-dependent diabetes mellitus, and exocrine pancreatic insufficiency. {ECO:0000269|PubMed:24212882}. Note=The disease is caused by mutations affecting the gene represented in this entry. In some families with pancreatic agenesis, disease causing mutations affect the sequence and activity of an enhancer region of 400-bp located 25 kb downstream of PTF1A (PubMed:24212882). {ECO:0000269|PubMed:24212882}.	EDTA Blood Sample
Pankreas ve Beyincik Agenezisi, 609069, Otozomal Resesif; PACA (Kalıcı Neonatal Diyabetes Mellitus-Pankreatik ve Serebellar Agenezi Sendromu) (Prenatal) (PTF1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pancreatic and cerebellar agenesis, 609069, Autosomal recessive; PACA (Permanent neonatal diabetes mellitus-pancreatic and cerebellar agenesis syndrome) (PTF1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTF1A	PTF1A, PACA, PAGEN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pancreatic agenesis 2 (PAGEN2) [MIM:615935]: A disease characterized by isolated hypoplasia or agenesis of the pancreas, pancreatic beta-cell failure resulting in neonatal insulin-dependent diabetes mellitus, and exocrine pancreatic insufficiency. {ECO:0000269|PubMed:24212882}. Note=The disease is caused by mutations affecting the gene represented in this entry. In some families with pancreatic agenesis, disease causing mutations affect the sequence and activity of an enhancer region of 400-bp located 25 kb downstream of PTF1A (PubMed:24212882). {ECO:0000269|PubMed:24212882}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pankreas Kanseri (Ailesel Pankreas Karsinomu) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PANCREATIC CANCER (Familial pancreatic carcinoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri (Ailesel Pankreas Karsinomu) (MLPA)	PANCREATIC CANCER (Familial pancreatic carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri (Ailesel Pankreas Karsinomu) (MLPA)	PANCREATIC CANCER (Familial pancreatic carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri 2, 613347 (Ailesel Pankreas Karsinomu) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer 2, 613347 (Familial pancreatic carcinoma) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri 2, 613347 (Ailesel Pankreas Karsinomu) (MLPA)	Pancreatic cancer 2, 613347 (Familial pancreatic carcinoma) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri, 260350, Otozomal Dominant, Somatik Mutasyon, Multifaktöriyel (STK11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, 260350, Autosomal dominant, Somatic mutation, Multifactorial (STK11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STK11	STK11, PJS, LKB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, 260350, Somatik Mosaik, Otozomal Dominant, Multifaktöriyel (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, 260350, somatic mosaic, autosomal dominant, multifactorial (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, 260350, Somatik Mosaik, Otozomal Dominant, Multifaktöriyel (MLPA)	Pancreatic cancer, 260350, somatic mosaic, autosomal dominant, multifactorial) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		20	60	.	.	30	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, Somatik (ACVR1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, somatic (ACVR1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACVR1B	ACVR1B, ACVRLK4, ALK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Note=ACVRIB is abundantly expressed in systemic sclerosis patient fibroblasts and production of collagen is also induced by activin-A/HHBA. This suggests that the activin/ACRV1B signaling mechanism is involved in systemic sclerosis. {ECO:0000269|PubMed:21377836}.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, Somatik, 260350 (SMAD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, somatic, 260350 (SMAD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, Yatkınlık, 1, 606856 (Ailesel Pankreas Karsinomu) (PALLD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, susceptibility to, 1, 606856 (Familial pancreatic carcinoma) (PALLD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PALLD	PALLD, KIAA0992, PNCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatic cancer 1 (PNCA1) [MIM:606856]: A malignant neoplasm of the pancreas. Tumors can arise from both the exocrine and endocrine portions of the pancreas, but 95% of them develop from the exocrine portion, including the ductal epithelium, acinar cells, connective tissue, and lymphatic tissue. {ECO:0000269|PubMed:17194196, ECO:0000269|PubMed:17415588}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri, Yatkınlık, 3, 613348 (Ailesel Pankreas Karsinomu) (PALB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, susceptibility to, 3, 613348 (Familial pancreatic carcinoma) (PALB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PALB2	PALB2, FANCN, PNCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:17287723, ECO:0000269|PubMed:22241545}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Breast cancer susceptibility is strongly associated with PALB2 truncating mutations. Conversely, rare missense mutations do not strongly influence breast cancer risk (PubMed:22241545). {ECO:0000269|PubMed:22241545}.	EDTA Blood Sample

Pankreas Kanseri, Yatkınlık, 4, 614320; PNCA4 (Ailesel Pankreas Karsinomu) (BRCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer, susceptibility to, 4, 614320; PNCA4 (Familial pancreatic carcinoma) (BRCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA1	BRCA1, PSCP, BROVCA1, PNCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10323242, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:18285836, ECO:0000269|PubMed:7545954, ECO:0000269|PubMed:7894491, ECO:0000269|PubMed:7894493, ECO:0000269|PubMed:7939630, ECO:0000269|PubMed:8554067, ECO:0000269|PubMed:8723683, ECO:0000269|PubMed:8776600, ECO:0000269|PubMed:9482581, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9760198}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Mutations in BRCA1 are thought to be responsible for 45% of inherited breast cancer. Moreover, BRCA1 carriers have a 4-fold increased risk of colon cancer, whereas male carriers face a 3- fold increased risk of prostate cancer. Cells lacking BRCA1 show defects in DNA repair by homologous recombination.	EDTA Blood Sample
Pankreas Kanseri, Yatkınlık, 4, 614320; PNCA4 (Ailesel Pankreas Karsinomu) (MLPA)	Pancreatic cancer, susceptibility to, 4, 614320; PNCA4 (Familial pancreatic carcinoma) (MLPA)	BRCA1	BRCA1, PSCP, BROVCA1, PNCA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10323242, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:18285836, ECO:0000269|PubMed:7545954, ECO:0000269|PubMed:7894491, ECO:0000269|PubMed:7894493, ECO:0000269|PubMed:7939630, ECO:0000269|PubMed:8554067, ECO:0000269|PubMed:8723683, ECO:0000269|PubMed:8776600, ECO:0000269|PubMed:9482581, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9760198}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Mutations in BRCA1 are thought to be responsible for 45% of inherited breast cancer. Moreover, BRCA1 carriers have a 4-fold increased risk of colon cancer, whereas male carriers face a 3- fold increased risk of prostate cancer. Cells lacking BRCA1 show defects in DNA repair by homologous recombination.	EDTA Blood Sample
Pankreas Kanseri / Melanom Sendromu, 606719, Otozomal Dominant (Ailesel Atipik Multipl Mol Melanom Sendromu) (CDKN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic cancer/melanoma syndrome, 606719, Autosomal dominant (Familial atypical multiple mole melanoma syndrome) (CDKN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDKN2A	CDKN2A, MTS1, P16, MLM, CMM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Li-Fraumeni syndrome (LFS) [MIM:151623]: Autosomal dominant familial cancer syndrome that in its classic form is defined by the existence of a proband affected by a sarcoma before 45 years with a first degree relative affected by any tumor before 45 years and another first degree relative with any tumor before 45 years or a sarcoma at any age. Other clinical definitions for LFS have been proposed (PubMed:8118819 and PubMed:8718514) and called Li-Fraumeni like syndrome (LFL). In these families affected relatives develop a diverse set of malignancies at unusually early ages. Four types of cancers account for 80% of tumors occurring in TP53 germline mutation carriers: breast cancers, soft tissue and bone sarcomas, brain tumors (astrocytomas) and adrenocortical carcinomas. Less frequent tumors include choroid plexus carcinoma or papilloma before the age of 15, rhabdomyosarcoma before the age of 5, leukemia, Wilms tumor, malignant phyllodes tumor, colorectal and gastric cancers. {ECO:0000269	PubMed:10494981}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreas Kanseri, Somatik (RBBP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic carcinoma, somatic (RBBP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBBP8	RBBP8, RIM, SCKL2, JWDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Jawad syndrome (JWDS) [MIM:251255]: A syndrome characterized by congenital microcephaly, moderately severe mental retardation, and symmetrical digital anomalies. Digital malformations of variable degree include hallux valgus, syndactyly of toes 4 and 5, short fifth fingers, single flexion crease of fifth fingers, polydactyly and syndydactyly. {ECO:0000269	PubMed:21998596}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreas Kanseri, Somatik, 260350 (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic carcinoma, somatic, 260350 (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pankreatik Lipaz Eksikliği, 614338, Otozomal Resesif; PNLIPD (Kombine Pankreatik Lipaz-Kolipaz Eksikliği) (PNLIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatic lipase deficiency, 614338, Autosomal recessive; PNLIPD (Combined pancreatic lipase-colipase deficiency) (PNLIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNLIP	PNLIP, PNLIPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pankreatit, Kronik, Yatkınlık, 167800, Otozomal Dominant (Ailesel Kronik Pankreatit) (CTRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatitis, chronic, susceptibility to, 167800, Autosomal dominant (Hereditary chronic pancreatitis) (CTRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTRC	CTRC, CLCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pankreatit, Herediter, 167800, Otozomal Dominant (Ailesel Kronik Pankreatit) (PRSS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatitis, hereditary, 167800, Autosomal dominant (Hereditary chronic pancreatitis) (PRSS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRSS1	PRSS1, TRY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. {ECO:0000269|PubMed:10204851, ECO:0000269|PubMed:10381903, ECO:0000269|PubMed:10930381, ECO:0000269|PubMed:11073545, ECO:0000269|PubMed:11788572, ECO:0000269|PubMed:11866271, ECO:0000269|PubMed:14695529, ECO:0000269|PubMed:15776435, ECO:0000269|PubMed:8841182, ECO:0000269|PubMed:9322498, ECO:0000269|PubMed:9633818}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatit, Herediter, 167800, Otozomal Dominant (Ailesel Kronik Pankreatit) (MLPA)	Pancreatitis, hereditary, 167800, Autosomal dominant (Hereditary chronic pancreatitis) (MLPA)	PRSS1	PRSS1, TRY1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. {ECO:0000269|PubMed:10204851, ECO:0000269|PubMed:10381903, ECO:0000269|PubMed:10930381, ECO:0000269|PubMed:11073545, ECO:0000269|PubMed:11788572, ECO:0000269|PubMed:11866271, ECO:0000269|PubMed:14695529, ECO:0000269|PubMed:15776435, ECO:0000269|PubMed:8841182, ECO:0000269|PubMed:9322498, ECO:0000269|PubMed:9633818}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatit, Herediter, 167800, Otozomal Dominant; PCTT (Ailesel Kronik Pankreatit) (SPINK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatitis, hereditary, 167800, Autosomal dominant; PCTT (Hereditary chronic pancreatitis) (SPINK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. {ECO:0000269|PubMed:10691414, ECO:0000269|PubMed:10835640, ECO:0000269|PubMed:12974284}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pankreatit, Herediter, 167800, Otozomal Dominant; PCTT (Ailesel Kronik Pankreatit) (MLPA)	Pancreatitis, hereditary, 167800, Autosomal dominant; PCTT (Hereditary chronic pancreatitis) (MLPA)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. {ECO:0000269|PubMed:10691414, ECO:0000269|PubMed:10835640, ECO:0000269|PubMed:12974284}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

Pankreatit, İdyopatik, 167800, Otozomal Dominant (Ailesel Kronik Pankreatit) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pancreatitis, idiopathic, 167800, Autosomal dominant (Hereditary chronic pancreatitis) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Pankreatit, İdyopatik, 167800, Otozomal Dominant (Ailesel Kronik Pankreatit) (MLPA)	Pancreatitis, idiopathic, 167800, Autosomal dominant (Hereditary chronic pancreatitis) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407	EDTA Blood Sample
Panhipopituitarizm, X'e Bağlı, 312000, X'e Bağlı, PHPX (Panhipopituitarizm, X'e Bağlı) (SOX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Panthypopituitarism, X-linked, 312000, X-linked; PHPX (Panhypopituitarism) (SOX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX3	SOX3, MRGH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, with isolated growth hormone deficiency (MRXGH) [MIM:300123]: A disorder characterized by the association of variable degrees of mental retardation with panhypopituitarism, variable combinations of hypothyroidism, delayed pubertal development, and short stature due to growth hormone deficiency. {ECO:0000269|PubMed:12428212}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Panik Bozukluğu, Yatkınlık, 167870, Otozomal Dominant (COMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Panic disorder, susceptibility to, 167870, Autosomal dominant (COMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COMT	COMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Koroid Pleksusun Papillaması; Cpp (Koroid Pleksus Karsinomu) (TP53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PAPILLOMA OF CHOROID PLEXUS; CPP (Choroid plexus carcinoma) (TP53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP53	TP53, P53, LFS1 , BCC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Koroid Pleksusun Papillaması; Cpp (Koroid Pleksus Karsinomu) (MLPA)	PAPILLOMA OF CHOROID PLEXUS; CPP (Choroid plexus carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Koroid Pleksusun Papillaması; Cpp (Koroid Pleksus Karsinomu) (MLPA)	PAPILLOMA OF CHOROID PLEXUS; CPP (Choroid plexus carcinoma) (MLPA)	TP53	TP53, P53, LFS1 , BCC7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Papillon-Lefevre Sendromu, 245000, Otozomal Resesif; PALS (Papillon-Lefevre Sendromu) (CTSC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Papillon-Lefevre syndrome, 245000, Autosomal recessive; PALS (Papillon-Lefevre syndrome) (CTSC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSC	CTSC, CPPI, PALS, PLS, HMS, PDON1, JPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periodontitis, aggressive, 1 (AP1) [MIM:170650]: A disease characterized by severe and protracted gingival infections, generalized or localized, leading to tooth loss. Amounts of microbial deposits are generally inconsistent with the severity of periodontal tissue destruction and the progression of attachment and bone loss may be self arresting. {ECO:0000269|PubMed:10662808, ECO:0000269|PubMed:14974080}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Papillorenal Sendrom, 120330, Otozomal Dominant; PAPRS (Renal Koloboma Sendromu) (PAX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Papillorenal syndrome, 120330, Autosomal dominant; PAPRS (Renal coloboma syndrome) (PAX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX2	PAX2, PAPRS, FSGS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Papillorenal syndrome (PAPRS) [MIM:120330]: An autosomal dominant disorder characterized by both ocular and renal anomalies, but may also include vesicoureteral reflux, high frequency hearing loss, central nervous system anomalies, and/or genital anomalies. Eye anomalies in this disorder consist of a wide and sometimes excavated dysplastic optic disk with the emergence of the retinal vessels from the periphery of the disk, designated optic nerve coloboma or 'morning glory' anomaly. Associated findings may include a small corneal diameter, retinal coloboma, scleral staphyloma, optic nerve cyst, microphthalmia, and pigmentary macular dysplasia. The kidneys are small and abnormally formed (renal hypodysplasia), and have fewer than the normal number of glomeruli, which are enlarged (oligomeganephronia). These ocular and renal anomalies result in decreased visual acuity and retinal detachment, as well as hypertension, proteinuria, and renal insufficiency that frequently progresses to end-stage renal disease. {ECO:0000269|PubMed:15652857, ECO:0000269|PubMed:19954729, ECO:0000269|PubMed:22213154, ECO:0000269|PubMed:9760197}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Papillorenal Sendrom, 120330, Otozomal Dominant; PAPRS (Renal Koloboma Sendromu) (Prenatal) (PAX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Papillorenal syndrome, 120330, Autosomal dominant; PAPRS (Renal coloboma syndrome) (PAX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX2	PAX2, PAPRS, FSGS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Papillorenal syndrome (PAPRS) [MIM:120330]: An autosomal dominant disorder characterized by both ocular and renal anomalies, but may also include vesicoureteral reflux, high frequency hearing loss, central nervous system anomalies, and/or genital anomalies. Eye anomalies in this disorder consist of a wide and sometimes excavated dysplastic optic disk with the emergence of the retinal vessels from the periphery of the disk, designated optic nerve coloboma or 'morning glory' anomaly. Associated findings may include a small corneal diameter, retinal coloboma, scleral staphyloma, optic nerve cyst, microphthalmia, and pigmentary macular dysplasia. The kidneys are small and abnormally formed (renal hypodysplasia), and have fewer than the normal number of glomeruli, which are enlarged (oligomeganephronia). These ocular and renal anomalies result in decreased visual acuity and retinal detachment, as well as hypertension, proteinuria, and renal insufficiency that frequently progresses to end-stage renal disease. {ECO:0000269|PubMed:15652857, ECO:0000269|PubMed:19954729, ECO:0000269|PubMed:22213154, ECO:0000269|PubMed:9760197}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Paraganglioma ve Gastrik Stromal Sarkom, 606864 (Carney-Stratakis Sendromu) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paraganglioma and gastric stromal sarcoma, 606864 (Carney-Stratakis syndrome) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Paraganglioma ve Gastrik Stromal Sarkom, 606864 (Carney-Stratakis Sendromu) (SDHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paraganglioma and gastric stromal sarcoma, 606864 (Carney-Stratakis syndrome) (SDHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHC	SDHC, PGL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 3 (PGL3) [MIM:605373]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11062460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Paraganglioma ve Gastrik Stromal Sarkom, 606864 (Carney-Stratakis Sendromu) (SDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paraganglioma and gastric stromal sarcoma, 606864 (Carney-Stratakis syndrome) (SDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHB	SDHB, SDH2, SDHIP, PGL4, CWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 4 (PGL4) [MIM:115310]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11404820, ECO:0000269|PubMed:11897817, ECO:0000269|PubMed:14715873, ECO:0000269|PubMed:14974914, ECO:0000269|PubMed:15328326}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paraganglioma ve feokromositoma (5p15.3, 14q23.3) (MLPA)	Paraganglioma and pheochromocytoma (5p15.3, 14q23.3) (MLPA)	5p15.3, 14q23.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Paragangliomalar 1, ± Sağırlık, 168000, Otozomal Dominant; PGL1 (Kalıtsal Feokromositoma-Paraganglioma) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paragangliomas 1, with or without deafness, 168000, Autosomal dominant; PGL1 (Hereditary pheochromocytoma-paraganglioma) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Paragangliomalar 2, 601650, Otozomal Dominant; PGL2 (Kalıtsal Feokromositoma-Paraganglioma) (SDHAF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paragangliomas 2, 601650, Autosomal dominant; PGL2 (Hereditary pheochromocytoma-paraganglioma) (SDHAF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHAF2	SDHAF2, SDH5, PGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 2 (PGL2) [MIM:601650]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:19628817}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Paragangliomalar 3, 605373, Otozomal Dominant; PGL3 (Kalıtsal Feokromositoma-Paraganglioma) (SDHC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paragangliomas 3, 605373, Autosomal dominant; PGL3 (Hereditary pheochromocytoma-paraganglioma) (SDHC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHC	SDHC, PGL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 3 (PGL3) [MIM:605373]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11062460}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paragangliomalar 4, 115310, Otozomal Dominant; PGL4 (Kalıtsal Feokromositoma-Paraganglioma) (SDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paragangliomas 4, 115310, Autosomal dominant; PGL4 (Hereditary pheochromocytoma-paraganglioma) (SDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHB	SDHB, SDH2, SDHIP, PGL4, CWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 4 (PGL4) [MIM:115310]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11404820, ECO:0000269|PubMed:11897817, ECO:0000269|PubMed:14715873, ECO:0000269|PubMed:14974914, ECO:0000269|PubMed:15328326}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paramyotonia Konjenita, 168300, Otozomal Dominant; PMC (Paramyotonia Konjenita - Von Eulenburg Tipi) (SCN4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paramyotonia congenita, 168300, Autosomal dominant; PMC (Paramyotonia congenita of Von Eulenburg) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paramyotonia Konjenita, 168300, Otozomal Dominant; PMC (Paramyotonia Konjenita - Von Eulenburg Tipi) (MLPA)	Paramyotonia congenita, 168300, Autosomal dominant; PMC (Paramyotonia congenita of Von Eulenburg) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paramyotonia Konjenita, 168300, Otozomal Dominant; PMC (Paramyotonia Konjenita - Von Eulenburg Tipi) (Prenatal) (SCN4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Paramyotonia congenita, 168300, Autosomal dominant; PMC (Paramyotonia congenita of Von Eulenburg) (SCN4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Paramyotonia Konjenita, 168300, Otozomal Dominant; PMC (Paramyotonia Konjenita - Von Eulenburg Tipi) (Prenatal) (MLPA)	Paramyotonia congenita, 168300, Autosomal dominant; PMC (Paramyotonia congenita of Von Eulenburg) (Prenatal) (MLPA)	SCN4A	SCN4A, HYPP, NAC1A, HOKPP2, CMS16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Paramyotonia congenita of von Eulenburg (PMC) [MIM:168300]: An autosomal dominant channelopathy characterized by myotonia, increased by exposure to cold, intermittent flaccid paresis, not necessarily dependent on cold or myotonia, lability of serum potassium, non-progressive nature and lack of atrophy or hypertrophy of muscles. In some patients, myotonia is not increased by cold exposure (paramyotonia without cold paralysis). Patients may have a combination phenotype of PMC and HYPP. {ECO:0000269|PubMed:10369308, ECO:0000269|PubMed:10727489, ECO:0000269|PubMed:1310898, ECO:0000269|PubMed:1316765, ECO:0000269|PubMed:1338909, ECO:0000269|PubMed:15318338, ECO:0000269|PubMed:15790667, ECO:0000269|PubMed:16786525, ECO:0000269|PubMed:18166706, ECO:0000269|PubMed:19077043, ECO:0000269|PubMed:20076800, ECO:0000269|PubMed:8242056, ECO:0000269|PubMed:8308722, ECO:0000269|PubMed:8388676, ECO:0000269|PubMed:8580427}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Parastrematik Kısalık, 168400, Otozomal Dominant (Parastrematik Dwarfizm) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parastremmatic dwarfism, 168400, Autosomal dominant (Parastremmatic dwarfism) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269|PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parastrematik Kısalık, 168400, Otozomal Dominant (Parastrematik Dwarfizm) (Prenatal) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Parastremmatic dwarfism, 168400, Autosomal dominant (Parastremmatic dwarfism) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269|PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Paratiroid Adenomu-Kistik Değişiklikler, 145001, Otozomal Dominant (Hiperparatiroidizm-Çene Tümörü Sendromu) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parathyroid adenoma with cystic changes, 145001, Autosomal dominant (Hyperparathyroidism-jaw tumor syndrome) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. (ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paratiroid Adenomu, Somatik (MEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parathyroid adenoma, somatic (MEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEN1	MEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Familial isolated hyperparathyroidism (FIHP) [MIM:145000]: Autosomal dominant disorder characterized by hypercalcemia, elevated parathyroid hormone (PTH) levels, and uniglandular or multiglandular parathyroid tumors. (ECO:0000269|PubMed:10634381, ECO:0000269|PubMed:10664521, ECO:0000269|PubMed:12016470, ECO:0000269|PubMed:12699448, ECO:0000269|PubMed:9792884, ECO:0000269|PubMed:9843042, ECO:0000269|PubMed:9888389). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Paratiroid Karsinomu, 608266 (Paratiroid Karsinomu) (CDC73 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parathyroid carcinoma, 608266 (Parathyroid carcinoma) (CDC73 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC73	CDC73, HRPT2, C1orf28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperparathyroidism-jaw tumor syndrome (HPT-JT) [MIM:145001]: Autosomal dominant, multiple neoplasia syndrome primarily characterized by hyperparathyroidism due to parathyroid tumors. Thirty percent of individuals with HPT-JT may also develop ossifying fibromas, primarily of the mandible and maxilla, which are distinct from the brown tumors associated with severe hyperparathyroidism. Kidney lesions may also occur in HPT-JT as bilateral cysts, renal hamartomas or Wilms tumors. (ECO:0000269|PubMed:12434154, ECO:0000269|PubMed:15613436, ECO:0000269|PubMed:16487440). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramina 1, 168500, Otozomal Dominant; PFM1 (Geniş Parietal Foramina) (MSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parietal foramina 1, 168500, Autosomal dominant; PFM1 (Enlarged parietal foramina) (MSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX2	MSX2, CRS2, HOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramina 1, 168500, Otozomal Dominant; PFM1 (Geniş Parietal Foramina) (MLPA)	Parietal foramina 1, 168500, Autosomal dominant; PFM1 (Enlarged parietal foramina) (MLPA)	MSX2	MSX2, CRS2, HOX8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramen 2, 609597, Otozomal Dominant; PFM2 (Geniş Parietal Foramina) (ALX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parietal foramina 2, 609597, Autosomal dominant; PFM2 (Enlarged parietal foramina) (ALX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. (ECO:0000305|PubMed:11017806, ECO:0000305|PubMed:11903336). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramen 2, 609597, Otozomal Dominant; PFM2 (Geniş Parietal Foramina) (MLPA)	Parietal foramina 2, 609597, Autosomal dominant; PFM2 (Enlarged parietal foramina) (MLPA)	ALX4	ALX4, PFM2, FPP, FND2, CRS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. (ECO:0000305|PubMed:11017806, ECO:0000305|PubMed:11903336). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramina-Kleidokraniyal Displazi, 168550, Otozomal Dominant; PFMCCD (Parietal Foramen-Kleidokraniyal Displazi) (MSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parietal foramina with cleidocranial dysplasia, 168550, Autosomal dominant; PFMCCD (Parietal foramina with cleidocranial dysplasia) (MSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX2	MSX2, CRS2, HOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramina-Kleidokraniyal Displazi, 168550, Otozomal Dominant; PFMCCD (Parietal Foramen-Kleidokraniyal Displazi) (MLPA)	Parietal foramina with cleidocranial dysplasia, 168550, Autosomal dominant; PFMCCD (Parietal foramina with cleidocranial dysplasia) (MLPA)	MSX2	MSX2, CRS2, HOX8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pariyetal Foramina-Kleidokraniyal Displazi, 168550, Otozomal Dominant; PFMCCD (Parietal Foramen-Kleidokraniyal Displazi) (Prenatal) (MSX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Parietal foramina with cleidocranial dysplasia, 168550, Autosomal dominant; PFMCCD (Parietal foramina with cleidocranial dysplasia) (MSX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MSX2	MSX2, CRS2, HOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pariyetal Foramina-Kleidokraniyal Displazi, 168550, Otozomal Dominant; PFMCCD (Parietal Foramen-Kleidokraniyal Displazi) (Prenatal) (MLPA)	Parietal foramina with cleidocranial dysplasia, 168550, Autosomal dominant; PFMCCD (Parietal foramina with cleidocranial dysplasia) (Prenatal) (MLPA)	MSX2	MSX2, CRS2, HOX8	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Parietal foramina 1 (PFM1) [MIM:168500]: Autosomal dominant disease characterized by oval defects of the parietal bones caused by deficient ossification around the parietal notch, which is normally obliterated during the fifth fetal month. (ECO:0000269|PubMed:10742103, ECO:0000269|PubMed:10767351). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Parkes Weber Sendromu, 608355 (Parkes Weber Sendromu) (RASA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkes Weber syndrome, 608355 (Parkes Weber syndrome) (RASA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RASA1	RASA1, GAP, CMAVM, PKWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Capillary malformation-arteriovenous malformation (CMAVM) [MIM:608354]: A disorder characterized by atypical capillary malformations that are multiple, small, round to oval in shape and pinkish red in color. These capillary malformations are associated with either arteriovenous malformation, arteriovenous fistula, or Parkes Weber syndrome. (ECO:0000269|PubMed:14639529, ECO:0000269|PubMed:24038909). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkes Weber Sendromu, 608355 (Parkes Weber Sendromu) (Prenatal) (RASA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Parkes Weber syndrome, 608355 (Parkes Weber syndrome) (RASA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RASA1	RASA1, GAP, CMAVM, PKWS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Capillary malformation-arteriovenous malformation (CMAVM) [MIM:608354]: A disorder characterized by atypical capillary malformations that are multiple, small, round to oval in shape and pinkish red in color. These capillary malformations are associated with either arteriovenous malformation, arteriovenous fistula, or Parkes Weber syndrome. (ECO:0000269|PubMed:14639529, ECO:0000269|PubMed:24038909). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Parkinson Hastalığı 1, 168601, Otozomal Dominant; PARK1 (Parkinson-Piramidal Sendromu) (SNCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 1, 168601, Autosomal dominant; PARK1 (Parkinsonian-pyramidal syndrome) (SNCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNCA	SNCA, NACP, PARK1, PARK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dementia Lewy body (DLB) [MIM:127750]: A neurodegenerative disorder characterized by mental impairment leading to dementia, parkinsonism, fluctuating cognitive function, visual hallucinations, falls, syncopal episodes, and sensitivity to neuroleptic medication. Brainstem or cortical intraneuronal accumulations of aggregated proteins (Lewy bodies) are the only essential pathologic features. Patients may also have hippocampal and neocortical senile plaques, sometimes in sufficient number to fulfill the diagnostic criteria for Alzheimer disease. (ECO:0000269	PubMed:14755719). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 11, 607688; PARK11 (Kalıtsal Geç Başlangıçlı Parkinson Hastalığı) (GIGYF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 11, 607688; PARK11 (Hereditary late-onset Parkinson disease) (GIGYF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GIGYF2	GIGYF2, KIAA0642, PARK11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 11 (PARK11) [MIM:607688]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability, as well as by a clinically significant response to treatment with levodopa. The pathology involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. (ECO:0000269	PubMed:18358451). Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 13, 610297; PARK13 (Genç Başlangıçlı Parkinson Hastalığı) (HTRA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 13, 610297; PARK13 (Young-onset Parkinson disease) (HTRA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTRA2	HTRA2, OMI, PARK13, PRSS25, MGC48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 13 (PARK13) [MIM:610297]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability, as well as by a clinically significant response to treatment with levodopa. The pathology involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. (ECO:0000269	PubMed:15961413). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 14, Otozomal Resesif, 612953, Otozomal Resesif; PARK14 (Yetişkin Başlangıçlı Distoni-Parkinsonizm) (PLA2G6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 14, autosomal recessive, 612953, Autosomal recessive; PARK14 (Adult-onset dystonia-parkinsonism) (PLA2G6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. (ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı 14, Otozomal Resesif, 612953, Otozomal Resesif; PARK14 (Yetişkin Başlangıçlı Distoni-Parkinsonizm) (MLPA)	Parkinson disease 14, autosomal recessive, 612953, Autosomal recessive; PARK14 (Adult-onset dystonia-parkinsonism) (MLPA)	PLA2G6	PLA2G6, IPLA2, INAD1, NBIA2B, NBIA2A, PARK14	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 2A (NBIA2A) [MIM:256600]: A neurodegenerative disease characterized by pathologic axonal swelling and spheroid bodies in the central nervous system. Onset is within the first 2 years of life with death by age 10 years. (ECO:0000269|PubMed:16783378, ECO:0000269|PubMed:17033970, ECO:0000269|PubMed:23749988). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Parkinson Hastalığı; 15 Otozomal Resesif, 260300, Otozomal Resesif; PARK15 (Parkinson-Piramidal Sendromi (FBXO7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 15, autosomal recessive, 260300, Autosomal recessive; PARK15 (Parkinsonian-pyramidal syndrome) (FBXO7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBXO7	FBXO7, FBX7, FBX, PKPS, PARK15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 15 (PARK15) [MIM:260300]: A neurodegenerative disorder characterized by parkinsonian and pyramidal signs. Clinical manifestations include tremor, bradykinesia, rigidity, postural instability, spasticity, mainly in the lower limbs, and hyperreflexia. {ECO:0000269	PubMed:18513678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 17, 614203, Otozomal Dominant; PARK17 (Kalıtsal Geç Başlangıçlı Parkinson Hastalığı) (VPS35 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 17, 614203, Autosomal dominant; PARK17 (Hereditary late-onset Parkinson disease) (VPS35 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS35	VPS35, MEM3, PARK17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı 18, 614251, Otozomal Dominant; PARK18 (Kalıtsal Geç Başlangıçlı Parkinson Hastalığı) (EIF4G1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 18, 614251, Autosomal dominant; PARK18 (Hereditary late-onset Parkinson disease) (EIF4G1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF4G1	EIF4G1, EIF4G, PARK18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 18 (PARK18) [MIM:614251]: An autosomal dominant, late-onset form of Parkinson disease. Parkinson disease is a complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability, as well as by a clinically significant response to treatment with levodopa. The pathology involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. {ECO:0000269	PubMed:21907011}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 19a, Gençlikte Başlayan, 615528, Otozomal Resesif; PARK19A (Genç Başlangıçlı Parkinson Hastalığı) (DNAJC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 19a, juvenile-onset, 615528, Autosomal recessive; PARK19A (Young-onset Parkinson disease) (DNAJC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC6	DNAJC6, DJC6, KIAA0473, PARK19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı 19b, Erken Başlangıçlı, 615528, Otozomal Resesif (Genç Başlangıçlı Parkinson Hastalığı) (DNAJC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 19b, early-onset, 615528, Autosomal recessive (Young-onset Parkinson disease) (DNAJC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJC6	DNAJC6, DJC6, KIAA0473, PARK19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı 20, Erken Başlangıçlı, 615530, Otozomal Resesif; PARK20 (Atipik Jüvenil Parkinsonizmi (SYNJ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 20, early-onset, 615530, Autosomal recessive; PARK20 (Atypical juvenile parkinsonism) (SYNJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNJ1	SYNJ1, PARK20, EIEE53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 20, early-onset (PARK20) [MIM:615530]: An early-onset form of Parkinson disease, a complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability, as well as by a clinically significant response to treatment with levodopa. The pathology involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. PARK20 is characterized by young adult-onset of parkinsonism. Additional features may include seizures, cognitive decline, abnormal eye movements, and dystonia. {ECO:0000269|PubMed:23804563, ECO:0000269|PubMed:23804577}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 22 Otozomal Dominant, 616710, PARK22 (CHCHD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 22, autosomal dominant, 616710, PARK22 (CHCHD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHCHD2	CHCHD2, PARK22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Parkinson Hastalığı 23, Otozomal Resesif, Erken Başlangıçlı, 616840, Otozomal Resesif; PARK23 (Genç Başlangıçlı Parkinson Hastalığı) (VPS13C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 23, autosomal recessive, early onset, 616840, Autosomal recessive; PARK23 (Young-onset Parkinson disease) (VPS13C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS13C	VPS13C, KIAA1421, PARK23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı 4, 605543, Otozomal Dominant; PARK4 (Kalıtsal Geç Başlangıçlı Parkinson Hastalığı) (SNCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 4, 605543, Autosomal dominant; PARK4 (Hereditary late-onset Parkinson disease) (SNCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNCA	SNCA, NACP, PARK1, PARK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dementia Lewy body (DLB) [MIM:127750]: A neurodegenerative disorder characterized by mental impairment leading to dementia, parkinsonism, fluctuating cognitive function, visual hallucinations, falls, syncopal episodes, and sensitivity to neuroleptic medication. Brainstem or cortical intraneuronal accumulations of aggregated proteins (Lewy bodies) are the only essential pathologic features. Patients may also have hippocampal and neocortical senile plaques, sometimes in sufficient number to fulfill the diagnostic criteria for Alzheimer disease. {ECO:0000269	PubMed:14755719}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 5, Yatkınlık, 613643; PARK5 (Genç Başlangıçlı Parkinson Hastalığı) (UCHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 5, susceptibility to, 613643; PARK5 (Young-onset Parkinson disease) (UCHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UCHL1	UCHL1, PARK5, SPG79, NDGOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 5 (PARK5) [MIM:613643]: A complex neurodegenerative disorder with manifestations ranging from typical Parkinson disease to dementia with Lewy bodies. Clinical features include parkinsonian symptoms (resting tremor, rigidity, postural instability and bradykinesia), dementia, diffuse Lewy body pathology, autonomic dysfunction, hallucinations and paranoia. {ECO:0000269	PubMed:9774100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı 5, Yatkınlık, 613643; PARK5 (Genç Başlangıçlı Parkinson Hastalığı) (MLPA)	Parkinson disease 5, susceptibility to, 613643; PARK5 (Young-onset Parkinson disease) (MLPA)	UCHL1	UCHL1, PARK5, SPG79, NDGOA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Parkinson disease 5 (PARK5) [MIM:613643]: A complex neurodegenerative disorder with manifestations ranging from typical Parkinson disease to dementia with Lewy bodies. Clinical features include parkinsonian symptoms (resting tremor, rigidity, postural instability and bradykinesia), dementia, diffuse Lewy body pathology, autonomic dysfunction, hallucinations and paranoia. {ECO:0000269	PubMed:9774100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı 6, Erken Başlayan, 605909, Otozomal Resesif; PARK6 (Genç Başlangıçlı Parkinson Hastalığı) (PINK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 6, early onset, 605909, Autosomal recessive; PARK6 (Young-onset Parkinson disease) (PINK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PINK1	PINK1, PARK6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 6 (PARK6) [MIM:605909]: A neurodegenerative disorder characterized by parkinsonian signs such as rigidity, resting tremor and bradykinesia. A subset of patients manifest additional symptoms including hyperreflexia, autonomic instability, dementia and psychiatric disturbances. Symptoms show diurnal fluctuation and can improve after sleep. {ECO:0000269|PubMed:15087508, ECO:0000269|PubMed:15349860, ECO:0000269|PubMed:15349870, ECO:0000269|PubMed:15505171, ECO:0000269|PubMed:15596610, ECO:0000269|PubMed:15955953, ECO:0000269|PubMed:15970950, ECO:0000269|PubMed:16009891, ECO:0000269|PubMed:16207217, ECO:0000269|PubMed:16251723, ECO:0000269|PubMed:16401616, ECO:0000269|PubMed:16482571, ECO:0000269|PubMed:16632486, ECO:0000269|PubMed:16966503, ECO:0000269|PubMed:17030667, ECO:0000269|PubMed:18286320, ECO:0000269|PubMed:23956510}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 7, Otozomal Resesif Erken Başlangıçlı, 606324, Otozomal Resesif; PARK7 (Genç Başlangıçlı Parkinson Hastalığı) (PARK7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 7, autosomal recessive early-onset, 606324, Autosomal recessive; PARK7 (Young-onset Parkinson disease) (PARK7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PARK7	DJ1, PARK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 7 (PARK7) [MIM:606324]: A neurodegenerative disorder characterized by resting tremor, postural tremor, bradykinesia, muscular rigidity, anxiety and psychotic episodes. PARK7 has onset before 40 years, slow progression and initial good response to levodopa. Some patients may show traits reminiscent of amyotrophic lateral sclerosis- parkinsonism/dementia complex (Guam disease). {ECO:0000269|PubMed:12446870, ECO:0000269|PubMed:12953260, ECO:0000269|PubMed:15254937, ECO:0000269|PubMed:15365989}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Parkinson Hastalığı 8, 607060, Otozomal Dominant; PARK8 (Kalıtsal Geç Başlangıçlı Parkinson Hastalığı) (LRRK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease 8, 607060, Autosomal dominant; PARK8 (Hereditary late-onset Parkinson disease) (LRRK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRRK2	LRRK2, PARK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 8 (PARK8) [MIM:607060]: A slowly progressive neurodegenerative disorder characterized by bradykinesia, rigidity, resting tremor, postural instability, neuronal loss in the substantia nigra, and the presence of neurofibrillary MAPT (tau)-positive and Lewy bodies in some patients. {ECO:0000269|PubMed:15541308, ECO:0000269|PubMed:15541309, ECO:0000269|PubMed:15680455, ECO:0000269|PubMed:15680456, ECO:0000269|PubMed:15680457, ECO:0000269|PubMed:15726496, ECO:0000269|PubMed:15732108, ECO:0000269|PubMed:15811454, ECO:0000269|PubMed:15852371, ECO:0000269|PubMed:15880653, ECO:0000269|PubMed:15925109, ECO:0000269|PubMed:15929036, ECO:0000269|PubMed:16102999, ECO:0000269|PubMed:16157901, ECO:0000269|PubMed:16157908, ECO:0000269|PubMed:16157909, ECO:0000269|PubMed:16172858, ECO:0000269|PubMed:16240353, ECO:0000269|PubMed:16247070, ECO:0000269|PubMed:16250030, ECO:0000269|PubMed:16251215, ECO:0000269|PubMed:16272164, ECO:0000269|PubMed:16272257, ECO:0000269|PubMed:16298482, ECO:0000269|PubMed:16333314, ECO:0000269|PubMed:16533964, ECO:0000269|PubMed:18213618,	EDTA Blood Sample
Parkinson Hastalığı, Geç Başlangıçlı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (GLUD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, age of onset, modifier, 168600, Isolated cases, Multifactorial (GLUD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLUD2	GLUD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı, Jüvenil, Tip 2, 600116, Otozomal Resesif; PARK2 (Genç Başlangıçlı Parkinson Hastalığı) (PRKN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, juvenile, type 2, 600116, Autosomal recessive; PARK2 (Young-onset Parkinson disease) (PRKN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKN	PRKN, PARK2, PDJ, LPRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Parkinson Hastalığı, Geç Başlangıçlı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (GBA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, late-onset, susceptibility to, 168600, Isolated cases, Multifactorial (GBA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA	GBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Parkinson disease (PARK) [MIM:168600]: A complex neurodegenerative disorder characterized by bradykinesia, resting tremor, muscular rigidity and postural instability. Additional features are characteristic postural abnormalities, dysautonomia, dystonic cramps, and dementia. The pathology of Parkinson disease involves the loss of dopaminergic neurons in the substantia nigra and the presence of Lewy bodies (intraneuronal accumulations of aggregated proteins), in surviving neurons in various areas of the brain. The disease is progressive and usually manifests after the age of 50 years, although early-onset cases (before 50 years) are known. The majority of the cases are sporadic suggesting a multifactorial etiology based on environmental and genetic factors. However, some patients present with a positive family history for the disease. Familial forms of the disease usually begin at earlier ages and are associated with atypical clinical features. {ECO:0000269|PubMed:12847165, ECO:0000269|PubMed:16148263, ECO:0000269|PubMed:19286955}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı, Geç Başlangıçlı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (ATXN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, late-onset, susceptibility to, 168600, Isolated cases, Multifactorial (ATXN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN2	ATXN2, ATX2, SCA2, ASL13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 2 (SCA2) [MIM:183090]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA2 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA2 is characterized by hyporeflexia, myoclonus and action tremor and dopamine-responsive parkinsonism. In some patients, SCA2 presents as pure familial parkinsonism without cerebellar signs. {ECO:0000269|PubMed:8896555, ECO:0000269|PubMed:8896556, ECO:0000269|PubMed:8896557}. Note=The disease is caused by mutations affecting the gene represented in this entry. SCA2 is caused by expansion of a CAG repeat resulting in about 36 to 52 repeats in some patients. Longer expansions result in earlier the expansion, onset of the disease.	EDTA Blood Sample
Parkinson Hastalığı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, susceptibility to, 168600, Isolated cases, Multifactorial (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (ADH1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, susceptibility to, 168600, Isolated cases, Multifactorial (ADH1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADH1C	ADH1C, ADH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Parkinson Hastalığı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (MLPA)	Parkinson disease, susceptibility to, 168600, Isolated cases, Multifactorial (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson Hastalığı, Yatkınlık, 168600, Sporadik Vakalar, Multifaktöryel (Spinoserebellar Ataksi Tip 17) (TBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinson disease, susceptibility to, 168600, Isolated cases, Multifactorial (Spinocerebellar ataxia type 17) (TBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBP	TBP, SCA17, HDL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 17 (SCA17) [MIM:607136]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA17 is an autosomal dominant cerebellar ataxia (ADCA) characterized by widespread cerebral and cerebellar atrophy, dementia and extrapyramidal signs. The molecular defect in SCA17 is the expansion of a CAG repeat in the coding region of TBP. Longer expansions result in earlier onset and more severe clinical manifestations of the disease. {ECO:0000269|PubMed:11313753, ECO:0000269|PubMed:11448935, ECO:0000269|PubMed:11939898}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson-Demans Sendromu (Progresif Supranükleer Palsi) (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	PARKINSON-DEMENTIA SYNDROME (Progressive supranuclear palsy) (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson-Demans Sendromu (Progresif Supranükleer Palsi) (MLPA)	PARKINSON-DEMENTIA SYNDROME (Progressive supranuclear palsy) (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinson hastalığı, ailesel (Miks 2) (PARK2 6q25.2, UCHL1 4p14, GCH1 14q22.1, LRRK2 12q12) (MLPA)	Parkinson's disease, familial (Mix 2) (PARK2 6q25.2, UCHL1 4p14, GCH1 14q22.1, LRRK2 12q12) (MLPA)	PARK2 6q25.2, UCHL1 4p14, GCH1 14q22.1, LRRK2 12q12	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Parkinsonizm-Spastisite, X'e Bağlı, 300911, X'e Bağlı Resesif (X'e Bağlı Parkinsonizm-Spastisite Sendromu) (ATP6AP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinsonism with spasticity, X-linked, 300911, X-linked recessive (X-linked parkinsonism-spasticity syndrome) (ATP6AP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6AP2	ATP6AP2, ATP6M8-9, XMRE, MRXSH, XPDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, X-linked, with epilepsy (MRXE) [MIM:300423]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRXE patients manifest mild to moderate mental retardation associated with epilepsy, delays in motor milestones and speech acquisition in infancy. {ECO:0000269|PubMed:15746149}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parkinsonizm-Distoni, İnfantil, 613135, Otozomal Resesif; PARK9 (İnfantil Distoni-Parkinsonizm) (SLC6A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Parkinsonism-dystonia, infantile, 613135, Autosomal recessive; PARK9 (infantile dystonia-parkinsonism) (SLC6A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC6A3	SLC6A3, DAT1, PKDYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Paroksismal Aşırı Ağrı Bozukluğu, 167400, Otozomal Dominant (Paroksismal Aşırı Ağrı Bozukluğu) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paroxysmal extreme pain disorder, 167400, Autosomal dominant (Paroxysmal extreme pain disorder) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Paroksismal Noktumal Hemoglobinüri 2, 615399, Otozomal Dominant, Somatik Mutasyon; PNH2 (Paroksismal Noktumal Hemoglobinüri) (PIGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paroxysmal nocturnal hemoglobinuria 2, 615399, Autosomal dominant, Somatic mutation; PNH2 (Paroxysmal nocturnal hemoglobinuria) (PIGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGT	PIGT, NDAP, PNH2, MCAHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Paroksismal Noktumal Hemoglobinüri, Somatik, 300818; PNH1 (Paroksismal Noktumal Hemoglobinüri) (PIGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paroxysmal nocturnal hemoglobinuria, somatic, 300818; PNH1 (Paroxysmal nocturnal hemoglobinuria) (PIGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIGA	PIGA, PNH1, MCAHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paroxysmal nocturnal hemoglobinuria 1 (PNH1) [MIM:300818]: A disorder characterized by hemolytic anemia with hemoglobinuria, thromboses in large vessels, and a deficiency in hematopoiesis. Red blood cell breakdown with release of hemoglobin into the urine is manifested most prominently by dark-colored urine in the morning. {ECO:0000269|PubMed:10087994, ECO:0000269|PubMed:12037021, ECO:0000269|PubMed:8167330, ECO:0000269|PubMed:8306954, ECO:0000269|PubMed:8500164}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Paroksismal Nonkinesijenik Diskinezi 1; Pnkd1 (Paroksismal Non-Kinesijenik Diskinezi) (MR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PAROXYSMAL NONKINESIGENIC DYSKINESIA 1; PNKD1 (Paroxysmal non-kinesigenic dyskinesia) (MR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MR1	MR1, TAHCCP2, KIPP1184, BRP17, PNKD, FPD1, PDC, DYT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Paroksismal Nonkinesijenik Diskinezi 1; Pnkd1 (Paroksismal Non-Kinesijenik Diskinezi) (Prenatal) (MR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PAROXYSMAL NONKINESIGENIC DYSKINESIA 1; PNKD1 (Paroxysmal non-kinesigenic dyskinesia) (MR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MR1	MR1, TAHCCP2, KIPP1184, BRP17, PNKD, FPD1, PDC, DYT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) Kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Paroksismal Nonkinesijenik Diskinezi, 118800, Otozomal Dominant (Paroksismal Non-Kinesijenik Diskinezi) (PNKD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Paroxysmal nonkinesigeric dyskinesia, 118800, Autosomal dominant (Paroxysmal non-kinesigenic dyskinesia) (PNKD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNKD	MR1, TAHCCP2, KIPP1184, BRP17, PNKD, FPD1, PDC, DYT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 8 (DYT8) [MIM:118800]: A paroxysmal non-kinesigenic dystonia/dyskinesia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 8 is characterized by attacks of involuntary movements brought on by stress, alcohol, fatigue or caffeine. The attacks generally last between a few seconds and four hours or longer. The attacks may begin in one limb and spread throughout the body, including the face. {ECO:0000269|PubMed:15282732, ECO:0000269|PubMed:15824259, ECO:0000269|PubMed:16632198, ECO:0000269|PubMed:16717228, ECO:0000269|PubMed:16972263}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Paroksismal Nonkinesijenik Diskinezi, 118800, Otozomal Dominant (Paroksismal Non-Kinesijenik Diskinezi) (Prenatal) (PNKD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Paroxysmal nonkinesigeric dyskinesia, 118800, Autosomal dominant (Paroxysmal non-kinesigenic dyskinesia) (PNKD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNKD	MR1, TAHCCP2, KIPP1184, BRP17, PNKD, FPD1, PDC, DYT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dystonia 8 (DYT8) [MIM:118800]: A paroxysmal non-kinesigenic dystonia/dyskinesia. Dystonia is defined by the presence of sustained involuntary muscle contractions, often leading to abnormal postures. Dystonia type 8 is characterized by attacks of involuntary movements brought on by stress, alcohol, fatigue or caffeine. The attacks generally last between a few seconds and four hours or longer. The attacks may begin in one limb and spread throughout the body, including the face. {ECO:0000269|PubMed:15282732, ECO:0000269|PubMed:15824259, ECO:0000269|PubMed:16632198, ECO:0000269|PubMed:16717228, ECO:0000269|PubMed:16972263}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Parsiyel Lipodistrofi, Konjenital Katarakt ve Nörodejenerasyon Sendromu, 606721, Otozomal Dominant (CAV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721, Autosomal dominant (CAV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. {ECO:0000269|PubMed:18237401}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Parsiyel Lipodistrofi, Konjenital Katarakt ve Nörodejenerasyon Sendromu, 606721, Otozomal Dominant (Prenatal) (CAV1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome, 606721, Autosomal dominant (CAV1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. {ECO:0000269|PubMed:18237401}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Partington Sendromu, 309510, X'e Bağlı Resesif; PRTS (Partington Sendromu) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Partington syndrome, 309510, X-linked recessive; PRTS (Partington syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Partington Sendromu, 309510, X'e Bağlı Resesif; PRTS (Partington Sendromu) (MLPA)	Partington syndrome, 309510, X-linked recessive; PRTS (Partington syndrome) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Partington Sendromu, 309510, X'e Bağlı Resesif; PRTS (Partington Sendromu) (Prenatal) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Partington syndrome, 309510, X-linked recessive; PRTS (Partington syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Partington Sendromu, 309510, X'e Bağlı Resesif; PRTS (Partington Sendromu) (Prenatal) (MLPA)	Partington syndrome, 309510, X-linked recessive; PRTS (Partington syndrome) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Patent Duktus Arteriosus 2, 617035, Otozomal Dominant; PDA2 (Ailesel Patent Arter Kanalı) (TFAP2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Patent ductus arteriosus 2, 617035, Autosomal dominant; PDA2 (Familial patent arterial duct) (TFAP2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFAP2B	TFAP2B, CHAR, PDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Char syndrome (CHAR) [MIM:169100]: An autosomal dominant disorder characterized by patent ductus arteriosus (PDA), facial dysmorphism and hand anomalies. {ECO:0000269|PubMed:10802654, ECO:0000269|PubMed:11505339}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Patent Duktus Arteriosus 3, 617039, Otozomal Dominant; PDA3 (Ailesel Patent Arter Kanalı) (PRDM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Patent ductus arteriosus 3, 617039, Autosomal dominant; PDA3 (Familial patent arterial duct) (PRDM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRDM6	PRDM6, PRISM, PDA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hasta İzlemi ve İzlem Danışmanlığı.	Patient followup (.)	.	.	Klinik değerlendirme/ Clinical Evaluation	Tüm Tıbbi Dokümanlar ve Aile Fotoğrafları		0	0	.	.	0	Postnatal	.	Medical Documents and Family Photos
PCWH Sendromu, 609136, Otozomal Dominant (Nörolojik Waardenburg-Shah Sendromu) (SOX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PCWH syndrome, 609136, Autosomal dominant (Neurologic Waardenburg-Shah syndrome) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome and Hirschsprung disease (PCWH) [MIM:609136]: A complex neurocristopathy that includes features of 4 distinct syndromes: peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome and Hirschsprung disease. {ECO:0000269|PubMed:10762540, ECO:0000269|PubMed:15004559, ECO:0000269|PubMed:19208381, ECO:0000269|PubMed:21898658}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
PCWH Sendromu, 609136, Otozomal Dominant (Nörolojik Waardenburg-Shah Sendromu) (Prenatal) (SOX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PCWH syndrome, 609136, Autosomal dominant (Neurologic Waardenburg-Shah syndrome) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome and Hirschsprung disease (PCWH) [MIM:609136]: A complex neurocristopathy that includes features of 4 distinct syndromes: peripheral demyelinating neuropathy, central dysmyelinating leukodystrophy, Waardenburg syndrome and Hirschsprung disease. {ECO:0000269|PubMed:10762540, ECO:0000269|PubMed:15004559, ECO:0000269|PubMed:19208381, ECO:0000269|PubMed:21898658}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
PDGFRB (5q32) Breakapart (FISH)	PDGFRB (5q32) Breakapart (FISH)	5q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Soyulan Deri Sendromu 1, 270300 Soyulması Otozomal Resesif; PSS1 (Soyulan Deri Sendromu Tip B) (CDSN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin syndrome 1, 270300, Autosomal recessive; PSS1 (Peeling skin syndrome type B) (CDSN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDSN	CDSN, HTSS1, HYPT2, PSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypotrichosis 2 (HYPT2) [MIM:146520]: A condition characterized by the presence of less than the normal amount of hair. Affected individuals have normal hair in early childhood but experience progressive hair loss limited to the scalp beginning in the middle of the first decade and almost complete baldness by the third decade. Body hair, beard, eyebrows, axillary hair, teeth, and nails develop normally. {ECO:0000269|PubMed:12754508}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Soyulan Deri Sendromu 2, 609796, Otozomal Resesif; PSS2 (Akral Soyulmuş Deri Sendromu) (TGM5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin syndrome 2, 609796, Autosomal recessive; PSS2 (Acral peeling skin syndrome) (TGM5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGM5	TGM5, TGX, PSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peeling skin syndrome 2 (PSS2) [MIM:609796]: A non-inflammatory and localized form of peeling skin syndrome, a genodermatosis characterized by the continuous shedding of the outer layers of the epidermis. In PSS2 patients, skin peeling is painless and strictly limited to the dorsa of the hands and feet. It is accompanied by painless erythema and spontaneous non-scarring healing. Ultrastructural and histological analysis shows a level of blistering high in the epidermis at the stratum granulosum-stratum corneum junction. {ECO:0000269|PubMed:16380904}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Soyulan Deri Sendromu 3, 616265, Otozomal Resesif; PSS3 (Soyulmuş Deri Sendromu Tip A) (CHST8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin syndrome 3, 616265, Autosomal recessive; PSS3 (Peeling skin syndrome type A) (CHST8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHST8	CHST8, GALNAC4ST1, PSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peeling skin syndrome 3 (PSS3) [MIM:616265]: A form of peeling skin syndrome, a genodermatosis characterized by generalized, continuous shedding of the outer layers of the epidermis. Two main PSS subtypes have been suggested. Patients with non-inflammatory PSS (type A) manifest white scaling, with painless and easy removal of the skin, irritation when in contact with water, dust and sand, and no history of erythema, pruritis or atopy. Inflammatory PSS (type B) is associated with generalized erythema, pruritus and atopy. It is an ichthyosiform erythroderma characterized by lifelong patchy peeling of the entire skin with onset at birth or shortly after. Several patients have been reported with high IgE levels. PSS3 is characterized by generalized white scaling occurring over the upper and lower extremities. Symptoms start during the second half of the first decade of life. {ECO:0000269|PubMed:22289416}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Soyulan Deri Sendromu 4, 607936, Otozomal Resesif; PSS4 (Eksfoliatif İktiyoz) (CSTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin syndrome 4, 607936, Autosomal recessive; PSS4 (Exfoliative ichthyosis) (CSTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSTA	CSTA, STFA, STF1, AREI, PSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Soyulan Deri Sendromu 5, 617115, Otozomal Resesif; PSS5 (Eksfoliatif İktiyoz) (SERPINB8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin syndrome 5, 617115, Autosomal recessive; PSS5 (Exfoliative ichthyosis) (SERPINB8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINB8	SERPINB8, PI8, CAP2, PSS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Soyulan Deri-Lökonişi, Akrak Punktat Keratoz, Keilitis, Parmak Ucu Yastıklıkları, 616295, Otozomal Resesif; PLACK (Cilt Soyulması-Lökonişi-Akral Punktat Keratoz-Keilitis-Parmak Ucu Pedleri Sendromu) (CAST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295, Autosomal recessive; PLACK (Peeling skin-leukonuchia-acral punctate keratoses-cheilitis-knuckle pads syndrome) (CAST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAST	CAST, PLACK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads (PLACK) [MIM:616295]: An autosomal recessive disease characterized by generalized, continuous shedding of the outer layers of the epidermis, leukonychia, acral punctate keratosis, cheilitis, knuckle pads with multiple hyperkeratotic micropapules involving the interphalangeal joints, and palmoplantar keratoderma. [ECO:0000269	PubMed:25683118]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
PEHO Sendromu, 260565, Otozomal Resesif (PEHO Sendromu) (ZNHT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PEHO syndrome, 260565, Autosomal recessive (PEHO syndrome) (ZNHT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNHT3	TRIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
PEHO Sendromu, 260565, Otozomal Resesif (PEHO Sendromu) (Prenatal) (ZNHT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PEHO syndrome, 260565, Autosomal recessive (PEHO syndrome) (ZNHT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZNHT3	TRIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pelger-Huet Anomalisi, 169400, Otozomal Dominant; PHA (LBR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pelger-Huet anomaly, 169400, Autosomal dominant; PHA (LBR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pelger-Huet anomaly (PHA) [MIM:169400]: An autosomal dominant inherited abnormality of granulocytes, characterized by abnormal ovoid shape, reduced nuclear segmentation and an apparently lossar chromatin structure. Some individuals occasionally have skeletal anomalies, developmental delay, and seizures. [ECO:0000269	PubMed:14617022]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pelger-Huet Anomalisi, 169400, Otozomal Dominant; PHA (Prenatal) (LBR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pelger-Huet anomaly, 169400, Autosomal dominant; PHA (LBR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pelger-Huet anomaly (PHA) [MIM:169400]: An autosomal dominant inherited abnormality of granulocytes, characterized by abnormal ovoid shape, reduced nuclear segmentation and an apparently lossar chromatin structure. Some individuals occasionally have skeletal anomalies, developmental delay, and seizures. [ECO:0000269	PubMed:14617022]. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pelizaeus-Merzbacher Hastalığı, 312080, X'e Bağlı Resesif; PMD (Pelizaeus-Merzbacher Hastalığı) (PLP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pelizaeus-Merzbacher disease, 312080, X-linked recessive; PMD (Pelizaeus-Merzbacher disease) (PLP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLP1	PLP1, PMD, HLD1, SPG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 1 (HLD1) [MIM:312080]: A X-linked recessive disorder of the central nervous system in which myelin is not formed properly. Clinically characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. (ECO:0000269|PubMed:10417279, ECO:0000269|PubMed:10425042, ECO:0000269|PubMed:11093273, ECO:0000269|PubMed:11786921, ECO:0000269|PubMed:1376966, ECO:0000269|PubMed:1384324, ECO:0000269|PubMed:15712223, ECO:0000269|PubMed:1707231, ECO:0000269|PubMed:1708672, ECO:0000269|PubMed:1715570, ECO:0000269|PubMed:2479017, ECO:0000269|PubMed:2480601, ECO:0000269|PubMed:2773936, ECO:0000269|PubMed:7531827, ECO:0000269|PubMed:7539213, ECO:0000269|PubMed:7541731, ECO:0000269|PubMed:7573159, ECO:0000269|PubMed:7679906, ECO:0000269|PubMed:7683951, ECO:0000269|PubMed:7684886, ECO:0000269|PubMed:8037216, ECO:0000269|PubMed:8909455, ECO:0000269|PubMed:9008538, ECO:0000269|PubMed:9143933, ECO:0000269|PubMed:9482656, ECO:0000269|PubMed:9633722, ECO:0000269|PubMed:9747038, ECO:0000269|PubMed:9788732.	EDTA Blood Sample
Pelizaeus-Merzbacher Hastalığı, 312080, X'e Bağlı Resesif; PMD (Pelizaeus-Merzbacher Hastalığı) (MLPA)	Pelizaeus-Merzbacher disease, 312080, X-linked recessive; PMD (Pelizaeus-Merzbacher disease) (MLPA)	PLP1	PLP1, PMD, HLD1, SPG2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 1 (HLD1) [MIM:312080]: A X-linked recessive disorder of the central nervous system in which myelin is not formed properly. Clinically characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. (ECO:0000269|PubMed:10417279, ECO:0000269|PubMed:10425042, ECO:0000269|PubMed:11093273, ECO:0000269|PubMed:11786921, ECO:0000269|PubMed:1376966, ECO:0000269|PubMed:1384324, ECO:0000269|PubMed:15712223, ECO:0000269|PubMed:1707231, ECO:0000269|PubMed:1708672, ECO:0000269|PubMed:1715570, ECO:0000269|PubMed:2479017, ECO:0000269|PubMed:2480601, ECO:0000269|PubMed:2773936, ECO:0000269|PubMed:7531827, ECO:0000269|PubMed:7539213, ECO:0000269|PubMed:7541731, ECO:0000269|PubMed:7573159, ECO:0000269|PubMed:7679906, ECO:0000269|PubMed:7683951, ECO:0000269|PubMed:7684886, ECO:0000269|PubMed:8037216, ECO:0000269|PubMed:8909455, ECO:0000269|PubMed:9008538, ECO:0000269|PubMed:9143933, ECO:0000269|PubMed:9482656, ECO:0000269|PubMed:9633722, ECO:0000269|PubMed:9747038, ECO:0000269|PubMed:9788732.	EDTA Blood Sample
Pelizaeus-Merzbacher Hastalığı, 312080, X'e Bağlı Resesif; PMD (Pelizaeus-Merzbacher Hastalığı) (Prenatal) (PLP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pelizaeus-Merzbacher disease, 312080, X-linked recessive; PMD (Pelizaeus-Merzbacher disease) (PLP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLP1	PLP1, PMD, HLD1, SPG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 1 (HLD1) [MIM:312080]: A X-linked recessive disorder of the central nervous system in which myelin is not formed properly. Clinically characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. (ECO:0000269|PubMed:10417279, ECO:0000269|PubMed:10425042, ECO:0000269|PubMed:11093273, ECO:0000269|PubMed:11786921, ECO:0000269|PubMed:1376966, ECO:0000269|PubMed:1384324, ECO:0000269|PubMed:15712223, ECO:0000269|PubMed:1707231, ECO:0000269|PubMed:1708672, ECO:0000269|PubMed:1715570, ECO:0000269|PubMed:2479017, ECO:0000269|PubMed:2480601, ECO:0000269|PubMed:2773936, ECO:0000269|PubMed:7531827, ECO:0000269|PubMed:7539213, ECO:0000269|PubMed:7541731, ECO:0000269|PubMed:7573159, ECO:0000269|PubMed:7679906, ECO:0000269|PubMed:7683951, ECO:0000269|PubMed:7684886, ECO:0000269|PubMed:8037216, ECO:0000269|PubMed:8909455, ECO:0000269|PubMed:9008538, ECO:0000269|PubMed:9143933, ECO:0000269|PubMed:9482656, ECO:0000269|PubMed:9633722, ECO:0000269|PubMed:9747038, ECO:0000269|PubMed:9788732.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pelizaeus-Merzbacher Hastalığı, 312080, X'e Bağlı Resesif; PMD (Pelizaeus-Merzbacher Hastalığı) (Prenatal) (MLPA)	Pelizaeus-Merzbacher disease, 312080, X-linked recessive; PMD (Pelizaeus-Merzbacher disease) (Prenatal) (MLPA)	PLP1	PLP1, PMD, HLD1, SPG2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukodystrophy, hypomyelinating, 1 (HLD1) [MIM:312080]: A X-linked recessive disorder of the central nervous system in which myelin is not formed properly. Clinically characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. (ECO:0000269|PubMed:10417279, ECO:0000269|PubMed:10425042, ECO:0000269|PubMed:11093273, ECO:0000269|PubMed:11786921, ECO:0000269|PubMed:1376966, ECO:0000269|PubMed:1384324, ECO:0000269|PubMed:15712223, ECO:0000269|PubMed:1707231, ECO:0000269|PubMed:1708672, ECO:0000269|PubMed:1715570, ECO:0000269|PubMed:2479017, ECO:0000269|PubMed:2480601, ECO:0000269|PubMed:2773936, ECO:0000269|PubMed:7531827, ECO:0000269|PubMed:7539213, ECO:0000269|PubMed:7541731, ECO:0000269|PubMed:7573159, ECO:0000269|PubMed:7679906, ECO:0000269|PubMed:7683951, ECO:0000269|PubMed:7684886, ECO:0000269|PubMed:8037216, ECO:0000269|PubMed:8909455, ECO:0000269|PubMed:9008538, ECO:0000269|PubMed:9143933, ECO:0000269|PubMed:9482656, ECO:0000269|PubMed:9633722, ECO:0000269|PubMed:9747038, ECO:0000269|PubMed:9788732.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pemfigoid, Yatkınlık, 142857 (HLA-DRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pemphigoid, susceptibility to, 142857 (HLA-DRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DRB1	HLA-DRB1, SS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Pendred Sendromu, 274600, Otozomal Resesif; PDS (Pendred Sendromu) (SLC26A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pendred syndrome, 274600, Autosomal recessive; PDS (Pendred syndrome) (SLC26A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A4	SLC26A4, PDS, DFNB4, EVA, TDH2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal recessive, 4 (DFNB4) [MIM:600791]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB4 is associated with an enlarged vestibular aqueduct. {ECO:0000269|PubMed:10190331, ECO:0000269|PubMed:10700480, ECO:0000269|PubMed:11748854, ECO:0000269|PubMed:12676893, ECO:0000269|PubMed:14508505, ECO:0000269|PubMed:14679580, ECO:0000269|PubMed:19204907, ECO:0000269|PubMed:20108392, ECO:0000269|PubMed:20597900, ECO:0000269|PubMed:9500541}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pendred Sendromu, 274600, Otozomal Resesif; PDS (Pendred Sendromu) (MLPA)	Pendred syndrome, 274600, Autosomal recessive; PDS (Pendred syndrome) (MLPA)	SLC26A4	SLC26A4, PDS, DFNB4, EVA, TDH2B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Deafness, autosomal recessive, 4 (DFNB4) [MIM:600791]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. DFNB4 is associated with an enlarged vestibular aqueduct. {ECO:0000269|PubMed:10190331, ECO:0000269|PubMed:10700480, ECO:0000269|PubMed:11748854, ECO:0000269|PubMed:12676893, ECO:0000269|PubMed:14508505, ECO:0000269|PubMed:14679580, ECO:0000269|PubMed:19204907, ECO:0000269|PubMed:20108392, ECO:0000269|PubMed:20597900, ECO:0000269|PubMed:9500541}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
PEPCK Eksikliği, Mitokondriyal, 261650, Otozomal Resesif (Fosfoenolpiruvat Karboksikinaz Eksikliği) (PCK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PEPCK deficiency, mitochondrial, 261650, Autosomal recessive (Phosphoenolpyruvate carboxykinase deficiency) (PCK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCK2	PCK2, PEPCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial phosphoenolpyruvate carboxykinase deficiency (M-PEPCKD) [MIM:261650]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
PEPCK Eksikliği, Mitokondriyal, 261650, Otozomal Resesif (Fosfoenolpiruvat Karboksikinaz Eksikliği) (Prenatal) (PCK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PEPCK deficiency, mitochondrial, 261650, Autosomal recessive (Phosphoenolpyruvate carboxykinase deficiency) (PCK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCK2	PCK2, PEPCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial phosphoenolpyruvate carboxykinase deficiency (M-PEPCKD) [MIM:261650]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pepsinojen 3, Grup I; Pga3 (PGA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PEPSINOGEN 3, GROUP I; PGA3 (PGA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Periferik Kandan Kromozom Analizi	Peripheral blood- Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Heparinli Kan (2-4 ml)	*	15	45	.	.	21	Postnatal	.	Heparinized blood sample
Periyodik Ateş, Ailesel, 142680, Otozomal Dominant (Tümör Nekroz Faktörü Reseptörü 1 İlişkili Periyodik Sendrom) (TNFRSF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periodic fever, familial, 142680, Autosomal dominant (Tumor necrosis factor receptor 1 associated periodic syndrome) (TNFRSF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF1A	TNFRSF1A, TNFR1, TNFAR, FPF, MSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hibernian fever (FHF) [MIM:142680]: A hereditary periodic fever syndrome characterized by recurrent fever, abdominal pain, localized tender skin lesions and myalgia. Reactive amyloidosis is the main complication and occurs in 25% of cases. {ECO:0000269|PubMed:10199409, ECO:0000269|PubMed:10602757, ECO:0000269|PubMed:11443543, ECO:0000269|PubMed:13130484, ECO:0000269|PubMed:14610673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Periyodik Ateş, Mensturasyona Bağlı, 614674, Otozomal Dominant (HTR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periodic fever, menstrual cycle dependent, 614674, Autosomal dominant (HTR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR1A	HTR1A, ADRB2RL1, PFMCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Periodontitis 1, Jüvenil, 170650, Otozomal Resesif (Papillon-Lefèvre Sendromu) (CTSC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periodontitis 1, juvenile, 170650, Autosomal recessive (Papillon-Lefèvre syndrome) (CTSC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSC	CTSC, CPPI, PALS, PLS, HMS, PDON1, JPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periodontitis, aggressive, 1 (AP1) [MIM:170650]: A disease characterized by severe and protracted gingival infections, generalized or localized, leading to tooth loss. Amounts of microbial deposits are generally inconsistent with the severity of periodontal tissue destruction and the progression of attachment and bone loss may be self arresting. {ECO:0000269|PubMed:10662808, ECO:0000269|PubMed:14974080}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Periferal Nöropati, Miyopati, Ses Kısıklığı ve İşitme Kaybı, 614369, Otozomal Dominant; PNMHH (Periferal Nöropati-Miyopati-Ses Kısıklığı-İşitme Kaybı Sendromu) (MYH14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peripheral neuropathy, myopathy, hoarseness, and hearing loss, 614369, Autosomal dominant; PNMHH (Peripheral neuropathy-myopathy-hoarseness-hearing loss syndrome) (MYH14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH14	MYH14, KIAA2034, DFNA4A, PNMHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 4A (DFNA4A) [MIM:600652]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:15015131, ECO:0000269|PubMed:16222661}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Periventriküler Heterotopi, Mikrosefali, 608097, Otozomal Resesif; ARPHM (Nodüler Nöronal Heterotopi) (ARFGEF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periventricular heterotopia with microcephaly, 608097, Autosomal recessive; ARPHM (Nodular neuronal heterotopia) (ARFGEF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARFGEF2	ARFGEF2, BIG2, PVNH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periventricular nodular heterotopia 2 (PVNH2) [MIM:608097]: A developmental disorder characterized by the presence of periventricular nodules of cerebral gray matter, resulting from a failure of neurons to migrate normally from the lateral ventricular proliferative zone, where they are formed, to the cerebral cortex. PVNH2 is an autosomal recessive form characterized by microcephaly (small brain), severe developmental delay and recurrent infections. No anomalies extrinsic to the central nervous system, such as dysmorphic features or grossly abnormal endocrine or other conditions, are associated with PVNH2. {ECO:0000269	PubMed:14647276}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Periventriküler Heterotopi, Mikrosefali, 608097, Otozomal Resesif; ARPHM (Nodüler Nöronal Heterotopi) (Prenatal) (ARFGEF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Periventricular heterotopia with microcephaly, 608097, Autosomal recessive; ARPHM (Nodular neuronal heterotopia) (ARFGEF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARFGEF2	ARFGEF2, BIG2, PVNH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Periventricular nodular heterotopia 2 (PVNH2) [MIM:608097]: A developmental disorder characterized by the presence of periventricular nodules of cerebral gray matter, resulting from a failure of neurons to migrate normally from the lateral ventricular proliferative zone, where they are formed, to the cerebral cortex. PVNH2 is an autosomal recessive form characterized by microcephaly (small brain), severe developmental delay and recurrent infections. No anomalies extrinsic to the central nervous system, such as dysmorphic features or grossly abnormal endocrine or other conditions, are associated with PVNH2. {ECO:0000269	PubMed:14647276}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Periventriküler Nodüler Heterotopi 1; Pvnh1 (Nodüler Nöronal Heterotopi) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1 (Nodular neuronal heterotopia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Periventriküler Nodüler Heterotopi 1; Pvnh1 (Nodüler Nöronal Heterotopi) (MLPA)	PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1 (Nodular neuronal heterotopia) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Periventriküler Nodüler Heterotopi 1; Pvnh1 (Nodüler Nöronal Heterotopi) (Prenatal) (FLNA geni) (Tüm Gen Dizi Analizi) (Prenatal)	PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1 (Nodular neuronal heterotopia) (FLNA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Periventriküler Nodüler Heterotopi 1; Pvnh1 (Nodüler Nöronal Heterotopi) (Prenatal) (MLPA)	PERIVENTRICULAR NODULAR HETEROTOPIA 1; PVNH1 (Nodular neuronal heterotopia) (Prenatal) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269	PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Periventriküler Nodüler Heterotopi 6, 615544, Otozomal Dominant; PVNH6 (Nodüler Nöronal Heterotopi) (ERMARD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periventricular nodular heterotopia 6, 615544, Autosomal dominant; PVNH6 (Nodular neuronal heterotopia) (ERMARD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERMARD	ERMARD, C6orf70, PVNH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periventricular nodular heterotopia 6 (PVNH6) [MIM:615544]: A form of periventricular nodular heterotopia, a disorder resulting from a defect in the pattern of neuronal migration in which ectopic collections of neurons lie along the lateral ventricles of the brain or just beneath, contiguously or in isolated patches. PVNH6 results in delayed psychomotor development, delayed speech, strabismus, and onset of seizures with hypsarrhythmia in early infancy. {ECO:0000269	PubMed:24056535}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Periventriküler Nodüler Heterotopi 6, 615544, Otozomal Dominant; PVNH6 (Nodüler Nöronal Heterotopi) (Prenatal) (ERMARD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Periventricular nodular heterotopia 6, 615544, Autosomal dominant; PVNH6 (Nodular neuronal heterotopia) (ERMARD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERMARD	ERMARD, C6orf70, PVNH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Periventricular nodular heterotopia 6 (PVNH6) [MIM:615544]: A form of periventricular nodular heterotopia, a disorder resulting from a defect in the pattern of neuronal migration in which ectopic collections of neurons lie along the lateral ventricles of the brain or just beneath, contiguously or in isolated patches. PVNH6 results in delayed psychomotor development, delayed speech, strabismus, and onset of seizures with hypsarrhythmia in early infancy. {ECO:0000269	PubMed:24056535}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Periventriküler Nodüler Heterotopi 7, 617201, Otozomal Dominant; PVNH7 (Nodüler Nöronal Heterotopi (NEDD4L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Periventricular nodular heterotopia 7, 617201, Autosomal dominant; PVNH7 (Nodular neuronal heterotopia) (NEDD4L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEDD4L	NEDD4L, KIAA0439, RSP5, PVNH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Periventriküler Nodüler Heterotopi 7, 617201, Otozomal Dominant; PVNH7 (Nodüler Nöronal Heterotopi) (Prenatal) (NEDD4L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Periventricular nodular heterotopia 7, 617201, Autosomal dominant; PVNH7 (Nodular neuronal heterotopia) (NEDD4L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEDD4L	NEDD4L, KIAA0439, RSP5, PVNH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perlman Sendromu, 267000, Otozomal Resesif (Perlman Sendromu) (DIS3L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perlman syndrome, 267000, Autosomal recessive (Perlman syndrome) (DIS3L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIS3L2	DIS3L2, PRLMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Perlman syndrome (PRLMNS) [MIM:267000]: An autosomal recessive congenital overgrowth syndrome. Affected children are large at birth, are hypotonic, and show organomegaly, characteristic facial dysmorphisms (inverted V-shaped upper lip, prominent forehead, deep-set eyes, broad and flat nasal bridge, and low-set ears), renal anomalies (nephromegaly and hydronephrosis), frequent neurodevelopmental delay, and high neonatal mortality. Perlman syndrome is associated with a high risk of Wilms tumor. Histologic examination of the kidneys in affected children shows frequent nephroblastomatosis, which is a precursor lesion for Wilms tumor. {ECO:0000269|PubMed:22306653, ECO:0000269|PubMed:23486540, ECO:0000269|PubMed:23576526}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Perlman Sendromu, 267000, Otozomal Resesif (Perlman Sendromu) (Prenatal) (DIS3L2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perlman syndrome, 267000, Autosomal recessive (Perlman syndrome) (DIS3L2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DIS3L2	DIS3L2, PRLMNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Perlman syndrome (PRLMNS) [MIM:267000]: An autosomal recessive congenital overgrowth syndrome. Affected children are large at birth, are hypotonic, and show organomegaly, characteristic facial dysmorphisms (inverted V-shaped upper lip, prominent forehead, deep-set eyes, broad and flat nasal bridge, and low-set ears), renal anomalies (nephromegaly and hydronephrosis), frequent neurodevelopmental delay, and high neonatal mortality. Perlman syndrome is associated with a high risk of Wilms tumor. Histologic examination of the kidneys in affected children shows frequent nephroblastomatosis, which is a precursor lesion for Wilms tumor. {ECO:0000269|PubMed:22306653, ECO:0000269|PubMed:23486540, ECO:0000269|PubMed:23576526}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisomal Açil-CoA Oksidaz Eksikliği, 264470, Otozomal Resesif (Perokszisomal Açil-CoA Oksidaz Eksikliği) (ACOX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisomal acyl-CoA oxidase deficiency, 264470, Autosomal recessive (Peroxisomal acyl-CoA oxidase deficiency) (ACOX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACOX1	ACOX1, ACOX, SCOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Peroksisomal Açil-CoA Oksidaz Eksikliği, 264470, Otozomal Resesif (Perokszisomal Açil-CoA Oksidaz Eksikliği) (Prenatal) (ACOX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisomal acyl-CoA oxidase deficiency, 264470, Autosomal recessive (Peroxisomal acyl-CoA oxidase deficiency) (ACOX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACOX1	ACOX1, ACOX, SCOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisomal Yağ Açil-CoA Redüktaz 1 Bozukluğu, 616154, Otozomal Resesif; PFCRD (Ağır Entelektüel Yetersizlik-Epilepsi-Katarakt Sendromu, Peroksisomal Yağ Açil-CoA Redüktaz 1 Eksikliğine Bağlı) (FAR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154, Autosomal recessive; PFCRD (Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency) (FAR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAR1	FAR1, MLSTD2, PFCRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisomal fatty acyl-CoA reductase 1 disorder (PFCRD) [MIM:616154]: An autosomal recessive metabolic disorder clinically characterized by severe intellectual disability, early-onset epilepsy, microcephaly, congenital cataracts, growth retardation, and spasticity. {ECO:0000269|PubMed:25439727}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Peroksisomal Yağ Açil-CoA Redüktaz 1 Bozukluğu, 616154, Otozomal Resesif; PFCRD (Ağır Entelektüel Yetersizlik-Epilepsi-Katarakt Sendromu, Peroksisomal Yağ Açil-CoA Redüktaz 1 Eksikliğine Bağlı) (Prenatal) (FAR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisomal fatty acyl-CoA reductase 1 disorder, 616154, Autosomal recessive; PFCRD (Severe intellectual disability-epilepsy-cataract syndrome due to fatty acyl-CoA reductase 1 deficiency) (FAR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAR1	FAR1, MLSTD2, PFCRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisomal fatty acyl-CoA reductase 1 disorder (PFCRD) [MIM:616154]: An autosomal recessive metabolic disorder clinically characterized by severe intellectual disability, early-onset epilepsy, microcephaly, congenital cataracts, growth retardation, and spasticity. {ECO:0000269|PubMed:25439727}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 10A (Zellweger), 614882, Otozomal Resesif; PBD10A (Zellweger Sendromu) (PEX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 10A (Zellweger), 614882, Autosomal recessive; PBD10A (Zellweger syndrome) (PEX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX3	PEX3, PBD10A, PBD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 10A (PBD10A) [MIM:614882]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:10848631, ECO:0000269|PubMed:10958759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 10A (Zellweger), 614882, Otozomal Resesif; PBD10A (Zellweger Sendromu) (Prenatal) (PEX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 10A (Zellweger), 614882, Autosomal recessive; PBD10A (Zellweger syndrome) (PEX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX3	PEX3, PBD10A, PBD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 10A (PBD10A) [MIM:614882]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:10848631, ECO:0000269|PubMed:10958759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenezis Bozukluğu 10B, 617370; PBD10B (Zellweger Sendromu) (PEX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 10B, 617370; PBD10B (Zellweger syndrome) (PEX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX3	PEX3, PBD10A, PBD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 10A (PBD10A) [MIM:614882]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:10848631, ECO:0000269|PubMed:10958759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Peroksisom Biyogenezis Bozukluğu 10B, 617370; PBD10B (Zellweger Sendromu) (Prenatal) (PEX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 10B, 617370; PBD10B (Zellweger syndrome) (PEX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX3	PEX3, PBD10A, PBD10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 10A (PBD10A) [MIM:614882]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:10848631, ECO:0000269|PubMed:10958759}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 11A (Zellweger), 614883, Otozomal Resesif; PBD11A (Zellweger Sendromu) (PEX13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 11A (Zellweger), 614883, Autosomal recessive; PBD11A (Zellweger syndrome) (PEX13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX13	PEX13, ZWS, NALD, PBD11A, PBD11B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 13 (PBD-CG13) [MIM:614883]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:10332040, ECO:0000269|PubMed:19449432}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 11A (Zellweger), 614883, Otozomal Resesif; PBD11A (Zellweger Sendromu) (Prenatal) (PEX13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 11A (Zellweger), 614883, Autosomal recessive; PBD11A (Zellweger syndrome) (PEX13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX13	PEX13, ZWS, NALD, PBD11A, PBD11B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 13 (PBD-CG13) [MIM:614883]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:10332040, ECO:0000269|PubMed:19449432}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 11B, 614885, Otozomal Resesif; PBD11B (Neonatal Adrenolökodistrofi) (PEX13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 11B, 614885, Autosomal recessive; PBD11B (Neonatal adrenoleukodystrophy) (PEX13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX13	PEX13, ZWS, NALD, PBD11A, PBD11B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 13 (PBD-CG13) [MIM:614883]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:10332040, ECO:0000269|PubMed:19449432}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Peroksisom Biyogenez Bozukluğu 11B, 614885, Otozomal Resesif; PBD11B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 11B, 614885, Autosomal recessive; PBD11B (Neonatal adrenoleukodystrophy) (PEX13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX13	PEX13, ZWS, NALD, PBD11A, PBD11B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 13 (PBD-CG13) [MIM:614883]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:10332040, ECO:0000269|PubMed:19449432}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 12A (Zellweger), 614886, Otozomal Resesif; PBD12A (Zellweger Sendromu) (PEX19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 12A (Zellweger), 614886, Autosomal recessive; PBD12A (Zellweger syndrome) (PEX19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX19	PEX19, PXF, HK33, D1S2223E, PBD12A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 12A (PBD12A) [MIM:614886]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269	PubMed:10051604}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 12A (Zellweger), 614886, Otozomal Resesif; PBD12A (Zellweger Sendromu) (PEX19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 12A (Zellweger), 614886, Autosomal recessive; PBD12A (Zellweger syndrome) (PEX19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX19	PEX19, PXF, HK33, D1S2223E, PBD12A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 12A (PBD12A) [MIM:614886]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269	PubMed:10051604}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 13A (Zellweger), 614887, Otozomal Resesif; PBD13A (Zellweger Sendromu) (PEX14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 13A (Zellweger), 614887, Autosomal recessive; PBD13A (Zellweger syndrome) (PEX14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX14	PEX14, PBD13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group K (PBD-CGK) [MIM:614887]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:15146459}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 13A (Zellweger), 614887, Otozomal Resesif; PBD13A (Zellweger Sendromu) (Prenatal) (PEX14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 13A (Zellweger), 614887, Autosomal recessive; PBD13A (Zellweger syndrome) (PEX14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX14	PEX14, PBD13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group K (PBD-CGK) [MIM:614887]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:15146459}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenezis Bozukluğu 14B, 614920, Otozomal Resesif; PEX14B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX11B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 14B, 614920, Autosomal recessive; PEX14B (Neonatal adrenoleukodystrophy) (PEX11B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX11B	PEX11B, PEX14B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 14B (PBD14B) [MIM:614920]: An autosomal recessive peroxisome biogenesis disorder characterized clinically by mild intellectual disability, congenital cataracts, progressive hearing loss, and polyneuropathy. Additionally, recurrent migraine-like episodes following mental stress or physical exertion, not a common feature in peroxisome disorders, are observed. {ECO:0000269	PubMed:22581968}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenezis Bozukluğu 14B, 614920, Otozomal Resesif; PEX14B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX11B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 14B, 614920, Autosomal recessive; PEX14B (Neonatal adrenoleukodystrophy) (PEX11B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX11B	PEX11B, PEX14B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 14B (PBD14B) [MIM:614920]: An autosomal recessive peroxisome biogenesis disorder characterized clinically by mild intellectual disability, congenital cataracts, progressive hearing loss, and polyneuropathy. Additionally, recurrent migraine-like episodes following mental stress or physical exertion, not a common feature in peroxisome disorders, are observed. {ECO:0000269	PubMed:22581968}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 1A (Zellweger), 214100, Otozomal Resesif; PBD1A (Zellweger Sendromu) (PEX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 1A (Zellweger), 214100, Autosomal recessive; PBD1A (Zellweger syndrome) (PEX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX1	PEX1, ZWS1, PBD1A, PBD1B, HML#1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 1 (PBD-CG1) [MIM:214100]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 1A (Zellweger), 214100, Otozomal Resesif; PBD1A (Zellweger Sendromu) (Prenatal) (PEX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 1A (Zellweger), 214100, Autosomal recessive; PBD1A (Zellweger syndrome) (PEX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX1	PEX1, ZWS1, PBD1A, PBD1B, HML#1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 1 (PBD-CG1) [MIM:214100]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 1B (NALD / IRD), 601539, Otozomal Resesif; PBD1B (Neonatal Adrenolökodistrofi) (PEX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 1B (NALD/IRD), 601539, Autosomal recessive; PBD1B (Neonatal adrenoleukodystrophy) (PEX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX1	PEX1, ZWS1, PBD1A, PBD1B, HML#1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 1 (PBD-CG1) [MIM:214100]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 1B (NALD / IRD), 601539, Otozomal Resesif; PBD1B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 1B (NALD/IRD), 601539, Autosomal recessive; PBD1B (Neonatal adrenoleukodystrophy) (PEX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX1	PEX1, ZWS1, PBD1A, PBD1B, HML#1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 1 (PBD-CG1) [MIM:214100]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 2A (Zellweger), 214110, Otozomal Resesif; PBD2A (Zellweger Sendromu) (PEX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 2A (Zellweger), 214110, Autosomal recessive; PBD2A (Zellweger syndrome) (PEX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX5	PEX5, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10482504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Peroksizom Biyogenez Bozukluğu 2A (Zellweger), 214110, Otozomal Resesif; PBD2A (Zellweger Sendromu) (Prenatal) (PEX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 2A (Zellweger), 214110, Autosomal recessive; PBD2A (Zellweger syndrome) (PEX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX5	PEX5, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10482504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 2B, 202370, Otozomal Resesif; PBD2B (Neonatal Adrenolökodistrofi) (PEX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 2B, 202370, Autosomal recessive; PBD2B (Neonatal adrenoleukodystrophy) (PEX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX5	PEX5, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10482504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 2B, 202370, Otozomal Resesif; PBD2B (Neonatal Adrenolökodistrofi) (PEX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 2B, 202370, Autosomal recessive; PBD2B (Neonatal adrenoleukodystrophy) (PEX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX5	PEX5, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10482504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 3A (Zellweger), 614859, Otozomal Resesif; PBD3A (Zellweger Sendromu) (PEX12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 3A (Zellweger), 614859, Autosomal recessive; PBD3A (Zellweger syndrome) (PEX12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX12	PEX12, PBD3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 3B (PBD3B) [MIM:266510]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:10562279, ECO:0000269|PubMed:19105186}.;	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 3A (Zellweger), 614859, Otozomal Resesif; PBD3A (Zellweger Sendromu) (PEX12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 3A (Zellweger), 614859, Autosomal recessive; PBD3A (Zellweger syndrome) (PEX12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX12	PEX12, PBD3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 3B (PBD3B) [MIM:266510]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:10562279, ECO:0000269|PubMed:19105186}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 3B, 266510, Otozomal Resesif; PBD3B (Neonatal Adrenolökodistrofi) (PEX12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 3B, 266510, Autosomal recessive; PBD3B (Neonatal adrenoleukodystrophy) (PEX12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX12	PEX12, PBD3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 3B (PBD3B) [MIM:266510]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:10562279, ECO:0000269|PubMed:19105186}.;	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 3B, 266510, Otozomal Resesif; PBD3B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 3B, 266510, Autosomal recessive; PBD3B (Neonatal adrenoleukodystrophy) (PEX12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX12	PEX12, PBD3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 3B (PBD3B) [MIM:266510]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269|PubMed:10562279, ECO:0000269|PubMed:19105186}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 4A (Zellweger), 614862, Otozomal Resesif; PBD4A (Zellweger Sendromu) (PEX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 4A (Zellweger), 614862, Autosomal recessive; PBD4A (Zellweger syndrome) (PEX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX6	PEX6, PXAAA1, PAF2, PBD4A, PDB4B, HML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 4 (PBD-CG4) [MIM:614862]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 4A (Zellweger), 614862, Otozomal Resesif; PBD4A (Zellweger Sendromu) (PEX6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 4A (Zellweger), 614862, Autosomal recessive; PBD4A (Zellweger syndrome) (PEX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX6	PEX6, PXAAA1, PAF2, PBD4A, PDB4B, HML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 4 (PBD-CG4) [MIM:614862]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 4B, 614863, Otozomal Resesif; PBD4B (Neonatal Adrenolökodistrofi) (PEX6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 4B, 614863, Autosomal recessive; PBD4B (Neonatal adrenoleukodystrophy) (PEX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX6	PEX6, PXAAA1, PAF2, PBD4A, PDB4B, HML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 4 (PBD-CG4) [MIM:614862]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186, ECO:0000269|PubMed:26387595}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Peroksizom Biyogenez Bozukluğu 4B, 614863, Otozomal Resesif; PBD4B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 4B, 614863, Autosomal recessive; PBD4B (Neonatal adrenoleukodystrophy) (PEX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX6	PEX6, PXAAA1, PAF2, PBD4A, PDB4B, HML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 4 (PBD-CG4) [MIM:614862]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:26387595, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 5A (Zellweger), 614866, Otozomal Resesif; PBD5A (Zellweger Sendromu) (PEX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 5A (Zellweger), 614866, Autosomal recessive; PBD5A (Zellweger syndrome) (PEX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX2	PEX2, PAF1, PMP35, PBD5A, PBD5B, PXMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 5 (PBD-CG5) [MIM:614866]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksizom Biyogenez Bozukluğu 5A (Zellweger), 614866, Otozomal Resesif; PBD5A (Zellweger Sendromu) (Prenatal) (PEX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 5A (Zellweger), 614866, Autosomal recessive; PBD5A (Zellweger syndrome) (PEX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX2	PEX2, PAF1, PMP35, PBD5A, PBD5B, PXMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 5 (PBD-CG5) [MIM:614866]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 5B, 614867, Otozomal Resesif; PBD5B (Neonatal Adrenoleukodistrofi) (PEX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 5B, 614867, Autosomal recessive; PBD5B (Neonatal adrenoleukodystrophy) (PEX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX2	PEX2, PAF1, PMP35, PBD5A, PBD5B, PXMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 5 (PBD-CG5) [MIM:614866]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 5B, 614867, Otozomal Resesif; PBD5B (Neonatal Adrenoleukodistrofi) (Prenatal) (PEX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 5B, 614867, Autosomal recessive; PBD5B (Neonatal adrenoleukodystrophy) (PEX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX2	PEX2, PAF1, PMP35, PBD5A, PBD5B, PXMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 5 (PBD-CG5) [MIM:614866]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 6A (Zellweger), 614870, Otozomal Resesif; PBD6A (Zellweger Sendromu) (PEX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 6A (Zellweger), 614870, Autosomal recessive; PBD6A (Zellweger syndrome) (PEX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX10	PEX10, NALD, PBD6A, PBD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 7 (PBD-CG7) [MIM:614870]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 6A (Zellweger), 614870, Otozomal Resesif; PBD6A (Zellweger Sendromu) (Prenatal) (PEX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 6A (Zellweger), 614870, Autosomal recessive; PBD6A (Zellweger syndrome) (PEX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX10	PEX10, NALD, PBD6A, PBD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 7 (PBD-CG7) [MIM:614870]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 6B, 614871, Otozomal Resesif; PBD6B (Neonatal Adrenoleukodistrofi) (PEX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 6B, 614871, Autosomal recessive; PBD6B (Neonatal adrenoleukodystrophy) (PEX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX10	PEX10, NALD, PBD6A, PBD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 7 (PBD-CG7) [MIM:614870]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186}, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 6B, 614871, Otozomal Resesif; PBD6B (Neonatal Adrenoleukodistrofi) (Prenatal) (PEX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 6B, 614871, Autosomal recessive; PBD6B (Neonatal adrenoleukodystrophy) (PEX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX10	PEX10, NALD, PBD6A, PBD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 7 (PBD-CG7) [MIM:614870]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269|PubMed:19105186}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 7A (Zellweger), 614872, Otozomal Resesif; PBD7A (Zellweger Sendromu) (PEX26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 7A (Zellweger), 614872, Autosomal recessive; PBD7A (Zellweger syndrome) (PEX26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX26	PEX26, PBD7A, PBD7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 7A (PBD7A) [MIM:614872]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:12851857}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 7A (Zellweger), 614872, Otozomal Resesif; PBD7A (Zellweger Sendromu) (Prenatal) (PEX26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 7A (Zellweger), 614872, Autosomal recessive; PBD7A (Zellweger syndrome) (PEX26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX26	PEX26, PBD7A, PBD7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 7A (PBD7A) [MIM:614872]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:12851857}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 7B, 614873, Otozomal Resesif; PBD7B (Neonatal Adrenoleukodistrofi) (PEX26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 7B, 614873, Autosomal recessive; PBD7B (Neonatal adrenoleukodystrophy) (PEX26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX26	PEX26, PBD7A, PBD7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 7A (PBD7A) [MIM:614872]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269|PubMed:12851857}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Peroksisom Biyogenez Bozukluğu 7B, 614873, Otozomal Resesif; PBD7B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 7B, 614873, Autosomal recessive; PBD7B (Neonatal adrenoleukodystrophy) (PEX26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX26	PEX26, PBD7A, PBD7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 7A (PBD7A) [MIM:614872]: A fatal peroxisome biogenesis disorder belonging to the Zellweger disease spectrum and clinically characterized by severe neurologic dysfunction with profound psychomotor retardation, severe hypotonia and neonatal seizures, craniofacial abnormalities, liver dysfunction, and biochemically by the absence of peroxisomes. Additional features include cardiovascular and skeletal defects, renal cysts, ocular abnormalities, and hearing impairment. Most severely affected individuals with the classic form of the disease (classic Zellweger syndrome) die within the first year of life. {ECO:0000269	PubMed:12851857}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 8A (Zellweger), 614876, Otozomal Resesif; PBD8A (Zellweger Sendromu) (PEX16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 8A (Zellweger), 614876, Autosomal recessive; PBD8A (Zellweger syndrome) (PEX16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX16	PEX16, PBD8A, PBD8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 9 (PBD-CG9) [MIM:614876]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:9837814}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 8A (Zellweger), 614876, Otozomal Resesif; PBD8A (Zellweger Sendromu) (Prenatal) (PEX16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 8A (Zellweger), 614876, Autosomal recessive; PBD8A (Zellweger syndrome) (PEX16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX16	PEX16, PBD8A, PBD8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 9 (PBD-CG9) [MIM:614876]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:9837814}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 8B, 614877, Otozomal Resesif; PBD8B (Neonatal Adrenolökodistrofi) (PEX16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 8B, 614877, Autosomal recessive; PBD8B (Neonatal adrenoleukodystrophy) (PEX16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX16	PEX16, PBD8A, PBD8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder complementation group 9 (PBD-CG9) [MIM:614876]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:9837814}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 8B, 614877, Otozomal Resesif; PBD8B (Neonatal Adrenolökodistrofi) (Prenatal) (PEX16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 8B, 614877, Autosomal recessive; PBD8B (Neonatal adrenoleukodystrophy) (PEX16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX16	PEX16, PBD8A, PBD8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder complementation group 9 (PBD-CG9) [MIM:614876]: A peroxisomal disorder arising from a failure of protein import into the peroxisomal membrane or matrix. The peroxisome biogenesis disorders (PBD group) are genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies. Include disorders are: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum (PBD-ZSS). {ECO:0000269	PubMed:9837814}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 9B, 614879; PBD9B (Refsum Hastalığı) (PEX7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peroxisome biogenesis disorder 9B, 614879; PBD9B (Refsum disease) (PEX7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX7	PEX7, RCDP1, PBD9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhizomelic chondrodysplasia punctata 1 (RCDP1) [MIM:215100]: A peroxisome biogenesis disorder. It is characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269	PubMed:9090381}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peroksisom Biyogenez Bozukluğu 9B, 614879; PBD9B (Refsum Hastalığı) (Prenatal) (PEX7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peroxisome biogenesis disorder 9B, 614879; PBD9B (Refsum disease) (PEX7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX7	PEX7, RCDP1, PBD9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rhizomelic chondrodysplasia punctata 1 (RCDP1) [MIM:215100]: A peroxisome biogenesis disorder. It is characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269	PubMed:9090381}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 1, 233400, Otozomal Resesif; PRLTS1 (Perrault Sendromu) (HSD17B4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perrault syndrome 1, 233400, Autosomal recessive; PRLTS1 (Perrault syndrome) (HSD17B4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD17B4	HSD17B4, PRLTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Perrault syndrome 1 (PRLTS1) [MIM:233400]: A sex-influenced disorder characterized by sensorineural deafness in both males and females and ovarian dysgenesis in females. Some patients also have neurologic manifestations, including mild mental retardation and cerebellar and peripheral nervous system involvement. {ECO:0000269	PubMed:20673864}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Perrault Sendromu 1, 233400, Otozomal Resesif; PRLTS1 (Perrault Sendromu) (Prenatal) (HSD17B4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perrault syndrome 1, 233400, Autosomal recessive; PRLTS1 (Perrault syndrome) (HSD17B4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSD17B4	HSD17B4, PRLTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Perrault syndrome 1 (PRLTS1) [MIM:233400]: A sex-influenced disorder characterized by sensorineural deafness in both males and females and ovarian dysgenesis in females. Some patients also have neurologic manifestations, including mild mental retardation and cerebellar and peripheral nervous system involvement. {ECO:0000269	PubMed:20673864}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 2, 614926, Otozomal Resesif; PRLTS2 (Perrault Sendromu) (HARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perrault syndrome 2, 614926, Autosomal recessive; PRLTS2 (Perrault syndrome) (HARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HARS2	HARS2, HARSL, HARSR, HO3, PRLTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Perrault Sendromu 2, 614926, Otozomal Resesif; PRLTS2 (Perrault Sendromu) (Prenatal) (HARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perrault syndrome 2, 614926, Autosomal recessive; PRLTS2 (Perrault syndrome) (HARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HARS2	HARS2, HARSL, HARSR, HO3, PRLTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 3, 614129, Otozomal Resesif; PRLTS3 (Perrault Sendromu) (CLPP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perrault syndrome 3, 614129, Autosomal recessive; PRLTS3 (Perrault syndrome) (CLPP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLPP	CLPP, PRLTS3, DFNB81	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Perrault Sendromu 3, 614129, Otozomal Resesif; PRLTS3 (Perrault Sendromu) (Prenatal) (CLPP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perrault syndrome 3, 614129, Autosomal recessive; PRLTS3 (Perrault syndrome) (CLPP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLPP	CLPP, PRLTS3, DFNB81	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 4, 615300, Otozomal Resesif; PRLTS4 (Perrault Sendromu) (LARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perrault syndrome 4, 615300, Autosomal recessive; PRLTS4 (Perrault syndrome) (LARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LARS2	LARS2, PRLTS4, HLASA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Perrault Sendromu 4, 615300, Otozomal Resesif; PRLTS4 (Perrault Sendromu) (Prenatal) (LARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perrault syndrome 4, 615300, Autosomal recessive; PRLTS4 (Perrault syndrome) (LARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LARS2	LARS2, PRLTS4, HLASA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 5, 616138, Otozomal Resesif; PRLTS5 (Perrault Sendromu) (TWNK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perrault syndrome 5, 616138, Autosomal recessive; PRLTS5 (Perrault syndrome) (TWNK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Perrault Sendromu 5, 616138, Otozomal Resesif; PRLTS5 (Perrault Sendromu) (MLPA)	Perrault syndrome 5, 616138, Autosomal recessive; PRLTS5 (Perrault syndrome) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Perrault Sendromu 5, 616138, Otozomal Resesif; PRLTS5 (Perrault Sendromu) (Prenatal) (TWNK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perrault syndrome 5, 616138, Autosomal recessive; PRLTS5 (Perrault syndrome) (TWNK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perrault Sendromu 5, 616138, Otozomal Resesif; PRLTS5 (Perrault Sendromu) (Prenatal) (MLPA)	Perrault syndrome 5, 616138, Autosomal recessive; PRLTS5 (Perrault syndrome) (Prenatal) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Perry Sendromu, 168605, Otozomal Dominant (Perry Sendromu) (DCTN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Perry syndrome, 168605, Autosomal dominant (Perry syndrome) (DCTN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCTN1	DCTN1, HMN7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis (ALS) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:15326253, ECO:0000269|PubMed:16240349}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Perry Sendromu, 168605, Otozomal Dominant (Perry Sendromu) (Prenatal) (DCTN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Perry syndrome, 168605, Autosomal dominant (Perry syndrome) (DCTN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCTN1	DCTN1, HMN7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Amyotrophic lateral sclerosis (ALS) [MIM:105400]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:15326253, ECO:0000269|PubMed:16240349}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kalıcı Hiperplastik Primer Vitreus, Otozomal Resesif, 221900, Otozomal Resesif; PHPVAR (Persistan Hiperplastik Primer Vitreus) (ATOH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Persistent hyperplastic primary vitreous, autosomal recessive, 221900, Autosomal recessive; PHPVAR (Persistent hyperplastic primary vitreous) (ATOH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATOH7	ATOH7, PHPVAR, NCRNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Persistent hyperplastic primary vitreous, autosomal recessive (PHPVAR) [MIM:221900]: A developmental eye malformation associated with microphthalmia, cataract, glaucoma, and congenital retinal non-attachment. It is due to failure of the primary vitreous to regress in utero, resulting in the presence of a retrolental fibrovascular membrane with persistence of the posterior portion of the tunica vasculosa lentis and hyaloid artery. Disease manifestations range from a trivial remnant of hyaloid vessels to a dense fibrovascular mass causing lens opacity and retinal detachment. {ECO:0000269|PubMed:21441919, ECO:0000269|PubMed:22068589, ECO:0000269|PubMed:22645276}. Note=The disease is caused by mutations affecting the gene represented in this entry. A 6.5 kb deletion that spans a remote cis regulatory element 20 kb upstream from ATOH7 has been found in PHPVAR patients (PubMed:21441919). {ECO:0000269|PubMed:21441919},.	EDTA Blood Sample
Kalıcı Müllerian Kanal Sendromu, Tip 1, 261550, Otozomal Resesif (Persistant Müllerian Kanal Sendromu) (AMH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Persistent Mullerian duct syndrome, type I, 261550, Autosomal recessive (Persistent Müllerian duct syndrome) (AMH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMH	AMH, MIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Persistent Muellerian duct syndrome 1 (PMDS1) [MIM:261550]: A form of male pseudohermaphroditism characterized by a failure of Muellerian duct regression in otherwise normal males. {ECO:0000269|PubMed:8162013, ECO:0000269|PubMed:8872466}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kalıcı Müllerian Kanal Sendromu, Tip 2, 261550, Otozomal Resesif; PMDS (Persistant Müllerian Kanal Sendromu) (AMHR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Persistent Mullerian duct syndrome, type II, 261550, Autosomal recessive; PMDS (Persistent Müllerian duct syndrome) (AMHR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMHR2	AMHR2, AMHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Persistent Muellerian duct syndrome 2 (PMDS2) [MIM:261550]: A form of male pseudohermaphroditism characterized by a failure of Muellerian duct regression in otherwise normal males. {ECO:0000269|PubMed:11549681, ECO:0000269|PubMed:8872466}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Persistan Truncus Arteriosus, 217095 (Konotrunkal Kalp Malformasyonları) (NKX2-6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Persistent truncus arteriosus, 217095 (Conotruncal heart malformations) (NKX2-6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-6	NKX2-6, CSX2, CTHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Conotruncal heart malformations (CTHM) [MIM:217095]: A group of congenital heart defects involving the outflow tracts. Examples include truncus arteriosus communis, double-outlet right ventricle and transposition of great arteries. Truncus arteriosus communis is characterized by a single outflow tract instead of a separate aorta and pulmonary artery. In transposition of the great arteries, the aorta arises from the right ventricle and the pulmonary artery from the left ventricle. In double outlet of the right ventricle, both the pulmonary artery and aorta arise from the right ventricle. {ECO:0000269|PubMed:15649947, ECO:0000269|PubMed:14421281, ECO:0000269|PubMed:25195019, ECO:0000269|PubMed:25380965}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Persistan Truncus Arteriosus, 217095 (Trunkus Arteriozus) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Persistent truncus arteriosus, 217095 (Truncus arteriosus) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peters Plus Sendromu, 261540, Otozomal Resesif (Peters Plus Sendromu) (B3GLCT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peters-plus syndrome, 261540, Autosomal recessive (Peters plus syndrome) (B3GLCT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GLCT	B3GLCT, B3GALTL, B3GTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Peters Plus Sendromu, 261540, Otozomal Resesif (Peters Plus Sendromu) (Prenatal) (B3GLCT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Peters-plus syndrome, 261540, Autosomal recessive (Peters plus syndrome) (B3GLCT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B3GLCT	B3GLCT, B3GALTL, B3GTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pettigrew Sendromu, Syf (X-e Bağlı Entelektüel Yetersizlik-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı-Nöbet Sendromu) (AP1S2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PETTIGREW SYNDROME; PGS (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (AP1S2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP1S2	AP1S2, MRX59, MRXSF, MRXS21, MRXSS, PGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pettigrew Sendromu, Syf (X-e Bağlı Entelektüel Yetersizlik-Dandy-Walker Malformasyonu-Bazal Ganglion Hastalığı–Nöbet Sendromu) (Prenatal) (AP1S2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PETTIGREW SYNDROME; PGS (X-linked intellectual disability-Dandy-Walker malformation-basal ganglia disease-seizures syndrome) (AP1S2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AP1S2	AP1S2, MRX59, MRXSF, MRXS21, MRXSS, PGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Peutz-Jeghers sendromu (STK11 19p13.3) (MLPA)	Peutz-Jeghers syndrome (STK11 19p13.3) (MLPA)	STK11 19p13.3	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Peutz-Jeghers Sendromu, 175200, Otozomal Dominant; PJS (Peutz-Jeghers Sendromu) (STK11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Peutz-Jeghers syndrome, 175200, Autosomal dominant; PJS (Peutz-Jeghers syndrome) (STK11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STK11	STK11, PJS, LKB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peutz-Jeghers syndrome (PJS) [MIM:175200]: An autosomal dominant disorder characterized by melanocytic macules of the lips, multiple gastrointestinal hamartomatous polyps and an increased risk for various neoplasms, including gastrointestinal cancer. {ECO:0000269|PubMed:10408777, ECO:0000269|PubMed:12372054, ECO:0000269|PubMed:21411391, ECO:0000269|PubMed:9425897, ECO:0000269|PubMed:9428765, ECO:0000269|PubMed:9760200, ECO:0000269|PubMed:9837816}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Peutz-Jeghers Sendromu, 175200, Otozomal Dominant; PJS (Peutz-Jeghers Sendromu) (MLPA)	Peutz-Jeghers syndrome, 175200, Autosomal dominant; PJS (Peutz-Jeghers syndrome) (MLPA)	STK11	STK11, PJS, LKB1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Peutz-Jeghers syndrome (PJS) [MIM:175200]: An autosomal dominant disorder characterized by melanocytic macules of the lips, multiple gastrointestinal hamartomatous polyps and an increased risk for various neoplasms, including gastrointestinal cancer. {ECO:0000269|PubMed:10408777, ECO:0000269|PubMed:12372054, ECO:0000269|PubMed:21411391, ECO:0000269|PubMed:9425897, ECO:0000269|PubMed:9428765, ECO:0000269|PubMed:9760200, ECO:0000269|PubMed:9837816}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pfeiffer syndrome, 101600, autosomal dominant (Pfeiffer syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pfeiffer syndrome, 101600, autosomal dominant (Pfeiffer syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (MLPA)	Pfeiffer syndrome, 101600, Autosomal dominant (Pfeiffer syndrome) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pfeiffer syndrome, 101600, autosomal dominant (Pfeiffer syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly of brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (Prenatal) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pfeiffer syndrome, 101600, Autosomal dominant (Pfeiffer syndrome) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pfeiffer Sendromu, 101600, Otozomal Dominant (Pfeiffer Sendromu) (Prenatal) (MLPA)	Pfeiffer syndrome, 101600, Autosomal dominant (Pfeiffer syndrome) (Prenatal) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269	PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Phelan-Mcdermid Sendromu, 606232, Sporadik Vakalar; PHMDS (Monozomi 22q13 (Phelan-Mcdermid Sendromu)) (2200)	Phelan-McDermid syndrome, 606232, Isolated cases; PHMDS (Monosomy 22q13) (440)	SHANK3	SHANK3, PSAP2, PROSAP2, KIAA1650, DEL22q13.3, SCZD15	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	DISEASE: Phelan-McDermid syndrome (PHMDS) [MIM:606232]: A developmental disorder with variable features. Common features include neonatal hypotonia, global developmental delay, normal to accelerated growth, absent to severely delayed speech, autistic behavior, and minor dysmorphic features. {ECO:0000269|PubMed:22892527, ECO:0000269|PubMed:23758760, ECO:0000269|PubMed:24132240}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample and Heparinized blood sample
Phelan-Mcdermid Sendromu, 606232, Sporadik Vakalar; PHMDS (Monozomi 22q13 (Phelan-Mcdermid Sendromu)) (Prenatal)	Phelan-McDermid syndrome, 606232, Isolated cases; PHMDS (Monosomy 22q13) (Prenatal)	SHANK3	SHANK3, PSAP2, PROSAP2, KIAA1650, DEL22q13.3, SCZD15	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phelan-McDermid syndrome (PHMDS) [MIM:606232]: A developmental disorder with variable features. Common features include neonatal hypotonia, global developmental delay, normal to accelerated growth, absent to severely delayed speech, autistic behavior, and minor dysmorphic features. {ECO:0000269|PubMed:22892527, ECO:0000269|PubMed:23758760, ECO:0000269|PubMed:24132240}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fenilketonüri, 261600, Otozomal Resesif; PKU (Fenilketonüri) (PAH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phenylketonuria, 261600, Autosomal recessive; PKU (Phenylketonuria) (PAH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAH	PAH, PKU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phenylketonuria (PKU) [MIM:261600]: Autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. {ECO:0000269|PubMed:10200057, ECO:0000269|PubMed:10679941, ECO:0000269|PubMed:11180595, ECO:0000269|PubMed:11385716, ECO:0000269|PubMed:11461196, ECO:0000269|PubMed:12501224, ECO:0000269|PubMed:1355066, ECO:0000269|PubMed:1363837, ECO:0000269|PubMed:1363838, ECO:0000269|PubMed:1671810, ECO:0000269|PubMed:1672290, ECO:0000269|PubMed:1672294, ECO:0000269|PubMed:1679030, ECO:0000269|PubMed:1709636, ECO:0000269|PubMed:1975559, ECO:0000269|PubMed:2014802, ECO:0000269|PubMed:22513348, ECO:0000269|PubMed:22526846, ECO:0000269|PubMed:23792259, ECO:0000269|PubMed:2564729, ECO:0000269|PubMed:2615549, ECO:0000269|PubMed:2840952, ECO:0000269|PubMed:3935954	EDTA Blood Sample
Fenilketonüri, 261600, Otozomal Resesif; PKU (Fenilketonüri) (MLPA)	Phenylketonuria, 261600, Autosomal recessive; PKU (Phenylketonuria) (MLPA)	PAH	PAH, PKU1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Phenylketonuria (PKU) [MIM:261600]: Autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. {ECO:0000269|PubMed:10200057, ECO:0000269|PubMed:10679941, ECO:0000269|PubMed:11180595, ECO:0000269|PubMed:11385716, ECO:0000269|PubMed:11461196, ECO:0000269|PubMed:12501224, ECO:0000269|PubMed:1355066, ECO:0000269|PubMed:1363837, ECO:0000269|PubMed:1363838, ECO:0000269|PubMed:1671810, ECO:0000269|PubMed:1672290, ECO:0000269|PubMed:1672294, ECO:0000269|PubMed:1679030, ECO:0000269|PubMed:1709636, ECO:0000269|PubMed:1975559, ECO:0000269|PubMed:2014802, ECO:0000269|PubMed:22513348, ECO:0000269|PubMed:22526846, ECO:0000269|PubMed:23792259, ECO:0000269|PubMed:2564729, ECO:0000269|PubMed:2615549, ECO:0000269|PubMed:2840952, ECO:0000269|PubMed:3935954	EDTA Blood Sample
Fenilketonüri, 261600, Otozomal Resesif; PKU (Fenilketonüri) (PAH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phenylketonuria, 261600, Autosomal recessive; PKU (Phenylketonuria) (PAH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAH	PAH, PKU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phenylketonuria (PKU) [MIM:261600]: Autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. {ECO:0000269|PubMed:10200057, ECO:0000269|PubMed:10679941, ECO:0000269|PubMed:11180595, ECO:0000269|PubMed:11385716, ECO:0000269|PubMed:11461196, ECO:0000269|PubMed:12501224, ECO:0000269|PubMed:1355066, ECO:0000269|PubMed:1363837, ECO:0000269|PubMed:1363838, ECO:0000269|PubMed:1671810, ECO:0000269|PubMed:1672290, ECO:0000269|PubMed:1672294, ECO:0000269|PubMed:1679030, ECO:0000269|PubMed:1709636, ECO:0000269|PubMed:1975559, ECO:0000269|PubMed:2014802, ECO:0000269|PubMed:22513348, ECO:0000269|PubMed:22526846, ECO:0000269|PubMed:23792259, ECO:0000269|PubMed:2564729, ECO:0000269|PubMed:2615549, ECO:0000269|PubMed:2840952, ECO:0000269|PubMed:3935954	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fenilketonüri, 261600, Otozomal Resesif; PKU (Fenilketonüri) (Prenatal) (MLPA)	Phenylketonuria, 261600, Autosomal recessive; PKU (Phenylketonuria) (Prenatal) (MLPA)	PAH	PAH, PKU1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phenylketonuria (PKU) [MIM:261600]: Autosomal recessive inborn error of phenylalanine metabolism, due to severe phenylalanine hydroxylase deficiency. It is characterized by blood concentrations of phenylalanine persistently above 1200 mumol (normal concentration 100 mumol) which usually causes mental retardation (unless low phenylalanine diet is introduced early in life). They tend to have light pigmentation, rashes similar to eczema, epilepsy, extreme hyperactivity, psychotic states and an unpleasant 'mousy' odor. {ECO:0000269|PubMed:10200057, ECO:0000269|PubMed:10679941, ECO:0000269|PubMed:11180595, ECO:0000269|PubMed:11385716, ECO:0000269|PubMed:11461196, ECO:0000269|PubMed:12501224, ECO:0000269|PubMed:1355066, ECO:0000269|PubMed:1363837, ECO:0000269|PubMed:1363838, ECO:0000269|PubMed:1671810, ECO:0000269|PubMed:1672290, ECO:0000269|PubMed:1672294, ECO:0000269|PubMed:1679030, ECO:0000269|PubMed:1709636, ECO:0000269|PubMed:1975559, ECO:0000269|PubMed:2014802, ECO:0000269|PubMed:22513348, ECO:0000269|PubMed:22526846, ECO:0000269|PubMed:23792259, ECO:0000269|PubMed:2564729, ECO:0000269|PubMed:2615549, ECO:0000269|PubMed:2840952, ECO:0000269|PubMed:3935954	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma (VHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (VHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VHL	VHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. {ECO:0000269|PubMed:11886208}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (SDHD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (SDHD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHD	SDHD, PGL1, CWS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal carcinoid tumor (ICT) [MIM:114900]: A yellow, well-differentiated, circumscribed tumor that arises from enterochromaffin cells in the small intestine or, less frequently, in other parts of the gastrointestinal tract. {ECO:0000269|PubMed:12007193}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (SDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (SDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDHB	SDHB, SDH2, SDHP, PGL4, CWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Paragangliomas 4 (PGL4) [MIM:115310]: A neural crest tumor usually derived from the chromoreceptor tissue of a paraganglion. Paragangliomas can develop at various body sites, including the head, neck, thorax and abdomen. Most commonly, they are located in the head and neck region, specifically at the carotid bifurcation, the jugular foramen, the vagal nerve, and in the middle ear. {ECO:0000269|PubMed:11404820, ECO:0000269|PubMed:11897817, ECO:0000269|PubMed:14715873, ECO:0000269|PubMed:14974914, ECO:0000269|PubMed:15328326}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (RET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (RET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RET	RET, MEN2A, HSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (KIF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (KIF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1B	KIF1B, CMT2A, CMT2A1, NBLST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pheochromocytoma (PCC) [MIM:171300]: A catecholamine- producing tumor of chromaffin tissue of the adrenal medulla or sympathetic paraganglia. The cardinal symptom, reflecting the increased secretion of epinephrine and norepinephrine, is hypertension, which may be persistent or intermittent. (ECO:0000269	PubMed:18334619). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (MLPA)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (MLPA)	VHL	VHL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. (ECO:0000269	PubMed:11986208). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Feokromositoma, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (MLPA)	Pheochromocytoma, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (MLPA)	RET	RET, MEN2A, HSCR1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Feokromasitoma, Modifiye Edici, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (GDNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, modifier of, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (GDNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDNF	GDNF, HSCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Feokromasitoma, Modifiye Edici, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (MLPA)	Pheochromocytoma, modifier of, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (MLPA)	GDNF	GDNF, HSCR3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Congenital central hypoventilation syndrome (CCHS) [MIM:209880]: Rare disorder characterized by abnormal control of respiration in the absence of neuromuscular or lung disease, or an identifiable brain stem lesion. A deficiency in autonomic control of respiration results in inadequate or negligible ventilatory and arousal responses to hypercapnia and hypoxemia. (ECO:0000269	PubMed:9497256). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Feokromasitoma, Yatkınlık, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (TMEM127 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, susceptibility to, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (TMEM127 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM127	TMEM127	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Feokromasitoma, Yatkınlık, 171300, Otozomal Dominant (Kalıtsal Feokromositoma-Paraganglioma) (MAX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pheochromocytoma, susceptibility to, 171300, Autosomal dominant (Hereditary pheochromocytoma-paraganglioma) (MAX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAX	MAX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Fosfoenolpiruvat Karboksikinaz Eksikliği, Mitokondriyal (Fosfoenolpiruvat Karboksikinaz Eksikliği) (PCK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PHOSPHOENOLPYRUVATE CARBOXYKINASE DEFICIENCY, MITOCHONDRIAL (Phosphoenolpyruvate carboxykinase deficiency) (PCK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCK2	PCK2, PEPCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial phosphoenolpyruvate carboxykinase deficiency (M-PEPCKD) [MIM:261650]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfoenolpiruvat Karboksikinaz Eksikliği, Mitokondriyal (Fosfoenolpiruvat Karboksikinaz Eksikliği) (Prenatal) (PCK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PHOSPHOENOLPYRUVATE CARBOXYKINASE DEFICIENCY, MITOCHONDRIAL (Phosphoenolpyruvate carboxykinase deficiency) (PCK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCK2	PCK2, PEPCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial phosphoenolpyruvate carboxykinase deficiency (M-PEPCKD) [MIM:261650]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfoenolpiruvat Karboksikinaz-1, Sitosolik, Yetersizlik, 261680, Otozomal Resesif (Fosfoenolpiruvat Karboksikinaz Eksikliği) (PCK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency, 261680, Autosomal recessive (Phosphoenolpyruvate carboxykinase deficiency) (PCK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCK1	PCK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cytosolic phosphoenolpyruvate carboxykinase deficiency (C-PEPCKD) [MIM:261680]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfoenolpiruvat Karboksikinaz-1, Sitosolik, Yetersizlik, 261680, Otozomal Resesif (Fosfoenolpiruvat Karboksikinaz Eksikliği) (Prenatal) (PCK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoenolpyruvate carboxykinase-1, cytosolic, deficiency, 261680, Autosomal recessive (Phosphoenolpyruvate carboxykinase deficiency) (PCK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCK1	PCK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cytosolic phosphoenolpyruvate carboxykinase deficiency (C-PEPCKD) [MIM:261680]: Metabolic disorder resulting from impaired gluconeogenesis. It is a rare disease with less than 10 cases reported in the literature. Clinical characteristics include hypotonia, hepatomegaly, failure to thrive, lactic acidosis and hypoglycemia. Autopsy reveals fatty infiltration of both the liver and kidneys. The disorder is transmitted as an autosomal recessive trait. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfogliserat Dehidrojenaz Yetmezliği, 601815, Otozomal Resesif, PHGDHD (3-Fosfogliserat Dehidrogenaz Eksikliği, İnfantil Formlu) (PHGDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoglycerate dehydrogenase deficiency, 601815, Autosomal recessive, PHGDHD (3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form) (PHGDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHGDH	PHGDH, NLS1, PHGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neu-Laxova syndrome 1 (NLS1) [MIM:256520]: A lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies including lissencephaly, cerebellar hypoplasia and/or abnormalitygenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. (ECO:0000269	PubMed:24836451). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfogliserat Dehidrojenaz Yetmezliği, 601815, Otozomal Resesif, PHGDHD (3-Fosfogliserat Dehidrogenaz Eksikliği, İnfantil / Juvenil Formi) (Prenatal) (PHGDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoglycerate dehydrogenase deficiency, 601815, Autosomal recessive, PHGDHD (3-phosphoglycerate dehydrogenase deficiency, infantile/juvenile form) (PHGDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHGDH	PHGDH, NLS1, PHGDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neu-Laxova syndrome 1 (NLS1) [MIM:256520]: A lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies including lissencephaly, cerebellar hypoplasia and/or abnormalitygenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. (ECO:0000269	PubMed:24836451). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfogliserat Kinaz 1 Eksikliği, 300653, X'e Bağlı Resesif (Glikojen Depo Hastalığı, Fosfogliserat Kinaz 1 Eksikliğine Bağlı) (PGK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoglycerate kinase 1 deficiency, 300653, X-linked recessive (Glycogen storage disease due to phosphoglycerate kinase 1 deficiency) (PGK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGK1	PGK1, PGKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phosphoglycerate kinase 1 deficiency (PGK1D) [MIM:300653]: A condition with a highly variable clinical phenotype that includes hemolytic anemia, rhabdomyolysis, myopathy and neurologic involvement. Patients can express one or more of these manifestations. (ECO:0000269|PubMed:1547346, ECO:0000269|PubMed:1586722, ECO:0000269|PubMed:2001457, ECO:0000269|PubMed:6933565, ECO:0000269|PubMed:6941312, ECO:0000269|PubMed:8043870, ECO:0000269|PubMed:8615693, ECO:0000269|PubMed:8673469, ECO:0000269|PubMed:9744480). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfogliserat Kinaz 1 Eksikliği, 300653, X'e Bağlı Resesif (Glikojen Depo Hastalığı, Fosfogliserat Kinaz 1 Eksikliğine Bağlı) (Prenatal) (PGK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoglycerate kinase 1 deficiency, 300653, X-linked recessive (Glycogen storage disease due to phosphoglycerate kinase 1 deficiency) (PGK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PGK1	PGK1, PGKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phosphoglycerate kinase 1 deficiency (PGK1D) [MIM:300653]: A condition with a highly variable clinical phenotype that includes hemolytic anemia, rhabdomyolysis, myopathy and neurologic involvement. Patients can express one or more of these manifestations. (ECO:0000269|PubMed:1547346, ECO:0000269|PubMed:1586722, ECO:0000269|PubMed:2001457, ECO:0000269|PubMed:6933565, ECO:0000269|PubMed:6941312, ECO:0000269|PubMed:8043870, ECO:0000269|PubMed:8615693, ECO:0000269|PubMed:8673469, ECO:0000269|PubMed:9744480). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfolipaz A2, Grup 4 A, Eksikliği, (PLA2G4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phospholipase A2, group IV A, deficiency of (PLA2G4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G4A	PLA2G4A, PLA2G4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=PLA2G4A mutations resulting in phospholipase A2 deficiency have been found in a patient affected by recurrent episodes of multiple complicated ulcers of the small intestine, not due to cyclooxygenase inhibitors use. Disease features also include platelet dysfunction, and globally decreased eicosanoid synthesis (PubMed:18451993). (ECO:0000269	PubMed:18451993).	EDTA Blood Sample
Fosfolipaz A2, Grup 4 A, Eksikliği, (Prenatal) (PLA2G4A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phospholipase A2, group IV A, deficiency of (PLA2G4A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLA2G4A	PLA2G4A, PLA2G4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=PLA2G4A mutations resulting in phospholipase A2 deficiency have been found in a patient affected by recurrent episodes of multiple complicated ulcers of the small intestine, not due to cyclooxygenase inhibitors use. Disease features also include platelet dysfunction, and globally decreased eicosanoid synthesis (PubMed:18451993). (ECO:0000269	PubMed:18451993).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Fosforibozilprofosfat Sentetaz Süperaktivitesi, 300681, X'e Bağlı Resesif (Fosforiboziloprofosfat Sentetaz Süperaktivitesi) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoribosylpyrophosphate synthetase superactivity, 300681, X-linked recessive (Phosphoribosylpyrophosphate synthetase superactivity) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phosphoribosylpyrophosphate synthetase superactivity (PRPS1 superactivity) [MIM:300661]: Familial disorder characterized by excessive purine production, gout and uric acid urolithiasis. {ECO:0000269|PubMed:7593598, ECO:0000269|Ref.12}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fosforiboziloprofosfat Sentetaz Süperaktivitesi, 300661, X'e Bağlı Resesif (Fosforiboziloprofosfat Sentetaz Süperaktivitesi) (Prenatal) (PRPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoribosylpyrophosphate synthetase superactivity, 300661, X-linked recessive (Phosphoribosylpyrophosphate synthetase superactivity) (PRPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRPS1	PRPS1, CMTX5, DFNX1, DFN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phosphoribosylpyrophosphate synthetase superactivity (PRPS1 superactivity) [MIM:300661]: Familial disorder characterized by excessive purine production, gout and uric acid urolithiasis. {ECO:0000269|PubMed:7593598, ECO:0000269|Ref.12}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Karaciğer ve Kasdaki Fosforilaz Kinaz Eksikliği, Otozomal Resesif, 261750, Otozomal Resesif, GSD9B (Glikojen Depo Hastalığı, Karaciğer ve Kas Fosforilaz Kinaz Eksikliği) (PHKB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750, Autosomal recessive; GSD9B (Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency) (PHKB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHKB	PHKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 9B (GSD9B) [MIM:261750]: A metabolic disorder characterized by hepatomegaly, only slightly elevated transaminases and plasma lipids, clinical improvement with increasing age, and remarkably no clinical muscle involvement. Biochemical observations suggest that this mild phenotype is caused by an incomplete holoenzyme that lacks the beta suburit, but that may possess residual activity. {ECO:0000269	PubMed:9402963}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Karaciğer ve Kasdaki Fosforilaz Kinaz Eksikliği, Otozomal Resesif, 261750, Otozomal Resesif, GSD9B (Glikojen Depo Hastalığı, Karaciğer ve Kas Fosforilaz Kinaz Eksikliği) (Prenatal) (PHKB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750, Autosomal recessive; GSD9B (Glycogen storage disease due to liver and muscle phosphorylase kinase deficiency) (PHKB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHKB	PHKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Glycogen storage disease 9B (GSD9B) [MIM:261750]: A metabolic disorder characterized by hepatomegaly, only slightly elevated transaminases and plasma lipids, clinical improvement with increasing age, and remarkably no clinical muscle involvement. Biochemical observations suggest that this mild phenotype is caused by an incomplete holoenzyme that lacks the beta suburit, but that may possess residual activity. {ECO:0000269	PubMed:9402963}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfoserin Aminotransferaz Eksikliği, 610992, Otozomal Resesif; PSATD (Fosfoserin Aminotransferaz Eksikliği) (PSAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoserine aminotransferase deficiency, 610992, Autosomal recessive; PSATD (Phosphoserine aminotransferase deficiency) (PSAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSAT1	PSAT1, PSAT, EPIP, PSATD, NLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neu-Laxova syndrome 2 (NLS2) [MIM:616038]: A form of Neu- Laxova syndrome, a lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies. These include lissencephaly, cerebellar hypoplasia and/or abnormal/agenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269	PubMed:25152457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfoserin Aminotransferaz Eksikliği, 610992, Otozomal Resesif; PSATD (Fosfoserin Aminotransferaz Eksikliği) (Prenatal) (PSAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoserine aminotransferase deficiency, 610992, Autosomal recessive; PSATD (Phosphoserine aminotransferase deficiency) (PSAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSAT1	PSAT1, PSAT, EPIP, PSATD, NLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Neu-Laxova syndrome 2 (NLS2) [MIM:616038]: A form of Neu- Laxova syndrome, a lethal, autosomal recessive multiple malformation syndrome characterized by ichthyosis, marked intrauterine growth restriction, microcephaly, short neck, limb deformities, hypoplastic lungs, edema, and central nervous system anomalies. These include lissencephaly, cerebellar hypoplasia and/or abnormal/agenesis of the corpus callosum. Abnormal facial features include severe proptosis with ectropion, hypertelorism, micrognathia, flattened nose, and malformed ears. {ECO:0000269	PubMed:25152457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fosfoserin Fosfataz Eksikliği, 614023, Otozomal Resesif; PSPHD (3-Fosfoserin Fosfataz Eksikliği) (PSPH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Phosphoserine phosphatase deficiency, 614023, Autosomal recessive; PSPHD (3-phosphoserine phosphatase deficiency) (PSPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSPH	PSPH, PSP, PSPHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Phosphoserine phosphatase deficiency (PSPHD) [MIM:614023]: A disorder that results in pre- and postnatal growth retardation, moderate psychomotor retardation and facial features suggestive of Williams syndrome. {ECO:0000269	PubMed:14673469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Fosfoserin Fosfataz Eksikliği, 614023, Otozomal Resesif; PSPHD (3-Fosfoserin Fosfataz Eksikliği) (Prenatal) (PSPH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Phosphoserine phosphatase deficiency, 614023, Autosomal recessive; PSPHD (3-phosphoserine phosphatase deficiency) (PSPH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSPH	PSPH, PSP, PSPHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Phosphoserine phosphatase deficiency (PSPHD) [MIM:614023]: A disorder that results in pre- and postnatal growth retardation, moderate psychomotor retardation and facial features suggestive of Williams syndrome. {ECO:0000269	PubMed:14673469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pick Hastalığı, 172700, Otozomal Dominant, Sporadik Vakalar (Frontotemporal Demans) (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pick disease, 172700, Autosomal dominant, Isolated cases (Frontotemporal dementia) (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pick Hastalığı, 172700, Otozomal Dominant, Sporadik Vakalar (Frontotemporal Demans) (MLPA)	Pick disease, 172700, Autosomal dominant, Isolated cases (Frontotemporal dementia) (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. {ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pick Hastalığı, 172700, Otozomal Dominant, Sporadik Vakalar; FTD (Frontotemporal Demans) (PSEN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pick disease, 172700, Autosomal dominant, Isolated cases; FTD (Frontotemporal dementia) (PSEN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSEN1	PSEN1, AD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Frontotemporal dementia (FTD) [MIM:600274]: A form of dementia characterized by pathologic finding of frontotemporal lobar degeneration, presenile dementia with behavioral changes, deterioration of cognitive capacities and loss of memory. In some cases, parkinsonian symptoms are prominent. Neuropathological changes include frontotemporal atrophy often associated with atrophy of the basal ganglia, substantia nigra, amygdala. In most cases, protein tau deposits are found in glial cells and/or neurons. {ECO:0000269	PubMed:11094121}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Piebaldizm, 172800, Otozomal Dominant (Piebaldizm) (SNAI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Piebaldism, 172800, Autosomal dominant (Piebaldism) (SNAI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNAI2	SNAI2, SLUG, WS2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Piebald trait (PBT) [MIM:172800]: Autosomal dominant genetic developmental abnormality of pigmentation characterized by congenital patches of white skin and hair that lack melanocytes. {ECO:0000269	PubMed:12955764}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piebaldizm, 172800, Otozomal Dominant; PBT (Piebaldizm) (KIT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Piebaldism, 172800, Autosomal dominant; PBT (Piebaldism) (KIT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIT	KIT, PBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Piebald trait (PBT) [MIM:172800]: Autosomal dominant genetic developmental abnormality of pigmentation characterized by congenital patches of white skin and hair that lack melanocytes. {ECO:0000269|PubMed:11074500, ECO:0000269|PubMed:1370874, ECO:0000269|PubMed:1376329, ECO:0000269|PubMed:1717985, ECO:0000269|PubMed:7687262, ECO:0000269|PubMed:8680409, ECO:0000269|PubMed:9450866, ECO:0000269|PubMed:9699740}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pierpont Sendromu, 602342, Otozomal Dominant (TBL1XR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pierpont syndrome, 602342, Autosomal dominant (TBL1XR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBL1XR1	TBL1XR1, TBLR1, IRA1, C21, MRD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pierpont Sendromu, 602342, Otozomal Dominant (Prenatal) (TBL1XR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pierpont syndrome, 602342, Autosomal dominant (TBL1XR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBL1XR1	TBL1XR1, TBLR1, IRA1, C21, MRD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pierson Sendromu, 609049, Otozomal Resesif (Pierson Sendromu) (LAMB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pierson syndrome, 609049, Autosomal recessive (Pierson syndrome) (LAMB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMB2	LAMB2, LAMS, NPHS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pierson syndrome (PIERS5) [MIM:609049]: Characterized by nephrotic syndrome with neonatal onset, diffuse mesangial sclerosis and eye abnormalities with microcoria as the leading clinical feature. Death usually occurs within the first weeks of life. Disease severity depends on the mutation type: nontruncating LAMB2 mutations may display variable phenotypes ranging from a milder variant of Pierson syndrome to isolated congenital nephrotic syndrome. {ECO:0000269|PubMed:15367484, ECO:0000269|PubMed:16912710}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pierson Sendromu, 609049, Otozomal Resesif (Pierson Sendromu) (Prenatal) (LAMB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pierson syndrome, 609049, Autosomal recessive (Pierson syndrome) (LAMB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMB2	LAMB2, LAMS, NPHS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pierson syndrome (PIERS5) [MIM:609049]: Characterized by nephrotic syndrome with neonatal onset, diffuse mesangial sclerosis and eye abnormalities with microcoria as the leading clinical feature. Death usually occurs within the first weeks of life. Disease severity depends on the mutation type: nontruncating LAMB2 mutations may display variable phenotypes ranging from a milder variant of Pierson syndrome to isolated congenital nephrotic syndrome. {ECO:0000269|PubMed:15367484, ECO:0000269|PubMed:16912710}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pigmenter Bozukluk, Retiküler, Sistemik Bulgular, X'e Bağlı, 301220, X'e Bağlı Resesif; PDR (X'e Bağlı Retiküler Pigment Bozukluğu) (POLA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pigmentary disorder, reticulate, with systemic manifestations, X-linked, 301220, X-linked recessive; PDR (X-linked reticulate pigmentary disorder) (POLA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLA1	POLA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Pigmentli Nodüler Adrenokortikal Hastalık, Primer, 1, 610489. Otozomal Dominant; PPNAD1 (Primer Pigmentli Nodüler Adrenokortikal Hastalık) (PRKAR1A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Pigmented nodular adrenocortical disease, primary, 1, 610489, Autosomal dominant; PPNAD1 (Primary pigmented nodular adrenocortical disease) (PRKAR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAR1A	PRKAR1A, TSE1, CNC1, CAR, PPNAD1, ACRDYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrodysostosis 1, with or without hormone resistance (ACRDYS1) [MIM:101800]: A form of skeletal dysplasia characterized by short stature, severe brachydactyly, facial dysostosis, and nasal hypoplasia. Affected individuals often have advanced bone age and obesity. Laboratory studies show resistance to multiple hormones, including parathyroid, thyrotropin, calcitonin, growth hormone-releasing hormone, and gonadotropin. However, not all patients show endocrine abnormalities. {ECO:0000269|PubMed:21651393, ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:22723333, ECO:0000269|PubMed:23043190, ECO:0000269|PubMed:23425300}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Pigmentli Nodüler Adrenokortikal Hastalık, Primer, 2, 610475. Otozomal Dominant; PPNAD2 (Primer Pigmentli Nodüler Adrenokortikal Hastalık) (PDE11A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Pigmented nodular adrenocortical disease, primary, 2, 610475, Autosomal dominant; PPNAD2 (Primary pigmented nodular adrenocortical disease) (PDE11A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE11A	PDE11A, PDE11A1, PDE11A2, PDE11A3, PPNAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary pigmented nodular adrenocortical disease 2 (PPNAD2) [MIM:610475]: A rare bilateral adrenal defect causing ACTH-independent Cushing syndrome. Macroscopic appearance of the adrenals is characteristic with small pigmented micronodules observed in the cortex. Adrenal glands show overall normal size and weight, and multiple small yellow-to-dark brown nodules surrounded by a cortex with a uniform appearance. Microscopically, there are moderate diffuse cortical hyperplasia with mostly nonpigmented nodules, multiple capsular deficits and massive circumscribed and infiltrating extra-adrenal cortical excrescences with micronodules. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. {ECO:0000269	PubMed:16767104}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pigmentli Nodüler Adrenokortikal Hastalık, Primer, 3, 614190; PPNAD3 (Primer Pigmentli Nodüler Adrenokortikal Hastalık) (PDE8B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pigmented nodular adrenocortical disease, primary, 3, 614190; PPNAD3 (Primary pigmented nodular adrenocortical disease) (PDE8B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE8B	PDE8B, PPNAD3, ADSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Striatal degeneration autosomal dominant (ADSD) [MIM:609161]: A movement disorder affecting the striatal part of the basal ganglia and characterized by bradykinesia, dysarthria and muscle rigidity. These symptoms resemble idiopathic Parkinson disease, but tremor is not present. {ECO:0000269	PubMed:20085714}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pigmentli Paravenöz Korioretinal Atrofi, 172870, Otozomal Dominant; PPCRA (Pigmentli Paravenöz Retinokoroid Atrofi) (CRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pigmented paravenous chorioretinal atrophy, 172870, Autosomal dominant; PPCRA (Pigmented paravenous retinochoroidal atrophy) (CRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRB1	CRB1, RP12, LCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269	PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Pigmentli Paravenöz Korioretinal Atrofi, 172870, Otozomal Dominant; PPCRA (Pigmentli Paravenöz Retinokoroid Atrofi) (MLPA)	Pigmented paravenous chorioretinal atrophy, 172870, Autosomal dominant; PPCRA (Pigmented paravenous retinochoroidal atrophy) (MLPA)	CRB1	CRB1, RP12, LCA8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269	PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Pilomatrikom, Somatik, 132600 (Pilomatrikoma) (CTNNB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pilomatricoma, somatic, 132600 (Pilomatrixoma) (CTNNB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTNNB1	CTNNB1, MRD19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Pineal Hiperplazi, İnsülin Dirençli Diyabetes Mellitus ve Somatik Anomaliler (INSR geni) (Tüm Gen Dizi Analizi) (Postnatal)	PINEAL HYPERPLASIA, INSULIN-RESISTANT DIABETES MELLITUS, AND SOMATIC ABNORMALITIES (INSR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	INSR	INSR, HHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Pitt-Hopkins Benzeri Sendrom 1, 610042 (Pitt-Hopkins Benzeri Sendrom) (CNTNAP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Pitt-Hopkins like syndrome 1, 610042 (Pitt-Hopkins-like syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Autism 15 (AUTS15) [MIM:612100]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pitt-Hopkins Benzeri Sendrom 1, 610042 (Pitt-Hopkins Benzeri Sendrom) (Prenatal) (CNTNAP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Pitt-Hopkins like syndrome 1, 610042 (Pitt-Hopkins-like syndrome) (CNTNAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CNTNAP2	CNTNAP2, CASPR2, NRXN4, CDFE, AUTS15, PTHSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Autism 15 (AUTS15) [MIM:612100]: A complex multifactorial, pervasive developmental disorder characterized by impairments in reciprocal social interaction and communication, restricted and stereotyped patterns of interests and activities, and the presence of developmental abnormalities by 3 years of age. Most individuals with autism also manifest moderate mental retardation. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pitt-Hopkins Sendromu, 610954, Otozomal Dominant; PTHS (Pitt-Hopkins Sendromu) (TCF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pitt-Hopkins syndrome, 610954, Autosomal dominant; PTHS (Pitt-Hopkins syndrome) (TCF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCF4	TCF4, SEF2, ITF2, PTHS, FECD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pitt-Hopkins syndrome (PTHS) [MIM:610954]: A syndrome characterized by mental retardation, wide mouth and distinctive facial features, and intermittent hyperventilation followed by apnea. Features include intellectual disability with severe speech impairment, normal growth parameters at birth, postnatal microcephaly, breathing anomalies, severe motor developmental delay, motor incoordination, ocular anomalies, constipation, seizures, typical behavior and subtle brain abnormalities. {ECO:0000269|PubMed:17436254, ECO:0000269|PubMed:17436255, ECO:0000269|PubMed:18728071, ECO:0000269|PubMed:19235238, ECO:0000269|PubMed:20194619, ECO:0000269|PubMed:22045651}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pitt-Hopkins Sendromu, 610954, Otozomal Dominant; PTHS (Pitt-Hopkins Sendromu) (Prenatal) (TCF4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pitt-Hopkins syndrome, 610954, Autosomal dominant; PTHS (Pitt-Hopkins syndrome) (TCF4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCF4	TCF4, SEF2, ITF2, PTHS, FECD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pitt-Hopkins syndrome (PTHS) [MIM:610954]: A syndrome characterized by mental retardation, wide mouth and distinctive facial features, and intermittent hyperventilation followed by apnea. Features include intellectual disability with severe speech impairment, normal growth parameters at birth, postnatal microcephaly, breathing anomalies, severe motor developmental delay, motor incoordination, ocular anomalies, constipation, seizures, typical behavior and subtle brain abnormalities. {ECO:0000269|PubMed:17436254, ECO:0000269|PubMed:17436255, ECO:0000269|PubMed:18728071, ECO:0000269|PubMed:19235238, ECO:0000269|PubMed:20194619, ECO:0000269|PubMed:22045651}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pitt-Hopkins Benzeri Sendrom 2, 614325, Otozomal Resesif; PTHSL2 (Pitt-Hopkins Benzeri Sendrom) (NRXN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pitt-Hopkins-like syndrome 2, 614325, Autosomal recessive; PTHSL2 (Pitt-Hopkins-like syndrome) (NRXN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRXN1	NRXN1, PTHSL2, SCZD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pitt-Hopkins Benzeri Sendrom 2, 614325, Otozomal Resesif; PTHSL2 (Pitt-Hopkins Benzeri Sendrom) (Prenatal) (NRXN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pitt-Hopkins-like syndrome 2, 614325, Autosomal recessive; PTHSL2 (Pitt-Hopkins-like syndrome) (NRXN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NRXN1	NRXN1, PTHSL2, SCZD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hipofizer ACTH Salgılayan Adenom (GNAI2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary ACTH-secreting adenoma (GNAI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAI2	GNAI2, GNAI2B, GIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipofiz Adenomları, ACTH Salgılayan, 219090, Otozomal Resesif (Cushing Hastalığı) (AIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary adenoma, ACTH-secreting, 219090, Autosomal recessive (Cushing disease) (AIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIP	AIP, XAP2, ARA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: ACTH-secreting pituitary adenoma (ASPA) [MIM:219090]: A pituitary adenoma resulting in excessive production of adrenocorticotropic hormone. This leads to hypersecretion of cortisol by the adrenal glands and ACTH-dependent Cushing syndrome. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. {ECO:0000269	PubMed:17360484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Adenomları, ACTH Salgılayan, 219090, Otozomal Resesif (Cushing Hastalığı) (MLPA)	Pituitary adenoma, ACTH-secreting, 219090, Autosomal recessive (Cushing disease) (MLPA)	AIP	AIP, XAP2, ARA9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: ACTH-secreting pituitary adenoma (ASPA) [MIM:219090]: A pituitary adenoma resulting in excessive production of adrenocorticotropic hormone. This leads to hypersecretion of cortisol by the adrenal glands and ACTH-dependent Cushing syndrome. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. {ECO:0000269	PubMed:17360484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Adenomları, Büyüme Hormonu Salgılayan 2, 300943; PAGH1 (Akromegali) (GPR101 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary adenoma, growth hormone-secreting 2, 300943; PAGH1 (Acromegaly) (GPR101 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR101	GPR101, PAGH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pituitary adenoma, growth hormone-secreting, 2 (PAGH2) [MIM:300943]: A growth hormone-secreting, benign neoplasm of the anterior pituitary gland, also known as somatotropinoma. It clinically results in acromegaly, a condition characterized by coarse facial features, protruding jaw, and enlarged extremities. Excessive production of growth hormone in children or adolescents before the closure of epiphyses causes gigantism, a condition characterized by abnormally tall stature. {ECO:0000269	PubMed:25470569}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Hipofiz Adenomları, Büyüme Hormonu Salgılayan, 102200, Otozomal Dominant, Somatik Mutasyon (Akromegali) (AIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary adenoma, growth hormone-secreting, 102200, Autosomal dominant, Somatic mutation (Acromegaly) (AIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIP	AIP, XAP2, ARA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal		Biopsy of affected site-transport media
Hipofiz Adenomu, Büyüme Hormonu Salgılayan, 2; Pagh2 (Akromegali) (GPR101 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PITUITARY ADENOMA, GROWTH HORMONE-SECRETING, 2; PAGH2 (Acromegaly) (GPR101 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPR101	GPR101, PAGH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pituitary adenoma, growth hormone-secreting, 2 (PAGH2) [MIM:300943]: A growth hormone-secreting, benign neoplasm of the anterior pituitary gland, also known as somatotropinoma. It clinically results in acromegaly, a condition characterized by coarse facial features, protruding jaw, and enlarged extremities. Excessive production of growth hormone in children or adolescents before the closure of epiphyses causes gigantism, a condition characterized by abnormally tall stature. {ECO:0000269	PubMed:25470569}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Hipofiz Adenomları, Prolaktin Salgılayan, 600634 (Ailesel İzole Hipofiz Adenomları) (AIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary adenoma, prolactin-secreting, 600634 (Familial isolated pituitary adenoma) (AIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIP	AIP, XAP2, ARA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: ACTH-secreting pituitary adenoma (ASPA) [MIM:219090]: A pituitary adenoma resulting in excessive production of adrenocorticotropic hormone. This leads to hypersecretion of cortisol by the adrenal glands and ACTH-dependent Cushing syndrome. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. {ECO:0000269	PubMed:17360484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Adenomları, Prolaktin Salgılayan, 600634 (Ailesel İzole Hipofiz Adenomları) (MLPA)	Pituitary adenoma, prolactin-secreting, 600634 (Familial isolated pituitary adenoma) (MLPA)	AIP	AIP, XAP2, ARA9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: ACTH-secreting pituitary adenoma (ASPA) [MIM:219090]: A pituitary adenoma resulting in excessive production of adrenocorticotropic hormone. This leads to hypersecretion of cortisol by the adrenal glands and ACTH-dependent Cushing syndrome. Clinical manifestations of Cushing syndrome include facial and truncal obesity, abdominal striae, muscular weakness, osteoporosis, arterial hypertension, diabetes. {ECO:0000269	PubMed:17360484}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 1, 613038, Otozomal Resesif, Otozomal Dominant; CPHD1 (Kombine Hipofiz Hormon Eksiklikleri, Kalıtsal Formları) (POU1F1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 1, 613038, Autosomal recessive, Autosomal dominant; CPHD1 (Combined pituitary hormone deficiencies, genetic forms) (POU1F1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POU1F1	POU1F1, PIT1, CPHD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 1 (CPHD1) [MIM:613038]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD1 is characterized by pleiotropic deficiencies of growth hormone, prolactin and thyroid- stimulating hormone, while the production of adrenocorticotropic hormone, luteinizing hormone, and follicle-stimulating hormone are preserved. In infancy severe growth deficiency from birth as well as distinctive facial features with prominent forehead, marked midfacial hypoplasia with depressed nasal bridge, deep-set eyes, and a short nose with anteverted nostrils and hypoplastic pituitary gland by MRI examination can be seen. Some cases present with severe mental retardation along with short stature. {ECO:0000269|PubMed:11297581, ECO:0000269|PubMed:1472057, ECO:0000269|PubMed:1509262, ECO:0000269|PubMed:1509263, ECO:0000269|PubMed:15928241, ECO:0000269|PubMed:16968807, ECO:0000269|PubMed:7852536, ECO:0000269|PubMed:8768831, ECO:0000269|PubMed:9485179, ECO:0000269|PubMed:9626142}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 1, 613038, Otozomal Resesif, Otozomal Dominant; CPHD1 (Kombine Hipofiz Hormon Eksiklikleri, Kalıtsal Formları) (MLPA)	Pituitary hormone deficiency, combined, 1, 613038, Autosomal recessive, Autosomal dominant; CPHD1 (Combined pituitary hormone deficiencies, genetic forms) (MLPA)	POU1F1	POU1F1, PIT1, CPHD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 1 (CPHD1) [MIM:613038]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD1 is characterized by pleiotropic deficiencies of growth hormone, prolactin and thyroid- stimulating hormone, while the production of adrenocorticotropic hormone, luteinizing hormone, and follicle-stimulating hormone are preserved. In infancy severe growth deficiency from birth as well as distinctive facial features with prominent forehead, marked midfacial hypoplasia with depressed nasal bridge, deep-set eyes, and a short nose with anteverted nostrils and hypoplastic pituitary gland by MRI examination can be seen. Some cases present with severe mental retardation along with short stature. {ECO:0000269|PubMed:11297581, ECO:0000269|PubMed:1472057, ECO:0000269|PubMed:1509262, ECO:0000269|PubMed:1509263, ECO:0000269|PubMed:15928241, ECO:0000269|PubMed:16968807, ECO:0000269|PubMed:7852536, ECO:0000269|PubMed:8768831, ECO:0000269|PubMed:9485179, ECO:0000269|PubMed:9626142}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 2, 262600, Otozomal Resesif; CPHD2 (Panhipopituitarizm, X'e Bağlı) (PROP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 2, 262600, Autosomal recessive; CPHD2 (Panhypopituitarism) (PROP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROP1	PROP1, CPHD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 2, 262600, Otozomal Resesif, CPHD2 (Panhipopituitarizm, X'e Bağlı) (MLPA)	Pituitary hormone deficiency, combined, 2, 262600, Autosomal recessive; CPHD2 (Panhypopituitarism) (MLPA)	PROP1	PROP1, CPHD2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 3, 221750, Otozomal Resesif; CPHD3 (Edinilmiş Kombine Hipofizer Hormon Eksikliği-Vertebra Anormallikleri) (LHX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 3, 221750, Autosomal recessive; CPHD3 (Non-acquired combined pituitary hormone deficiency with spine abnormalities) (LHX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHX3	LHX3, CPHD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 3 (CPHD3) [MIM:221750]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD3 is characterized by a complete deficit in all but one (adrenocorticotropin) anterior pituitary hormone and a rigid cervical spine leading to limited head rotation. {ECO:0000269|PubMed:10835633, ECO:0000269|PubMed:17327381}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine 3, 221750, Otozomal Resesif; CPHD3 (Edinilmiş Kombine Hipofizer Hormon Eksikliği-Vertebra Anormallikleri) (MLPA)	Pituitary hormone deficiency, combined, 3, 221750, Autosomal recessive; CPHD3 (Non-acquired combined pituitary hormone deficiency with spine abnormalities) (MLPA)	LHX3	LHX3, CPHD3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 3 (CPHD3) [MIM:221750]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD3 is characterized by a complete deficit in all but one (adrenocorticotropin) anterior pituitary hormone and a rigid cervical spine leading to limited head rotation. {ECO:0000269|PubMed:10835633, ECO:0000269|PubMed:17327381}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 4, 262700, Otozomal Dominant; CPHD4 (Kısa Boy-Hipofiz ve Beyin Bozuklukları-Küçük Sella Trusica Sendromu) (LHX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 4, 262700, Autosomal dominant; CPHD4 (Short stature-pituitary and cerebellar defects-small sella turcica syndrome) (LHX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHX4	LHX4, CPHD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 4 (CPHD4) [MIM:262700]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD4 is characterized by complete or partial deficiencies of growth hormone, thyroid- stimulating hormone, luteinizing hormone, follicle stimulating hormone and adrenocorticotropic hormone. Clinical features include short stature, cerebellar defects, and small sella turcica. {ECO:0000269|PubMed:17527005, ECO:0000269|PubMed:18073311}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 4, 262700, Otozomal Dominant; CPHD4 (Kısa Boy-Hipofiz ve Beyin Bozuklukları-Küçük Sella Trusica Sendromu) (MLPA)	Pituitary hormone deficiency, combined, 4, 262700, Autosomal dominant; CPHD4 (Short stature-pituitary and cerebellar defects-small sella turcica syndrome) (MLPA)	LHX4	LHX4, CPHD4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pituitary hormone deficiency, combined, 4 (CPHD4) [MIM:262700]: Combined pituitary hormone deficiency is defined as the impaired production of growth hormone and one or more of the other five anterior pituitary hormones. CPHD4 is characterized by complete or partial deficiencies of growth hormone, thyroid- stimulating hormone, luteinizing hormone, follicle stimulating hormone and adrenocorticotropic hormone. Clinical features include short stature, cerebellar defects, and small sella turcica. {ECO:0000269|PubMed:17527005, ECO:0000269|PubMed:18073311}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 182230, Otozomal Resesif, Otozomal Dominant, 5 (Septo-Optik Displazi Spektrumu) (HESX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 5, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (HESX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HESX1	HESX1, RPX, CPHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. {ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 182230, Otozomal Resesif, Otozomal Dominant, 5 (Septo-Optik Displazi Spektrumu) (MLPA)	Pituitary hormone deficiency, combined, 5, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (MLPA)	HESX1	HESX1, RPX, CPHD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. {ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Hormonu Eksikliği, Kombine, 6, 613986, Otozomal Dominant; CPHD6 (Kombine Hipofizer Hormon Eksiklikleri, Kalıtsal Formları) (OTX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary hormone deficiency, combined, 6, 613986, Autosomal dominant; CPHD6 (Combined pituitary hormone deficiencies, genetic forms) (OTX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTX2	OTX2, MCOPS5, CPHD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 5 (MCOPS5) [MIM:610125]: Patients manifest unilateral or bilateral microphthalmia/clinical anophthalmia and variable additional features including pituitary dysfunction, coloboma, microcornea, cataract, retinal dystrophy, hypoplasia or agenesis of the optic nerve, agenesis of the corpus callosum, developmental delay, joint laxity, hypotonia, and seizures. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. {ECO:0000269|PubMed:15846561, ECO:0000269|PubMed:20396904, ECO:0000269|PubMed:22577225, ECO:0000269|PubMed:24167467}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hipofiz Tümörü, İnvaziv (PRKCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pituitary tumor, invasive (PRKCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCA	PRKCA, PKCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pitriazis Rubra Pilaris, 173200, Otozomal Dominant; PRP (Pitriasis Rubra Pilaris) (CARD14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pityriasis rubra pilaris, 173200, Autosomal dominant; PRP (Pityriasis rubra pilaris) (CARD14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARD14	CARD14, CARMA2, BIMP2, PSORS2, PSS1, PRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pityriasis rubra pilaris (PRP) [MIM:173200]: A rare, papulosquamous skin disease characterized by the appearance of keratotic follicular papules, well-demarcated salmon-colored erythematous plaques covered with fine powdery scales interspersed with distinct islands of uninvolved skin, and palmoplantar keratoderma. Most cases are sporadic. The rare familial cases show autosomal dominant inheritance with incomplete penetrance and variable expression. Familial PRP usually presents at birth or appears during the first years of life and runs a chronic course. It is characterized by prominent follicular hyperkeratosis, diffuse palmoplantar keratoderma, and erythema. {ECO:0000269	PubMed:22703878}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Plasenta Dekolmanı (NOS3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Placental abruption (NOS3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOS3	NOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Variation in NOS3 seem to be associated with susceptibility to coronary spasm. {ECO:0000269|PubMed:11740345, ECO:0000269|PubMed:9737729}.	EDTA Blood Sample
Plazma Fibronektin Eksikliği, 614101, Otozomal Dominant (FN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Plasma fibronectin deficiency, 614101, Autosomal dominant (FN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FN1	FN1, FN, LETS, FNZ, GFND2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glomerulopathy with fibronectin deposits 2 (GFND2) [MIM:601894]: Genetically heterogeneous autosomal dominant disorder characterized clinically by proteinuria, microscopic hematuria, and hypertension that leads to end-stage renal failure in the second to fifth decade of life. {ECO:0000269|PubMed:18268355}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Plazma Trigliserit Düzeyi QTL, Düşük, 615881, Otozomal Dominant; TGQTL (ANGPTL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Plasma triglyceride level QTL, low, 615881, Autosomal dominant; TGQTL (ANGPTL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANGPTL4	ANGPTL4, PGAR, HFARP, FIAF, TGQTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Plazminojen Aktivatör İnhibitörü 1 Eksikliği, 613329, Otozomal Resesif, Otozomal Dominant (Konjenital Plazminojen Aktivatör İnhibitörü Tip 1 Eksikliği) (SERPINE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Plasminogen activator inhibitor-1 deficiency, 613329, Autosomal recessive, Autosomal dominant (Congenital plasminogen activator inhibitor type 1 deficiency) (SERPINE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINE1	PAI1, PLANH1, SERPINE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Plasminogen activator inhibitor-1 deficiency (PAI-1D) [MIM:613329]: A hematologic disorder characterized by increased bleeding after trauma, injury, or surgery. Affected females have menorrhagia. The bleeding defect is due to increased fibrinolysis of fibrin blood clots due to deficiency of plasminogen activator inhibitor-1, which inhibits tissue and urinary activators of plasminogen. {ECO:0000269|PubMed:9207454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Plazminojen Eksikliği, Tip 1, 217090, Otozomal Resesif (Hipoplazminojenemi) (PLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Plasminogen deficiency, type I, 217090, Autosomal recessive (Hypoplasminogenemia) (PLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLG	PLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Plasminogen deficiency (PLGD) [MIM:217090]: A disorder characterized by decreased serum plasminogen activity. Two forms of the disorder are distinguished: type I deficiency is additionally characterized by decreased plasminogen antigen levels and clinical symptoms, whereas type 2 deficiency, also known as dysplasminogenemia, is characterized by normal, or slightly reduced antigen levels, and absence of clinical manifestations. Plasminogen deficiency type 1 results in markedly impaired extracellular fibrinolysis and chronic mucosal pseudomembranous lesions due to subepithelial fibrin deposition and inflammation. The most common clinical manifestation of type 1 deficiency is ligneous conjunctivitis in which pseudomembranes formation on the palpebral surfaces of the eye progresses to white, yellow-white, or red thick masses with a wood-like consistency that replace the normal mucosa. {ECO:0000269|PubMed:10233898, ECO:0000269|PubMed:1427790, ECO:0000269|PubMed:1986355, ECO:0000269|PubMed:6216475, ECO:0000269|PubMed:6238949, ECO:0000269|PubMed:8392398, ECO:0000269|PubMed:9242524, ECO:0000269|PubMed:9858247}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombosit Anormallikler, Eozinofili ve İmmün Aracılı İnflamatuvar Hastalık (ARPC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platelet abnormalities with eosinophilia and immune-mediated inflammatory disease (ARPC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARPC2	ARPC2, ARC34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trombosit Bozukluğu, Ailesel, Miyeloid Malignitelyle Birlikte, Ailesel, 601399, Otozomal Dominant; FPDMM (Ailesel Trombosit Sendromu-Akut Myelositer Lösemi Eğilimi) (RUNX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platelet disorder, familial, with associated myeloid malignancy, 601399, Autosomal dominant; FPDMM (Familial platelet syndrome with predisposition to acute myelogenous leukemia) (RUNX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUNX1	RUNX1, CBFA2, AML1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial platelet disorder with associated myeloid malignancy (FPDMM) [MIM:601399]: Autosomal dominant disease characterized by qualitative and quantitative platelet defects, and propensity to develop acute myelogenous leukemia. {ECO:0000269|PubMed:10508512}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombosit Glikoprotein 4 Eksikliği, 608404, Otozomal Resesif (CD36 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platelet glycoprotein IV deficiency, 608404, Autosomal recessive (CD36 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD36	CD36, CHDS7, BDPLT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Platelet glycoprotein IV deficiency (PG4D) [MIM:608404]: A disorder characterized by macrothrombocytopenia without notable hemostatic problems and bleeding tendency. Platelet glycoprotein IV deficiency can be divided into 2 subgroups. The type I phenotype is characterized by platelets and monocytes/macrophages exhibiting complete CD36 deficiency. The type II phenotype lacks the surface expression of CD36 in platelets, but expression in monocytes/macrophages is near normal. {ECO:0000269|PubMed:11950861, ECO:0000269|PubMed:7533783}. Note=The disease is caused by mutations affecting the gene represented in this entry. Patients also have postprandial hypertriglyceridemia, insulin resistance and hypertension increasing atherosclerotic risk. {ECO:0000269|PubMed:18753675}.	EDTA Blood Sample
Trombosit PLC Beta-2 Eksikliği (PLCB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platelet PLC beta-2 deficiency (PLCB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCB2	PLCB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Trombosit Aktive Edici Faktör Asetilhidrolaz Eksikliği, 614278; PAFAD (PLA2G7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platelet-activating factor acetylhydrolase deficiency, 614278; PAFAD (PLA2G7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLA2G7	PLA2G7, PAFAH, PAFAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atopic hypersensitivity (ATOPY) [MIM:147050]: A condition characterized by predisposition to develop hypersensitivity reactions. Atopic individuals can develop eczema, allergic rhinitis and allergic asthma. {ECO:0000269|PubMed:10733466}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Platispondik İskelet Displazisi, Torrance Tipi, 151210, Otozomal Dominant; PLSDT (Platispondilik Displazi, Torrance Tip) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Platyspondylic skeletal dysplasia, Torrance type, 151210, autosomal dominant; PLSDT (Platyspondylic dysplasia, Torrance type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Platispondik İskelet Displazisi, Torrance Tipi, 151210, Otozomal Dominant; PLSDT (Platispondilik Displazi, Torrance Tip) (MLPA)	Platyspondylic skeletal dysplasia, Torrance type, 151210, autosomal dominant; PLSDT (Platyspondylic dysplasia, Torrance type) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Platispondik İskelet Displazisi, Torrance Tipi, 151210, Otozomal Dominant; PLSDT (Platispondilik Displazi, Torrance Tip) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Platyspondylic skeletal dysplasia, Torrance type, 151210, autosomal dominant; PLSDT (Platyspondylic dysplasia, Torrance type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Platispondik İskelet Displazisi, Torrance Tipi, 151210, Otozomal Dominant; PLSDT (Platispondilik Displazi, Torrance Tip) (Prenatal) (MLPA)	Platyspondylic skeletal dysplasia, Torrance type, 151210, autosomal dominant; PLSDT (Platyspondylic dysplasia, Torrance type) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Plevra Perfon Sıvılarından Direkt Yöntemle Kromozom Analizi ve FISH	Pleural-peritoneal fluid- Chromosome Analysis-Direct sampling and FISH	.	.	Kromozom analizi/ Karyotype analysis/ FISH	Heparinli enjektör içinde en az 5 cc		15	45	.	.	21	Postnatal	.	Heparinized sample - At least 5 ml
Plevropulmoner Blastom, 601200, Otozomal Dominant; PPB (Plevropulmoner Blastom-Ailesel Tümör Yatkınlık Sendromu) (DICER1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pleuropulmonary blastoma, 601200, Autosomal dominant; PPB (Pleuropulmonary blastoma family tumor susceptibility syndrome) (DICER1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DICER1	DICER1, HERNA, KIAA0928, MNG1, RMSE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Goiter multinodular 1, with or without Sertoli-Leydig cell tumors (MNG1) [MIM:138800]: A common disorder characterized by nodular overgrowth of the thyroid gland. Some individuals may also develop Sertoli-Leydig cell tumors, usually of the ovary. {ECO:0000269|PubMed:21205968}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pnömokok Hastalık, İnvaz#, Karşı Koruma, 610799 (Tüberküloz) (TIRAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pneumococcal disease, invasive, protection against, 610799 (Tuberculosis) (TIRAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIRAP	TIRAP, BACTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Prömotoraks, Primer Spontan, 173600, Otozomal Dominant (Ailesel Spontan Prömotoraks) (FLCN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Pneumothorax, primary spontaneous, 173600, Autosomal dominant (Familial spontaneous pneumothorax) (FLCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLCN	FLCN, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Birt-Hogg-Dube syndrome (BHD) [MIM:135150]: A rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of Birt- Hogg-Dube syndrome skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. Birt-Hogg-Dube syndrome is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in Birt-Hogg- Dube syndrome patients. (ECO:0000269|PubMed:12204536, ECO:0000269|PubMed:15852235, ECO:0000269|PubMed:18234728, ECO:0000269|PubMed:19785621). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prömotoraks, Primer Spontan, 173600, Otozomal Dominant (Ailesel Spontan Prömotoraks) (MLPA)	Pneumothorax, primary spontaneous, 173600, Autosomal dominant (Familial spontaneous pneumothorax) (MLPA)	FLCN	FLCN, BHD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Birt-Hogg-Dube syndrome (BHD) [MIM:135150]: A rare autosomal dominant genodermatosis characterized by hair follicle hamartomas (fibrofolliculomas), kidney tumors, and spontaneous pneumothorax. Fibrofolliculomas are part of the triad of Birt- Hogg-Dube syndrome skin lesions that also includes trichodiscomas and acrochordons. Onset of this dermatologic condition is invariably in adulthood. Birt-Hogg-Dube syndrome is associated with a variety of histologic types of renal tumors, including chromophobe renal cell carcinoma (RCC), benign renal oncocytoma, clear-cell RCC and papillary type I RCC. Multiple lipomas, angiolipomas, and parathyroid adenomas are also seen in Birt-Hogg- Dube syndrome patients. (ECO:0000269|PubMed:12204536, ECO:0000269|PubMed:15852235, ECO:0000269|PubMed:18234728, ECO:0000269|PubMed:19785621). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Poikiloderma, Nötropenik, 604173, Otozomal Resesif; PN (Poikiloderma-Nötropeni) (USB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Poikiloderma with neutropenia, 604173, Autosomal recessive; PN (Poikiloderma with neutropenia) (USB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USB1	C16orf57, PN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Poikiloderma with neutropenia (PN) [MIM:604173]: A genodermatosis characterized by poikiloderma, pachyonychia and chronic neutropenia. The disorder starts as a papular erythematous rash on the limbs during the first year of life. It gradually spreads centripetally and, as the papular rash resolves, hypo- and hyperpigmentation result, with development of telangiectasias. Another skin manifestation is pachyonychia, but alopecia and leukoplakia are distinctively absent. Patients have recurrent pneumonias that usually result in reactive airway disease and/or chronic cough. One of the most important extracutaneous symptoms is an increased susceptibility to infections, mainly affecting the respiratory system, primarily due to a chronic neutropenia and to neutrophil functional defects. Bone marrow abnormalities accuunt for neutropenia and may evolve into myelodysplasia associated with the risk of leukemic transformation. Poikiloderma with neutropenia shows phenotypic overlap with Rothmund-Thomson syndrome. (ECO:0000269|PubMed:20004881, ECO:0000269|PubMed:20503306). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Poikiloderma; Kalıtsal Fibrozlu, Tendon Kontraktürleri, Miyopati ve Pulmoner Fibroz, 615704, Otozomal Dominant; POIKTMP (Kalıtsal Sklerozan Poikiloderma-Tendon ve Akciğer Tutulumu) (FAM111B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704, Autosomal dominant; POIKTMP (Hereditary sclerosing poikiloderma with tendon and pulmonary involvement) (FAM111B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM111B	FAM111B, POIKTMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Poliarteritis Nodosa, Çocuklukta Başlayan, 615688, Otozomal Resesif; PAN (Vaskülit, ADA2 Eksikliğine Bağlı) (ADA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polyarteritis nodosa, childhood-onset, 615688, Autosomal recessive; PAN (Vasculitis due to ADA2 deficiency) (ADA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADA2	CECR1, PAN, SNEDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polikistik Böbrek Hastalığı 3, 600666, Otozomal Dominant (GANAB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic kidney disease 3, 600666, Autosomal dominant (GANAB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GANAB	GANAB, PKD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polikistik Böbrek Hastalığı 3, 600666, Otozomal Dominant (Prenatal) (GANAB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic kidney disease 3, 600666, Autosomal dominant (GANAB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GANAB	GANAB, PKD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Böbrek ve Karaciğer Hastalığı, 263200, Otozomal Resesif (Polikistik Böbrek Hastalığı, Otozomal Resesif) (PKHD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic kidney and hepatic disease, 263200, Autosomal recessive (Autosomal recessive polycystic kidney disease) (PKHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKHD1	FCYT, PKHD1, ARPKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polikistik Böbrek ve Karaciğer Hastalığı, 263200, Otozomal Resesif (Polikistik Böbrek Hastalığı, Otozomal Resesif) (MLPA)	Polycystic kidney and hepatic disease, 263200, Autosomal recessive (Autosomal recessive polycystic kidney disease) (MLPA)	PKHD1	FCYT, PKHD1, ARPKD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Polikistik Böbrek ve Karaciğer Hastalığı, 263200, Otozomal Resesif (Polikistik Böbrek Hastalığı, Otozomal Resesif) (PKHD1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic kidney and hepatic disease, 263200, Autosomal recessive (Autosomal recessive polycystic kidney disease) (PKHD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PKHD1	FCYT, PKHD1, ARPKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Böbrek ve Karaciğer Hastalığı, 263200, Otozomal Resesif (Polikistik Böbrek Hastalığı, Otozomal Resesif) (Prenatal) (MLPA)	Polycystic kidney and hepatic disease, 263200, Autosomal recessive (Autosomal recessive polycystic kidney disease) (Prenatal) (MLPA)	PKHD1	FCYT, PKHD1, ARPKD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Böbrek Hastalığı, Yetişkin Tip 1, 173900, Otozomal Dominant; PKD1 (PKD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic kidney disease, adult type I, 173900, Autosomal dominant; PKD1 (PKD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKD1	PKD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polycystic kidney disease 2 (PKD2) [MIM:613095]: A disorder characterized by progressive formation and enlargement of cysts in both kidneys, typically leading to end-stage renal disease in adult life. Cysts also occurs in the liver and other organs. It represents approximately 15% of the cases of autosomal dominant polycystic kidney disease. PKD2 is clinically milder than PKD1 but it has a deleterious impact on overall life expectancy. (ECO:0000269|PubMed:10411676, ECO:0000269|PubMed:10541293, ECO:0000269|PubMed:10835625, ECO:0000269|PubMed:11968093, ECO:0000269|PubMed:12707387, ECO:0000269|PubMed:14093477, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:21115670, ECO:0000269|PubMed:9326320). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polikistik Böbrek Hastalığı 2, 613095; PKD2 (PKD2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic kidney disease 2, 613095; PKD2 (PKD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKD2	PKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polycystic kidney disease 2 (PKD2) [MIM:613095]: A disorder characterized by progressive formation and enlargement of cysts in both kidneys, typically leading to end-stage renal disease in adult life. Cysts also occurs in the liver and other organs. It represents approximately 15% of the cases of autosomal dominant polycystic kidney disease. PKD2 is clinically milder than PKD1 but it has a deleterious impact on overall life expectancy. (ECO:0000269|PubMed:10411676, ECO:0000269|PubMed:10541293, ECO:0000269|PubMed:10835625, ECO:0000269|PubMed:11968093, ECO:0000269|PubMed:12707387, ECO:0000269|PubMed:14093477, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:21115670, ECO:0000269|PubMed:9326320). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polikistik Böbrek Hastalığı 2, 613095; PKD2 (MLPA)	Polycystic kidney disease 2, 613095; PKD2 (MLPA)	PKD2	PKD2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Polycystic kidney disease 2 (PKD2) [MIM:613095]: A disorder characterized by progressive formation and enlargement of cysts in both kidneys, typically leading to end-stage renal disease in adult life. Cysts also occurs in the liver and other organs. It represents approximately 15% of the cases of autosomal dominant polycystic kidney disease. PKD2 is clinically milder than PKD1 but it has a deleterious impact on overall life expectancy. (ECO:0000269|PubMed:10411676, ECO:0000269|PubMed:10541293, ECO:0000269|PubMed:10835625, ECO:0000269|PubMed:11968093, ECO:0000269|PubMed:12707387, ECO:0000269|PubMed:14093477, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:21115670, ECO:0000269|PubMed:9326320). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polikistik Böbrek Hastalığı 2, 613095; PKD2 (Prenatal) (PKD2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic kidney disease 2, 613095; PKD2 (PKD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PKD2	PKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polycystic kidney disease 2 (PKD2) [MIM:613095]: A disorder characterized by progressive formation and enlargement of cysts in both kidneys, typically leading to end-stage renal disease in adult life. Cysts also occurs in the liver and other organs. It represents approximately 15% of the cases of autosomal dominant polycystic kidney disease. PKD2 is clinically milder than PKD1 but it has a deleterious impact on overall life expectancy. (ECO:0000269|PubMed:10411676, ECO:0000269|PubMed:10541293, ECO:0000269|PubMed:10835625, ECO:0000269|PubMed:11968093, ECO:0000269|PubMed:12707387, ECO:0000269|PubMed:14093477, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:21115670, ECO:0000269|PubMed:9326320). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Polikistik Böbrek Hastalığı 2, 613095; PKD2 (Prenatal) (MLPA)	Polycystic kidney disease 2, 613095; PKD2 (Prenatal) (MLPA)	PKD2	PKD2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polycystic kidney disease 2 (PKD2) [MIM:613095]: A disorder characterized by progressive formation and enlargement of cysts in both kidneys, typically leading to end-stage renal disease in adult life. Cysts also occurs in the liver and other organs. It represents approximately 15% of the cases of autosomal dominant polycystic kidney disease. PKD2 is clinically milder than PKD1 but it has a deleterious impact on overall life expectancy. {ECO:0000269|PubMed:10411676, ECO:0000269|PubMed:10541293, ECO:0000269|PubMed:10835625, ECO:0000269|PubMed:11968093, ECO:0000269|PubMed:12707387, ECO:0000269|PubMed:14993477, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:21115670, ECO:0000269|PubMed:9326320}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Böbrek Hastalığı, Yetişkin Tip 1, 173900, Otozomal Dominant; PKD1 (MLPA)	Polycystic kidney disease, adult type I, 173900, Autosomal dominant; PKD1 (MLPA)	PKD1	PKD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Polycystic kidney disease 1 (PKD1) [MIM:173900]: A disorder characterized by renal cysts, liver cysts and intracranial aneurysm. Clinical variability is due to differences in the rate of loss of glomerular filtration, the age of reaching end-stage renal disease and the occurrence of hypertension, symptomatic extrarenal cysts, and subarachnoid hemorrhage from intracranial "berry" aneurysm. {ECO:0000269|PubMed:10200984, ECO:0000269|PubMed:10364515, ECO:0000269|PubMed:10577909, ECO:0000269|PubMed:10647901, ECO:0000269|PubMed:10729710, ECO:0000269|PubMed:10854095, ECO:0000269|PubMed:10923040, ECO:0000269|PubMed:10987650, ECO:0000269|PubMed:11012875, ECO:0000269|PubMed:11058904, ECO:0000269|PubMed:11115377, ECO:0000269|PubMed:11216660, ECO:0000269|PubMed:11316854, ECO:0000269|PubMed:11558899, ECO:0000269|PubMed:11571556, ECO:0000269|PubMed:11691639, ECO:0000269|PubMed:11773467, ECO:0000269|PubMed:11857740, ECO:0000269|PubMed:11967008, ECO:0000269|PubMed:12007219, ECO:0000269|PubMed:12070253, ECO:0000269|PubMed:12220456, ECO:0000269|PubMed:12842373, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:18837007, ECO:0000269|PubMed:21115670.	EDTA Blood Sample
Polikistik Böbrek Hastalığı, Yetişkin Tip 1, 173900, Otozomal Dominant; PKD1 (Prenatal) (MLPA)	Polycystic kidney disease, adult type I, 173900, Autosomal dominant; PKD1 (Prenatal) (MLPA)	PKD1	PKD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polycystic kidney disease 1 (PKD1) [MIM:173900]: A disorder characterized by renal cysts, liver cysts and intracranial aneurysm. Clinical variability is due to differences in the rate of loss of glomerular filtration, the age of reaching end-stage renal disease and the occurrence of hypertension, symptomatic extrarenal cysts, and subarachnoid hemorrhage from intracranial "berry" aneurysm. {ECO:0000269|PubMed:10200984, ECO:0000269|PubMed:10364515, ECO:0000269|PubMed:10577909, ECO:0000269|PubMed:10647901, ECO:0000269|PubMed:10729710, ECO:0000269|PubMed:10854095, ECO:0000269|PubMed:10923040, ECO:0000269|PubMed:10987650, ECO:0000269|PubMed:11012875, ECO:0000269|PubMed:11058904, ECO:0000269|PubMed:11115377, ECO:0000269|PubMed:11216660, ECO:0000269|PubMed:11316854, ECO:0000269|PubMed:11558899, ECO:0000269|PubMed:11571556, ECO:0000269|PubMed:11691639, ECO:0000269|PubMed:11773467, ECO:0000269|PubMed:11857740, ECO:0000269|PubMed:11967008, ECO:0000269|PubMed:12007219, ECO:0000269|PubMed:12070253, ECO:0000269|PubMed:12220456, ECO:0000269|PubMed:12842373, ECO:0000269|PubMed:15772804, ECO:0000269|PubMed:18837007, ECO:0000269|PubMed:21115670.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Böbrek Hastalığı, Otozomal Dominant (ADPKD) (PKD1, PKD2) (MLPA)	Polycystic Kidney Disease, Autosomal Dominant (ADPKD) (PKD1, PKD2) (MLPA)	PKD1, PKD2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Polikistik Böbrek Hastalığı, Otozomal Dominant (ADPKD) (PKD1, PKD2) (MLPA) (Prenatal)	Polycystic Kidney Disease, Autosomal Dominant (ADPKD) (PKD1, PKD2) (MLPA) (Prenatal)	PKD1, PKD2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polycystic Kidney Hastalığı, Otozomal Resesif; Arpkd (Polikistik Böbrek Hastalığı, Otozomal Resesif) (PKHD1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE; ARPKD (Autosomal recessive polycystic kidney disease) (PKHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKHD1	FCYT, PKHD1, ARPKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polycystic Kidney Hastalığı, Otozomal Resesif; Arpkd (Polikistik Böbrek Hastalığı, Otozomal Resesif) (Prenatal) (PKHD1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	POLYCYSTIC KIDNEY DISEASE, AUTOSOMAL RECESSIVE; ARPKD (Autosomal recessive polycystic kidney disease) (PKHD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PKHD1	FCYT, PKHD1, ARPKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Lipomembranöz Osteodisplazi-Sklerozan Lökoensefalopati (PLOSL), 221770, Otozomal Resesif (Nasu-Hakola Hastalığı) (TYROBP gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL), 221770, Autosomal recessive (Nasu-Hakola disease) (TYROBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYROBP	TYROBP, PLOSL, DAP12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL) [MIM:221770]: Recessively inherited disease characterized by a combination of psychotic symptoms rapidly progressing to presenile dementia and bone cysts restricted to wrists and ankles. PLOSL has a global distribution, although most of the patients have been diagnosed in Finland and Japan, with an estimated population prevalence of 2x10(-6) in the Finns. {ECO:0000269|PubMed:10888890, ECO:0000269|PubMed:12370476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polikistik Lipomembranöz Osteodisplazi-Sklerozan Lökoensefalopati (PLOSL), 221770, Otozomal Resesif (Nasu-Hakola Hastalığı) (Prenatal) (TYROBP gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL), 221770, Autosomal recessive (Nasu-Hakola disease) (TYROBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TYROBP	TYROBP, PLOSL, DAP12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (PLOSL) [MIM:221770]: Recessively inherited disease characterized by a combination of psychotic symptoms rapidly progressing to presenile dementia and bone cysts restricted to wrists and ankles. PLOSL has a global distribution, although most of the patients have been diagnosed in Finland and Japan, with an estimated population prevalence of 2x10(-6) in the Finns. {ECO:0000269|PubMed:10888890, ECO:0000269|PubMed:12370476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Lipomembranöz Osteodisplazi-Sklerozan Lökoensefalopati, PLOSL, 221770, Otozomal Resesif (Nasu-Hakola Hastalığı) (TREM2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY; PLOSL, 221770, Autosomal recessive (Nasu-Hakola disease) (TREM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREM2	TREM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Polikistik Karaciğer Hastalığı 1, 174050, Otozomal Dominant; PCLD1 (İzole Polikistik Karaciğer Hastalığı) (PRKCSH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic liver disease 1, 174050, Autosomal dominant; PCLD1 (Isolated polycystic liver disease) (PRKCSH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCSH	PRKCSH, G19P1, PCLD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polycystic liver disease (PCLD) [MIM:174050]: A hepatobiliary disease characterized by overgrowth of biliary epithelium and supportive connective tissue, resulting in multiple liver cysts. {ECO:0000269|PubMed:12529853, ECO:0000269|PubMed:12577059}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polikistik Karaciğer Hastalığı 1, 174050, Otozomal Dominant; PCLD1 (İzole Polikistik Karaciğer Hastalığı) (Prenatal) (PRKCSH gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic liver disease 1, 174050, Autosomal dominant; PCLD1 (Isolated polycystic liver disease) (PRKCSH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRKCSH	PRKCSH, G19P1, PCLD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Polycystic liver disease (PCLD) [MIM:174050]: A hepatobiliary disease characterized by overgrowth of biliary epithelium and supportive connective tissue, resulting in multiple liver cysts. {ECO:0000269|PubMed:12529853, ECO:0000269|PubMed:12577059}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polikistik Karaciğer Hastalığı 2, 617004, Otozomal Dominant; PCLD2 (İzole Polikistik Karaciğer Hastalığı) (SEC63 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polycystic liver disease 2, 617004, Autosomal dominant; PCLD2 (Isolated polycystic liver disease) (SEC63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEC63	SEC63, PCLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polikistik Karaciğer Hastalığı 2, 617004, Otozomal Dominant; PCLD2 (İzole Polikistik Karaciğer Hastalığı) (Prenatal) (SEC63 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Polycystic liver disease 2, 617004, Autosomal dominant; PCLD2 (Isolated polycystic liver disease) (SEC63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEC63	SEC63, PCLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polisitemia Vera, Somatik, 263300; PV (Polisitemi Vera) V617F Mutasyonu ve (Ekzon 12-14) (JAK2 gen) (Dizi Analizi) (Postnatal)	Polycythemia vera, somatic, 263300; PV (Polycythemia vera) V617F Mutation and (Exon 12-14) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	JAK2, THCYT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Polidaktili, Postaksiyel, Tip A6, 615226, Otozomal Resesif; PAPA6 (Postaksiyel Polidaktili Tip A) (ZNF141 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polydactyly, postaxial, type A6, 615226, Autosomal recessive; PAPA6 (Postaxial polydactyly type A) (ZNF141 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF141	ZNF141, D4S90, PAPA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polidaktili, Postaksiyel, Tip A1 ve B, 174200, Otozomal Dominant; PAPA1 (Postaksiyel Polidaktili Tip B) (GLI3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Polydactyly, postaxial, types A1 and B, 174200, Autosomal dominant; PAPA1 (Postaxial polydactyly type B) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polidaktili, Postaksiyel, Tip A1 ve B, 174200, Otozomal Dominant; PAPA1 (Postaksiyel Polidaktili Tip B) (MLPA)	Polydactyly, postaxial, types A1 and B, 174200, Autosomal dominant; PAPA1 (Postaxial polydactyly type B) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Polidaktili, Preaksiyel Tip 2, 174500, Otozomal Dominant; PPD2 (Trifalangeal Basparmak-Polisindaktili Sendromu) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polydactyly, preaxial type II, 174500, Autosomal dominant; PPD2 (Triphalangeal thumb-polysyndactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159}.;	EDTA Blood Sample
Polidaktili, Preaksiyel, Tip 4, 174700, Otozomal Dominant (Polipolisindaktili) (GLI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polydactyly, preaxial, type IV, 174700, Autosomal dominant (Polysyndactyly) (GLI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLI3	GLI3, PAPA, PAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polidaktili, Preaksiyel, Tip 4, 174700, Otozomal Dominant (Polipolisindaktili) (MLPA)	Polydactyly, preaxial, type IV, 174700, Autosomal dominant (Polysyndactyly) (MLPA)	GLI3	GLI3, PAPA, PAPB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pallister-Hall syndrome (PHS) [MIM:146510]: An autosomal dominant disorder characterized by a wide range of clinical manifestations. Clinical features include hypothalamic hamartoma, pituitary dysfunction, central or postaxial polydactyly, and syndactyly. Malformations are frequent in the viscera, e.g. anal atresia, bifid uvula, congenital heart malformations, pulmonary or renal dysplasia. {ECO:0000269	PubMed:10441570}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Poliendokrin-Polinöropati Sendromu, 616113, Otozomal Resesif; PEPNS (Poliendokrin-Polinöropati Sendromu) (DMXL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyendocrine-polyneuropathy syndrome, 616113, Autosomal recessive; PEPNS (Polyendocrine-polyneuropathy syndrome) (DMXL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DMXL2	DMXL2, RC3, KIAA0856, PEPNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyendocrine-polyneuropathy syndrome (PEPNS) [MIM:616113]: A progressive endocrine and neurodevelopmental disorder manifesting early in childhood with growth retardation and recurrent episodes of profound asymptomatic hypoglycemia. PEPNS is characterized by central hypothyroidism, hypogonadotropic hypogonadism, incomplete puberty, progressive non-autoimmune insulin-dependent diabetes mellitus, peripheral demyelinating sensorimotor polyneuropathy, and cerebellar and pyramidal signs. {ECO:0000269	PubMed:25248098}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Poliglikozan Body Hastalığı, Yetişkin Bir Şekilde, 263570, Otozomal Resesif; APBN (Glikojen Depo Hastalığı-Glikojen Dallandırıcı Enzim Eksikliğine Bağlı) (GBE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyglucosan body disease, adult form, 263570, Autosomal recessive; APBN (Glycogen storage disease due to glycogen branching enzyme deficiency) (GBE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBE1	GBE1, GSD4, APBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyglucosan body neuropathy, adult form (APBN) [MIM:263570]: A late-onset, slowly progressive disorder affecting the central and peripheral nervous systems. Patients typically present after age 40 years with a variable combination of cognitive impairment, pyramidal tetraparesis, peripheral neuropathy, and neurogenic bladder. Other manifestations include cerebellar dysfunction and extrapyramidal signs. The pathologic hallmark of APBN is the widespread accumulation of round, intracellular polyglucosan bodies throughout the nervous system, which are confined to neuronal and astrocytic processes. {ECO:0000269	PubMed:10762170}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Poliglikozan Body Hastalığı, Yetişkin Bir Şekilde, 263570, Otozomal Resesif; APBN (Glikojen Depo Hastalığı-Glikojen Dallandırıcı Enzim Eksikliğine Bağlı) (Prenatal) (GBE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polyglucosan body disease, adult form, 263570, Autosomal recessive; APBN (Glycogen storage disease due to glycogen branching enzyme deficiency) (GBE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GBE1	GBE1, GSD4, APBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Polyglucosan body neuropathy, adult form (APBN) [MIM:263570]: A late-onset, slowly progressive disorder affecting the central and peripheral nervous systems. Patients typically present after age 40 years with a variable combination of cognitive impairment, pyramidal tetraparesis, peripheral neuropathy, and neurogenic bladder. Other manifestations include cerebellar dysfunction and extrapyramidal signs. The pathologic hallmark of APBN is the widespread accumulation of round, intracellular polyglucosan bodies throughout the nervous system, which are confined to neuronal and astrocytic processes. {ECO:0000269	PubMed:10762170}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Poliglikozan Body Miyopatisi 1, ± İmmün Yetmezlik, 615895, Otozomal Resesif; PGBM1 (Otoinflamatuvar Sendromu-Piyojenik Bakteriyel Enfeksiyon ve Amilopektinozis) (RBCK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895, Autosomal recessive; PGBM1 (Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis) (RBCK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBCK1	RBCK1, HOIL1, PGBM1, PBMEI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyglucosan body myopathy 1 with or without immunodeficiency (PGBM1) [MIM:615895]: A disease characterized by polyglucosan storage myopathy associated with early-onset progressive muscle weakness and progressive dilated cardiomyopathy, which may necessitate cardiac transplant in severe cases. Some patients present with severe immunodeficiency, invasive bacterial infections and chronic autoinflammation. {ECO:0000269|PubMed:23104095, ECO:0000269|PubMed:23798481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Poliglikozan Body Miyopatisi 1, ± İmmün Yetmezlik, 615895, Otozomal Resesif; PGBM1 (Otoinflamatuvar Sendromu-Piyojenik Bakteriyel Enfeksiyon ve Amilopektinozis) (Prenatal) (RBCK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polyglucosan body myopathy 1 with or without immunodeficiency, 615895, Autosomal recessive; PGBM1 (Autoinflammatory syndrome with pyogenic bacterial infection and amylopectinosis) (RBCK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBCK1	RBCK1, HOIL1, PGBM1, PBMEI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Polyglucosan body myopathy 1 with or without immunodeficiency (PGBM1) [MIM:615895]: A disease characterized by polyglucosan storage myopathy associated with early-onset progressive muscle weakness and progressive dilated cardiomyopathy, which may necessitate cardiac transplant in severe cases. Some patients present with severe immunodeficiency, invasive bacterial infections and chronic autoinflammation. {ECO:0000269	PubMed:23104095, ECO:0000269	PubMed:23798481}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Poliglikozan Body Miyopatisi 2, 616199, Otozomal Resesif; PGBM2 (Poliglukozan Cisimcik Miyopati Tip 2) (GYG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyglucosan body myopathy 2, 616199, Autosomal recessive; PGBM2 (Polyglucosan body myopathy type 2) (GYG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GYG1	GYG1, GSD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Glycogen storage disease 15 (GSD15) [MIM:613507]: A metabolic disorder resulting in muscle weakness, associated with the glycogen depletion in skeletal muscle, and cardiac arrhythmia, associated with the accumulation of abnormal storage material in the heart. The skeletal muscle shows a marked predominance of slow-twitch, oxidative muscle fibers and mitochondrial proliferation. {ECO:0000269	PubMed:20357282}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Poliglikozan Body Miyopati 2, 616199, Otozomal Resesif; PGBM2 (Poliglukozan Cisimcik Miyopati Tip 2) (Prenatal) (GYG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polyglucosan body myopathy 2, 616199, Autosomal recessive; PGBM2 (Polyglucosan body myopathy type 2) (GYG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GYG1	GYG1, GSD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Glycogen storage disease 15 (GSD15) [MIM:613507]: A metabolic disorder resulting in muscle weakness, associated with the glycogen depletion in skeletal muscle, and cardiac arrhythmia, associated with the accumulation of abnormal storage material in the heart. The skeletal muscle shows a marked predominance of slow-twitch, oxidative muscle fibers and mitochondrial proliferation. {ECO:0000269	PubMed:20357282}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polihidramnyos, Megalensefali ve Semptomatik Epilepsi, 611087, Otozomal Resesif; PMSE (STRADA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyhydramnios, megalencephaly, and symptomatic epilepsy, 611087, Autosomal recessive; PMSE (STRADA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STRADA	STRADA, STRAD, LYKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A homozygous 7-kb deletion involving STRADA is a cause of a syndrome characterized by polyhydramnios, megalencephaly and symptomatic epilepsy.;	EDTA Blood Sample
Polimikrogiri, Optik Sinir Hipoplazisi 613180, Otozomal Resesif (Polimikrogiri-Optik Sinir Hipoplazisi) (TUBA8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria with optic nerve hypoplasia, 613180, Autosomal recessive (Polymicrogyria with optic nerve hypoplasia) (TUBA8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBA8	TUBA8, TUBAL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polymicrogyria, with optic nerve hypoplasia (PMGONH) [MIM:613180]: A disease characterized by extensive polymicrogyria, optic nerve hypoplasia, severe developmental delay, hypotonia, seizures, a dysplastic or absent corpus callosum and colpocephaly. Polymicrogyria is a malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269	PubMed:19896110}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polimikrogiri, Optik Sinir Hipoplazisi 613180, Otozomal Resesif (Polimikrogiri-Optik Sinir Hipoplazisi) (Prenatal) (TUBA8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polymicrogyria with optic nerve hypoplasia, 613180, Autosomal recessive (Polymicrogyria with optic nerve hypoplasia) (TUBA8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBA8	TUBA8, TUBAL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Polymicrogyria, with optic nerve hypoplasia (PMGONH) [MIM:613180]: A disease characterized by extensive polymicrogyria, optic nerve hypoplasia, severe developmental delay, hypotonia, seizures, a dysplastic or absent corpus callosum and colpocephaly. Polymicrogyria is a malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269	PubMed:19896110}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polimikrogiri, Bilateral Frontoparyetal, 606854, Otozomal Resesif; BFPP (Bilateral Polimikrogiri) (ADGRG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria, bilateral frontoparietal, 606854, Autosomal recessive; BFPP (Bilateral polymicrogyria) (ADGRG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRG1	ADGRG1, GPR56, TM7XN1, BFPP, BPPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polymicrogyria, bilateral frontoparietal (BFPP) [MIM:606854]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination, most severe in the frontoparietal regions. BFPP clinical manifestations include developmental and psychomotor delay, cerebellar and pyramidal signs, truncal ataxia, seizures, hyperreflexia. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269|PubMed:15044805, ECO:0000269|PubMed:16240336, ECO:0000269|PubMed:21723461}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polimikrogiri, Bilateral Frontoparyetal, 606854, Otozomal Resesif; BFPP (Bilateral Polimikrogiri) (Prenatal) (ADGRG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polymicrogyria, bilateral frontoparietal, 606854, Autosomal recessive; BFPP (Bilateral polymicrogyria) (ADGRG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADGRG1	ADGRG1, GPR56, TM7XN1, BFPP, BPPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Polymicrogyria, bilateral frontoparietal (BFPP) [MIM:606854]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination, most severe in the frontoparietal regions. BFPP clinical manifestations include developmental and psychomotor delay, cerebellar and pyramidal signs, truncal ataxia, seizures, hyperreflexia. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269	PubMed:15044805, ECO:0000269	PubMed:16240336, ECO:0000269	PubMed:21723461}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Polimikrogiri, Bilateral Perisilvian, 615752: BPPR (Bilateral Polimikrogiri) (ADGRG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria, bilateral perisylvian, 615752: BPPR (Bilateral polymicrogyria) (ADGRG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRG1	ADGRG1, GPR56, TM7XN1, BPPP, BPPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polymicrogyria, bilateral frontoparietal (BFPP) [MIM:606854]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination, most severe in the frontoparietal regions. BFPP clinical manifestations include developmental and psychomotor delay, cerebellar and pyramidal signs, truncal ataxia, seizures, hyperreflexia. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269|PubMed:15044805, ECO:0000269|PubMed:16240336, ECO:0000269|PubMed:21723461}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polimikrogiri, Bilateral Perisilvian, 615752: BPPR (Bilateral Polimikrogiri) (Prenatal) (ADGRG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polymicrogyria, bilateral perisylvian, 615752: BPPR (Bilateral polymicrogyria) (ADGRG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADGRG1	ADGRG1, GPR56, TM7XN1, BPPP, BPPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Polymicrogyria, bilateral frontoparietal (BFPP) [MIM:606854]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination, most severe in the frontoparietal regions. BFPP clinical manifestations include developmental and psychomotor delay, cerebellar and pyramidal signs, truncal ataxia, seizures, hyperreflexia. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269|PubMed:15044805, ECO:0000269|PubMed:16240336, ECO:0000269|PubMed:21723461}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polimikrogiri İkili Temporooksipital, 612691, Otozomal Resesif; BTOP (Bilateral Polimikrogiri) (FIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria, bilateral temporooccipital, 612691, Autosomal recessive; BTOP (Bilateral polymicrogyria) (FIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIG4	FIG4, KIAA0274, SAC3, ALS11, YVS, BTOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Yunis-Varon syndrome (YVS) [MIM:216340]: A severe autosomal recessive disorder characterized by skeletal defects, including cleidocranial dysplasia and digital anomalies, and severe neurologic involvement with neuronal loss. Enlarged cytoplasmic vacuoles are found in neurons, muscle, and cartilage. The disorder is usually lethal in infancy. {ECO:0000269|PubMed:23673387}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polimikrogiri, Perisilvyan, Serebellar Hipoplazi ve Atrofiyle, 616531, Otozomal Resesif; PMOYCHA (Bilateral Polimikrogiri) (PI4KA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531, Autosomal recessive; PMOYCHA (Bilateral polymicrogyria) (PI4KA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PI4KA	PI4KA, PIK4CA, PMGYCHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Polimikrogiri, Perisilvyan, Serebellar Hipoplazi ve Atrofiyle, 616531, Otozomal Resesif; PMOYCHA (Bilateral Polimikrogiri) (Prenatal) (PI4KA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polymicrogyria, perisylvian, with cerebellar hypoplasia and arthrogryposis, 616531, Autosomal recessive; PMOYCHA (Bilateral polymicrogyria) (PI4KA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PI4KA	PI4KA, PIK4CA, PMGYCHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polimikrogiri, Simetrik Veya Asimetrik, 610031, Otozomal Dominant (Bilateral Polimikrogiri) (TUBB2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polymicrogyria, symmetric or asymmetric, 610031, Autosomal dominant (Bilateral polymicrogyria) (TUBB2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB2B	TUBB2B, PMGYSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polymicrogyria, symmetric or asymmetric (PMGYSA) [MIM:610031]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269|PubMed:19456910}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polimikrogiri, Simetrik Veya Asimetrik, 610031, Otozomal Dominant (Bilateral Polimikrogiri) (Prenatal) (TUBB2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polymicrogyria, symmetric or asymmetric, 610031, Autosomal dominant (Bilateral polymicrogyria) (TUBB2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB2B	TUBB2B, PMGYSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Polymicrogyria, symmetric or asymmetric (PMGYSA) [MIM:610031]: A malformation of the cortex in which the brain surface is irregular and characterized by an excessive number of small gyri with abnormal lamination. Polymicrogyria is a heterogeneous disorder, considered to be the result of postmigratory abnormal cortical organization. {ECO:0000269|PubMed:19456910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polinöropati, İşitme Kaybı, Ataksi, Retinitis Pigmentoza ve Katarakt, 612674 İşitme, Otozomal Resesif; PHARC (Polinöropati-İşitme Kaybı-Ataksi-Retinitis Pigmentoza-Katarakt Sendromu) (ABHD12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674, Autosomal recessive; PHARC (Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome) (ABHD12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABHD12	ABHD12, PHARC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (PHARC) [MIM:612674]: A slowly progressive neurologic disorder with a variable phenotype resembling Refsum disease. Clinical features include sensorineural hearing loss, visual problems related to cataracts, retinitis pigmentosa, pes cavus, ataxic and/or spastic gait disturbances with a progressive sensorimotor peripheral neuropathy. Other features include hyporeflexia, hyperreflexia, extensor plantar responses. {ECO:0000269|PubMed:20797687, ECO:0000269|PubMed:22938382, ECO:0000269|PubMed:24027063}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Polinöropati, İşitme Kaybı, Ataksi, Retinitis Pigmentoza ve Katarakt, 612674 İşitme, Otozomal Resesif; PHARC (Polinöropati-İşitme Kaybı-Ataksi-Retinitis Pigmentoza-Katarakt Sendromu) (Prenatal) (ABHD12 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract, 612674, Autosomal recessive; PHARC (Polyneuropathy-hearing loss-ataxia-retinitis pigmentosa-cataract syndrome) (ABHD12 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABHD12	ABHD12, PHARC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract (PHARC) [MIM:612674]: A slowly progressive neurologic disorder with a variable phenotype resembling Refsum disease. Clinical features include sensorineural hearing loss, visual problems related to cataracts, retinitis pigmentosa, pes cavus, ataxic and/or spastic gait disturbances with a progressive sensorimotor peripheral neuropathy. Other features include hyporeflexia, hyperreflexia, extensor plantar responses. {ECO:0000269|PubMed:20797687, ECO:0000269|PubMed:22938382, ECO:0000269|PubMed:24027063}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Polipozis Sendromu, Kalıtsal, 2, Karışık 610069 (Kalıtsal Karışık Polipozis Sendromu) (BMPR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyposis syndrome, hereditary mixed, 2, 610069 (Hereditary mixed polyposis syndrome) (BMPR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1A	BMPR1A, ACVRLK3, ALK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polipozis Sendromu, Kalıtsal, 2, Karışık 610069 (Kalıtsal Karışık Polipozis Sendromu) (MLPA)	Polyposis syndrome, hereditary mixed, 2, 610069 (Hereditary mixed polyposis syndrome) (MLPA)	BMPR1A	BMPR1A, ACVRLK3, ALK3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polipozis Sendromu, Kalıtsal Miks tip 1, 601228. Otozomal Dominant (Kalıtsal Karışık Polipozis Sendromu) (Kromozom 15q13-q14 duplikasyon sendromu) (2309	Polyposis syndrome, mixed hereditary 1, 601228. Autosomal dominant (Hereditary mixed polyposis syndrome) (CHROMOSOME 15q13-q14 DUPLICATION SYNDROME) (440	.	HMPS1, CRAC1, CRCS4, DUP15q, C15DUPq	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Polipozis, Jüvenil İntestinal, 174900, Otozomal Dominant (Jüvenil Polipozis Sendromu) (SMAD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyposis, juvenile intestinal, 174900, Autosomal dominant (Juvenile polyposis syndrome) (SMAD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polipozis, Jüvenil İntestinal, 174900, Otozomal Dominant (Jüvenil Polipozis Sendromu) (BMPR1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Polyposis, juvenile intestinal, 174900, Autosomal dominant (Juvenile polyposis syndrome) (BMPR1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR1A	BMPR1A, ACVRLK3, ALK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polipozis, Jüvenil İntestinal, 174900, Otozomal Dominant (Jüvenil Polipozis Sendromu) (MLPA)	Polyposis, juvenile intestinal, 174900, Autosomal dominant (Juvenile polyposis syndrome) (MLPA)	SMAD4	SMAD4, MADH4, DPC4, SMAD4, JIP, MYHRS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. {ECO:0000269|PubMed:16959974}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Polipozis, Jüvenil İntestinal, 174900, Otozomal Dominant (Jüvenil Polipozis Sendromu) (MLPA)	Polyposis, juvenile intestinal, 174900, Autosomal dominant (Juvenile polyposis syndrome) (MLPA)	BMPR1A	BMPR1A, ACVRLK3, ALK3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Polyposis syndrome, mixed hereditary 2 (HMPS2) [MIM:610069]: A disease is characterized by atypical juvenile polyps, colonic adenomas, and colorectal carcinomas. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 1A, 607596, Otozomal Resesif; PCH1A (Pontoserebellar Hipoplazi Tip 1) (VRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 1A, 607596, Autosomal recessive; PCH1A (Pontocerebellar hypoplasia type 1) (VRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VRK1	VRK1, PCH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 1A (PCH1A) [MIM:607596]: A disorder characterized by an abnormally small cerebellum and brainstem, central and peripheral motor dysfunction from birth, gliosis and spinal cord anterior horn cells degeneration resembling infantile spinal muscular atrophy. Additional features include muscle hypotonia, congenital contractures and respiratory insufficiency that is evident at birth. {ECO:0000269|PubMed:19646678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 1A, 607596, Otozomal Resesif; PCH1A (Pontoserebellar Hipoplazi Tip 1) (Prenatal) (VRK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 1A, 607596, Autosomal recessive; PCH1A (Pontocerebellar hypoplasia type 1) (VRK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VRK1	VRK1, PCH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 1A (PCH1A) [MIM:607596]: A disorder characterized by an abnormally small cerebellum and brainstem, central and peripheral motor dysfunction from birth, gliosis and spinal cord anterior horn cells degeneration resembling infantile spinal muscular atrophy. Additional features include muscle hypotonia, congenital contractures and respiratory insufficiency that is evident at birth. {ECO:0000269|PubMed:19646678}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2A, 277470, Otozomal Resesif; PCH2A (Pontoserebellar Hipoplazi Tip 2) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 2A, 277470, Autosomal recessive; PCH2A (Pontocerebellar hypoplasia type 2) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2A, 277470, Otozomal Resesif; PCH2A (Pontoserebellar Hipoplazi Tip 2) (Prenatal) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 2A, 277470, Autosomal recessive; PCH2A (Pontocerebellar hypoplasia type 2) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2B, 612389, Otozomal Resesif; PCH2B (Pontoserebellar Hipoplazi Tip 3) (TSEN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 2B, 612389, Autosomal recessive; PCH2B (Pontocerebellar hypoplasia type 2) (TSEN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN2	TSEN2, SEN2, PCH2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Pontoserebellar Hipoplazi Tip 2B, 612389, Otozomal Resesif; PCH2B (Pontoserebellar Hipoplazi Tip 3) (Prenatal) (TSEN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 2B, 612389, Autosomal recessive; PCH2B (Pontocerebellar hypoplasia type 2) (TSEN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN2	TSEN2, SEN2, PCH2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2C, 612390; PCH2C (Pontoserebellar Hipoplazi Tip 4) (TSEN34 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 2C, 612390; PCH2C (Pontocerebellar hypoplasia type 2) (TSEN34 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN34	TSEN34, PCH2C, LENG5, SEN34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 2C (PCH2C) [MIM:612390]: A disorder characterized by an abnormally small cerebellum and brainstem, and progressive microcephaly from birth combined with extrapyramidal dyskinesia. Severe chorea occurs and epilepsy is frequent. There are no signs of spinal cord anterior horn cells degeneration. {ECO:0000269	PubMed:18711368}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2C, 612390; PCH2C (Pontoserebellar Hipoplazi Tip 4) (Prenatal) (TSEN34 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 2C, 612390; PCH2C (Pontocerebellar hypoplasia type 2) (TSEN34 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN34	TSEN34, PCH2C, LENG5, SEN34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 2C (PCH2C) [MIM:612390]: A disorder characterized by an abnormally small cerebellum and brainstem, and progressive microcephaly from birth combined with extrapyramidal dyskinesia. Severe chorea occurs and epilepsy is frequent. There are no signs of spinal cord anterior horn cells degeneration. {ECO:0000269	PubMed:18711368}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2D, 613811, Otozomal Resesif; PCH2D (Pontoserebellar Hipoplazi Tip 5) (SEPSECS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 2D, 613811, Autosomal recessive; PCH2D (Pontocerebellar hypoplasia type 2) (SEPSECS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEPSECS	SEPSECS, SLA, LP, PCH2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 2D, 613811, Otozomal Resesif; PCH2D (Pontoserebellar Hipoplazi Tip 5) (Prenatal) (SEPSECS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 2D, 613811, Autosomal recessive; PCH2D (Pontocerebellar hypoplasia type 2) (SEPSECS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SEPSECS	SEPSECS, SLA, LP, PCH2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 4, 225753, Otozomal Resesif (Pontoserebellar Hipoplazi Tip 4) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 4, 225753, Autosomal recessive (Pontocerebellar hypoplasia type 4) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 4, 225753, Otozomal Resesif (Pontoserebellar Hipoplazi Tip 4) (Prenatal) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 4, 225753, Autosomal recessive (Pontocerebellar hypoplasia type 4) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 5, 610204, Otozomal Resesif (Pontoserebellar Hipoplazi Tip 5) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia type 5, 610204, Autosomal recessive (Pontocerebellar hypoplasia type 5) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pontoserebellar Hipoplazi Tip 5, 610204, Otozomal Resesif (Pontoserebellar Hipoplazi Tip 5) (Prenatal) (TSEN54 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia type 5, 610204, Autosomal recessive (Pontocerebellar hypoplasia type 5) (TSEN54 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN54	TSEN54, SEN54, PCH2A, PCH4, PCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 4 (PCH4) [MIM:225753]: A disorder characterized by an abnormally small cerebellum and brainstem, severe neonatal encephalopathy, microcephaly, myoclonus and muscular hypertonia. There is a severe inferior olivary and pontine neuronal loss and a diffuse white matter gliosis. {ECO:0000269|PubMed:18711368, ECO:0000269|PubMed:21824568}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Otozomal Resesif Tip 10, 615803 (Pontoserebellar Hipoplazi Tip 10) (CLP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 10, 615803, Autosomal recessive (Pontocerebellar hypoplasia type 10) (CLP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLP1	CLP1, HEAB, PCH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 10 (PCH10) [MIM:615803]: A form of pontocerebellar hypoplasia, a disorder characterized by structural defects of the pons and cerebellum, evident upon brain imaging. PCH10 features include cortical dysgenesis marked by a simplified gyral pattern, cortical atrophy, mild or focal cerebellar vermian volume loss, delayed myelination, progressive microcephaly, global growth and developmental delays, severe intellectual disabilities, and seizures refractory to treatment. {ECO:0000269|PubMed:24766809, ECO:0000269|PubMed:24766810}. Note=The disease is caused by mutations affecting the gene represented in this entry. Neurodegeneration is due to defects in tRNA splicing (PubMed:24766809, PubMed:24766810). {ECO:0000269|PubMed:24766809, ECO:0000269|PubMed:24766810}.	EDTA Blood Sample
Pontoserebellar Hipoplazi, Otozomal Resesif Tip 10, 615803 (Pontoserebellar Hipoplazi Tip 10) (Prenatal) (CLP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 10, 615803, Autosomal recessive (Pontocerebellar hypoplasia type 10) (CLP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CLP1	CLP1, HEAB, PCH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 10 (PCH10) [MIM:615803]: A form of pontocerebellar hypoplasia, a disorder characterized by structural defects of the pons and cerebellum, evident upon brain imaging. PCH10 features include cortical dysgenesis marked by a simplified gyral pattern, cortical atrophy, mild or focal cerebellar vermian volume loss, delayed myelination, progressive microcephaly, global growth and developmental delays, severe intellectual disabilities, and seizures refractory to treatment. {ECO:0000269|PubMed:24766809, ECO:0000269|PubMed:24766810}. Note=The disease is caused by mutations affecting the gene represented in this entry. Neurodegeneration is due to defects in tRNA splicing (PubMed:24766809, PubMed:24766810). {ECO:0000269|PubMed:24766809, ECO:0000269|PubMed:24766810}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 1B, 614678, Otozomal Resesif; PCH1B (Pontoserebellar Hipoplazi Tip 2) (EXOSC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 1B, 614678, Autosomal recessive; PCH1B (Pontocerebellar hypoplasia type 1) (EXOSC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXOSC3	EXOSC3, RRP40, PCH1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 1B (PCH1B) [MIM:614678]: A severe autosomal recessive neurologic disorder characterized by a combination of cerebellar and spinal motor neuron degeneration beginning at birth. There is diffuse muscle weakness, progressive microcephaly, global developmental delay, and brainstem involvement. {ECO:0000269	PubMed:22544365}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 1B, 614678, Otozomal Resesif; PCH1B (Pontoserebellar Hipoplazi Tip 2) (Prenatal) (EXOSC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 1B, 614678, Autosomal recessive; PCH1B (Pontocerebellar hypoplasia type 1) (EXOSC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EXOSC3	EXOSC3, RRP40, PCH1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 1B (PCH1B) [MIM:614678]: A severe autosomal recessive neurologic disorder characterized by a combination of cerebellar and spinal motor neuron degeneration beginning at birth. There is diffuse muscle weakness, progressive microcephaly, global developmental delay, and brainstem involvement. {ECO:0000269	PubMed:22544365}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 1C, 616081, Otozomal Resesif; PCH1C (Pontoserebellar Hipoplazi Tip 3) (EXOSC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 1C, 616081, Autosomal recessive; PCH1C (Pontocerebellar hypoplasia type 1) (EXOSC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXOSC8	EXOSC8, OIP2, RRP43, PCH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 1C (PCH1C) [MIM:616081]: A severe autosomal recessive neurodegenerative disease characterized by cerebellar and corpus callosum hypoplasia, abnormal myelination of the central nervous system, and spinal motor neuron disease. Affected individuals manifest failure to thrive, severe muscle weakness, spasticity and psychomotor retardation. Vision and hearing are impaired. {ECO:0000269|PubMed:24989451}. Note=The disease is caused by mutations affecting the gene represented in this entry. EXOSC8 dysfunction causes myelin disruption through an imbalanced supply of myelin proteins due to dysregulation of their ARE-containing mRNAs (PubMed:24989451). {ECO:0000269|PubMed:24989451}..	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 1C, 616081, Otozomal Resesif; PCH1C (Pontoserebellar Hipoplazi Tip 3) (Prenatal) (EXOSC8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 1C, 616081, Autosomal recessive; PCH1C (Pontocerebellar hypoplasia type 1) (EXOSC8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EXOSC8	EXOSC8, OIP2, RRP43, PCH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 1C (PCH1C) [MIM:616081]: A severe autosomal recessive neurodegenerative disease characterized by cerebellar and corpus callosum hypoplasia, abnormal myelination of the central nervous system, and spinal motor neuron disease. Affected individuals manifest failure to thrive, severe muscle weakness, spasticity and psychomotor retardation. Vision and hearing are impaired. {ECO:0000269|PubMed:24989451}. Note=The disease is caused by mutations affecting the gene represented in this entry. EXOSC8 dysfunction causes myelin disruption through an imbalanced supply of myelin proteins due to dysregulation of their ARE-containing mRNAs (PubMed:24989451). {ECO:0000269|PubMed:24989451}..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 2E, 615851, Otozomal Resesif; PCH2E (Progresif Serebello-Beyin Atrofisi) (VPS53 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 2E, 615851, Autosomal recessive; PCH2E (Progressive cerebello-cerebral atrophy) (VPS53 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS53	VPS53, HCCS1, PCH2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 2E (PCH2E) [MIM:615851]: A neurodegenerative disorder characterized by progressive cerebello- cerebral atrophy, profound mental retardation, progressive microcephaly, spasticity, and early-onset epilepsy. {ECO:0000269	PubMed:24577744}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 2E, 615851, Otozomal Resesif; PCH2E (Progresif Serebello-Beyin Atrofisi) (Prenatal) (VPS53 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 2E, 615851, Autosomal recessive; PCH2E (Progressive cerebello-cerebral atrophy) (VPS53 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VPS53	VPS53, HCCS1, PCH2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 2E (PCH2E) [MIM:615851]: A neurodegenerative disorder characterized by progressive cerebello- cerebral atrophy, profound mental retardation, progressive microcephaly, spasticity, and early-onset epilepsy. {ECO:0000269	PubMed:24577744}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 2F, 617026, Otozomal Resesif; PCH2F (Pontoserebellar Hipoplazi Tip 6) (TSEN15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 2F, 617026, Autosomal recessive; PCH2F (Pontocerebellar hypoplasia type 2) (TSEN15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSEN15	TSEN15, SEN15, C1orf19, PCH2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 2F, 617026, Otozomal Resesif; PCH2F (Pontoserebellar Hipoplazi Tip 6) (Prenatal) (TSEN15 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 2F, 617026, Autosomal recessive; PCH2F (Pontocerebellar hypoplasia type 2) (TSEN15 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSEN15	TSEN15, SEN15, C1orf19, PCH2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Pontoserebellar Hipoplazi, Tip 3, 608027, Otozomal Resesif; PCH3 (Pontoserebellar Hipoplazi Tip 3) (PCLO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 3, 608027, Autosomal recessive; PCH3 (Pontocerebellar hypoplasia type 3) (PCLO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCLO	PCLO, PCH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 3 (PCH3) [MIM:608027]: A form of pontocerebellar hypoplasia, a disorder characterized by structural defects of the pons and cerebellum. Brain MRI shows an abnormally small cerebellum and brainstem, decreased cerebral white matter, and a thin corpus callosum. PCH3 features include seizures, short stature, optic atrophy, progressive microcephaly, severe developmental delay. {ECO:0000269	PubMed:25832664}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 3, 608027, Otozomal Resesif; PCH3 (Pontoserebellar Hipoplazi Tip 3) (Prenatal) (PCLO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 3, 608027, Autosomal recessive; PCH3 (Pontocerebellar hypoplasia type 3) (PCLO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCLO	PCLO, PCH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 3 (PCH3) [MIM:608027]: A form of pontocerebellar hypoplasia, a disorder characterized by structural defects of the pons and cerebellum. Brain MRI shows an abnormally small cerebellum and brainstem, decreased cerebral white matter, and a thin corpus callosum. PCH3 features include seizures, short stature, optic atrophy, progressive microcephaly, severe developmental delay. {ECO:0000269	PubMed:25832664}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Otozomal Resesif Tip 6, 611523; PCH6 (Pontoserebellar Hipoplazi Tip 6) (RARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 6, 611523, Autosomal recessive; PCH6 (Pontocerebellar hypoplasia type 6) (RARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RARS2	RARS2, RARSL, PCH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 6 (PCH6) [MIM:611523]: A disorder characterized by an abnormally small cerebellum and brainstem, infantile encephalopathy, generalized hypotonia, lethargy and poor feeding. Recurrent apnea, intractable seizures occur early in the course of this condition. {ECO:0000269	PubMed:17847012}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pontoserebellar Hipoplazi, Otozomal Resesif Tip 6, 611523; PCH6 (Pontoserebellar Hipoplazi Tip 6) (Prenatal) (RARS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 6, 611523, Autosomal recessive; PCH6 (Pontocerebellar hypoplasia type 6) (RARS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RARS2	RARS2, RARSL, PCH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 6 (PCH6) [MIM:611523]: A disorder characterized by an abnormally small cerebellum and brainstem, infantile encephalopathy, generalized hypotonia, lethargy and poor feeding. Recurrent apnea, intractable seizures occur early in the course of this condition. {ECO:0000269	PubMed:17847012}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 8, 614961, Otozomal Resesif; PCH8 (Pontoserebellar Hipoplazi Tip 8) (CHMP1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 8, 614961, Autosomal recessive; PCH8 (Pontocerebellar hypoplasia type 8) (CHMP1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHMP1A	CHMP1A, PCOLN3, PRSM1, PCH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pontocerebellar hypoplasia 8 (PCH8) [MIM:614961]: An autosomal recessive neurodevelopmental disorder characterized by severe psychomotor retardation, abnormal movements, hypotonia, spasticity, and variable visual defects. Brain MRI shows pontocerebellar hypoplasia, decreased cerebral white matter, and a thin corpus callosum. {ECO:0000269	PubMed:23023333}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 8, 614961, Otozomal Resesif; PCH8 (Pontoserebellar Hipoplazi Tip 8) (Prenatal) (CHMP1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 8, 614961, Autosomal recessive; PCH8 (Pontocerebellar hypoplasia type 8) (CHMP1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHMP1A	CHMP1A, PCOLN3, PRSM1, PCH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pontocerebellar hypoplasia 8 (PCH8) [MIM:614961]: An autosomal recessive neurodevelopmental disorder characterized by severe psychomotor retardation, abnormal movements, hypotonia, spasticity, and variable visual defects. Brain MRI shows pontocerebellar hypoplasia, decreased cerebral white matter, and a thin corpus callosum. {ECO:0000269	PubMed:23023333}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 9, 615809, Otozomal Resesif; PCH9 (Pontoserebellar Hipoplazi Tip 9) (AMPD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pontocerebellar hypoplasia, type 9, 615809, Autosomal recessive; PCH9 (Pontocerebellar hypoplasia type 9) (AMPD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMPD2	AMPD2, SPG63, PCH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 63, autosomal recessive (SPG63) [MIM:615686]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:24482476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pontoserebellar Hipoplazi, Tip 9, 615809, Otozomal Resesif; PCH9 (Pontoserebellar Hipoplazi Tip 9) (Prenatal) (AMPD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pontocerebellar hypoplasia, type 9, 615809, Autosomal recessive; PCH9 (Pontocerebellar hypoplasia type 9) (AMPD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AMPD2	AMPD2, SPG63, PCH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 63, autosomal recessive (SPG63) [MIM:615686]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:24482476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Popliteal Pterjiyum Sendromu 1, 119500, Otozomal Dominant; PPS (Otozomal Dominant Popliteal Pterjium Sendromu) (IRF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Popliteal pterygium syndrome 1, 119500, Autosomal dominant; PPS (Autosomal dominant popliteal pterygium syndrome) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. {ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Popliteal Pterjiyum Sendromu 1, 119500, Otozomal Dominant; PPS (Otozomal Dominant Popliteal Pterjium Sendromu) (Prenatal) (IRF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Popliteal pterygium syndrome 1, 119500, Autosomal dominant; PPS (Autosomal dominant popliteal pterygium syndrome) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. {ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Popliteal Pterjiyum Sendromu, Bartsocas-Papas Tipi, 263650, Otozomal Resesif (Bartsocas-Papas Sendromu) (RIPK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650, Autosomal recessive (Bartsocas-Papas syndrome) (RIPK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIPK4	RIPK4, NKRD3, DIK, PPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Popliteal pterygium syndrome, lethal type (PPS-L) [MIM:263650]: An autosomal recessive disorder characterized by multiple popliteal pterygia leading to severe arthrogryposis, ankyloblepharon filiforme adnatum, filiform bands between the jaws, synostosis of the carpal/tarsal and phalangeal bones in the hands and feet, digital hypoplasia/aplasia, complete soft-tissue syndactyly, lack of nails, lack of scalp hair, eyebrows and eyelashes, blepharophimosis, cleft lip and/or palate, and hypoplastic external genitalia. Early lethality is common, although survival into childhood and beyond has been reported. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Popliteal Pterjiyum Sendromu, Bartsocas-Papas Tipi, 263650, Otozomal Resesif (Bartsocas-Papas Sendromu) (Prenatal) (RIPK4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650, Autosomal recessive (Bartsocas-Papas syndrome) (RIPK4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RIPK4	RIPK4, NKRD3, DIK, PPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Popliteal pterygium syndrome, lethal type (PPS-L) [MIM:263650]: An autosomal recessive disorder characterized by multiple popliteal pterygia leading to severe arthrogryposis, ankyloblepharon filiforme adnatum, filiform bands between the jaws, synostosis of the carpal/tarsal and phalangeal bones in the hands and feet, digital hypoplasia/aplasia, complete soft-tissue syndactyly, lack of nails, lack of scalp hair, eyebrows and eyelashes, blepharophimosis, cleft lip and/or palate, and hypoplastic external genitalia. Early lethality is common, although survival into childhood and beyond has been reported. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porensefali 1, 175780, Otozomal Dominant; POREN1 (Porensefali) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porencephaly 1, 175780, Autosomal dominant; POREN1 (Porencephaly) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porencephaly 1 (POREN1) [MIM:175780]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres, neurologic manifestations, facial paresis, and visual defects. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 1 is usually unilateral and results from focal destructive lesions such as fetal vascular occlusion or birth trauma. {ECO:0000269|PubMed:15905400, ECO:0000269|PubMed:16107487, ECO:0000269|PubMed:19194877}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porensefali 1, 175780, Otozomal Dominant; POREN1 (Porensefali) (Prenatal) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porencephaly 1, 175780, Autosomal dominant; POREN1 (Porencephaly) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A1	COL4A1, POREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Porencephaly 1 (POREN1) [MIM:175780]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres, neurologic manifestations, facial paresis, and visual defects. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 1 is usually unilateral and results from focal destructive lesions such as fetal vascular occlusion or birth trauma. {ECO:0000269|PubMed:15905400, ECO:0000269|PubMed:16107487, ECO:0000269|PubMed:19194877}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porensefali 2, 614483, Otozomal Dominant; POREN2 (Porensefali) (COL4A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porencephaly 2, 614483, Autosomal dominant; POREN2 (Porencephaly) (COL4A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A2	COL4A2, POREN2, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porencephaly 2 (POREN2) [MIM:614483]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 2, or schizencephalic porencephaly, is usually symmetric and represents a primary defect in the development of the cerebral ventricles. {ECO:0000269	PubMed:22209246}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porensefali 2, 614483, Otozomal Dominant; POREN2 (Porensefali) (Prenatal) (COL4A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porencephaly 2, 614483, Autosomal dominant; POREN2 (Porencephaly) (COL4A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL4A2	COL4A2, POREN2, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Porencephaly 2 (POREN2) [MIM:614483]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 2, or schizencephalic porencephaly, is usually symmetric and represents a primary defect in the development of the cerebral ventricles. {ECO:0000269	PubMed:22209246}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Poretti-Boltshauser Sendromu, 615960, Otozomal Resesif; PTBHS (Ataksi-Mental Retardasyon-Okulomotor Apraksi-Serebellar Kistler Sendromu) (LAMA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Poretti-Boltshauser syndrome, 615960, Autosomal recessive; PTBHS (Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome) (LAMA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAMA1	LAMA1, PTBHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Poretti-Boltshauser syndrome (PTBHS) [MIM:615960]: An autosomal recessive disorder characterized by cerebellar dysplasia, cerebellar vermis atrophy, cerebellar cysts in most patients, high myopia, variable retinal dystrophy, and eye movement abnormalities including strabismus, ocular apraxia, nystagmus. Affected individuals have ataxia, delayed motor development, language impairment, and intellectual disability with variable severity. [ECO:0000269	PubMed:25105227]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Poretti-Boltshauser Sendromu, 615960, Otozomal Resesif; PTBHS (Ataksi-Mental Retardasyon-Okulomotor Apraksi-Serebellar Kistler Sendromu) (Prenatal) (LAMA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Poretti-Boltshauser syndrome, 615960, Autosomal recessive; PTBHS (Ataxia-intellectual disability-oculomotor apraxia-cerebellar cysts syndrome) (LAMA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LAMA1	LAMA1, PTBHS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Poretti-Boltshauser syndrome (PTBHS) [MIM:615960]: An autosomal recessive disorder characterized by cerebellar dysplasia, cerebellar vermis atrophy, cerebellar cysts in most patients, high myopia, variable retinal dystrophy, and eye movement abnormalities including strabismus, ocular apraxia, nystagmus. Affected individuals have ataxia, delayed motor development, language impairment, and intellectual disability with variable severity. [ECO:0000269	PubMed:25105227]. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porokeratoz 1, Çeşitli Tipler, 175800, Otozomal Dominant; POROK1 (Mibelli`nin Porokeratozisi) (PMVK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porokeratosis 1, multiple types, 175800, Autosomal dominant; POROK1 (Porokeratosis of Mibelli) (PMVK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMVK	PMVK, PMK, POROK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porokeratosis 1, multiple types (POROK1) [MIM:175800]: A form of porokeratosis, a disorder of faulty keratinization characterized by one or more atrophic patches surrounded by a distinctive hyperkeratotic ridgelike border called the cornoid lamella. The keratotic lesions can progress to overt cutaneous neoplasms, typically squamous cell carcinomas. Multiple clinical variants of porokeratosis are recognized, including porokeratosis of Mibelli, linear porokeratosis, disseminated superficial actinic porokeratosis, palmoplantar porokeratosis, and punctate porokeratosis. Different clinical presentations can be observed among members of the same family. Individuals expressing more than one variant have also been reported. [ECO:0000269	PubMed:26202976]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Porokeratoz 3, Multipl Tipler 175900, Otozomal Dominant; POROK3 (Mibelli`nin Porokeratozisi) (MVK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porokeratosis 3, multiple types, 175900, Autosomal dominant; POROK3 (Porokeratosis of Mibelli) (MVK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MVK	MVK, MVLK, POROK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porokeratosis 3, multiple types (POROK3) [MIM:175900]: A form of porokeratosis, a disorder of faulty keratinization characterized by one or more atrophic patches surrounded by a distinctive hyperkeratotic ridgelike border called the cornoid lamella. The keratotic lesions can progress to overt cutaneous neoplasms, typically squamous cell carcinomas. Multiple clinical variants of porokeratosis are recognized, including porokeratosis of Mibelli, linear porokeratosis, disseminated superficial actinic porokeratosis, palmoplantar porokeratosis, and punctate porokeratosis. Different clinical presentations can be observed among members of the same family. Individuals expressing more than one variant have also been reported. [ECO:0000269|PubMed:22983302, ECO:0000269|PubMed:24781643]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Porokeratoz 7, Çeşitli Tipler, 614714, Otozomal Dominant; POROK7 (Disseminе Yüzeysel Aktinik Porokeratozis) (MVD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porokeratosis 7, multiple types, 614714, Autosomal dominant; POROK7 (Disseminated superficial actinic porokeratosis) (MVD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MVD	MVD, MPD, POROK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Porokeratoz 8, Yaygın Yüzeysel Aktinik Tipi, 616063, Otozomal Dominant; POROK8 (Disseminе Yüzeysel Aktinik Porokeratozis) (SLC17A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porokeratosis 8, disseminated superficial actinic type, 616063, Autosomal dominant; POROK8 (Disseminated superficial actinic porokeratosis) (SLC17A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A9	SLC17A9, C20orf59, POROK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Porokeratoz 9, Çeşitli Tipler, 616631; POROK9 (Disseminе Yüzeysel Aktinik Porokeratozis9 (FDPS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porokeratosis 9, multiple types, 616631; POROK9 (Disseminated superficial actinic porokeratosis) (FDPS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FDPS	FDPS, FPS, POROK9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porokeratosis 9, multiple types (POROK9) [MIM:616631]: A form of porokeratosis, a disorder of faulty keratinization characterized by one or more atrophic patches surrounded by a distinctive hyperkeratotic ridgelike border called the cornoid lamella. The keratotic lesions can progress to overt cutaneous neoplasms, typically squamous cell carcinomas. Multiple clinical variants of porokeratosis are recognized, including porokeratosis of Mibelli, linear porokeratosis, disseminated superficial actinic porokeratosis, palmoplantar porokeratosis, and punctate porokeratosis. Different clinical presentations can be observed among members of the same family. Individuals expressing more than one variant have also been reported. [ECO:0000269	PubMed:26202976]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Porfiria Kutanea Tarda, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Aileseli) (UROD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria cutanea tarda, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (UROD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UROD	UROD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. (ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porfiria Kutanea Tarda, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Aileseli) (MLPA)	Porphyria cutanea tarda, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (MLPA)	UROD	UROD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. (ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porfiria Kutanea Tarda, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Aileseli) (Prenatal) (UROD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria cutanea tarda, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (UROD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UROD	UROD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. (ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria Kutanea Tarda, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Aileseli) (Prenatal) (MLPA)	Porphyria cutanea tarda, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (Prenatal) (MLPA)	UROD	UROD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. (ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Porfiria Kutanea Tarda, Yatkınlık, 176100, Otozomal Dominant (Porfiria Kutanea Tarda, Allesel) (HFE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria cutanea tarda, susceptibility to, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Variegate porphyria (VP) [MIM:176200]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Variegate porphyria is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Iron overload due to HFE variants is a precipitating or exacerbating factor in variegate porphyria.	EDTA Blood Sample
Porfiria Kutanea Tarda, Yatkınlık, 176100, Otozomal Dominant (Porfiria Kutanea Tarda, Allesel) (Prenatal) (HFE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria cutanea tarda, susceptibility to, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (HFE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Variegate porphyria (VP) [MIM:176200]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Variegate porphyria is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Iron overload due to HFE variants is a precipitating or exacerbating factor in variegate porphyria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria Paneli (Miks 1) (ALAD, HMBS, PPOX) (MLPA)	Porphyria Panel (Mix 1) (ALAD, HMBS, PPOX) (MLPA)	ALAD, HMBS, PPOX	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Porfiria Paneli (Miks 1) (ALAD, HMBS, PPOX) (MLPA) (Prenatal)	Porphyria Panel (Mix 1) (ALAD, HMBS, PPOX) (MLPA) (Prenatal)	ALAD, HMBS, PPOX	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria Paneli (Miks 2) (FECH, UROS, UROD, CPOX) (MLPA)	Porphyria Panel (Mix 2) (FECH, UROS, UROD, CPOX) (MLPA)	FECH, UROS, UROD, CPOX	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Porfiria Paneli (Miks 2) (FECH, UROS, UROD, CPOX) (MLPA) (Prenatal)	Porphyria Panel (Mix 2) (FECH, UROS, UROD, CPOX) (MLPA) (Prenatal)	FECH, UROS, UROD, CPOX	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria Variegata, 176200, Otozomal Dominant (Porfiria Variegata) (PPOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria variegata, 176200, Autosomal dominant (Porphyria variegata) (PPOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPOX	PPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Porfiria Variegata, 176200, Otozomal Dominant (Porfiria Variegata) (MLPA)	Porphyria variegata, 176200, Autosomal dominant (Porphyria variegata) (MLPA)	PPOX	PPOX	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Porfiria Variegata, 176200, Otozomal Dominant (Porfiria Variegata) (Prenatal) (PPOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria variegata, 176200, Autosomal dominant (Porphyria variegata) (PPOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPOX	PPOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria Variegata, 176200, Otozomal Dominant (Porfiria Variegata) (Prenatal) (MLPA)	Porphyria variegata, 176200, Autosomal dominant (Porphyria variegata) (MLPA)	PPOX	PPOX	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porphyria Variegata, Yatkınlık, 176200, Otozomal Dominant (Porfiria Variegata) (HFE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria variegata, susceptibility to, 176200, Autosomal dominant (Porphyria variegata) (HFE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Variegate porphyria (VP) [MIM:176200]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Variegate porphyria is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Iron overload due to HFE variants is a precipitating or exacerbating factor in variegate porphyria.	EDTA Blood Sample
Porphyria Variegata, Yatkınlık, 176200, Otozomal Dominant (Porfiria Variegata) (Prenatal) (HFE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria variegata, susceptibility to, 176200, Autosomal dominant (Porphyria variegata) (HFE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HFE	HFE, HLA-H, HFE1, MVCD7, TFQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Variegate porphyria (VP) [MIM:176200]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Variegate porphyria is the most common form of porphyria in South Africa. It is characterized by skin hyperpigmentation and hypertrichosis, abdominal pain, tachycardia, hypertension and neuromuscular disturbances. High fecal levels of protoporphyrin and coproporphyrin, increased urine uroporphyrins and iron overload are typical markers of the disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Iron overload due to HFE variants is a precipitating or exacerbating factor in variegate porphyria.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut Hepatik, 612740, Otozomal Resesif (Porfiria, ALA Dehidrataz Eksikliğine Bağlı) (ALAD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria, acute hepatic, 612740, Autosomal recessive (Porphyria due to ALA dehydratase deficiency) (ALAD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALAD	ALAD, ALADH, PBGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Porfiria, Akut Hepatik, 612740, Otozomal Resesif (Porfiria, ALA Dehidrataz Eksikliğine Bağlı) (MLPA)	Porphyria, acute hepatic, 612740, Autosomal recessive (Porphyria due to ALA dehydratase deficiency) (MLPA)	ALAD	ALAD, ALADH, PBGS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Porfiria, Akut Hepatik, 612740, Otozomal Resesif (Porfiria, ALA Dehidrataz Eksikliğine Bağlı) (Prenatal) (ALAD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria, acute hepatic, 612740, Autosomal recessive (Porphyria due to ALA dehydratase deficiency) (ALAD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALAD	ALAD, ALADH, PBGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut Hepatik, 612740, Otozomal Resesif (Porfiria, ALA Dehidrataz Eksikliğine Bağlı) (Prenatal) (MLPA)	Porphyria, acute hepatic, 612740, Autosomal recessive (Porphyria due to ALA dehydratase deficiency) (Prenatal) (MLPA)	ALAD	ALAD, ALADH, PBGS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acute hepatic porphyria (AHEPP) [MIM:612740]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AHP is characterized by attacks of gastrointestinal disturbances, abdominal colic, paralyses and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors. {ECO:0000269|PubMed:10706561, ECO:0000269|PubMed:1309003, ECO:0000269|PubMed:1569184, ECO:0000269|PubMed:2063868}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut, Intermittant, 176000, Otozomal Dominant (Akut Intermittant Porfiria) (HMBS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria, acute intermittent, 176000, Autosomal dominant (Acute intermittent porphyria) (HMBS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMBS	HMBS, PBGD, UPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Porfiria, Akut, Intermittant, 176000, Otozomal Dominant (Akut Intermittant Porfiria) (MLPA)	Porphyria, acute intermittent, 176000, Autosomal dominant (Acute intermittent porphyria) (MLPA)	HMBS	HMBS, PBGD, UPS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Porfiria, Akut, Intermittant, 176000, Otozomal Dominant (Akut Intermittant Porfiria) (Prenatal) (HMBS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria, acute intermittent, 176000, Autosomal dominant (Acute intermittent porphyria) (HMBS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HMBS	HMBS, PBGD, UPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut, Intermittant, 176000, Otozomal Dominant (Akut Intermittant Porfiria) (Prenatal) (MLPA)	Porphyria, acute intermittent, 176000, Autosomal dominant (Acute intermittent porphyria) (Prenatal) (MLPA)	HMBS	HMBS, PBGD, UPS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut, Intermittant, Nonerithroid Varyant, 176000, Otozomal Dominant; AIP (Akut Intermittant Porfiria) (HMBS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria, acute intermittent, nonerythroid variant, 176000, Autosomal dominant; AIP (Acute intermittent porphyria) (HMBS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMBS	HMBS, PBGD, UPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Porfiria, Akut, Intermittant, Nonerithroid Varyant, 176000, Otozomal Dominant; AIP (Akut Intermittant Porfiria) (MLPA)	Porphyria, acute intermittent, nonerythroid variant, 176000, Autosomal dominant; AIP (Acute intermittent porphyria) (MLPA)	HMBS	HMBS, PBGD, UPS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Porfiria, Akut, Intermittant, Nonerithroid Varyant, 176000, Otozomal Dominant; AIP (Akut Intermittant Porfiria) (Prenatal) (HMBS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria, acute intermittent, nonerythroid variant, 176000, Autosomal dominant; AIP (Acute intermittent porphyria) (HMBS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HMBS	HMBS, PBGD, UPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Akut, Intermittant, Nonerithroid Varyant, 176000, Otozomal Dominant; AIP (Akut Intermittant Porfiria) (Prenatal) (MLPA)	Porphyria, acute intermittent, nonerythroid variant, 176000, Autosomal dominant; AIP (Acute intermittent porphyria) (Prenatal) (MLPA)	HMBS	HMBS, PBGD, UPS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Porfiria, Konjenital Eritropoetik, 263700, Otozomal Resesif (Konjenital Eritropoetik Porfiria) (UROS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria, congenital erythropoietic, 263700, Autosomal recessive (Congenital erythropoietic porphyria) (UROS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UROS	UROS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Severe congenital erythropoietic porphyria is associated with non-immune hydrops fetalis, a generalized edema of the fetus with fluid accumulation in the body cavities due to non- immune causes. Non-immune hydrops fetalis is not a diagnosis in itself but a symptom, a feature of many genetic disorders, and the end-stage of a wide variety of disorders.;	EDTA Blood Sample
Porfiria, Konjenital Eritropoetik, 263700, Otozomal Resesif (Konjenital Eritropoetik Porfiria) (MLPA)	Porphyria, congenital erythropoietic, 263700, Autosomal recessive (Congenital erythropoietic porphyria) (MLPA)	UROS	UROS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Note=Severe congenital erythropoietic porphyria is associated with non-immune hydrops fetalis, a generalized edema of the fetus with fluid accumulation in the body cavities due to non- immune causes. Non-immune hydrops fetalis is not a diagnosis in itself but a symptom, a feature of many genetic disorders, and the end-stage of a wide variety of disorders.;	EDTA Blood Sample
Porfiria, Konjenital Eritropoetik, 263700, Otozomal Resesif (Konjenital Eritropoetik Porfiria) (Prenatal) (UROS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria, congenital erythropoietic, 263700, Autosomal recessive (Congenital erythropoietic porphyria) (UROS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UROS	UROS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Severe congenital erythropoietic porphyria is associated with non-immune hydrops fetalis, a generalized edema of the fetus with fluid accumulation in the body cavities due to non- immune causes. Non-immune hydrops fetalis is not a diagnosis in itself but a symptom, a feature of many genetic disorders, and the end-stage of a wide variety of disorders.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Konjenital Eritropoetik, 263700, Otozomal Resesif (Konjenital Eritropoetik Porfiria) (Prenatal) (MLPA)	Porphyria, congenital erythropoietic, 263700, Autosomal recessive (Congenital erythropoietic porphyria) (Prenatal) (MLPA)	UROS	UROS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Severe congenital erythropoietic porphyria is associated with non-immune hydrops fetalis, a generalized edema of the fetus with fluid accumulation in the body cavities due to non- immune causes. Non-immune hydrops fetalis is not a diagnosis in itself but a symptom, a feature of many genetic disorders, and the end-stage of a wide variety of disorders.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Hepatoeritropoetik, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Allesel) (UROD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Porphyria, hepatoerythropoietic, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (UROD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UROD	UROD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. {ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porfiria, Hepatoeritropoetik, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Allesel) (MLPA)	Porphyria, hepatoerythropoietic, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (MLPA)	UROD	UROD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. {ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Porfiria, Hepatoeritropoetik, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Allesel) (Prenatal) (UROD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Porphyria, hepatoerythropoietic, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (UROD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UROD	UROD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. {ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Porfiria, Hepatoeritropoetik, 176100, Otozomal Dominant (Porfiri, Kutanea Tarda, Allesel) (Prenatal) (MLPA)	Porphyria, hepatoerythropoietic, 176100, Autosomal dominant (Familial porphyria cutanea tarda) (Prenatal) (MLPA)	UROD	UROD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Familial porphyria cutanea tarda (FPCT) [MIM:176100]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Familial porphyria cutanea tarda is an autosomal dominant disorder characterized by light-sensitive dermatitis, with onset in later life. It is associated with the excretion of large amounts of uroporphyrin in the urine. Iron overload is often present in association with varying degrees of liver damage. {ECO:0000269|PubMed:10338097, ECO:0000269|PubMed:10477430, ECO:0000269|PubMed:11069625, ECO:0000269|PubMed:11295834, ECO:0000269|PubMed:11719352, ECO:0000269|PubMed:2243121, ECO:0000269|PubMed:2920211, ECO:0000269|PubMed:7706766, ECO:0000269|PubMed:8896428, ECO:0000269|PubMed:9792863}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Portal Hipertansiyon, Nonsirotik, 617068, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (DGUOK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Portal hypertension, noncirrhotic, 617068, Autosomal recessive (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Portal Hipertansiyon, Nonsirotik, 617068, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (MLPA)	Portal hypertension, noncirrhotic, 617068, Autosomal recessive (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Portal Hipertansiyon, Nonsirotik, 617068, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (Prenatal) (DGUOK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Portal hypertension, noncirrhotic, 617068, Autosomal recessive (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Portal Hipertansiyon, Nonsirotik, 617068, Otozomal Resesif (Mitokondriyal DNA Deplesyon Sendromu, DGUOK Eksikliğine Bağlı Hepatoserebral) (Prenatal) (MLPA)	Portal hypertension, noncirrhotic, 617068, Autosomal recessive (Mitochondrial DNA depletion syndrome, hepatocerebral form due to DGUOK deficiency) (Prenatal) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fetüs ve Otu Bebek Muayenesi	Postmortem examination	.	.	Klinik değerlendirme/ Clinical Evaluation	Tüm Tıbbi Dokümanlar ve Ale Fotoğrafları		0	0	.	.	0	Postnatal	.	.	Medical Documents and Family Photos
Fetüs - Yenidoğan Otopsisi + Genetik Muayene	Postmortem examination and autopsy	.	.	Klinik değerlendirme/ Clinical Evaluation	Tüm Tıbbi Dokümanlar ve Ale Fotoğrafları		0	0	.	.	0	Postnatal	.	.	Medical Documents and Family Photos
Potocki-Lupski Sendromu, 610883, Sporadik Vakalar (17p11.2 Mikroduplikasyon Sendromu) (22/00)	Potocki-Lupski syndrome, 610883, Isolated cases (17p11.2 microduplication syndrome) (446)	.	PTLS	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	.	EDTA Blood Sample and Heparinized blood sample
Potocki-Lupski Sendromu, 610883, Sporadik Vakalar (17p11.2 Mikroduplikasyon Sendromu) (Prenatal)	Potocki-Lupski syndrome, 610883, Isolated cases (17p11.2 microduplication syndrome) (Prenatal)	.	PTLS	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prader - Willi' Angelman Sendromu (15q11 - q13 Mikrodelesyonu) (FISH)	Prader-Willi / Angelman syndrome (15q11-q13 microdeletion) (FISH)	15q11-15q13	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	.	Heparinized blood sample
Prader - Willi' Angelman Sendromu (15q11 - q13 Mikrodelesyonu) (FISH) (Prenatal)	Prader-Willi / Angelman syndrome (15q11-q13 microdeletion) (Prenatal) (FISH)	15q11-15q13	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar (Prader-Willi Sendromu) (SNRPN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prader-Willi syndrome, 176270, Isolated cases (Prader-Willi syndrome) (SNRPN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNRPN	SNRPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar (Prader-Willi Sendromu) (MLPA)	Prader-Willi syndrome, 176270, Isolated cases (Prader-Willi syndrome) (MLPA)	SNRPN	SNRPN	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar (Prader-Willi Sendromu) (Prenatal) (SNRPN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Prader-Willi syndrome, 176270, Isolated cases (Prader-Willi syndrome) (SNRPN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNRPN	SNRPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar (Prader-Willi Sendromu) (MLPA)	Prader-Willi syndrome, 176270, Isolated cases (Prader-Willi syndrome) (Prenatal) (MLPA)	SNRPN	SNRPN	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar; PWS (Prader-Willi Sendromu) (NDN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prader-Willi syndrome, 176270, Isolated cases; PWS (Prader-Willi syndrome) (NDN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDN	NDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	.	EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar; PWS (Prader-Willi Sendromu) (MLPA)	Prader-Willi syndrome, 176270, Isolated cases; PWS (Prader-Willi syndrome) (MLPA)	NDN	NDN	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Prader-Willi Sendromu, 176270, Sporadik Vakalar; PWS (Prader-Willi Sendromu) (Prenatal) (NDN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Prader-Willi syndrome, 176270, Isolated cases; PWS (Prader-Willi syndrome) (NDN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NDN	NDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Prader-Willi Sendromu, 176270, Sporadik Vakalar; PWS (Prader-Willi Sendromu) (Prenatal) (MLPA)	Prader-Willi syndrome, 176270, Isolated cases; PWS (Prader-Willi syndrome) (Prenatal) (MLPA)	NDN	NDN	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Preaksiyel Eksiklik, Postaksiyel Polidaktili ve Hipospadias (Guttmacher Sendromu) (HOXA13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PREAXIAL DEFICIENCY, POSTAXIAL POLYDACTYLY, AND HYPOSPADIAS (Guttmacher syndrome) (HOXA13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA13	HOXA13, HOX1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hand-foot-genital syndrome (HFG) [MIM:140000]: A disorder characterized by limb and genitourinary anomalies. Clinical features include small feet with unusually short great toes and abnormal thumbs. Females with the disorder have duplication of the genital tract. {ECO:0000269|PubMed:10839976, ECO:0000269|PubMed:12073020}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Preaksiyel Eksiklik, Postaksiyel Polidaktili ve Hipospadias (Guttmacher Sendromu) (Prenatal) (HOXA13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	PREAXIAL DEFICIENCY, POSTAXIAL POLYDACTYLY, AND HYPOSPADIAS (Guttmacher syndrome) (HOXA13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXA13	HOXA13, HOX1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hand-foot-genital syndrome (HFG) [MIM:140000]: A disorder characterized by limb and genitourinary anomalies. Clinical features include small feet with unusually short great toes and abnormal thumbs. Females with the disorder have duplication of the genital tract. {ECO:0000269|PubMed:10839976, ECO:0000269|PubMed:12073020}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Puberte Prekoks, Santral, 1, 176400, Otozomal Dominant; CPPB1 (Santral Puberte Prekoks) (KISS1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Precocious puberty, central, 1, 176400, Autosomal dominant; CPPB1 (Central precocious puberty) (KISS1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KISS1R	KISS1R, GPR54, HH8, CPPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Precocious puberty, central 1 (CPPB1) [MIM:176400] A condition defined as the development of secondary sexual characteristics in boys and girls at a chronological age that is 2.5 standard deviations below the mean age at onset of puberty in the population. Central precocious puberty results from premature activation of the hypothalamic-pituitary-gonadal axis. {ECO:0000269	PubMed:18272894}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Puberte Prekoks, Santral, 2, 615346, Otozomal Dominant; CPPB2 (Santral Puberte Prekoks) (MKRN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Precocious puberty, central, 2, 615346, Autosomal dominant; CPPB2 (Central precocious puberty) (MKRN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MKRN3	MKRN3, ZFP127, ZNF127, CPPB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Puberte Prekoks, Santral, 2, 615346, Otozomal Dominant; CPPB2 (Santral Puberte Prekoks) (MLPA)	Precocious puberty, central, 2, 615346, Autosomal dominant; CPPB2 (Central precocious puberty) (MLPA)	MKRN3	MKRN3, ZFP127, ZNF127, CPPB2	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Puberte Prekoks, Erkek, 176410, Otozomal Dominant (Ailesel Erkek Sınırlı Puberte Prekoks) (LHCGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Precocious puberty, male, 176410, Autosomal dominant (Familial male-limited precocious puberty) (LHCGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LHCGR	LHCGR, LHR, LCGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Luteinizing hormone resistance (LHR) [MIM:238320]: An autosomal recessive disorder characterized by unresponsiveness to luteinizing hormone, defective sexual development in males, and defective follicular development and ovulation, amenorrhea and infertility in females. Two forms of the disorder have been defined in males. Type 1 is a severe form characterized by complete 46,XY male pseudohermaphroditism, low testosterone and high luteinizing hormone levels, total lack of responsiveness to luteinizing and chorionic gonadotropin hormones, lack of breast development, and absent development of secondary male sex characteristics. Type 2, a milder form, displays a broader range of phenotypic expression ranging from micropenis to severe hypospadias. {ECO:0000269|PubMed:12050206, ECO:0000269|PubMed:15372531, ECO:0000269|PubMed:15472221, ECO:0000269|PubMed:19551906, ECO:0000269|PubMed:7719343, ECO:0000269|PubMed:8559204, ECO:0000269|PubMed:9215288, ECO:0000269|PubMed:9514160, ECO:0000269|PubMed:9626144, ECO:0000269|PubMed:9626653}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Preeklampsi, Yatkınlık (AGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preeclampsia, susceptibility to (AGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGT	AGT, SERPINA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Essential hypertension (EHT) [MIM:145500]: A condition in which blood pressure is consistently higher than normal with no identifiable cause. {ECO:0000269|PubMed:1394429, ECO:0000269|PubMed:8513325}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Preeklampsi, Yatkınlık, 189800, Otozomal Dominant (Preeklampsi) (EPHX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preeclampsia, susceptibility to, 189800, Autosomal dominant (Preeclampsia) (EPHX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHX1	EPHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Familial hypercholanemia (FHCA) [MIM:607748]: A disorder characterized by elevated serum bile acid concentrations, itching, and fat malabsorption. {ECO:0000269|PubMed:12878321}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Preeklampsi / Eklampsi 4, 609404; PEE4 (Preeklampsi) (STOX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preeclampsia/eclampsia 4, 609404; PEE4 (Preeclampsia) (STOX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STOX1	STOX1, PEE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Preeklampsi / Eklampsi 5, 614595; PEE5 (Preeklampsi) (CORIN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preeclampsia/eclampsia 5, 614595; PEE5 (Preeclampsia) (CORIN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CORIN	CORIN, CRN, TMPRSS10, ATC2, PEE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pre-eclampsia/eclampsia 5 (PEE5) [MIM:614595]: A hypertensive disorder of pregnancy characterized by new hypertension (blood pressure 140/90 or greater) presenting after 20 weeks' gestation with clinically relevant proteinuria. It impacts 2 individuals, the mother and her child, both of whom can be severely affected. Preeclampsia is one of the causes of maternal mortality and morbidity worldwide. {ECO:0000269|PubMed:22437503}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Gebelik Kaybı, Tekrarlayan, 4, 270960, Otozomal Dominant (SYCP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pregnancy loss, recurrent, 4, 270960, Autosomal dominant (SYCP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYCP3	SYCP3, SCP3, COR1, SPGF4, RPRGL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 4 (SPGF4) [MIM:270960]: An infertility disorder characterized by azoospermia, a condition of having no sperm present in the ejaculate. Testicular histology shows arrest of spermatogenesis at the pachytene stage of primary spermatocytes. {ECO:0000269|PubMed:14643120}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Gebelik Kaybı, Tekrarlayan, Yatkınlık, 1, 614389, Otozomal Dominant; RPRGL1 (F5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pregnancy loss, recurrent, susceptibility to, 1, 614389, Autosomal dominant; RPRGL1 (F5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to activated protein C resistance (THPH2) [MIM:188055]: A hemostatic disorder due to defective degradation of factor V by activated protein C. It is characterized by a poor anticoagulant response to activated protein C resulting in tendency to thrombosis. {ECO:0000269|PubMed:11435304, ECO:0000269|PubMed:11858490, ECO:0000269|PubMed:14617013, ECO:0000269|PubMed:9454742}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Gebelik Kaybı, Tekrarlayan, Yatkınlık, 2, 614390, Otozomal Dominant (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pregnancy loss, recurrent, susceptibility to, 2, 614390, Autosomal dominant (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to thrombin defect (THPH1) [MIM:188050]: A multifactorial disorder of hemostasis characterized by abnormal platelet aggregation in response to various agents and recurrent thrombi formation. {ECO:0000269|PubMed:2825773}. Note=The disease is caused by mutations affecting the gene represented in this entry. A common genetic variation in the 3-prime untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increased risk of venous thrombosis.	EDTA Blood Sample
Gebelik Kaybı, Tekrarlayan, Yatkınlık, 3, 614391, Otozomal Dominant; RPRGL3 (ANXA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pregnancy loss, recurrent, susceptibility to, 3, 614391, Autosomal dominant; RPRGL3 (ANXA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANXA5	ANXA5, ENX2, RPRGL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	. DISEASE: Pregnancy loss, recurrent, 3 (RPRGL3) [MIM:614391]: A common complication of pregnancy, resulting in spontaneous abortion before the fetus has reached viability. The term includes all miscarriages from the time of conception until 24 weeks of gestation. Recurrent pregnancy loss is defined as 3 or more consecutive spontaneous abortions. {ECO:0000269|PubMed:17339269}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Preimplantasyon Embriyo Letalitesi 2, 617234, Otozomal Resesif (PADI6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preimplantation embryonic lethality 2, 617234, Autosomal recessive (PADI6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PADI6	PADI6, PREMBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Preimplantasyon Embriyo Letalitesi, 616814, Otozomal Resesif; PREMBL2 (TLE6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preimplantation embryonic lethality, 616814, Autosomal recessive; PREMBL2 (TLE6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLE6	TLE6, GRG6, PREMBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Erken Yaşlanma Sendromu, Penttinen Tipi, 601812, Otozomal Dominant; PENTT (Akroosteolizis-Keloid Benzeri Lezyonlar-Erken Yaşlanma Sendromu) (PDGFRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature aging syndrome, Penttinen type, 601812, Autosomal dominant; PENTT (Acrosteolysis-keloid-like lesions-premature aging syndrome) (PDGFRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDGFRB	PDGFRB, PDGFR, IBGC4, IMF1, PENTT, KOGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myeloproliferative disorder chronic with eosinophilia (MPD) [MIM:131440]: A hematologic disorder characterized by malignant eosinophils proliferation. Note=The gene represented in this entry may be involved in disease pathogenesis. Chromosomal aberrations involving PDGFRB have been found in many instances of chronic myeloproliferative disorder with eosinophilia. Translocation t(5;12) with ETV6 on chromosome 12 creating an PDGFRB-ETV6 fusion protein (PubMed:12181402). Translocation t(5;15)(q33;q22) with TPS3BP1 creating a PDGFRB-TPS3BP1 fusion protein (PubMed:15492236). Translocation t(1.5)(q23;q33) that forms a PDE4DIP-PDGFRB fusion protein (PubMed:12907457). Translocation t(5:9)(q33-34;q23) with CEP85L that fuses the 5'-end of CEP85L (isoform 4) to the 3'-end of PDGFRB (PubMed:21938754). {ECO:0000269|PubMed:12181402, ECO:0000269|PubMed:12907457, ECO:0000269|PubMed:15492236, ECO:0000269|PubMed:21938754}.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 1, 311360, X'e Bağlı; POF1 (Tekrar Sayısı)	Premature ovarian failure 1, 311360, X-linked; POF1(Repeat Analysis)	FMR1	FMR1, FRAXA, POF1	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Fragile X tremor/ataxia syndrome (FXTAS) [MIM:300623]: In FXTAS, the expanded repeats range in size from 55 to 200 repeats and are referred to as 'premutations'. Full repeat expansions with greater than 200 repeats results in fragile X mental retardation syndrome [MIM:300624]. Carriers of the premutation typically do not show the full fragile X syndrome phenotype, but comprise a subgroup that may have some physical features of fragile X syndrome or mild cognitive and emotional problems. {ECO:0000269|PubMed:11445641}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 1, 311360, X'e Bağlı; POF1 (MLPA)	Premature ovarian failure 1, 311360, X-linked; POF1 (MLPA)	FMR1	FMR1, FRAXA, POF1	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Fragile X tremor/ataxia syndrome (FXTAS) [MIM:300623]: In FXTAS, the expanded repeats range in size from 55 to 200 repeats and are referred to as 'premutations'. Full repeat expansions with greater than 200 repeats results in fragile X mental retardation syndrome [MIM:300624]. Carriers of the premutation typically do not show the full fragile X syndrome phenotype, but comprise a subgroup that may have some physical features of fragile X syndrome or mild cognitive and emotional problems. {ECO:0000269|PubMed:11445641}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Prematür Ovaryan Yetmezlik 10, 612885, Otozomal Resesif; POF10 (MCM8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 10, 612885, Autosomal recessive; POF10 (MCM8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MCM8	MCM8, POF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature ovarian failure 10 (POF10) [MIM:612885]: An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels of serum gonadotropins and low estradiol. [ECO:0000269	PubMed:25437880]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 11, 616946, Otozomal Dominant; POF11 (ERCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 11, 616946, Autosomal dominant; POF11 (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:9443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 12, 616947, Otozomal Resesif; POF12 (SYCE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 12, 616947, Autosomal recessive; POF12 (SYCE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYCE1	SYCE1, POF12, SPGF15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 2B, 300604; POF2B (POF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 2B, 300604; POF2B (POF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POF1B	FLJ22792, POF1B, POF2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature ovarian failure 2B (POF2B) [MIM:300604]: An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels of serum gonadotropins and low estradiol. [ECO:0000269	PubMed:16773570]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 3, 608996; POF3 (FOXL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 3, 608996; POF3 (FOXL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. {ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2}. Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 3, 608996; POF3 (MLPA)	Premature ovarian failure 3, 608996; POF3 (MLPA)	FOXL2	FOXL2, BPES, BPES1, PFRK, POF3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Blepharophimosis, ptosis, and epicanthus inversus syndrome (BPES) [MIM:110100]: A disorder characterized by eyelid dysplasia, small palpebral fissures, drooping eyelids and a skin fold curving in the mediolateral direction, inferior to the inner canthus. In type I BPSE (BPES1) eyelid abnormalities are associated with female infertility. Affected females show an ovarian deficit due to primary amenorrhea or to premature ovarian failure (POF). In type II BPSE (BPES2) affected individuals show only the eyelid defects. {ECO:0000269|PubMed:11175783, ECO:0000269|PubMed:11468277, ECO:0000269|PubMed:12400065, ECO:0000269|PubMed:12529855, ECO:0000269|PubMed:12630957, ECO:0000269|PubMed:12938087, ECO:0000269|PubMed:15257268, ECO:0000269|PubMed:16454982, ECO:0000269|PubMed:17089161, ECO:0000269|PubMed:18484667, ECO:0000269|PubMed:18642388, ECO:0000269|Ref.2}. Note=The disease is caused by mutations affecting the gene represented in this entry. There is a mutational hotspot in the region coding for the poly-Ala domain, since 30% of all mutations in the ORF lead to poly-Ala expansions, resulting mainly in BPES type II.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 4, 300510 (46,XX Gonadal Disgenezis) (BMP15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 4, 300510 (46,XX gonadal dysgenesis) (BMP15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMP15	BMP15, GDF9B, ODG2, POF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovarian dysgenesis 2 (ODG2) [MIM:300510]: A disorder characterized by lack of spontaneous pubertal development, primary amenorrhea, uterine hypoplasia, and hypergonadotropic hypogonadism as a result of streak gonads. [ECO:0000269	PubMed:15136966]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 5, 611548, Otozomal Dominant; POF5 (NOBOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 5, 611548, Autosomal dominant; POF5 (NOBOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOBOX	NOBOX, POF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 6, 612310; POF6 (FIGLA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 6, 612310; POF6 (FIGLA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIGLA	FIGLA, POF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature ovarian failure 6 (POF6) [MIM:612310]: An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels of serum gonadotropins and low estradiol. [ECO:0000269	PubMed:18499083]. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 7, 612964, Otozomal Dominant; POF7 (NR5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 7, 612964, Autosomal dominant; POF7 (NR5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. [ECO:0000269	PubMed:10383233]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 7, 612964, Otozomal Dominant; POF7 (MLPA)	Premature ovarian failure 7, 612964, Autosomal dominant; POF7 (MLPA)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, ADABP, POF7, SRXY3, SPGF8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. [ECO:0000269	PubMed:10383233]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 8, 615723, Otozomal Resesif; POF8 (STAG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 8, 615723, Autosomal recessive; POF8 (STAG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAG3	STAG3, POF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature ovarian failure 8 (POF8) [MIM:615723]: An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels of serum gonadotropins and low estradiol. {ECO:0000269|PubMed:24597867, ECO:0000303|PubMed:22428046}. Note=The disease is caused by mutations affecting the gene represented in this entry. A homozygous deletion in STAG3 predicted to result in frameshift and premature truncation, has been shown to be the cause of premature ovarian failure in a large consanguineous family.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik 9, 615724, Otozomal Resesif; POF9 (HFM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure 9, 615724, Autosomal recessive; POF9 (HFM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HFM1	HFM1, MER3, POF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prematür Ovaryan Yetmezlik, 300511 (DIAPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Premature ovarian failure, 300511 (DIAPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIAPH2	DIAPH2, DIA, POF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Premature ovarian failure 2A (POF2A) [MIM:300511]: An ovarian disorder defined as the cessation of ovarian function under the age of 40 years. It is characterized by oligomenorrhea or amenorrhea, in the presence of elevated levels of serum gonadotropins and low estradiol. {ECO:0000269	PubMed:9497258}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Preterm Prematüre Membran Rüptürü, Yatkınlık, 610504; PPROM (SERPINH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Preterm premature rupture of the membranes, susceptibility to, 610504; PPROM (SERPINH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINH1	SERPINH1, SERPINH2, PPROM, CBP2, CBP1, OI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteogenesis imperfecta 10 (OI10) [MIM:613848]: A form of osteogenesis imperfecta, a connective tissue disorder characterized by low bone mass, bone fragility and susceptibility to fractures after minimal trauma. Disease severity ranges from very mild forms without fractures to intrauterine fractures and perinatal lethality. Extraskeletal manifestations, which affect a variable number of patients, are dentinogenesis imperfecta, hearing loss, and blue sclerae. OI10 is an autosomal recessive form characterized by multiple bone deformities and fractures, generalized osteopenia, dentinogenesis imperfecta, and blue sclerae. {ECO:0000269	PubMed:20188343}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Primer Aldosteronizm, Nöbetler ve Nörolojik Anormallikler, 615474, Otozomal Dominant; PASNA (Aldosteron Üreten Adenoma - Nöbetler ve Nörolojik Anormallikler) (CACNA1D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Primary aldosteronism, seizures, and neurologic abnormalities, 615474, Autosomal dominant; PASNA (Aldosterone-producing adenoma with seizures and neurological abnormalities) (CACNA1D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1D	CACNA1D, CACNL1A2, CCHL1A2, SANDD, PASNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sinoatrial node dysfunction and deafness (SANDD) [MIM:614896]: A disease characterized by congenital severe to profound deafness without vestibular dysfunction, associated with episodic syncope due to intermittent pronounced bradycardia. {ECO:0000269	PubMed:21131953}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Primer Aldosteronizm, Nöbetler ve Nörolojik Anormallikler, 615474, Otozomal Dominant; PASNA (Aldosteron Üreten Adenoma - Nöbetler ve Nörolojik Anormallikler) (Prenatal) (CACNA1D geni) (Tüm Gen Dizi Analizi) (Prenatal)	Primary aldosteronism, seizures, and neurologic abnormalities, 615474, Autosomal dominant; PASNA (Aldosterone-producing adenoma with seizures and neurological abnormalities) (CACNA1D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CACNA1D	CACNA1D, CACNL1A2, CCHL1A2, SANDD, PASNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sinoatrial node dysfunction and deafness (SANDD) [MIM:614896]: A disease characterized by congenital severe to profound deafness without vestibular dysfunction, associated with episodic syncope due to intermittent pronounced bradycardia. {ECO:0000269	PubMed:21131953}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Primer Hiperoksalüri paneli (AGXT, GRHPR) (MLPA)	Primary hyperoxaluria panel (AGXT, GRHPR) (MLPA)	AGXT, GRHPR	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Primer Hiperoksalüri paneli (AGXT, GRHPR) (MLPA) (Prenatal)	Primary hyperoxaluria panel (AGXT, GRHPR) (MLPA) (Prenatal)	AGXT, GRHPR	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Primer Lateral Skleroz, Jüvenil, 606353, Otozomal Reseslf, PLSJ (Jüvenil Primer Lateral Skleroz) (ALS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Primary lateral sclerosis, juvenile, 606353, Autosomal recessive; PLSJ (Juvenile primary lateral sclerosis) (ALS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALS2	ALS2, ALSJ, PLSJ, IAHSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 2 (ALS2) [MIM:205100]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. (ECO:0000269	PubMed:11586298). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Primrose Sendromu, 259050, Otozomal Dominant; PRIMS (Entelektüel Yeterszilik-Katarakt-Kalsifiye Pirina-Miyopati Sendromu (Primrose Sendromu)) (ZBTB20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Primrose syndrome, 259050, Autosomal dominant; PRIMS (Intellectual disability-cataracts-calcified pinnae-myopathy syndrome) (ZBTB20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB20	ZBTB20, ZNF288, DPZF, PRIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primrose syndrome (PRIMS) [MIM:259050]: A disease characterized by macrocephaly, intellectual disability, disturbed behavior, dysmorphic facial features, ectopic calcifications, large calcified ear auricles, and progressive muscle wasting. (ECO:0000269	PubMed:25017102). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Primrose Sendromu, 259050, Otozomal Dominant; PRIMS (Entelektüel Yeterszilik-Katarakt-Kalsifiye Pirina-Miyopati Sendromu (Primrose Sendromu)) (Prenatal) (ZBTB20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Primrose syndrome, 259050, Autosomal dominant; PRIMS (Intellectual disability-cataracts-calcified pinnae-myopathy syndrome) (ZBTB20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZBTB20	ZBTB20, ZNF288, DPZF, PRIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Primrose syndrome (PRIMS) [MIM:259050]: A disease characterized by macrocephaly, intellectual disability, disturbed behavior, dysmorphic facial features, ectopic calcifications, large calcified ear auricles, and progressive muscle wasting. (ECO:0000269	PubMed:25017102). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prion Hastalığı, Uzun Süren, 606688, Otozomal Dominant (Sporadik Creutzfeldt-Jakob Hastalığı) (PRNP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prion disease with protracted course, 606688, Autosomal dominant (Sporadic Creutzfeldt-Jakob disease) (PRNP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prion Hastalığı, Uzun Süren, 606688, Otozomal Dominant (Sporadik Creutzfeldt-Jakob Hastalığı) (Prenatal) (PRNP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Prion disease with protracted course, 606688, Autosomal dominant (Sporadic Creutzfeldt-Jakob disease) (PRNP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRNP	PRNP, PRIP, KURU, CJD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progesteron Direnci, 264080, Otozomal Resesif (PGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progesterone resistance, 264080, Autosomal recessive (PGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PGR	PGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 2, 609283, Otozomal Dominant; PEOA2 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283, Autosomal dominant; PEOA2 (Autosomal dominant progressive external ophthalmoplegia) (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 2, 609283, Otozomal Dominant; PEOA2 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283, Autosomal dominant; PEOA2 (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 2, 609283, Otozomal Dominant; PEOA2 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (SLC25A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283, Autosomal dominant; PEOA2 (Autosomal dominant progressive external ophthalmoplegia) (SLC25A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 2, 609283, Otozomal Dominant; PEOA2 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 2, 609283, Autosomal dominant; PEOA2 (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	SLC25A4	SLC25A4, ANT1, T1, PEO3, PEO2, MTDPS12A, PEOA2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 2 (PEOA2) [MIM:609283]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269|PubMed:10926541, ECO:0000269|PubMed:11756613, ECO:0000269|PubMed:12112115, ECO:0000269|PubMed:15792871, ECO:0000269|PubMed:18575922). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 3, 609286, Otozomal Dominant; PEOA3 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (TWNK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286, Autosomal dominant; PEOA3 (Autosomal dominant progressive external ophthalmoplegia) (TWNK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 3, 609286, Otozomal Dominant; PEOA3 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286, Autosomal dominant; PEOA3 (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 3, 609286, Otozomal Dominant; PEOA3 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (TWNK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286, Autosomal dominant; PEOA3 (Autosomal dominant progressive external ophthalmoplegia) (TWNK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 3, 609286, Otozomal Dominant; PEOA3 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286, Autosomal dominant; PEOA3 (Autosomal dominant progressive external ophthalmoplegia) (Prenatal) (MLPA)	TWNK	C10orf2, TWINKLE, PEOA3, IOSCA, MTDPS7, PRLTS5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 4, 610131, Otozomal Dominant; PEOA4 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (POLG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131, Autosomal dominant; PEOA4 (Autosomal dominant progressive external ophthalmoplegia) (POLG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG2	POLG2, POLGB, PEOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4 (PEOA4) [MIM:610131]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269	PubMed:16695852). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Progresif Eksternal Oftalmopleji-Mitokondriyal DNA Delesyonlari, Otozomal Dominant 4, 610131, Otozomal Dominant; PEOA4 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (POLG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131, Autosomal dominant; PEOA4 (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	POLG2	POLG2, POLGB, PEOA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4 (PEOA4) [MIM:610131]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. (ECO:0000269	PubMed:16695852). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 4, 610131, Otozomal Dominant; PEOA4 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) ((POLG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131, Autosomal dominant; PEOA4 (Autosomal dominant progressive external ophthalmoplegia) (POLG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLG2	POLG2, POLGB, PEOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4 (PEOA4) [MIM:610131]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:16685452}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 4, 610131, Otozomal Dominant; PEOA4 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 4, 610131, Autosomal dominant; PEOA4 (Autosomal dominant progressive external ophthalmoplegia) (Prenatal) (MLPA)	POLG2	POLG2, POLGB, PEOA4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 4 (PEOA4) [MIM:610131]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:16685452}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 5, 613077, Otozomal Dominant; PEOA5 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (RRM2B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077, Autosomal dominant; PEOA5 (Autosomal dominant progressive external ophthalmoplegia) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPSBA, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPSBA) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 5, 613077, Otozomal Dominant; PEOA5 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077, Autosomal dominant; PEOA5 (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPSBA, MTDPS8B	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPSBA) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 5, 613077, Otozomal Dominant; PEOA5 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (RRM2B gen) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077, Autosomal dominant; PEOA5 (Autosomal dominant progressive external ophthalmoplegia) (RRM2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RRM2B	RRM2B, P53R2, PEOA5, MTDPSBA, MTDPS8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPSBA) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 5, 613077, Otozomal Dominant; PEOA5 (Otozomal Dominant Progresif Eksternal Oftalmopleji) (Prenatal) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 5, 613077, Autosomal dominant; PEOA5 (Autosomal dominant progressive external ophthalmoplegia) (Prenatal) (MLPA)	RRM2B	RRM2B, P53R2, PEOA5, MTDPSBA, MTDPS8B	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial DNA depletion syndrome 8A (MTDPSBA) [MIM:612075]: A disorder due to mitochondrial dysfunction characterized by various combinations of neonatal hypotonia, neurological deterioration, respiratory distress, lactic acidosis, and renal tubulopathy. {ECO:0000269|PubMed:17486094, ECO:0000269|PubMed:18504129}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 6, 615156, Otozomal Dominant; PEOA6 (Mitokondriyal DNA Delesyon Sendromu-Progresif Miyopati) (DNA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156, Autosomal dominant; PEOA6 (Mitochondrial DNA deletion syndrome with progressive myopathy) (DNA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNA2	DNA2, DNA2L, KIAA0083, PEOA6, SCKL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 (PEOA6) [MIM:615156]: A disorder characterized by muscle weakness, mainly affecting the lower limbs, external ophthalmoplegia, exercise intolerance, and mitochondrial DNA deletions on muscle biopsy. Symptoms may appear in childhood or adulthood and show slow progression. {ECO:0000269|PubMed:23352259}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 6, 615156, Otozomal Dominant; PEOA6 (Mitokondriyal DNA Delesyon Sendromu-Progresif Miyopati) (Prenatal) (DNA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 6, 615156, Autosomal dominant; PEOA6 (Mitochondrial DNA deletion syndrome with progressive myopathy) (DNA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNA2	DNA2, DNA2L, KIAA0083, PEOA6, SCKL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 (PEOA6) [MIM:615156]: A disorder characterized by muscle weakness, mainly affecting the lower limbs, external ophthalmoplegia, exercise intolerance, and mitochondrial DNA deletions on muscle biopsy. Symptoms may appear in childhood or adulthood and show slow progression. {ECO:0000269|PubMed:23352259}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 2, 616479, Otozomal Resesif; PEOB2 (Yetişkin Başlangıçlı Kronik Progresif Eksternal Oftalmopleji- Mitokondriyal Miyopati) (RNASEH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479, Autosomal recessive; PEOB2 (Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy) (RNASEH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASEH1	RNASEH1, PEOB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 2, 616479, Otozomal Resesif; PEOB2 (Yetişkin Başlangıçlı Kronik Progresif Eksternal Oftalmopleji- Mitokondriyal Miyopati) (Prenatal) (RNASEH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 2, 616479, Autosomal recessive; PEOB2 (Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy) (RNASEH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNASEH1	RNASEH1, PEOB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 3, 617069, Otozomal Resesif (Otozomal Resesif Progresif Eksternal Oftalmopleji) (TK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069, Autosomal recessive (Autosomal recessive progressive external ophthalmoplegia) (TK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TK2	TK2, MTDPS2, PEOB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Delesyonlari- Progresif Eksternal Oftalmopleji, Otozomal Resesif 3, 617069, Otozomal Resesif (Otozomal Resesif Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069, Autosomal recessive (Autosomal recessive progressive external ophthalmoplegia) (MLPA)	TK2	TK2, MTDPS2, PEOB3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Mitokondriyal DNA Delesyonlari- Progresif Eksternal Oftalmopleji, Otozomal Resesif 3, 617069, Otozomal Resesif (Otozomal Resesif Progresif Eksternal Oftalmopleji) (Prenatal) (TK2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069, Autosomal recessive (Autosomal recessive progressive external ophthalmoplegia) (TK2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TK2	TK2, MTDPS2, PEOB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Mitokondriyal DNA Delesyonlari- Progresif Eksternal Oftalmopleji, Otozomal Resesif 3, 617069, Otozomal Resesif (Otozomal Resesif Progresif Eksternal Oftalmopleji) (Prenatal) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 3, 617069, Autosomal recessive (Autosomal recessive progressive external ophthalmoplegia) (Prenatal) (MLPA)	TK2	TK2, MTDPS2, PEOB3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Dominant 4, 617070, Otozomal Resesif (Yetişkin Başlangıçlı Multipl Mitokondriyal DNA Delesyonsendromu, DGUOK Eksikliğine Bağlı) (DGUOK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070, Autosomal recessive (Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 4, 617070, Otozomal Resesif (Yetişkin Başlangıçlı Multipl Mitokondriyal DNA Delesyonsendromu, DGUOK Eksikliğine Bağlı) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070, Autosomal recessive (Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 4, 617070, Otozomal Resesif (Yetişkin Başlangıçlı Multipl Mitokondriyal DNA Delesyonsendromu, DGUOK Eksikliğine Bağlı) (Prenatal) (DGUOK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070, Autosomal recessive (Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency) (DGUOK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji- Mitokondriyal DNA Delesyonlari, Otozomal Resesif 4, 617070, Otozomal Resesif (Yetişkin Başlangıçlı Multipl Mitokondriyal DNA Delesyonsendromu, DGUOK Eksikliğine Bağlı) (Prenatal) (MLPA)	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal recessive 4, 617070, Autosomal recessive (Adult-onset multiple mitochondrial DNA deletion syndrome due to DGUOK deficiency) (Prenatal) (MLPA)	DGUOK	DGUOK, DGK, MTDPS3, PEOB4, NCPH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Progresif Eksternal Oftalmopleji, Otozomal Dominant 1, 157640, Otozomal Dominant (Otozomal Dominant Progresif Eksternal Oftalmopleji) (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia, autosomal dominant 1, 157640, Autosomal dominant (Autosomal dominant progressive external ophthalmoplegia) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Progresif Eksternal Oftalmopleji, Otozomal Dominant 1, 157640, Otozomal Dominant (Otozomal Dominant Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia, autosomal dominant 1, 157640, Autosomal dominant (Autosomal dominant progressive external ophthalmoplegia) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji, Otozomal Resesif 1, 258450, Otozomal Resesif; PFHB1A (Otozomal Resesif Progresif Eksternal Oftalmopleji (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive external ophthalmoplegia, autosomal recessive 1, 258450, Autosomal recessive; PFHB1A (Autosomal recessive progressive external ophthalmoplegia) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Eksternal Oftalmopleji, Otozomal Resesif 1, 258450, Otozomal Resesif; PFHB1A (Otozomal Resesif Progresif Eksternal Oftalmopleji) (MLPA)	Progressive external ophthalmoplegia, autosomal recessive 1, 258450, Autosomal recessive; PFHB1A (Autosomal recessive progressive external ophthalmoplegia) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Progresif Ailesel Kalp Boğu, Tip 1B, 604559, Otozomal Dominant; PFHB1B (Ailesel Progresif Kardiyak İletim Defekti) (TRPM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Progressive familial heart block, type IB, 604559, Autosomal dominant; PFHB1B (Familial progressive cardiac conduction defect) (TRPM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPM4	TRPM4, PFHB1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive familial heart block 1B (PFHB1B) [MIM:604559]: A cardiac bundle branch disorder characterized by progressive alteration of cardiac conduction through the His-Purkinje system, with a pattern of a right bundle-branch block and/or left anterior hemiblock occurring individually or together. It leads to complete atrio-ventricular block causing syncope and sudden death. {ECO:0000269|PubMed:19726882, ECO:0000269|PubMed:20562447, ECO:0000269|PubMed:21887725}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Proguanil Yavaş Metabolizma, 609535, Otozomal Resesif (Klopidogrel Direnci) (CYP2C19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proguanil poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (CYP2C19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C19	CYP2C, CYP2C19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Proguanil Yavaş Metabolizma, 609535, Otozomal Resesif (Klopidogrel Direnci) (MLPA)	Proguanil poor metabolizer, 609535, Autosomal recessive (Resistance to clopidogrel) (MLPA)	CYP2C19	CYP2C, CYP2C19	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Prolidaz Eksikliği, 170100, Otozomal Resesif (Prolidaz Eksikliği) (PEPD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prolidase deficiency, 170100, Autosomal recessive (Prolidase deficiency) (PEPD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEPD	PEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prolidase deficiency (PD) [MIM:170100]: A multisystem disorder associated with massive iminodipeptiduria and lack of or reduced prolidase activity in erythrocytes, leukocytes, or cultured fibroblasts. Clinical features include skin ulcers, developmental delay, recurrent infections, and a characteristic facies. {ECO:0000269|PubMed:12384772, ECO:0000269|PubMed:2365824, ECO:0000269|PubMed:8198124, ECO:0000269|PubMed:8900231}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Prolidaz Eksikliği, 170100, Otozomal Resesif (Prolidaz Eksikliği) (Prenatal) (PEPD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Prolidase deficiency, 170100, Autosomal recessive (Prolidase deficiency) (PEPD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEPD	PEPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Prolidase deficiency (PD) [MIM:170100]: A multisystem disorder associated with massive iminodipeptiduria and lack of or reduced prolidase activity in erythrocytes, leukocytes, or cultured fibroblasts. Clinical features include skin ulcers, developmental delay, recurrent infections, and a characteristic facies. {ECO:0000269|PubMed:12384772, ECO:0000269|PubMed:2365824, ECO:0000269|PubMed:8198124, ECO:0000269|PubMed:8900231}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Proliferatif Vaskülopati ve Hydrancephaly-Hidrosefali Sendromu, 225790, Otozomal Resesif; PVHH (Fowler Sendromu) (FLVCR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790, Autosomal recessive; PVHH (Fowler syndrome) (FLVCR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLVCR2	FLVCR2, C14orf59, CCT, PVHH, EPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Proliferatif Vaskülopati ve Hydrancephaly-Hidrosefali Sendromu, 225790, Otozomal Resesif; PVHH (Fowler Sendromu) (Prenatal) (FLVCR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome, 225790, Autosomal recessive; PVHH (Fowler syndrome) (FLVCR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLVCR2	FLVCR2, C14orf59, CCT, PVHH, EPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Uzamış Elektroretinal Cevap Supresyonu; Perrs (Bradyopsi) (RGS9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROLONGED ELECTRORETINAL RESPONSE SUPPRESSION, PERRS (Bradyopsia) (RGS9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RGS9	RGS9, PERRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prolonged electroretinal response suppression (PERRS) [MIM:609415]: Characterized by difficulty adjusting to sudden changes in luminance levels mediated by cones. {ECO:0000269	PubMed:14702087}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Proopiomelanokortin Eksikliği (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (POMC geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROOPIOMELANOCORTIN DEFICIENCY (Obesity due to pro-opiomelanocortin deficiency) (POMC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMC	POMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. {ECO:0000269	PubMed:12165561}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Proopiomelanokortin Eksikliği (Obezite, Proopiyomelanokortin Eksikliğine Bağlı) (MLPA)	PROOPIOMELANOCORTIN DEFICIENCY (Obesity due to pro-opiomelanocortin deficiency) (MLPA)	POMC	POMC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Obesity (OBESITY) [MIM:601665]: A condition characterized by an increase of body weight beyond the limitation of skeletal and physical requirements, as the result of excessive accumulation of body fat. {ECO:0000269	PubMed:12165561}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Properdin Eksikliği, X'e Bağlı, 312060, X'e Bağlı; CFPD (Properdin Eksikliği) (CFP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Properdin deficiency, X-linked, 312060, X-linked recessive; CFPD (Properdin deficiency) (CFP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFP	PFC, PFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Properdin deficiency (PFD) [MIM:312060]: Results in higher susceptibility to bacterial infections; especially to meningococcal infections. Three phenotypes have been reported: complete deficiency (type I), incomplete deficiency (type II), and dysfunction of properdin (type III). {ECO:0000269|PubMed:10909851, ECO:0000269|PubMed:8871668, ECO:0000269|PubMed:9710744}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (PCCB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (PCCB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCCB	PCCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Propionic acidemia type I (PA-2) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10447268, ECO:0000269|PubMed:12189489, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621, ECO:0000269|PubMed:2154743, ECO:0000269|PubMed:8411997, ECO:0000269|PubMed:9683601}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (PCCA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (PCCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCCA	PCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Propionic acidemia type I (PA-1) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10101253, ECO:0000269|PubMed:10329019, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (MLPA)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (MLPA)	PCCA	PCCA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Propionic acidemia type I (PA-1) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10101253, ECO:0000269|PubMed:10329019, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (Prenatal) (PCCB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (PCCB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCCB	PCCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Propionic acidemia type II (PA-2) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10447268, ECO:0000269|PubMed:12189489, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621, ECO:0000269|PubMed:2154743, ECO:0000269|PubMed:8411997, ECO:0000269|PubMed:9683601}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (Prenatal) (PCCA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (PCCA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCCA	PCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Propionic acidemia type I (PA-1) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10101253, ECO:0000269|PubMed:10329019, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Propiyonik Asidemi, 606054, Otozomal Resesif (Propiyonik Asidemi) (Prenatal) (MLPA)	Propionicacidemia, 606054, Autosomal recessive (Propionic acidemia) (Prenatal) (MLPA)	PCCA	PCCA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Propionic acidemia type I (PA-1) [MIM:606054]: Life-threatening disease characterized by episodic vomiting, lethargy and ketosis, neutropenia, periodic thrombocytopenia, hypogammaglobulinemia, developmental retardation, and intolerance to protein. {ECO:0000269|PubMed:10101253, ECO:0000269|PubMed:10329019, ECO:0000269|PubMed:12559849, ECO:0000269|PubMed:15059621}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Proprotein Konvertaz 1/3 Eksikliği (Obezite, Prohormon Konvertaz 1 Eksikliğine Bağlı) (PCSK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROPROTEIN CONVERTASE 1/3 DEFICIENCY (Obesity due to prohormone convertase I deficiency) (PCSK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCSK1	PCSK1, NEC1, PC1, PC3, BMIQ12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Proprotein convertase 1 deficiency (PC1 deficiency) [MIM:600955]: Characterized by obesity, hypogonadism, hypoadrenalism, reactive hypoglycemia as well as marked small-intestinal absorptive dysfunction It is due to impaired processing of prohormones. {ECO:0000269|PubMed:14617756, ECO:0000269|PubMed:17595246, ECO:0000269|PubMed:9207799}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Prostat Kanseri 1, 601518, Otozomal Dominant; HPC1 (Ailesel Prostat Kanseri) (RNASEL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer 1, 601518, Autosomal dominant; HPC1 (Familial prostate cancer) (RNASEL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNASEL	RNASEL, RNS4, PRCA1, HPC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prostate cancer, hereditary, 1 (HPC1) [MIM:601518]: A condition associated with familial predisposition to cancer of the prostate. Most prostate cancers are adenocarcinomas that develop in the acini of the prostatic ducts. Other rare histopathologic types of prostate cancer that occur in approximately 5% of patients include small cell carcinoma, mucinous carcinoma, prostatic ductal carcinoma, transitional cell carcinoma, squamous cell carcinoma, basal cell carcinoma, adenoid cystic carcinoma (basaloid), signet-ring cell carcinoma and neuroendocrine carcinoma. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000305|PubMed:11799394, ECO:0000305|PubMed:11941539, ECO:0000305|PubMed:12415269, ECO:0000305|PubMed:17344846}.;	EDTA Blood Sample
Prostat Kanseri, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (BRCA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, 176807, Autosomal dominant (Familial prostate cancer) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Prostat Kanseri, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (MLPA)	Prostate cancer, 176807, Autosomal dominant (Familial prostate cancer) (MLPA)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:10399947, ECO:0000269|PubMed:10978364, ECO:0000269|PubMed:11139248, ECO:0000269|PubMed:11241844, ECO:0000269|PubMed:11948477, ECO:0000269|PubMed:12145750, ECO:0000269|PubMed:12373604, ECO:0000269|PubMed:12442274, ECO:0000269|PubMed:12442275, ECO:0000269|PubMed:12938098, ECO:0000269|PubMed:14722926, ECO:0000269|PubMed:15026808, ECO:0000269|PubMed:15172753, ECO:0000269|PubMed:15365993, ECO:0000269|PubMed:8640237, ECO:0000269|PubMed:9150152, ECO:0000269|PubMed:9609997, ECO:0000269|PubMed:9654203, ECO:0000269|PubMed:9971877}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Prostat Kanseri, Ailesel, Yatkınlık, 176807, Otozomal Dominant (CHEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, familial, susceptibility to, 176807, Autosomal dominant (CHEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Prostat Kanseri, Ailesel, Yatkınlık, 176807, Otozomal Dominant (MLPA)	Prostate cancer, familial, susceptibility to, 176807, Autosomal dominant (MLPA)	CHEK2	CHEK2, RAD53, CHK2, CDS1, LFS2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269|PubMed:12094328, ECO:0000269|PubMed:21618645, ECO:0000269|PubMed:25619829}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269|PubMed:12094328}.;	EDTA Blood Sample
Prostat Kanseri, Kalıtsal, 12, 611868; HPC12 (Ailesel Prostat Kanseri) (EHBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, hereditary, 12, 611868; HPC12 (Familial prostate cancer) (EHBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EHBP1	EHBP1, KIAA0903, HPC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prostate cancer, hereditary, 12 [HPC12] [MIM:611868]: A condition associated with familial predisposition to cancer of the prostate. Most prostate cancers are adenocarcinomas that develop in the acini of the prostatic ducts. Other rare histopathologic types of prostate cancer that occur in approximately 5% of patients include small cell carcinoma, mucinous carcinoma, prostatic ductal carcinoma, transitional cell carcinoma, squamous cell carcinoma, basal cell carcinoma, adenoid cystic carcinoma (basaloid), signet-ring cell carcinoma and neuroendocrine carcinoma. {ECO:0000269	PubMed:18264098}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Prostat Kanseri, Kalıtsal, 13, 611928; HPC13 (Ailesel Prostat Kanseri) (MSMB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, hereditary, 13, 611928; HPC13 (Familial prostate cancer) (MSMB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSMB	MSMB, HPC13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prostat Kanseri, Kalıtsal, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (MSR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, hereditary, 176807, Autosomal dominant (Familial prostate cancer) (MSR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSR1	MSR1, SCARA1, SRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Barrett esophagus (BE) [MIM:614266]: A condition characterized by a metaplastic change in which normal esophageal squamous epithelium is replaced by a columnar and intestinal-type epithelium. Patients with Barrett esophagus have an increased risk of esophageal adenocarcinoma. The main cause of Barrett esophagus is gastroesophageal reflux. The retrograde movement of acid and bile salts from the stomach into the esophagus causes prolonged injury to the esophageal epithelium and induces chronic esophagitis, which in turn is believed to trigger the pathologic changes. {ECO:0000269	PubMed:21791690}. Note=The disease may be caused by mutations affecting the gene represented in this entry. Genetic variants in MSR1 have been found in individuals with Barrett esophagus and are thought to contribute to disease susceptibility.;	EDTA Blood Sample

Prostat Kanseri, Ailesel, 2, Yatkınlık, 614731; HPC2 (Ailesel Prostat Kanseri) (ELAC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, hereditary, 2, susceptibility to, 614731; HPC2 (Familial prostate cancer) (ELAC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELAC2	ELAC2, HPC2, COXPD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 17 (COXPD17) [MIM:615440]: An autosomal recessive disorder of mitochondrial dysfunction characterized by onset of severe hypertrophic cardiomyopathy in the first year of life. Other features include hypotonia, poor growth, lactic acidosis, and failure to thrive. The disorder may be fatal in early childhood. {ECO:0000269	PubMed:23849775}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Prostat Kanseri, Progresyon, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (HIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, progression of, 176807, Autosomal dominant (Familial prostate cancer) (HIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HIP1	HIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving HIP1 is found in a form of chronic myelomonocytic leukemia (CMML). Translocation t(5;7)(q33;q11.2) with PDGFRB (PubMed:9616134). The chimeric HIP1- PDGFRB transcript results from an in-frame fusion of the two genes (PubMed:9616134). The reciprocal PDGFRB-HIP1 transcript is not expressed (PubMed:9616134). {ECO:0000269	PubMed:9616134}.	EDTA Blood Sample
Prostat Kanseri, Somatik, 176807 (MAD1L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, somatic, 176807 (MAD1L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAD1L1	MAD1L1, TXBP181	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in MAD1L1 are involved in the development and/or progression of various types of cancer. {ECO:0000269|PubMed:10597320, ECO:0000269|PubMed:11423978}.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Prostat Kanseri, Somatik, 176807 (KLF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, somatic, 176807 (KLF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLF6	KLF6, COPEB, BCD1, ZF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Prostate cancer (PC) [MIM:176807]: A malignancy originating in tissues of the prostate. Most prostate cancers are adenocarcinomas that develop in the acini of the prostatic ducts. Other rare histopathologic types of prostate cancer that occur in approximately 5% of patients include small cell carcinoma, mucinous carcinoma, prostatic ductal carcinoma, transitional cell carcinoma, squamous cell carcinoma, basal cell carcinoma, adenoid cystic carcinoma (basaloid), signet-ring cell carcinoma and neuroendocrine carcinoma. {ECO:0000269|PubMed:11752579}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Prostat Kanseri, Yatkınlık, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (MX01 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, susceptibility to, 176807, Autosomal dominant (Familial prostate cancer) (MX01 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MX01	MX01	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prostat Kanseri, Yatkınlık, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (CDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, susceptibility to, 176807, Autosomal dominant (Familial prostate cancer) (CDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH1	CDH1, UVO, LCAM, ECAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hereditary diffuse gastric cancer (HDGC) [MIM:137215]: A cancer predisposition syndrome with increased susceptibility to diffuse gastric cancer. Diffuse gastric cancer is a malignant disease characterized by poorly differentiated infiltrating lesions resulting in thickening of the stomach. Malignant tumors start in the stomach, can spread to the esophagus or the small intestine, and can extend through the stomach wall to nearby lymph nodes and organs. It also can metastasize to other parts of the body. {ECO:0000269|PubMed:10319582, ECO:0000269|PubMed:12216071}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Heterozygous CDH1 germline mutations are responsible for familial cases of diffuse gastric cancer. Somatic mutations has also been found in patients with sporadic diffuse gastric cancer and lobular breast cancer.	EDTA Blood Sample
Prostat Kanseri, Yatkınlık, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (CD82 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, susceptibility to, 176807, Autosomal dominant (Familial prostate cancer) (CD82 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD82	CD82, SAR2, KAI1, ST6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Prostat Kanseri, Yatkınlık, 176807, Otozomal Dominant (Ailesel Prostat Kanseri) (AR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, susceptibility to, 176807, Autosomal dominant (Familial prostate cancer) (AR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end- organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022459, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7619368	EDTA Blood Sample
Prostat Kanseri, Yatkınlık, Somatik, 176807 (ZFHX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer, susceptibility to, somatic, 176807 (ZFHX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFHX3	ZFHX3, ATBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Prostat Kanseri / Beyin Kanseri Yatkınlık, Somatik, 603688 (EPHB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prostate cancer/brain cancer susceptibility, somatic, 603688 (EPHB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPHB2	EPHB2, EPHT3, DRT, ERK, PCBC, CAPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
PROTEİN KİNASE, AMP-AKTİVE EDİCİ, NONKATALİTİK, Gama-3; PRKAG3 (PRKAG3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, GAMMA-3; PRKAG3 (PRKAG3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAG3	PRKAG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Proteinüri, Düşük Molekül Ağırlıklı, Hiperkalsiüri ve Nefrokalsinozis (Dent Hastalığı) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROTEINURIA, LOW MOLECULAR WEIGHT, WITH HYPERCALCIURIA AND NEPHROCALCINOSIS (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. {ECO:0000269|PubMed:15086899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8559248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Proteinüri, Düşük Molekül Ağırlıklı, Hiperkalsiüri Nefrokalsinoz, 308990, X'ke Bağlı (Dent Hastalığı) (CLCN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis, 308990, X-linked recessive (Dent disease) (CLCN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLCN5	CLCN5, CLCK2, NPHL2, DENTS, NPHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephrolithiasis 2 (NPHL2) [MIM:300009]: An X-linked recessive renal disease belonging to the 'Dent disease complex', a group of disorders characterized by proximal renal tubular defect, hypercalciuria, nephrocalcinosis, and renal insufficiency. The spectrum of phenotypic features is remarkably similar in the various disorders, except for differences in the severity of bone deformities and renal impairment. Nephrolithiasis type 2 patients manifest hypercalciuria, hypophosphatemia, aminoaciduria, nephrocalcinosis and nephrolithiasis, renal insufficiency leading to renal failure in adulthood, rickets (33% of patients) and osteomalacia. {ECO:0000269|PubMed:15086899, ECO:0000269|PubMed:16247550, ECO:0000269|PubMed:16416111, ECO:0000269|PubMed:16822791, ECO:0000269|PubMed:17262170, ECO:0000269|PubMed:19657328, ECO:0000269|PubMed:8559248, ECO:0000269|PubMed:9187673, ECO:0000269|PubMed:9259268, ECO:0000269|PubMed:9602200, ECO:0000269|PubMed:9853249}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Proteus Sendromu, Somatik, 176920 (Proteus Sendromu) (AKT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proteus syndrome, somatic, 176920 (Proteus syndrome) (AKT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Protrombin Eksikliği, Konjenital (Konjenital Faktör 2 Eksikliği) (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	PROTHROMBIN DEFICIENCY, CONGENITAL (Congenital factor II deficiency) (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to thrombin defect (THPH1) [MIM:188050]: A multifactorial disorder of hemostasis characterized by abnormal platelet aggregation in response to various agents and recurrent thrombi formation. {ECO:0000269|PubMed:2825773}. Note=The disease is caused by mutations affecting the gene represented in this entry. A common genetic variation in the 3-prime untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increased risk of venous thrombosis.	EDTA Blood Sample

Protoporfiria, Eritropoietik, Otozomal Resesif, 177000, Otozomal Resesif; EPP (Otozomal Eritropoetik Protoporfiri) (FECH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Protoporphyria, erythropoietic, autosomal recessive, 177000, Autosomal recessive; EPP (Autosomal erythropoietic protoporphyria) (FECH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FECH	FECH, FCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Erythropoietic protoporphyria is marked by excessive protoporphyrin in erythrocytes, plasma, liver and feces, and by widely varying photosensitive skin changes ranging from a burning or pruritic sensation to erythema, edema and wheals. {ECO:0000269|PubMed:10942404, ECO:0000269|PubMed:11375302, ECO:0000269|PubMed:12063482, ECO:0000269|PubMed:12601550, ECO:0000269|PubMed:1376018, ECO:0000269|PubMed:15286165, ECO:0000269|PubMed:17196862, ECO:0000269|PubMed:1755842, ECO:0000269|PubMed:7910885, ECO:0000269|PubMed:8757534, ECO:0000269|PubMed:9585598, ECO:0000269|PubMed:9740232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Protoporfiria, Eritropoietik, Otozomal Resesif, 177000, Otozomal Resesif; EPP (Otozomal Eritropoetik Protoporfiri) (MLPA)	Protoporphyria, erythropoietic, autosomal recessive, 177000, Autosomal recessive; EPP (Autosomal erythropoietic protoporphyria) (MLPA)	FECH	FECH, FCE	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Erythropoietic protoporphyria is marked by excessive protoporphyrin in erythrocytes, plasma, liver and feces, and by widely varying photosensitive skin changes ranging from a burning or pruritic sensation to erythema, edema and wheals. {ECO:0000269|PubMed:10942404, ECO:0000269|PubMed:11375302, ECO:0000269|PubMed:12063482, ECO:0000269|PubMed:12601550, ECO:0000269|PubMed:1376018, ECO:0000269|PubMed:15286165, ECO:0000269|PubMed:17196862, ECO:0000269|PubMed:1755842, ECO:0000269|PubMed:7910885, ECO:0000269|PubMed:8757534, ECO:0000269|PubMed:9585598, ECO:0000269|PubMed:9740232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Protoporfiria, Eritropoietik, Otozomal Resesif, 177000, Otozomal Resesif; EPP (Otozomal Eritropoetik Protoporfiri) (Prenatal) (FECH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Protoporphyria, erythropoietic, autosomal recessive, 177000, Autosomal recessive; EPP (Autosomal erythropoietic protoporphyria) (FECH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FECH	FECH, FCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Erythropoietic protoporphyria is marked by excessive protoporphyrin in erythrocytes, plasma, liver and feces, and by widely varying photosensitive skin changes ranging from a burning or pruritic sensation to erythema, edema and wheals. {ECO:0000269|PubMed:10942404, ECO:0000269|PubMed:11375302, ECO:0000269|PubMed:12063482, ECO:0000269|PubMed:12601550, ECO:0000269|PubMed:1376018, ECO:0000269|PubMed:15286165, ECO:0000269|PubMed:17196862, ECO:0000269|PubMed:1755842, ECO:0000269|PubMed:7910885, ECO:0000269|PubMed:8757534, ECO:0000269|PubMed:9585598, ECO:0000269|PubMed:9740232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Protoporfiria, Eritropoietik, Otozomal Resesif, 177000, Otozomal Resesif; EPP (Otozomal Eritropoetik Protoporfiri) (Prenatal) (MLPA)	Protoporphyria, erythropoietic, autosomal recessive, 177000, Autosomal recessive; EPP (Autosomal erythropoietic protoporphyria) (Prenatal) (MLPA)	FECH	FECH, FCE	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Erythropoietic protoporphyria (EPP) [MIM:177000]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. Erythropoietic protoporphyria is marked by excessive protoporphyrin in erythrocytes, plasma, liver and feces, and by widely varying photosensitive skin changes ranging from a burning or pruritic sensation to erythema, edema and wheals. {ECO:0000269|PubMed:10942404, ECO:0000269|PubMed:11375302, ECO:0000269|PubMed:12063482, ECO:0000269|PubMed:12601550, ECO:0000269|PubMed:1376018, ECO:0000269|PubMed:15286165, ECO:0000269|PubMed:17196862, ECO:0000269|PubMed:1755842, ECO:0000269|PubMed:7910885, ECO:0000269|PubMed:8757534, ECO:0000269|PubMed:9585598, ECO:0000269|PubMed:9740232}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Protoporfiria, Eritropoietik, X'e Bağlı, 300752, X'e Bağlı Dominant; XLEPP (X'e Bağlı Eritropoietik Protoporfiri) (ALAS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Protoporphyria, erythropoietic, X-linked, 300752, X-linked dominant; XLEPP (X-linked erythropoietic protoporphyria) (ALAS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALAS2	ALAS2, ANH1, ASB, XLEPP, XLSA, ANH1, SIDBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Erythropoietic protoporphyria, X-linked dominant (XLDPT) [MIM:300752]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. XLDPT is characterized biochemically by a high proportion of zinc-protoporphyrin in erythrocytes, in which a mismatch between protoporphyrin production and the heme requirement of differentiating erythroid cells leads to overproduction of protoporphyrin in amounts sufficient to cause photosensitivity and liver disease. {ECO:0000269|PubMed:18760763}. Note=The disease is caused by mutations affecting the gene represented in this entry. Gain of function mutations in ALS2 are responsible for XLDPT, but they can also be a possible aggravating factor in congenital erythropoietic porphyria and other erythropoietic disorders caused by mutations in other genes (PubMed:21309041). {ECO:0000269|PubMed:21309041}.;	EDTA Blood Sample
Protoporfiria, Eritropoietik, X'e Bağlı, 300752, X'e Bağlı Dominant; XLEPP (X'e Bağlı Eritropoietik Protoporfiri) (Prenatal) (ALAS2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Protoporphyria, erythropoietic, X-linked, 300752, X-linked dominant; XLEPP (X-linked erythropoietic protoporphyria) (ALAS2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALAS2	ALAS2, ANH1, ASB, XLEPP, XLSA, ANH1, SIDBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Erythropoietic protoporphyria, X-linked dominant (XLDPT) [MIM:300752]: A form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. XLDPT is characterized biochemically by a high proportion of zinc-protoporphyrin in erythrocytes, in which a mismatch between protoporphyrin production and the heme requirement of differentiating erythroid cells leads to overproduction of protoporphyrin in amounts sufficient to cause photosensitivity and liver disease. {ECO:0000269|PubMed:18760763}. Note=The disease is caused by mutations affecting the gene represented in this entry. Gain of function mutations in ALS2 are responsible for XLDPT, but they can also be a possible aggravating factor in congenital erythropoietic porphyria and other erythropoietic disorders caused by mutations in other genes (PubMed:21309041). {ECO:0000269|PubMed:21309041}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Protrombin 20210 Mutasyonu (20120G>A) (. geni) (Dizi Analizi) (Postnatal)	Protrombin 20210 mutation (20120G>A) (. gene) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Proud Sendromu, 300004, X'e Bağlı (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Genitalya Sendromu (Proud Sendromu)) (ARX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proud syndrome, 300004, X-linked (Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Proud Sendromu, 300004, X'e Bağlı (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Genitalya Sendromu (Proud Sendromu)) (MLPA)	Proud syndrome, 300004, X-linked (Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269|PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Proud Sendromu, 300004, X'e Bağlı (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Genitalya Sendromu (Proud Sendromu)) (Prenatal) (ARX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Proud syndrome, 300004, X-linked (Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome) (ARX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Proud Sendromu, 300004, X'e Bağlı (Mikrosefali-Korpus Kallozum Agenezisi-Anormal Genitalya Sendromu (Proud Sendromu)) (Prenatal) (MLPA)	Proud syndrome, 300004, X-linked (Microcephaly-corpus callosum agenesis-abnormal genitalia syndrome) (Prenatal) (MLPA)	ARX	ARX, ISSX, PRTS, MRXS1, MRX36, MRXS4, MRX43, MRX87, MRX29, MRX32	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Agenesis of the corpus callosum, with abnormal genitalia (ACCAG) [MIM:300004]: A X-linked syndrome with variable expression in females. It is characterized by agenesis of corpus callosum, mental retardation and seizures. Manifestations in surviving males include severe acquired microencephaly, mental retardation, limb contractures, scoliosis, tapered fingers with hyperconvex nails, a characteristic face with large eyes, prominent supraorbital ridges, synophrys, optic atrophy, broad alveolar ridges, and seizures. Urologic anomalies include renal dysplasia, cryptorchidism, and hypospadias. {ECO:0000269	PubMed:14722918}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Proksimal Miyopati ve Oftalmopleji, 605637, Otozomal Resesif, Otozomal Dominant; MYPOP (Çocukluk Çağ Başlangıçlı Otozomal Resesif Miyopati-Eksternal Oftalmopleji (MYH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Proximal myopathy and ophthalmoplegia, 605637, Autosomal recessive, Autosomal dominant; MYPOP (Childhood-onset autosomal recessive myopathy with external ophthalmoplegia) (MYH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH2	MYH2, MYPOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, proximal, and ophthalmoplegia (MYPOP) [MIM:605637]: A muscular disorder characterized by mild-to-moderate muscle weakness, ophthalmoplegia, and contractures at birth in some patients. Muscle biopsies from patients show predominance of type 1 fibers and small or absent type 2A fibers. The disease is non-progressive or it progresses very slowly. Inheritance is autosomal dominant or recessive. {ECO:0000269	PubMed:11114175}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Proksimal Miyopati ve Oftalmopleji, 605637, Otozomal Resesif, Otozomal Dominant; MYPOP (Çocukluk Çağ Başlangıçlı Otozomal Resesif Miyopati-Eksternal Oftalmopleji) (Prenatal) (MYH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Proximal myopathy and ophthalmoplegia, 605637, Autosomal recessive, Autosomal dominant; MYPOP (Childhood-onset autosomal recessive myopathy with external ophthalmoplegia) (MYH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH2	MYH2, MYPOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, proximal, and ophthalmoplegia (MYPOP) [MIM:605637]: A muscular disorder characterized by mild-to-moderate muscle weakness, ophthalmoplegia, and contractures at birth in some patients. Muscle biopsies from patients show predominance of type 1 fibers and small or absent type 2A fibers. The disease is non-progressive or it progresses very slowly. Inheritance is autosomal dominant or recessive. {ECO:0000269	PubMed:11114175}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Prune Belly Sendromu, 100100, Otozomal Resesif, PBS (Prune Belly Sendromu) (CHRM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Prune belly syndrome, 100100, Autosomal recessive; PBS (Prune belly syndrome) (CHRM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRM3	CHRM3, PBS, EGBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Prune belly syndrome (PBS) [MIM:100100]: A syndrome characterized by thin abdominal musculature with overlying lax skin, cryptorchism, megacystis with disorganized detrusor muscle, and urinary tract abnormalities. {ECO:0000269	PubMed:22079972}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Prune Belly Sendromu, 100100, Otozomal Resesif, PBS (Prune Belly Sendromu) (Prenatal) (CHRM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Prune belly syndrome, 100100, Autosomal recessive; PBS (Prune belly syndrome) (CHRM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHRM3	CHRM3, PBS, EGBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Prune belly syndrome (PBS) [MIM:100100]: A syndrome characterized by thin abdominal musculature with overlying lax skin, cryptorchism, megacystis with disorganized detrusor muscle, and urinary tract abnormalities. {ECO:0000269	PubMed:22079972}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Psödoakondroplazi, 177170, Otozomal Dominant; PSACH (Psödoakondroplazi) (COMP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoachondroplasia, 177170, Autosomal dominant; PSACH (Pseudoachondroplasia) (COMP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COMP	COMP, EDM1, MED, PSACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 1 (EDM1) [MIM:132400]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:11565064, ECO:0000269|PubMed:21922596, ECO:0000269|PubMed:7670472, ECO:0000269|PubMed:9021009, ECO:0000269|PubMed:9184241, ECO:0000269|PubMed:9452026, ECO:0000269|PubMed:9463320, ECO:0000269|PubMed:9921895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psödoakondroplazi, 177170, Otozomal Dominant; PSACH (Psödoakondroplazi) (Prenatal) (COMP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pseudoachondroplasia, 177170, Autosomal dominant; PSACH (Pseudoachondroplasia) (COMP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COMP	COMP, EDM1, MED, PSACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 1 (EDM1) [MIM:132400]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:11565064, ECO:0000269|PubMed:21922596, ECO:0000269|PubMed:7670472, ECO:0000269|PubMed:9021009, ECO:0000269|PubMed:9184241, ECO:0000269|PubMed:9452026, ECO:0000269|PubMed:9463320, ECO:0000269|PubMed:9921895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Psödofolliculitis Barba, Yatkınlık, 612318 (KRT75 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudofolliculitis barbae, susceptibility to, 612318 (KRT75 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT75	KRT75, K6HF, PFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Loose anagen hair syndrome (LAHS) [MIM:606628]: In LAHS, anagen hairs are easily pulled from the scalp. The hair is relatively sparse and does not grow long. Hair of fair color and hair shafts of reduced caliber, and an early age of onset are features. Usually the hairs are not fragile and there are no areas of breakage. {ECO:0000269	PubMed:11939812}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohermafroditizm, Erkek, Jinekomasti, 264300, Otozomal Resesif (46,XY Cinsiyet Farklılaşma Bozukluğu, 17-Beta-Hidroksisteroid Dehidrogenaz 3 Eksikliğine Bağlı) (HSD17B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohermaphroditism, male, with gynecomastia, 264300, Autosomal recessive (46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency) (HSD17B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSD17B3	HSD17B3, EDH17B3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psödohermafroditizm, Erkek, Jinekomasti, 264300, Otozomal Resesif (46,XY Cinsiyet Farklılaşma Bozukluğu, 17-Beta-Hidroksisteroid Dehidrogenaz 3 Eksikliğine Bağlı) (MLPA)	Pseudohermaphroditism, male, with gynecomastia, 264300, Autosomal recessive (46,XY disorder of sex development due to 17-beta-hydroxysteroid dehydrogenase 3 deficiency) (MLPA)	HSD17B3	HSD17B3, EDH17B3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Psödohiperkalemi, Ailesel, 2, Eritrosit Kaçağına Bağlı, 609153, Otozomal Dominant (Ailesel Psödohiperkalemi) (ABCB6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohyperkalemia, familial, 2, due to red cell leak, 609153, Autosomal dominant (Familial pseudohyperkalemia) (ABCB6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCB6	ABCB6, MTABC3, MCOPCB7, LAN, DUH3, PSHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, isolated, with coloboma, 7 (MCOPCB7) [MIM:614497]: A disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues. Ocular abnormalities like opacities of the cornea and lens, scaring of the retina and choroid, and other abnormalities may also be present. Ocular colobomas are a set of malformations resulting from abnormal morphogenesis of the optic cup and stalk, and the fusion of the fetal fissure (optic fissure). {ECO:0000269	PubMed:22226084}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psödohipoaldosteronizm Tip 1, Otozomal Dominant, 177735, Otozomal Dominant (Renal Psödohipoaldosteronizm Tip 1) (NR3C2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism type I, autosomal dominant, 177735, Autosomal dominant (Renal pseudohypoaldosteronism type 1) (NR3C2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR3C2	NR3C2, MLR, MCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudohypoaldosteronism 1, autosomal dominant (PHA1A) [MIM:177735]: A salt wasting disease resulting from target organ unresponsiveness to mineralocorticoids. PHA1A is a mild form characterized by target organ defects confined to kidney. Patients may present with neonatal renal salt wasting with hyperkalaemic acidosis despite high aldosterone levels. These patients improve with age and usually become asymptomatic without treatment. {ECO:0000269	PubMed:16972228}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 1, 264350, Otozomal Resesif (Jeneralize Psödohipoaldosteronizm Tip 1) (SCNN1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type I, 264350, Autosomal recessive (Generalized pseudohypoaldosteronism type 1) (SCNN1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1G	SCNN1G, PHA1, BESC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269	PubMed:7550319}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 1, 264350, Otozomal Resesif (Jeneralize Psödohipoaldosteronizm Tip 1) (SCNN1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type I, 264350, Autosomal recessive (Generalized pseudohypoaldosteronism type 1) (SCNN1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1B	SCNN1B, BESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Liddle syndrome (LIDLS) [MIM:177200]: An autosomal dominant disorder characterized by hypertension, hypokalemic alkalosis, and suppression of plasma renin activity and aldosterone secretion. {ECO:0000269|PubMed:15483078, ECO:0000269|PubMed:7550319, ECO:0000269|PubMed:8524790, ECO:0000269|PubMed:8601645, ECO:0000269|PubMed:9626162, ECO:0000269|PubMed:9794716}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Psödohipoaldosteronizm, Tip 1, 264350, Otozomal Resesif, PHA1B (Jeneralize Psödohipoaldosteronizm Tip 1) (SCNN1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type I, 264350, Autosomal recessive, PHA1B (Generalized pseudohypoaldosteronism type 1) (SCNN1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCNN1A	SCNN1A, BESC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudohypoaldosteronism 1, autosomal recessive (PHA1B) [MIM:264350]: A rare salt wasting disease resulting from target organ unresponsiveness to mineralocorticoids. PHA1B is a severe form involving multiple organ systems, and characterized by an often fulminant presentation in the neonatal period with dehydration, hyponatremia, hyperkalemia, metabolic acidosis, failure to thrive and weight loss. {ECO:0000269|PubMed:10586178, ECO:0000269|PubMed:15853823, ECO:0000269|PubMed:18634878}. Note=The disease is caused by mutations affecting the gene represented in this entry. The degree of channel function impairment differentially affects the renin-aldosterone system and urinary Na/K ratios, resulting in distinct genotype-phenotype relationships in PHA1 patients. Loss-of-function mutations are associated with a severe clinical course and age-dependent hyperactivation of the renin-aldosterone system. This feature is not observed in patients with missense mutations that reduce but do not eliminate channel function. Markedly reduced channel activity results in impaired linear growth and delayed puberty (PubMed:18634878). {ECO:0000269|PubMed:18634878}.	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 2B, 614491, Otozomal Dominant; PHA2B (Psödohipoaldosteronizm Tip 2) (WNK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type IIB, 614491, Autosomal dominant; PHA2B (Pseudohypoaldosteronism type 2) (WNK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNK4	WNK4, PRKWNK4, PHA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudohypoaldosteronism 2B (PHA2B) [MIM:614491]: An autosomal dominant disorder characterized by hypertension, hyperkalemia, hyperchloremia, mild hyperchloremic metabolic acidosis, and correction of physiologic abnormalities by thiazide diuretics. {ECO:0000269|PubMed:11498583}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 2C, 614492, Otozomal Dominant; PHA2C (Psödohipoaldosteronizm Tip 2) (WNK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type IIC, 614492, Autosomal dominant; PHA2C (Pseudohypoaldosteronism type 2) (WNK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNK1	WNK1, PRKWNK1, KDP, PHA2C, HSAN2, HSN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory and autonomic, 2A (HSAN2A) [MIM:201300]: A form of hereditary sensory and autonomic neuropathy, a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN2A is an autosomal recessive disorder characterized by impairment of pain, temperature and touch sensation, onset of symptoms in infancy or early childhood, occurrence of distal extremity pathologies (paronychia, whitlows, ulcers, and Charcot joints), frequent amputations, sensory loss that affects all modalities of sensation (lower and upper limbs and perhaps the trunk as well), absence or diminution of tendon reflexes (usually in all limbs), minimal autonomic dysfunction, absence of sensory nerve action potentials, and virtual absence of myelinated fibers with decreased numbers of unmyelinated fibers in sural nerves. {ECO:0000269|PubMed:15060842, ECO:0000269|PubMed:15911806, ECO:0000269|PubMed:18521183}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 2D, 614495, Otozomal Resesif, Otozomal Dominant; PHA2D (Psödohipoaldosteronizm Tip 2) (KLHL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type IID, 614495, Autosomal recessive, Autosomal dominant; PHA2D (Pseudohypoaldosteronism type 2) (KLHL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL3	KLHL3, PHA2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 2E, 614496, Otozomal Dominant; PHA2E (Psödohipoaldosteronizm Tip 2) (CUL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoaldosteronism, type IIE, 614496, Autosomal dominant; PHA2E (Pseudohypoaldosteronism type 2) (CUL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CUL3	CUL3, PHA2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudohypoaldosteronism 2E (PHA2E) [MIM:614496]: An autosomal dominant disorder characterized by severe hypertension, hyperkalemia, hyperchloremia, hyperchloremic metabolic acidosis, and correction of physiologic abnormalities by thiazide diuretics. {ECO:0000269|PubMed:22266938}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm 1a, 103580, Otozomal Dominant; PHP1A (Psödohipoparatiroidizm Tip 1A) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoparathyroidism Ia, 103580, Autosomal dominant; PHP1A (Pseudohypoparathyroidism type 1A) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm 1a, 103580, Otozomal Dominant; PHP1A (Psödohipoparatiroidizm Tip 1A) (MLPA)	Pseudohypoparathyroidism Ia, 103580, Autosomal dominant; PHP1A (Pseudohypoparathyroidism type 1A) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm Ib, 603233, Otozomal Dominant (Psödohipoparatiroidizm Tip 1B) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoparathyroidism Ib, 603233, Autosomal dominant (Pseudohypoparathyroidism type 1B) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm Ib, 603233, Otozomal Dominant (Psödohipoparatiroidizm Tip 1B) (MLPA)	Pseudohypoparathyroidism Ib, 603233, Autosomal dominant (Pseudohypoparathyroidism type 1B) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm Ic 612462, Otozomal Dominant (Psödohipoparatiroidizm Tip 1C) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoparathyroidism Ic, 612462, Autosomal dominant (Pseudohypoparathyroidism type 1C) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm Ic 612462, Otozomal Dominant (Psödohipoparatiroidizm Tip 1C) (MLPA)	Pseudohypoparathyroidism Ic, 612462, Autosomal dominant (Pseudohypoparathyroidism type 1C) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödohipoparatiroidizm, Tip 1B, 603233, Otozomal Dominant (Psödohipoparatiroidizm Tipi 1B) (GNAS-AS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoparathyroidism, type IB, 603233, Autosomal dominant (Pseudohypoparathyroidism type 1B) (GNAS-AS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS-AS1	GNASAS1, GNASAS, SANG, NESPAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psödohipoparatiroidizm, Tip 1B, 603233, Otozomal Dominant; PHP1B (Psödohipoparatiroidizm Tipi 1B) (STX16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudohypoparathyroidism, type IB, 603233, Autosomal dominant; PHP1B (Pseudohypoparathyroidism type 1B) (STX16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STX16	STX16, SYN16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psödohipoaldosteronizm, Tip 1c, Php1c (Psödohipoparatiroidizm Tip 1C) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	PSEUDOHYPOPARATHYROIDISM, TYPE IC; PHP1C (Pseudohypoparathyroidism type 1C) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödomonas Aeruginosa, Kronik Enfeksiyona Karşı Hassasiyet, Kistik Fibrozda, 219700, Otozomal Resesif (Kistik Fibroz) (FCGR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudomonas aeruginosa, susceptibility to chronic infection by, in cystic fibrosis, 219700, Autosomal recessive (Cystic fibrosis) (FCGR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2A	FCGR2A, IGFR2, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psödopseudohipoparatiroidizm, 612463, Otozomal Dominant (Psödopsödohipoparatiroidizm) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudopseudohypoparathyroidism, 612463, Autosomal dominant (Pseudopseudohypoparathyroidism) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödopsödohipoparatiroidizm, 612463, Otozomal Dominant (Psödopsödohipoparatiroidizm) (MLPA)	Pseudopseudohypoparathyroidism, 612463, Autosomal dominant (Pseudopseudohypoparathyroidism) (MLPA)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödopsödohipoparatiroidizm; Pphp (Psödopsödohipoparatiroidizm) (GNAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	PSEUDOPSEUDOHYPOPARATHYROIDISM; PPHP (Pseudopseudohypoparathyroidism) (GNAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAS	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO, PHP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GNAS hyperfunction (GNASHYP) [MIM:139320]: This condition is characterized by increased trauma-related bleeding tendency, prolonged bleeding time, brachydactyly and mental retardation. Both the XLas isoforms and the ALEX protein are mutated which strongly reduces the interaction between them and this may allow unimpeded activation of the XLas isoforms. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödovajinal Perineoskrotal Hipospadyas, 264600, Otozomal Resesif; PPSH (46,XY Cinsiyet Farklılaşma Bozukluğu_5-Alfa-Redüktaz 2 Eksikliğine Bağlı (Psödovajinal Perineoskrotal Hipospadias)) (SRD5A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudovaginal perineoscrotal hypospadias, 264600, Autosomal recessive; PPSH (46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency) (SRD5A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRD5A2	SRD5A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudovaginal perineoscrotal hypospadias (PPSH) [MIM:264600]: A form of male pseudohermaphroditism in which 46,XY males show ambiguous genitalia at birth, including perineal hypospadias and a blind perineal pouch, and develop masculinization at puberty. The name of the disorder stems from the finding of a blind-ending perineal opening resembling a vagina and a severely hypospadiac penis with the urethra opening onto the perineum. {ECO:0000269|PubMed:10718838, ECO:0000269|PubMed:10999800, ECO:0000269|PubMed:1522235, ECO:0000269|PubMed:15528927, ECO:0000269|PubMed:15770495, ECO:0000269|PubMed:16098368, ECO:0000269|PubMed:16181229, ECO:0000269|PubMed:7543313, ECO:0000269|PubMed:8626825, ECO:0000269|PubMed:8768837, ECO:0000269|PubMed:9208814, ECO:0000269|PubMed:9745434, ECO:0000269|PubMed:9843052}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Psödovajinal Perineoskrotal Hipospadyas, 264600, Otozomal Resesif; PPSH (46,XY Cinsiyet Farklilişma Bozukluğu, 5-Alfa-Redüktaz 2 Eksikliğine Bağlı (Psödovajinal Perineoskrotal Hipospadias)) (MLPA)	Pseudovaginal perineoscrotal hypospadias, 264600, Autosomal recessive; PPSH (46,XY disorder of sex development due to 5-alpha-reductase 2 deficiency) (MLPA)	SRD5A2	SRD5A2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pseudovaginal perineoscrotal hypospadias (PPSH) [MIM:264600]: A form of male pseudohermaphroditism in which 46,XY males show ambiguous genitalia at birth, including perineal hypospadias and a blind perineal pouch, and develop masculinization at puberty. The name of the disorder stems from the finding of a blind-ending perineal opening resembling a vagina and a severely hypospadiac penis with the urethra opening onto the perineum. {ECO:0000269|PubMed:10718838, ECO:0000269|PubMed:10999800, ECO:0000269|PubMed:1522235, ECO:0000269|PubMed:15528927, ECO:0000269|PubMed:15770495, ECO:0000269|PubMed:16098368, ECO:0000269|PubMed:16181229, ECO:0000269|PubMed:7543315, ECO:0000269|PubMed:8626825, ECO:0000269|PubMed:8768837, ECO:0000269|PubMed:9208814, ECO:0000269|PubMed:9745434, ECO:0000269|PubMed:3843052}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psödoksantoma Elastikum Benzeri Hastalık-Birden Fazla Pıhtılaşma Faktörü Eksikliği 610842 (Vücut Deri Hiperfleksibilesi, K Vitamini Bağımlı Koagülasyon Faktörü Eksikliğine Bağlı) (GGCX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 (Body skin hyperlaxity due to vitamin K-dependent coagulation factor deficiency) (GGCX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GGCX	GGCX, VKCFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined deficiency of vitamin K-dependent clotting factors 1 (VKCFD1) [MIM:277450]: VKCFD leads to a bleeding tendency that is usually reversed by oral administration of vitamin K. {ECO:0000269|PubMed:11071668, ECO:0000269|PubMed:15287948, ECO:0000269|PubMed:9845520}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psödoksantoma Elasticum, 264800, Otozomal Resesif (Psödoksantoma Elastikum) (ABCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoxanthoma elasticum, 264800, Autosomal recessive (Pseudoxanthoma elasticum) (ABCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GACI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:10811882, ECO:0000269|PubMed:10835642, ECO:0000269|PubMed:10954200, ECO:0000269|PubMed:11427982, ECO:0000269|PubMed:11536079, ECO:0000269|PubMed:11702217, ECO:0000269|PubMed:15086542, ECO:0000269|PubMed:15098239, ECO:0000269|PubMed:15459974, ECO:0000269|PubMed:16086317, ECO:0000269|PubMed:17617515, ECO:0000269|PubMed:19339160, ECO:0000269|PubMed:20034067, ECO:0000269|PubMed:25615550}. Note=The disease is caused by mutations affecting the gene represented in this entry. Homozygous or compound heterozygous ABCC6 mutations have been found in the overwhelming majority of cases. Individuals carrying heterozygous mutations express limited manifestations of the pseudoxanthoma elasticum phenotype.	EDTA Blood Sample
Psödoksantoma Elasticum, 264800, Otozomal Resesif (Psödoksantoma Elastikum) (MLPA)	Pseudoxanthoma elasticum, 264800, Autosomal recessive (Pseudoxanthoma elasticum) (MLPA)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GACI2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:10811882, ECO:0000269|PubMed:10835642, ECO:0000269|PubMed:10954200, ECO:0000269|PubMed:11427982, ECO:0000269|PubMed:11536079, ECO:0000269|PubMed:11702217, ECO:0000269|PubMed:15086542, ECO:0000269|PubMed:15098239, ECO:0000269|PubMed:15459974, ECO:0000269|PubMed:16086317, ECO:0000269|PubMed:17617515, ECO:0000269|PubMed:19339160, ECO:0000269|PubMed:20034067, ECO:0000269|PubMed:25615550}. Note=The disease is caused by mutations affecting the gene represented in this entry. Homozygous or compound heterozygous ABCC6 mutations have been found in the overwhelming majority of cases. Individuals carrying heterozygous mutations express limited manifestations of the pseudoxanthoma elasticum phenotype.	EDTA Blood Sample
Psödoksantoma Elastikum, Forme Fruste, 177850, Otozomal Dominant; PXE (Psödoksantoma Elastikum) (ABCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoxanthoma elasticum, forme fruste, 177850, Autosomal dominant; PXE (Pseudoxanthoma elasticum) (ABCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GACI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:10811882, ECO:0000269|PubMed:10835642, ECO:0000269|PubMed:10954200, ECO:0000269|PubMed:11427982, ECO:0000269|PubMed:11536079, ECO:0000269|PubMed:11702217, ECO:0000269|PubMed:15086542, ECO:0000269|PubMed:15098239, ECO:0000269|PubMed:15459974, ECO:0000269|PubMed:16086317, ECO:0000269|PubMed:17617515, ECO:0000269|PubMed:19339160, ECO:0000269|PubMed:20034067, ECO:0000269|PubMed:25615550}. Note=The disease is caused by mutations affecting the gene represented in this entry. Homozygous or compound heterozygous ABCC6 mutations have been found in the overwhelming majority of cases. Individuals carrying heterozygous mutations express limited manifestations of the pseudoxanthoma elasticum phenotype.	EDTA Blood Sample
Psödoksantoma Elastikum, Forme Fruste, 177850, Otozomal Dominant; PXE (Psödoksantoma Elastikum) (MLPA)	Pseudoxanthoma elasticum, forme fruste, 177850, Autosomal dominant; PXE (Pseudoxanthoma elasticum) (MLPA)	ABCC6	ABCC6, ARA, ABC34, MLP1, PXE, GACI2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:10811882, ECO:0000269|PubMed:10835642, ECO:0000269|PubMed:10954200, ECO:0000269|PubMed:11427982, ECO:0000269|PubMed:11536079, ECO:0000269|PubMed:11702217, ECO:0000269|PubMed:15086542, ECO:0000269|PubMed:15098239, ECO:0000269|PubMed:15459974, ECO:0000269|PubMed:16086317, ECO:0000269|PubMed:17617515, ECO:0000269|PubMed:19339160, ECO:0000269|PubMed:20034067, ECO:0000269|PubMed:25615550}. Note=The disease is caused by mutations affecting the gene represented in this entry. Homozygous or compound heterozygous ABCC6 mutations have been found in the overwhelming majority of cases. Individuals carrying heterozygous mutations express limited manifestations of the pseudoxanthoma elasticum phenotype.	EDTA Blood Sample
Psödoksantoma Elastikum, Şiddetini Modifiye Edici, 264800, Otozomal Resesif (Psödoksantoma Elastikum) (XYLT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoxanthoma elasticum, modifier of severity of, 264800, Autosomal recessive (Pseudoxanthoma elasticum) (XYLT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XYLT2	XYLT2, XT2, SOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:16571645}. Note=The gene represented in this entry acts as a disease modifier. PXE patients carrying causative ABCC6 mutations, manifest a more severe disease course characterized by earlier onset, frequent skin lesions and higher organ involvement, in the presence of XYLT2 variants. {ECO:0000269|PubMed:16571645},;	EDTA Blood Sample

Psödoksantoma Elastikum, Şiddetini Modifiye Edici, 264800, Otozomal Resesif (Psödoksantoma Elastikum) (XYLT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pseudoxanthoma elasticum, modifier of severity of, 264800, Autosomal recessive (Pseudoxanthoma elasticum) (XYLT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XYLT1	XYLT1, XT1, DBQD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269	PubMed:16571645}. Note=The gene represented in this entry acts as a disease modifier.	EDTA Blood Sample
Psöriasis 14, Püstüler 614204, Otozomal Resesif; PSORS14 (Jeneralize Püstüler Psöriasis) (IL36RN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis 14, pustular, 614204, Autosomal recessive; PSORS14 (Generalized pustular psoriasis) (IL36RN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL36RN	IL36RN, IL1F5, FIL1D, IL1HY1, IL1RP3, PSORP, PSORS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriasis 14, pustular (PSORS14) [MIM:614204]: A life- threatening disease defined by repeated flares of sudden onset consisting of diffuse erythematous skin eruption characterized by rapid coverage with pustules, high-grade fever, asthenia, marked leukocytosis, and elevated serum levels of C-reactive protein. {ECO:0000269|PubMed:21839423, ECO:0000269|PubMed:21848462, ECO:0000269|PubMed:22903787}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psöriasis 15, Püstüler Yatkınlık, 616106, Otozomal Dominant; PSORS15 (Jeneralize Püstüler Psöriasis) (AP1S3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis 15, pustular, susceptibility to, 616106, Autosomal dominant; PSORS15 (Generalized pustular psoriasis) (AP1S3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP1S3	AP1S3, PSORS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psöriasis 2, 602723, Otozomal Dominant, PSORS2 (Pitriazis Rubra Pilaris) (CARD14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis 2, 602723, Autosomal dominant; PSORS2 (Pityriasis rubra pilaris) (CARD14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CARD14	CARD14, CARMA2, BIMP2, PSORS2, PSS1, PRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pityriasis rubra pilaris (PRP) [MIM:173200]: A rare, papulosquamous skin disease characterized by the appearance of keratotic follicular papules, well-demarcated salmon-colored erythematous plaques covered with fine powdery scales interspersed with distinct islands of uninvolved skin, and palmoplantar keratoderma. Most cases are sporadic. The rare familial cases show autosomal dominant inheritance with incomplete penetrance and variable expression. Familial PRP usually presents at birth or appears during the first years of life and runs a chronic course. It is characterized by prominent follicular hyperkeratosis, diffuse palmoplantar keratoderma, and erythema. {ECO:0000269	PubMed:22703878}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psöriasis Yatkınlık 1, 177900, Multifaktöriyel (HLA-C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis susceptibility 1, 177900, Multifactorial (HLA-C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-C	HLA-C, PSORS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Psöriasis Yatkınlık 13, 614070 (TRAF3IP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis susceptibility 13, 614070 (TRAF3IP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAF3IP2	TRAF3IP2, C6orf5, ACT1, CIK5, C6orf4, C6orf6, PSORS13, CAND9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Candidiasis, familial, 8 (CANDF8) [MIM:615527]: A primary immunodeficiency disorder with altered immune responses and impaired clearance of fungal infections, selective against Candida. It is characterized by persistent and/or recurrent infections of the skin, nails and mucous membranes caused by organisms of the genus Candida, mainly Candida albicans. {ECO:0000269	PubMed:24120361}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Psöriasis, Karşı Koruma, 605606 (IL23R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriasis, protection against, 605606 (IL23R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL23R	IL23R, IBD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Inflammatory bowel disease 17 (IBD17) [MIM:612261]: A chronic, relapsing inflammation of the gastrointestinal tract with a complex etiology. It is subdivided into Crohn disease and ulcerative colitis phenotypes. Crohn disease may affect any part of the gastrointestinal tract from the mouth to the anus, but most frequently it involves the terminal ileum and colon. Bowel inflammation is transmural and discontinuous; it may contain granulomas or be associated with intestinal or perianal fistulas. In contrast, in ulcerative colitis, the inflammation is continuous and limited to rectal and colonic mucosal layers; fistulas and granulomas are not observed. Both diseases include extraintestinal inflammation of the skin, eyes, or joints. {ECO:0000269|PubMed:17068223, ECO:0000269|PubMed:17804789}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Psoriatik Artrit, Yatkınlık, 607507 (NOD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriatic arthritis, susceptibility to, 607507 (NOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOD2	NOD2, CARD15, IBD1, CD, YAOS, BLAUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Blau syndrome (BLAUS) [MIM:186580]: A rare autosomal dominant disorder characterized by early-onset granulomatous arthritis, uveitis and skin rash. {ECO:0000269|PubMed:11528384, ECO:0000269|PubMed:15812565, ECO:0000269|PubMed:19116920, ECO:0000269|PubMed:19169908, ECO:0000269|PubMed:19479837, ECO:0000269|PubMed:20199415, ECO:0000269|PubMed:24960071, ECO:0000269|PubMed:25093298, ECO:0000269|PubMed:25692065, ECO:0000269|PubMed:25724124}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Psoriatik Artrit, Yatkınlık, 607507 (LTA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psoriatic arthritis, susceptibility to, 607507 (LTA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTA	LTA, TNFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet; arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). {ECO:0000269	PubMed:12746914}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Psikomotor Retardasyon, Epilepsi ve Kraniofasiyal Dismorfizm 614501, Otozomal Resesif; PMRED (SNIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501, Autosomal recessive; PMRED (SNIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNIP1	SNIP1, PMRED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psychomotor retardation, epilepsy, and craniofacial dysmorphism (PMRED) [MIM:614501]: A disease characterized by severe psychomotor retardation, intractable seizures, dysmorphic features, and a lumpy skull surface. Patients are hypotonic and have poor feeding in the neonatal period. {ECO:0000269	PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Psikomotor Retardasyon, Epilepsi ve Kraniofasiyal Dismorfizm 614501, Otozomal Resesif; PMRED (SNIP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Psychomotor retardation, epilepsy, and craniofacial dysmorphism, 614501, Autosomal recessive; PMRED (SNIP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNIP1	SNIP1, PMRED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTALI KAN		.	.	15	30	20	Prenatal	DISEASE: Psychomotor retardation, epilepsy, and craniofacial dysmorphism (PMRED) [MIM:614501]: A disease characterized by severe psychomotor retardation, intractable seizures, dysmorphic features, and a lumpy skull surface. Patients are hypotonic and have poor feeding in the neonatal period. {ECO:0000269	PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
PTEN Hamartoma Tümör Sendromu (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	PTEN hamartoma tumor syndrome (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
PTEN Hamartoma Tümör Sendromu (MLPA)	PTEN hamartoma tumor syndrome (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ptozis, Konjenital, 178300, Otozomal Dominant (Konjenital Ptozis) (ZFHX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ptosis, congenital, 178300, Autosomal dominant (Congenital ptosis) (ZFHX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFHX4	ZFHX4, ZFH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pulmoner Alveolar Mikrolitiazis, 265100, Otozomal Resesif (Pulmoner Alveolar Mikrolitiazis) (SLC34A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary alveolar microlithiasis, 265100, Autosomal recessive (Pulmonary alveolar microlithiasis) (SLC34A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC34A2	SLC34A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary alveolar microlithiasis (PALM) [MIM:265100]: Rare disease characterized by the deposition of calcium phosphate microliths throughout the lungs. Most patients are asymptomatic for several years or even for decades and generally, the diagnosis is incidental to clinical investigations unrelated to the disease. Cases with early-onset or rapid progression are rare. A 'sandstorm-appearing' chest roentgenogram is a typical diagnostic finding. The onset of this potentially lethal disease varies from the neonatal period to old age and the disease follows a long- term, progressive course, resulting in a slow deterioration of lung functions. Pulmonary alveolar microlithiasis is a recessive monogenic disease with full penetrance. {ECO:0000269	PubMed:16960801}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Akciğer Hastalığı, Kronik Obstrüktif, Yatkınlık, 606963 (HMOX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary disease, chronic obstructive, susceptibility to, 606963 (HMOX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HMOX1	HMOX1, HMOX1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heme oxygenase 1 deficiency (HMOX1D) [MIM:614034]: A disease characterized by impaired stress hematopoiesis, resulting in marked erythrocyte fragmentation and intravascular hemolysis, coagulation abnormalities, endothelial damage, and iron deposition in renal and hepatic tissues. Clinical features include persistent hemolytic anemia, asplenia, nephritis, generalized erythematous rash, growth retardation and hepatomegaly. {ECO:0000269	PubMed:9884342}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Akciğer Hastalığı, Kronik Obstrüktif, Yatkınlık, 606963 (Alfa-1-Antitripsin Eksikliği) (SERPINA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary disease, chronic obstructive, susceptibility to, 606963 (Alpha-1-antitrypsin deficiency) (SERPINA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA1	SERPINA1, PI, AAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (ALATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Akciğer Hastalığı, Kronik Obstrüktif, Yatkınlık, 606963 (Alfa-1-Antitripsin Eksikliği) (MLPA)	Pulmonary disease, chronic obstructive, susceptibility to, 606963 (Alpha-1-antitrypsin deficiency) (MLPA)	SERPINA1	SERPINA1, PI, AAT	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alpha-1-antitrypsin deficiency (ALATD) [MIM:613490]: A disorder whose most common manifestation is emphysema, which becomes evident by the third to fourth decade. A less common manifestation of the deficiency is liver disease, which occurs in children and adults, and may result in cirrhosis and liver failure. Environmental factors, particularly cigarette smoking, greatly increase the risk of emphysema at an earlier age. {ECO:0000269|PubMed:1905728, ECO:0000269|PubMed:2227940, ECO:0000269|PubMed:2390072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Fibrozis ve / Veya Kemik İliği Yetmezliği, Telomer İlişkili, 1, 614742, Otozomal Dominant; PFBMFT1 (İdiopatik Aplastik Anemi) (TERT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742, Autosomal dominant; PFBMFT1 (Idiopathic aplastic anemia) (TERT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERT	TERT, TCS1, EST2, DKCA2, DKCB4, PFBMFT1, CMM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pulmoner Fibrozis ve / Veya Kemik İliği Yetmezliği, Telomer İle İlgili, 2, Pfbmft2 (İdiopatik Aplastik Anemi) (TERC geni) (Tüm Gen Dizi Analizi) (Postnatal)	PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE, TELOMERE-RELATED, 2; PFBMFT2 (Idiopathic aplastic anemia) (TERC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pulmoner Fibrozis ve / Veya Kemik İliği Yetmezliği, Telomer İle İlgili, 2, Pfbmft2 (İdiopatik Aplastik Anemi) (MLPA)	PULMONARY FIBROSIS AND/OR BONE MARROW FAILURE, TELOMERE-RELATED, 2; PFBMFT2 (idiopathic aplastic anemia) (MLPA)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Pulmoner Fibrozis ve / Veya Kemik İliği Yetmezliği, Telomer İle İlgili, 3, 616373, Otozomal Dominant; PFBMFT3 (İdiopatik Pulmoner Fibrozis) (RTEL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373, Autosomal dominant; PFBMFT3 (Idiopathic pulmonary fibrosis) (RTEL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTEL1	RTEL1, C20orf41, NHL, KIAA1088, DKCB5, DKCA4, PFBMFT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 5 (DKCB5) [MIM:615190]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. DKCB5 is characterized by onset of bone marrow failure and immunodeficiency in early childhood. Most patients also have growth and developmental delay and cerebellar hypoplasia, consistent with a clinical diagnosis of Hoyeraal-Hreidarsson syndrome. {ECO:0000269|PubMed:23329068, ECO:0000269|PubMed:23453664, ECO:0000269|PubMed:23591994, ECO:0000269|PubMed:23959892, ECO:0000269|PubMed:24009516}. Note=The disease is caused by mutations affecting the gene represented in this entry. RTEL1 mutations have also been found in patients with a dyskeratosis congenita-like phenotype consisting of one feature of dyskeratosis congenita and short telomeres, in the absence of the typical DKC diagnostic triad (PubMed:23329068). {ECO:0000269|PubMed:23329068}.	EDTA Blood Sample
Pulmoner Fibrozis ve / Veya Kemik İliği Yetmezliği, Telomer İle İlgili, 4, 616371, Otozomal Dominant; PFBMFT4 (İdiopatik Pulmoner Fibrozis) (PARN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371, Autosomal dominant; PFBMFT4 (Idiopathic pulmonary fibrosis) (PARN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PARN	PARN, DAN, DKCB6, PFBMFT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal recessive, 6 (DKCB6) [MIM:616353]: A form of dyskeratosis congenita, a rare multisystem disorder caused by defective telomere maintenance. It is characterized by progressive bone marrow failure, and the clinical triad of reticulated skin hyperpigmentation, nail dystrophy, and mucosal leukoplakia. Common but variable features include premature graying, aplastic anemia, low platelets, osteoporosis, pulmonary fibrosis, and liver fibrosis among others. Early mortality is often associated with bone marrow failure, infections, fatal pulmonary complications, or malignancy. {ECO:0000269|PubMed:25893599}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pulmoner Fibroz, İdyopatik, 178500, Otozomal Dominant; IPF (İdyopatik Pulmoner Fibrozis) (SFTPA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis, idiopathic, 178500, Autosomal dominant; IPF (Idiopathic pulmonary fibrosis) (SFTPA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFTPA2	SFTPA2, SPA2, COLECS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. {ECO:0000269|PubMed:19105528}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Fibrozis, İdyopatik, Yatkınlık, 178500, Otozomal Dominant (İdyopatik Pulmoner Fibrozis) (MUC5B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis, idiopathic, susceptibility to, 178500, Autosomal dominant (Idiopathic pulmonary fibrosis) (MUC5B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MUC5B	MUC5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. {ECO:0000269|PubMed:21506748}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A common polymorphism in the promoter of MUC5B is associated with familial interstitial pneumonia and idiopathic pulmonary fibrosis, suggesting that dysregulated MUC5B expression in the lung may be involved in the pathogenesis of pulmonary fibrosis (PubMed:21506741). {ECO:0000269|PubMed:21506741}.;	EDTA Blood Sample
Pulmoner Fibrozis, İdyopatik, Yatkınlık, 178500, Otozomal Dominant; IPF (İdyopatik Pulmoner Fibrozis) (SFTPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis, idiopathic, susceptibility to, 178500, Autosomal dominant; IPF (Idiopathic pulmonary fibrosis) (SFTPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFTPA1	SFTPA1, SFTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary fibrosis, idiopathic (IPF) [MIM:178500]: A lung disease characterized by shortness of breath, radiographically evident diffuse pulmonary infiltrates, and varying degrees of inflammation and fibrosis on biopsy. In some cases, the disorder can be rapidly progressive and characterized by sequential acute lung injury with subsequent scarring and end-stage lung disease. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Pulmoner Fibrozis, İdyopatik, Yatkınlık, 614743, Otozomal Dominant; PFBMFT2 (İdiopatik Aplastik Anemi) (TERC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary fibrosis, idiopathic, susceptibility to, 614743, Autosomal dominant; PFBMFT2 (Idiopathic aplastic anemia) (TERC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Pulmoner Fibrozis, İdyopatik, Yatkınlık, 614743, Otozomal Dominant; PFBMFT2 (İdiopatik Aplastik Anemi) (MLPA)	Pulmonary fibrosis, idiopathic, susceptibility to, 614743, Autosomal dominant; PFBMFT2 (idiopathic aplastic anemia) (MLPA)	TERC	TERC, TRC3, TR, DKCA1, PFBMFT2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Pulmoner Hipertansiyon, Allesel Primer, 1, ± HHT, 178600, Otozomal Dominant; PPH1 (İdyopatik ve / Veya Allesel Pulmoner Arteriyel Hipertansiyon) (BMPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600, Autosomal dominant; PPH1 (Idiopathic and/or familial pulmonary arterial hypertension) (BMPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR2	BMPR2, PPH1, POVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Allesel Primer, 1, ± HHT, 178600, Otozomal Dominant; PPH1 (İdyopatik ve / Veya Allesel Pulmoner Arteriyel Hipertansiyon) (MLPA)	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600, Autosomal dominant; PPH1 (Idiopathic and/or familial pulmonary arterial hypertension) (MLPA)	BMPR2	BMPR2, PPH1, POVD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Allesel Primer, 1, ± HHT, 178600, Otozomal Dominant; PPH1 (İdyopatik ve / Veya Allesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (BMPR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600, Autosomal dominant; PPH1 (Idiopathic and/or familial pulmonary arterial hypertension) (BMPR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMPR2	BMPR2, PPH1, POVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Allesel Primer, 1, ± HHT, 178600, Otozomal Dominant; PPH1 (İdyopatik ve / Veya Allesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (MLPA)	Pulmonary hypertension, familial primary, 1, with or without HHT, 178600, Autosomal dominant; PPH1 (Idiopathic and/or familial pulmonary arterial hypertension) (Prenatal) (MLPA)	BMPR2	BMPR2, PPH1, POVD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Pulmoner Hipertansiyon, Neonatal, Yatkınlık, 615371; PHN (Karbamoil-Fosfat Sentetaz 1 Eksikliği) (CPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, neonatal, susceptibility to, 615371; PHN (Carbamoyl-phosphate synthetase 1 deficiency) (CPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPS1	CPS1, PHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Carbamoyl phosphate synthetase 1 deficiency (CPS1D) [MIM:237300]: An autosomal recessive disorder of the urea cycle causing hyperammonemia. It can present as a devastating metabolic disease dominated by severe hyperammonemia in neonates or as a more insidious late-onset condition, generally manifesting as life-threatening hyperammonemic crises under catabolic situations. Clinical features include protein intolerance, intermittent ataxia, seizures, lethargy, developmental delay and mental retardation. {ECO:0000269|PubMed:11388595, ECO:0000269|PubMed:11474210, ECO:0000269|PubMed:12655559, ECO:0000269|PubMed:12955727, ECO:0000269|PubMed:15164414, ECO:0000269|PubMed:15617192, ECO:0000269|PubMed:16737834, ECO:0000269|PubMed:17310273, ECO:0000269|PubMed:20578160, ECO:0000269|PubMed:21120950, ECO:0000269|PubMed:9711878}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Pulmoner Hipertansiyon, Neonatal, Yatkınlık, 615371; PHN (Karbamoil-Fosfat Sentetaz 1 Eksikliği) (Prenatal) (CPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, neonatal, susceptibility to, 615371; PHN (Carbamoyl-phosphate synthetase 1 deficiency) (CPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CPS1	CPS1, PHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Carbamoyl phosphate synthetase 1 deficiency (CPS1D) [MIM:237300]: An autosomal recessive disorder of the urea cycle causing hyperammonemia. It can present as a devastating metabolic disease dominated by severe hyperammonemia in neonates or as a more insidious late-onset condition, generally manifesting as life-threatening hyperammonemic crises under catabolic situations. Clinical features include protein intolerance, intermittent ataxia, seizures, lethargy, developmental delay and mental retardation. {ECO:0000269|PubMed:11388595, ECO:0000269|PubMed:11474210, ECO:0000269|PubMed:12655559, ECO:0000269|PubMed:12955727, ECO:0000269|PubMed:15164414, ECO:0000269|PubMed:15617192, ECO:0000269|PubMed:16737834, ECO:0000269|PubMed:17310273, ECO:0000269|PubMed:20578160, ECO:0000269|PubMed:21120950, ECO:0000269|PubMed:9711878}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 2, 615342, Otozomal Dominant; PPH2 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (SMAD9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, primary, 2, 615342, Autosomal dominant; PPH2 (Idiopathic and/or familial pulmonary arterial hypertension) (SMAD9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMAD9	MADH9, SMAD9, MADH6, PPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary hypertension, primary, 2 (PPH2) [MIM:615342]: A rare disorder characterized by plexiform lesions of proliferating endothelial cells in pulmonary arterioles. The lesions lead to elevated pulmonary arterial pression, right ventricular failure, and death. The disease can occur from infancy throughout life and it has a mean age at onset of 36 years. Penetrance is reduced. Although familial pulmonary hypertension is rare, cases secondary to known etiologies are more common and include those associated with the appetite-suppressant drugs. {ECO:0000269	PubMed:21898662}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 2, 615342, Otozomal Dominant; PPH2 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (SMAD9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, primary, 2, 615342, Autosomal dominant; PPH2 (Idiopathic and/or familial pulmonary arterial hypertension) (SMAD9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMAD9	MADH9, SMAD9, MADH6, PPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary hypertension, primary, 2 (PPH2) [MIM:615342]: A rare disorder characterized by plexiform lesions of proliferating endothelial cells in pulmonary arterioles. The lesions lead to elevated pulmonary arterial pression, right ventricular failure, and death. The disease can occur from infancy throughout life and it has a mean age at onset of 36 years. Penetrance is reduced. Although familial pulmonary hypertension is rare, cases secondary to known etiologies are more common and include those associated with the appetite-suppressant drugs. {ECO:0000269	PubMed:21898662}. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 3, 615343, Otozomal Dominant; PPH3 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (CAV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, primary, 3, 615343, Autosomal dominant; PPH3 (Idiopathic and/or familial pulmonary arterial hypertension) (CAV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. {ECO:0000269	PubMed:18237401}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 3, 615343, Otozomal Dominant; PPH3 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (CAV1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, primary, 3, 615343, Autosomal dominant; PPH3 (Idiopathic and/or familial pulmonary arterial hypertension) (CAV1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CAV1	CAV1, BSCL3, CGL3, PPH3, LCCNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Partial lipodystrophy, congenital cataracts, and neurodegeneration syndrome (LCCNS) [MIM:606721]: A form of familial partial lipodystrophy associated with congenital cataracts and neurodegeneration leading to cerebellar and spinal cord dysfunction. {ECO:0000269	PubMed:18237401}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 4, 615344, Otozomal Dominant; PPH4 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (KCNK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, primary, 4, 615344, Autosomal dominant; PPH4 (Idiopathic and/or familial pulmonary arterial hypertension) (KCNK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNK3	KCNK3, TASK, PPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, 4, 615344, Otozomal Dominant; PPH4 (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (KCNK3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, primary, 4, 615344, Autosomal dominant; PPH4 (Idiopathic and/or familial pulmonary arterial hypertension) (KCNK3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNK3	KCNK3, TASK, PPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, Fenfluramin veya Deksfenfluramin-Bağlantılı 178600, Otozomal Dominant (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (BMPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600, Autosomal dominant (idiopathic and/or familial pulmonary arterial hypertension) (BMPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR2	BMPR2, PPH1, POVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, Fenfluramin veya Deksfenfluramin-Bağlantılı 178600, Otozomal Dominant (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (MLPA)	Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600, Autosomal dominant (idiopathic and/or familial pulmonary arterial hypertension) (MLPA)	BMPR2	BMPR2, PPH1, POVD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, Fenfluramin veya Deksfenfluramin-Bağlantılı 178600, Otozomal Dominant (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (BMPR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600, Autosomal dominant (idiopathic and/or familial pulmonary arterial hypertension) (BMPR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BMPR2	BMPR2, PPH1, POVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Hipertansiyon, Primer, Fenfluramin veya Deksfenfluramin-Bağlantılı 178600, Otozomal Dominant (İdyopatik ve / Veya Ailesel Pulmoner Arteriyel Hipertansiyon) (Prenatal) (MLPA)	Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated, 178600, Autosomal dominant (idiopathic and/or familial pulmonary arterial hypertension) (Prenatal) (MLPA)	BMPR2	BMPR2, PPH1, POVD1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pulmoner Venooklüzif Hastalık 1, 265450, Otozomal Dominant; PVOD1 (Pulmoner Venooklüzif Hastalık) (BMPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary venocclusive disease 1, 265450, Autosomal dominant; PVOD1 (Pulmonary venocclusive disease) (BMPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BMPR2	BMPR2, PPH1, POVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pulmoner Venooklüzif Hastalık 1, 265450, Otozomal Dominant; PVOD1 (Pulmoner Venooklüzif Hastalık) (MLPA)	Pulmonary venocclusive disease 1, 265450, Autosomal dominant; PVOD1 (Pulmonary venocclusive disease) (MLPA)	BMPR2	BMPR2, PPH1, POVD1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pulmonary venoocclusive disease 1, autosomal dominant (PVOD1) [MIM:265450]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:12446270, ECO:0000269|PubMed:16429395}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Pulmoner Venooklüzif Hastalık 2, 234810, Otozomal Resesif; PVOD2 (Pulmoner Kapiller Hemanjiyomatozis) (EIF2AK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pulmonary venocclusive disease 2, 234810, Autosomal recessive; PVOD2 (Pulmonary capillary hemangiomatosis) (EIF2AK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2AK4	EIF2AK4, GCN2, KIAA1338, PVOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary venocclusive disease 2, autosomal recessive (PVOD2) [MIM:234810]: A disease characterized by widespread fibrous obstruction and intimal thickening of septal veins and preseptal venules, a low diffusing capacity for carbon monoxide, occult alveolar hemorrhage, and nodular ground-glass opacities, septal lines and lymph node enlargement showed by high-resolution computed tomography of the chest. It is frequently associated with pulmonary capillary dilatation and proliferation, and is a rare and devastating cause of pulmonary hypertension. {ECO:0000269|PubMed:24135949, ECO:0000269|PubMed:24292273}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Purpura, Transfüzyon Sonrası (Glanzmann Trombastenisi) (ITGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Purpura, posttransfusion (Glanzmann thrombasthenia) (ITGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB3	ITGB3, GP3A, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (6DPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. {ECO:0000269	PubMed:18065693}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piknodizostozis, 265800, Otozomal Resesif (Piknodizostozis) (CTSK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pycnodysostosis, 265800, Autosomal recessive (Pycnodysostosis) (CTSK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTSK	CTSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Piknodizostozis, 265800, Otozomal Resesif (Piknodizostozis) (Prenatal) (CTSK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pycnodysostosis, 265800, Autosomal recessive (Pycnodysostosis) (CTSK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CTSK	CTSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pyle Hastalığı, 265900, Otozomal Resesif; PYL (Pyle Hastalığı) (SFRP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyle disease, 265900, Autosomal recessive; PYL (Pyle disease) (SFRP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFRP4	SFRP4, FRPHE, PYL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Piyojenik Bakteriyel Enfeksiyonlar, Tekrarlayan, MYD88 Eksikliğine Bağlı, 612260 (Piyojenik Bakteriyel Enfeksiyonlar, Myd88 Eksikliğine Bağlı) (MYD88 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (Pyogenic bacterial infections due to MyD88 deficiency) (MYD88 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYD88	MYD88, MYD88D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: MYD88 deficiency (MYD88D) [MIM:612260]: Patients suffer from autosomal recessive, life-threatening, often recurrent pyogenic bacterial infections, including invasive pneumococcal disease, and die between 1 and 11 months of age. Surviving patients are otherwise healthy, with normal resistance to other microbes, and their clinical status improved with age. {ECO:0000269|PubMed:18669862, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Piyojenik Bakteriyel Enfeksiyonlar, Tekrarlayan, MYD88 Eksikliğine Bağlı, 612260 (Piyojenik Bakteriyel Enfeksiyonlar, Myd88 Eksikliğine Bağlı) (Prenatal) (MYD88 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyogenic bacterial infections, recurrent, due to MYD88 deficiency, 612260 (Pyogenic bacterial infections due to MyD88 deficiency) (MYD88 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYD88	MYD88, MYD88D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: MYD88 deficiency (MYD88D) [MIM:612260]: Patients suffer from autosomal recessive, life-threatening, often recurrent pyogenic bacterial infections, including invasive pneumococcal disease, and die between 1 and 11 months of age. Surviving patients are otherwise healthy, with normal resistance to other microbes, and their clinical status improved with age. {ECO:0000269|PubMed:18669862, ECO:0000269|PubMed:21057262, ECO:0000269|PubMed:24316379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piyojenik Steril Artrit, Piyoderma Gangrenosum ve Akne, 604416, Otozomal Dominant (Piyojenik Artrit-Piyoderma Gangrenozum-Akne Sendromu) (PSTPIP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416, Autosomal dominant (Pyogenic arthritis-pyoderma gangrenosum-acne syndrome) (PSTPIP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PSTPIP1	PSTPIP1, PSTPIP, CD2BP1, PAPAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: PAPA syndrome (PAPAS) [MIM:604416]: Characterized by autosomal dominant inheritance of early-onset, primarily affecting skin and joint tissues. Recurring inflammatory episodes lead to accumulation of sterile, pyogenic, neutrophil-rich material within the affected joints, ultimately resulting in significant destruction. {ECO:0000269|PubMed:11971877, ECO:0000269|PubMed:22161697}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piyojenik Steril Artrit, Piyoderma Gangrenosum ve Akne, 604416, Otozomal Dominant (Piyojenik Artrit-Piyoderma Gangrenozum-Akne Sendromu) (Prenatal) (PSTPIP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416, Autosomal dominant (Pyogenic arthritis-pyoderma gangrenosum-acne syndrome) (PSTPIP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PSTPIP1	PSTPIP1, PSTPIP, CD2BP1, PAPAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: PAPA syndrome (PAPAS) [MIM:604416]: Characterized by autosomal dominant inheritance of early-onset, primarily affecting skin and joint tissues. Recurring inflammatory episodes lead to accumulation of sterile, pyogenic, neutrophil-rich material within the affected joints, ultimately resulting in significant destruction. {ECO:0000269|PubMed:11971877, ECO:0000269|PubMed:22161697}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piridoksamin 5'-Fosfat Oksidaz Eksikliği, 610090, Otozomal Resesif; PNPOD (Piridoksal Fosfat Yanıtı Nöbetler) (PNPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyridoxamine 5'-phosphate oxidase deficiency, 610090, Autosomal recessive; PNPOD (Pyridoxal phosphate-responsive seizures) (PNPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPO	PNPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyridoxine-5'-phosphate oxidase deficiency (PNPO deficiency) [MIM:610090]: The main feature of neonatal epileptic encephalopathy is the onset within hours of birth of a severe seizure disorder that does not respond to anticonvulsant drugs and can be fatal. Seizures can cease with the administration of PLP, being resistant to treatment with pyridoxine., {ECO:0000269	PubMed:15772097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piridoksamin 5'-Fosfat Oksidaz Eksikliği, 610090, Otozomal Resesif; PNPOD (Piridoksal Fosfat Yanıtı Nöbetler) (Prenatal) (PNPO geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyridoxamine 5'-phosphate oxidase deficiency, 610090, Autosomal recessive; PNPOD (Pyridoxal phosphate-responsive seizures) (PNPO gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PNPO	PNPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyridoxine-5'-phosphate oxidase deficiency (PNPO deficiency) [MIM:610090]: The main feature of neonatal epileptic encephalopathy is the onset within hours of birth of a severe seizure disorder that does not respond to anticonvulsant drugs and can be fatal. Seizures can cease with the administration of PLP, being resistant to treatment with pyridoxine., {ECO:0000269	PubMed:15772097}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piropoikilositozis, 266140, Otozomal Resesif; HPP (Kalıtsal Piropoikilositozis) (SPTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyropoikilocytosis, 266140, Autosomal recessive; HPP (Hereditary pyropoikilocytosis) (SPTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTA1	SPTA1, EL2, SPHβ, HS3, HPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 2 (EL2) [MIM:130600]: A Rhesus-unlinked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. {ECO:0000269|PubMed:1541680, ECO:0000269|PubMed:1638030, ECO:0000269|PubMed:1679439, ECO:0000269|PubMed:1878597, ECO:0000269|PubMed:2384601, ECO:0000269|PubMed:2568861, ECO:0000269|PubMed:2568862, ECO:0000269|PubMed:2794061, ECO:0000269|PubMed:7772539, ECO:0000269|PubMed:8018926, ECO:0000269|PubMed:8193371, ECO:0000269|PubMed:8364215}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Piruvat Karboksilaz Eksikliği, 266150, Otozomal Resesif (Piruvat Karboksilaz Eksikliği, Benign Tip) (PC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate carboxylase deficiency, 266150, Autosomal recessive (Pyruvate carboxylase deficiency, benign type) (PC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PC	PC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate carboxylase deficiency (PC deficiency) [MIM:266150]: Leads to lactic acidosis, mental retardation and death. It occurs in three forms: mild or type A, severe neonatal or type B, and a very mild lactacidemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piruvat Karboksilaz Eksikliği, 266150, Otozomal Resesif (Piruvat Karboksilaz Eksikliği, Benign Tip) (Prenatal) (PC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate carboxylase deficiency, 266150, Autosomal recessive (Pyruvate carboxylase deficiency, benign type) (PC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PC	PC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate carboxylase deficiency (PC deficiency) [MIM:266150]: Leads to lactic acidosis, mental retardation and death. It occurs in three forms: mild or type A, severe neonatal or type B, and a very mild lactacidemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piruvat Dehidrogenaz E1-Alfa Eksikliği, 312170, Xe Bağlı Dominant; PDHAD (Piruvat Dehidrogenaz Eksikliği) (PDHA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate dehydrogenase E1-alpha deficiency, 312170, X-linked dominant; PDHAD (Pyruvate dehydrogenase deficiency) (PDHA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDHA1	PDHA1, PHE1A, PDHAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase E1-alpha deficiency (PDHAD) [MIM:312170]: An enzymatic defect causing primary lactic acidosis in children. It is associated with a broad clinical spectrum ranging from fatal lactic acidosis in the newborn to chronic neurologic dysfunction with structural abnormalities in the central nervous system without systemic acidosis. {ECO:0000269|PubMed:1293379, ECO:0000269|PubMed:1338114, ECO:0000269|PubMed:1551669, ECO:0000269|PubMed:1909401, ECO:0000269|PubMed:7545958, ECO:0000269|PubMed:7573035, ECO:0000269|PubMed:7757088, ECO:0000269|PubMed:7887409, ECO:0000269|PubMed:7967473, ECO:0000269|PubMed:8032855, ECO:0000269|PubMed:8504306, ECO:0000269|PubMed:8664900, ECO:0000269|PubMed:8844217, ECO:0000269|PubMed:9671272}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piruvat Dehidrogenaz E1-Alfa Eksikliği, 312170, Xe Bağlı Dominant; PDHAD (Piruvat Dehidrogenaz Eksikliği) (Prenatal) (PDHA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate dehydrogenase E1-alpha deficiency, 312170, X-linked dominant; PDHAD (Pyruvate dehydrogenase deficiency) (PDHA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDHA1	PDHA1, PHE1A, PDHAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase E1-alpha deficiency (PDHAD) [MIM:312170]: An enzymatic defect causing primary lactic acidosis in children. It is associated with a broad clinical spectrum ranging from fatal lactic acidosis in the newborn to chronic neurologic dysfunction with structural abnormalities in the central nervous system without systemic acidosis. {ECO:0000269|PubMed:1293379, ECO:0000269|PubMed:1338114, ECO:0000269|PubMed:1551669, ECO:0000269|PubMed:1909401, ECO:0000269|PubMed:7545958, ECO:0000269|PubMed:7573035, ECO:0000269|PubMed:7757088, ECO:0000269|PubMed:7887409, ECO:0000269|PubMed:7967473, ECO:0000269|PubMed:8032855, ECO:0000269|PubMed:8504306, ECO:0000269|PubMed:8664900, ECO:0000269|PubMed:8844217, ECO:0000269|PubMed:9671272}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piruvat Dehidrogenaz E1-Beta Eksikliği, 614111; PDHBD (Piruvat Dehidrogenaz Eksikliği) (PDHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate dehydrogenase E1-beta deficiency, 614111; PDHBD (Pyruvate dehydrogenase deficiency) (PDHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDHB	PDHB, PDHBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase E1-beta deficiency (PDHBD) [MIM:614111]: An enzymatic defect causing primary lactic acidosis in children. It is associated with a broad clinical spectrum ranging from fatal lactic acidosis in the newborn to chronic neurologic dysfunction with structural abnormalities in the central nervous system without systemic acidosis. {ECO:0000269	PubMed:15138885}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Piruvat Dehidrogenaz E1-Beta Eksikliği, 614111; PDHBD (Piruvat Dehidrogenaz Eksikliği) (Prenatal) (PDHB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate dehydrogenase E1-beta deficiency, 614111; PDHBD (Pyruvate dehydrogenase deficiency) (PDHB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDHB	PDHB, PDHBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase E1-beta deficiency (PDHBD) [MIM:614111]: An enzymatic defect causing primary lactic acidosis in children. It is associated with a broad clinical spectrum ranging from fatal lactic acidosis in the newborn to chronic neurologic dysfunction with structural abnormalities in the central nervous system without systemic acidosis. {ECO:0000269	PubMed:15138885}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piruvat Dehidrogenaz E2 Eksikliği, 245348, Otozomal Resesif; PDHDD (Piruvat Dehidrogenaz Eksikliği) (DLAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate dehydrogenase E2 deficiency, 245348, Autosomal recessive; PDHDD (Pyruvate dehydrogenase deficiency) (DLAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLAT	DLAT, PDCE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase E2 deficiency (PDHE2 deficiency) [MIM:245348]: Pyruvate dehydrogenase (PDH) deficiency is a major cause of primary lactic acidosis and neurological dysfunction in infancy and early childhood. In this form of PDH deficiency episodic dystonia is the major neurological manifestation, with other more common features of pyruvate dehydrogenase deficiency, such as hypotonia and ataxia, being less prominent. {ECO:0000269	PubMed:16049940}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Piruvat Dehidrogenaz E2 Eksikliği, 245348, Otozomal Resesif; PDHDD (Piruvat Dehidrogenaz Eksikliği) (DLAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate dehydrogenase E2 deficiency, 245348, Autosomal recessive; PDHDD (Pyruvate dehydrogenase deficiency) (DLAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLAT	DLAT, PDCE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase E2 deficiency (PDHE2 deficiency) [MIM:245348]: Pyruvate dehydrogenase (PDH) deficiency is a major cause of primary lactic acidosis and neurological dysfunction in infancy and early childhood. In this form of PDH deficiency episodic dystonia is the major neurological manifestation, with other more common features of pyruvate dehydrogenase deficiency, such as hypotonia and ataxia, being less prominent. {ECO:0000269	PubMed:16049940}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Pirüvat Dehidrojenaz Fosfataz Eksikliği, 608782, Otozomal Resesif; PDHPD (Piruvat Dehidrogenaz Eksikliği) (PDP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate dehydrogenase phosphatase deficiency, 608782, Autosomal recessive; PDHPD (Pyruvate dehydrogenase deficiency) (PDP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDP1	PDP1, PPM2C, PDP1, PDPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate dehydrogenase phosphatase deficiency (PDP deficiency) [MIM:608782]: Results in lactic acidosis leading to neurological dysfunction. {ECO:0000269	PubMed:15855260}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Pirüvat Dehidrojenaz Fosfataz Eksikliği, 608782, Otozomal Resesif; PDHPD (Piruvat Dehidrogenaz Eksikliği) (Prenatal) (PDP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate dehydrogenase phosphatase deficiency, 608782, Autosomal recessive; PDHPD (Pyruvate dehydrogenase deficiency) (PDP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDP1	PDP1, PPM2C, PDP1, PDPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate dehydrogenase phosphatase deficiency (PDP deficiency) [MIM:608782]: Results in lactic acidosis leading to neurological dysfunction. {ECO:0000269	PubMed:15855260}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piruvat Kinaz Eksikliği, 266200, Otozomal Resesif (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (PKLR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Pyruvate kinase deficiency, 266200, Autosomal recessive (Hemolytic anemia due to red cell pyruvate kinase deficiency) (PKLR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PKLR	PKLR, PK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269	PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Piruvat Kinaz Eksikliği, 266200, Otozomal Resesif (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (MLPA)	Pyruvate kinase deficiency, 266200, Autosomal recessive (Hemolytic anemia due to red cell pyruvate kinase deficiency) (MLPA)	PKLR	PKLR, PK1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269	PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Piruvat Kinaz Eksikliği, 266200, Otozomal Resesif (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (Prenatal) (PKLR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Pyruvate kinase deficiency, 266200, Autosomal recessive (Hemolytic anemia due to red cell pyruvate kinase deficiency) (PKLR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PKLR	PKLR, PK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269	PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Piruvat Kinaz Eksikliği, 266200, Otozomal Resesif (Hemolitik Anemi, Eritrosit Piruvat Kinaz Eksikliğine Bağlı) (Prenatal) (MLPA)	Pyruvate kinase deficiency, 266200, Autosomal recessive (Hemolytic anemia due to red cell pyruvate kinase deficiency) (Prenatal) (MLPA)	PKLR	PKLR, PK1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pyruvate kinase hyperactivity (PKHYP) [MIM:102900]: Autosomal dominant phenotype characterized by increase of red blood cell ATP. {ECO:0000269	PubMed:9090535}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
QF - PCR (13, 18, 21, X, Y)	QF-PCR (13, 18, 21, X, Y)	.	.	QF-PCR	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		1	4	.	.	3	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Quebec Trombosit Bozukluğu, 601709, Otozomal Dominant; QPD (Quebec Trombosit Bozukluğu) (PLAU geni) (Tüm Gen Dizi Analizi) (Postnatal)	Quebec platelet disorder, 601709, Autosomal dominant; QPD (Quebec platelet disorder) (PLAU gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAU	PLAU, URK, QPD, BDPLT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Quebec platelet disorder (QPD) [MIM:601709]: An autosomal dominant bleeding disorder due to a gain-of-function defect in fibrinolysis. Although affected individuals do not exhibit systemic fibrinolysis, they show delayed onset bleeding after challenge, such as surgery. The hallmark of the disorder is markedly increased PLAU levels within platelets, which causes intraplatelet plasmin generation and secondary degradation of alpha-granule proteins. {ECO:0000269	PubMed:20007542}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Soru İşareti Kulaklar, İnme, 612798, Otozomal Dominant; QME (Auriculocondylar Sendromu) (EDN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Question mark ears, isolated, 612798, Autosomal dominant; QME (Auriculocondylar syndrome) (EDN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN1	EDN1, ARCND3, QME, HDLCQ7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Auriculocondylar syndrome 3 (ARCND3) [MIM:615706]: A craniofacial malformation syndrome characterized by variable mandibular anomalies, including mild to severe micrognathia, temporomandibular joint ankylosis, cleft palate, and a characteristic ear malformation that consists of separation of the lobule from the external ear, giving the appearance of a question mark (question-mark ear). Other frequently described features include prominent cheeks, cupped and posteriorly rotated ears, preauricular tags, and microstomia. {ECO:0000269	PubMed:24268655}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amegakaryositik Trombositopeni-Radyoulnar Sinostoz 1, 605432, Otozomal Dominant; RUSAT1 (Radioulnar Sinostoz-Amegakaryositik Trombositopeni Sendromu) (HOXA11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432, Autosomal dominant; RUSAT1 (Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome) (HOXA11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXA11	HOXA11, HOX1I, RUSAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Radioulnar synostosis with amegakaryocytic thrombocytopenia (RSAT) [MIM:605432]: The syndrome consists of an unusual association of bone marrow failure and skeletal defects. Patients have the same skeletal defects, the proximal fusion of the radius and ulna, resulting in extremely limited pronation and supination of the forearm. Some patients have also symptomatic thrombocytopenia, with bruising and bleeding problems since birth, necessitating correction by bone marrow or umbilical-cord stem- cell transplantation. {ECO:0000269	PubMed:11101832}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Amegakaryositik Trombositopeni-Radyoulnar Sinostoz 1, 605432, Otozomal Dominant; RUSAT1 (Radioulnar Sinostoz-Amegakaryositik Trombositopeni Sendromu) (Prenatal) (HOXA11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Radioulnar synostosis with amegakaryocytic thrombocytopenia 1, 605432, Autosomal dominant; RUSAT1 (Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome) (HOXA11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HOXA11	HOXA11, HOX1I, RUSAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Radioulnar synostosis with amegakaryocytic thrombocytopenia (RSAT) [MIM:605432]: The syndrome consists of an unusual association of bone marrow failure and skeletal defects. Patients have the same skeletal defects, the proximal fusion of the radius and ulna, resulting in extremely limited pronation and supination of the forearm. Some patients have also symptomatic thrombocytopenia, with bruising and bleeding problems since birth, necessitating correction by bone marrow or umbilical-cord stem- cell transplantation. {ECO:0000269	PubMed:11101832}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Amegakaryositik Trombositopeni-Radyoulnar Sinostoz 2, 616738, Otozomal Dominant; RUSAT2 (Radioulnar Sinostoz-Amegakaryositik Trombositopeni Sendromu) (MECOM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738, Autosomal dominant; RUSAT2 (Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome) (MECOM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECOM	MECOM, EVI1, RUSAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving EVI1 is a cause of chronic myelogenous leukemia (CML). Translocation t(3;21)(q26;q22) with RUNX1/AML1. {ECO:0000269	PubMed:8313895;}.	EDTA Blood Sample
Amegakaryositik Trombositopeni-Radyoulnar Sinostoz 2, 616738, Otozomal Dominant; RUSAT2 (Radioulnar Sinostoz-Amegakaryositik Trombositopeni Sendromu) (Prenatal) (MECOM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Radioulnar synostosis with amegakaryocytic thrombocytopenia 2, 616738, Autosomal dominant; RUSAT2 (Radio-ulnar synostosis-amegakaryocytic thrombocytopenia syndrome) (MECOM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECOM	MECOM, EVI1, RUSAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving EVI1 is a cause of chronic myelogenous leukemia (CML). Translocation t(3;21)(q26;q22) with RUNX1/AML1. {ECO:0000269	PubMed:8313895;}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Raine Sendromu, 259775, Otozomal Resesif; RNS (Letal Osteosklerotik Kemik Displazisi (Raine Sendromu)) (FAM20C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Raine syndrome, 259775, Autosomal recessive; RNS (Lethal osteosclerotic bone dysplasia) (FAM20C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM20C	FAM20C, DMP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Raine syndrome (RNS) [MIM:259775]: Autosomal recessive osteosclerotic bone dysplasia with neonatal lethal outcome. Clinical features include generalized osteosclerosis, craniofacial dysplasia and micromelia. {ECO:0000269	PubMed:17924334, ECO:0000269	PubMed:22582013, ECO:0000269	PubMed:25789606}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Raine Sendromu, 259775, Otozomal Resesif; RNS (Letal Osteosklerotik Kemik Displazisi (Raine Sendromu)) (Prenatal) (FAM20C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Raine syndrome, 259775, Autosomal recessive; RNS (Lethal osteosclerotic bone dysplasia) (FAM20C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAM20C	FAM20C, DMP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Raine syndrome (RNS) [MIM:259775]: Autosomal recessive osteosclerotic bone dysplasia with neonatal lethal outcome. Clinical features include generalized osteosclerosis, craniofacial dysplasia and micromelia. {ECO:0000269	PubMed:17924334, ECO:0000269	PubMed:22582013, ECO:0000269	PubMed:25789606}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
RAPADILINO Sendromu, 266280, Otozomal Resesif (RAPADILINO Sendromu) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	RAPADILINO syndrome, 266280, Autosomal recessive (RAPADILINO syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. {ECO:0000269	PubMed:15964893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
RAPADILINO Sendromu, 266280, Otozomal Resesif (RAPADILINO Sendromu) (Prenatal) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	RAPADILINO syndrome, 266280, Autosomal recessive (RAPADILINO syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. {ECO:0000269	PubMed:15964893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rapid FISH (13, 18, 21, X, Y) (Prenatal)	Rapid FISH (13, 18, 21, X, Y) (Prenatal)	.	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		10	30	.	.	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

HIV1 Enfeksiyonundan AIDS'e Hızlı İlerleme, 609423 (CX3CR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rapid progression to AIDS from HIV1 infection, 609423 (CX3CR1 gen) (Sequence Analysis-All Coding Exons) (Postnatal)	CX3CR1	CX3CR1, GPR13, V28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 12 (ARMD12) [MIM:613784]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269	PubMed:15208270}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Rapp-Hodgkin Sendromu, 129400, Otozomal Dominant; RHS (Rapp-Hodgkin Sendromu) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rapp-Hodgkin syndrome, 129400, Autosomal dominant; RHS (Rapp-Hodgkin syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rapp-Hodgkin Sendromu, 129400, Otozomal Dominant; RHS (Rapp-Hodgkin Sendromu) (Prenatal) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rapp-Hodgkin syndrome, 129400, Autosomal dominant; RHS (Rapp-Hodgkin syndrome) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269	PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
RAS-İlişkili Otoimmün Lökoproliferatif Bozukluk, 614470, Otozomal Dominant; RALD (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	RAS-associated autoimmune leukoproliferative disorder, 614470, Autosomal dominant; RALD (RAS-associated autoimmune leukoproliferative disease) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
RAS-İlişkili Otoimmün Lökoproliferatif Bozukluk, 614470, Otozomal Dominant; RALD (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (MLPA)	RAS-associated autoimmune leukoproliferative disorder, 614470, Autosomal dominant; RALD (RAS-associated autoimmune leukoproliferative disease) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
RAS-İlişkili Otoimmün Lökoproliferatif Bozukluk, 614470, Otozomal Dominant; RALD (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (Prenatal) (KRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	RAS-associated autoimmune leukoproliferative disorder, 614470, Autosomal dominant; RALD (RAS-associated autoimmune leukoproliferative disease) (KRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
RAS-İlişkili Otoimmün Lökoproliferatif Bozukluk, 614470, Otozomal Dominant; RALD (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (Prenatal) (MLPA)	RAS-associated autoimmune leukoproliferative disorder, 614470, Autosomal dominant; RALD (RAS-associated autoimmune leukoproliferative disease) (Prenatal) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
RAS-İlişkili Otoimmün Lenfoproliferatif Sendrom Tip 4, Somatik, 614470 (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (RAS-associated autoimmune leukoproliferative disease) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
RAS-İlişkili Otoimmün Lenfoproliferatif Sendrom Tip 4, Somatik, 614470 (RAS-İlişkili Otoimmün Lökoproliferatif Hastalık) (Prenatal) (NRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470 (RAS-associated autoimmune leukoproliferative disease) (NRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Melanocytic nevus syndrome, congenital (CMNS) [MIM:137550]: A syndrome characterized by congenital pigmentary skin lesions which can occur at any site and can cover most of the body surface. These lesions may or may not be hairy. Congenital melanocytic nevi are associated with neuromelanosis (the presence of melanin-producing cells within the brain parenchyma or leptomeninges). Less commonly they are associated with malignant melanoma in childhood, both in the skin and the central nervous system. CMNS patients also tend to have a characteristic facial appearance, including wide or prominent forehead, periorbital fullness, small short nose with narrow nasal bridge, round face, full cheeks, prominent premaxilla, and everted lower lip. {ECO:0000269|PubMed:18633438, ECO:0000269|PubMed:23392294}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rekombinasyon Oranı QTL 1, 612042 (RNF212 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Recombination rate QTL 1, 612042 (RNF212 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF212	RNF212, ZHP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Reducing Body Miyopati, X'e Bağlı 1a, Şiddetli, İnfantil Veya Erken Çocuklukta Başlayan, 300717, X'e Bağlı Dominant (Reducing Body Miyopati) (FHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717, X-linked dominant (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Reducing Body Miyopati, X'e Bağlı 1a, Şiddetli, İnfantil Veya Erken Çocuklukta Başlayan, 300717, X'e Bağlı Dominant (Reducing Body Miyopati) (Prenatal) (FHL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Reducing body myopathy, X-linked 1a, severe, infantile or early childhood onset, 300717, X-linked dominant (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Reducing Body Myopati, X'e Bağlı 1b, Geç Çocukluk Veya Yetişkin Başlangıçlı, 300718, X'e Bağlı (Reducing Body Miyopati) (FHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718, X-linked (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Reducing Body Miyopati, X'e Bağlı 1b, Geç Çocukluk Veya Yetişkin Başlangıçlı, 300718, X'e Bağlı (Reducing Body Miyopati) (Prenatal) (FHL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Reducing body myopathy, X-linked 1b, with late childhood or adult onset, 300718, X-linked (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Reducing Body Myopathy, X'e Bağlı 1b-Geç Çocukluk veya Yetişkin Başlangıçlı; Rbmx1b (Reducing Body Miyopati) (FHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	REDUCING BODY MYOPATHY, X-LINKED 1B, WITH LATE CHILDHOOD OR ADULT ONSET; RBMX1B (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Reducing Body Myopathy, X'e Bağlı 1b-Geç Çocukluk veya Yetişkin Başlangıçlı; Rbmx1b (Reducing Body Miyopati) (Prenatal) (FHL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	REDUCING BODY MYOPATHY, X-LINKED 1B, WITH LATE CHILDHOOD OR ADULT ONSET; RBMX1B (Reducing body myopathy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269	PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Refsum Hastalığı, 266500, Otozomal Resesif (Refsum Hastalığı) (PHYH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Refsum disease, 266500, Autosomal recessive (Refsum disease) (PHYH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHYH	PHYH, PAHX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Refsum Hastalığı, 266500, Otozomal Resesif (Refsum Hastalığı) (Prenatal) (PHYH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Refsum disease, 266500, Autosomal recessive (Refsum disease) (PHYH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHYH	PHYH, PAHX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Refsum Hastalığı, Classic (Refsum Hastalığı) (PHYH geni) (Tüm Gen Dizi Analizi) (Postnatal)	REFSUM DISEASE, CLASSIC (Refsum disease) (PHYH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PHYH	PHYH, PAHX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Refsum Hastalığı, Classic (Refsum Hastalığı) (Prenatal) (PHYH geni) (Tüm Gen Dizi Analizi) (Prenatal)	REFSUM DISEASE, CLASSIC (Refsum disease) (PHYH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PHYH	PHYH, PAHX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal Karsinom, Kromofob, Somatik, 144700 (FLCN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal carcinoma, chromophobe, somatic, 144700 (FLCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLCN	FLCN, BHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample

Renal Hücreli Karsinom, 144700 (HNF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, 144700 (HNF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Hücreli Karsinom, 144700 (MLPA)	Renal cell carcinoma, 144700 (MLPA)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Hücreli Karsinom, 144700 (Kalıtsal Berrak Hücreli Renal Hücre Karsinomu) (RNF139 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, 144700 (Hereditary clear cell renal cell carcinoma) (RNF139 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF139	RNF139, TRC8, RCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. Note=The disease may be caused by mutations affecting the gene represented in this entry. A chromosomal aberration involving RNF139 has been found in a lymphoblastoid cell line established from a family with renal cell carcinoma and thyroid carcinoma. Translocation (3;8)(q14.2;q24.1) with FHIT. RNF139 is found to be fused to FHIT and disrupted within the steroi-sensing domain. In contrast, the FHIT coding region is maintained and expressed. Sporadic cases of renal carcinoma, where an acquired mutation in RNF139 results in the duplication of 12 nucleotides in the 5'-UTR, has also been identified.;	EDTA Blood Sample
Renal Hücreli Karsinom, 144700 (Kalıtsal Berrak Hücreli Renal Hücre Karsinomu) (HNF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, 144700 (Hereditary clear cell renal cell carcinoma) (HNF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hepatic adenomas familial (HEPAF) [MIM:142330]: Rare benign liver tumors of presumable epithelial origin that develop in an otherwise normal liver. Hepatic adenomas may be single or multiple. They consist of sheets of well-differentiated hepatocytes that contain fat and glycogen and can produce bile. Bile ducts or portal areas are absent. Kupffer cells, if present, are reduced in number and are non-functional. Conditions associated with adenomas are insulin-dependent diabetes mellitus and glycogen storage diseases (types I and 3). Note=The disease is caused by mutations affecting the gene represented in this entry. Bi-allelic inactivation of HNF1A, whether sporadic or associated with MODY3, may be an early step in the development of some hepatocellular carcinomas.	EDTA Blood Sample
Renal Hücreli Karsinom, 144700 (Kalıtsal Berrak Hücreli Renal Hücre Karsinomu) (MLPA)	Renal cell carcinoma, 144700 (Hereditary clear cell renal cell carcinoma) (MLPA)	HNF1A	HNF1A, TCF1, MODY3, IDDM20	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hepatic adenomas familial (HEPAF) [MIM:142330]: Rare benign liver tumors of presumable epithelial origin that develop in an otherwise normal liver. Hepatic adenomas may be single or multiple. They consist of sheets of well-differentiated hepatocytes that contain fat and glycogen and can produce bile. Bile ducts or portal areas are absent. Kupffer cells, if present, are reduced in number and are non-functional. Conditions associated with adenomas are insulin-dependent diabetes mellitus and glycogen storage diseases (types I and 3). Note=The disease is caused by mutations affecting the gene represented in this entry. Bi-allelic inactivation of HNF1A, whether sporadic or associated with MODY3, may be an early step in the development of some hepatocellular carcinomas.	EDTA Blood Sample
Renal Hücreli Karsinom, 144700; RCC (Kalıtsal Berrak Hücreli Renal Hücre Karsinomu) (DIRC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, 144700; RCC (Hereditary clear cell renal cell carcinoma) (DIRC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIRC2	DIRC2, RCC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving DIRC2 has been found in a family with renal carcinoma. Translocation t(2;3)(q35;q21) (PubMed:11912179). {ECO:0000269	PubMed:11912179}.;	EDTA Blood Sample
Renal Hücreli Karsinom, Berrak Hücreli, Somatik, 144700 (OGG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, clear cell, somatic, 144700 (OGG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OGG1	OGG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Renal Hücreli Karsinom, Papiller, 1, 300854 (Translokasyon Renal Hücreli Karsinom) (TFE3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, papillary, 1, 300854 (Translocation renal cell carcinoma) (TFE3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFE3	TFE3, RCCX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving TFE3 is found in patients with alveolar soft part sarcoma. Translocation t(X;17)(p11.q25) with ASPSCR1 forms a ASPSCR1-TFE3 fusion protein. {ECO:0000269	PubMed:11358836}.	EDTA Blood Sample
Renal Hücreli Karsinom, Papiller, 1, Ailesel ve Somatik, 605074 (MET geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, papillary, 1, familial and somatic, 605074 (MET gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MET	MET, DFNB97, OSFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Renal Hücreli Karsinom, Papiller, 605074; RCCP1 (Ailesel Papiller Böbrek Hücreli Karsinom) (PRCC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, papillary, 605074; RCCP1 (Hereditary papillary renal cell carcinoma) (PRCC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRCC	PRCC, RCCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renal Hücreli Karsinom, Somatik, 144700 (VHL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cell carcinoma, somatic, 144700 (VHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VHL	VHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Renal cell carcinoma (RCC) [MIM:144700]: Renal cell carcinoma is a heterogeneous group of sporadic or hereditary carcinoma derived from cells of the proximal renal tubular epithelium. It is subclassified into clear cell renal carcinoma (non-papillary carcinoma), papillary renal cell carcinoma, chromophobe renal cell carcinoma, collecting duct carcinoma with medullary carcinoma of the kidney, and unclassified renal cell carcinoma. Clear cell renal cell carcinoma is the most common subtype. {ECO:0000269	PubMed:11986208}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Renal Kistler ve Diyabet Sendromu, 137920, Otozomal Dominant; RCAD (Renal Kist - Diyabet Sendromu) (HNF1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal cysts and diabetes syndrome, 137920, Autosomal dominant; RCAD (Renal cysts and diabetes syndrome) (HNF1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Kistler ve Diyabet Sendromu, 137920, Otozomal Dominant; RCAD (Renal Kist - Diyabet Sendromu) (MLPA)	Renal cysts and diabetes syndrome, 137920, Autosomal dominant; RCAD (Renal cysts and diabetes syndrome) (MLPA)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Kistler ve Diyabet Sendromu, 137920, Otozomal Dominant; RCAD (Renal Kist - Diyabet Sendromu) (Prenatal) (MLPA)	Renal cysts and diabetes syndrome, 137920, Autosomal dominant; RCAD (Renal cysts and diabetes syndrome) (Prenatal) (MLPA)	HNF1B	HNF1B, TCF2, HNF2, MODY5, FJHN, HPC11	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269	PubMed:12161522}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal Displazi, Kistik, Yatkınlık, 601331, Otozomal Dominant; CYSRD (BICC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal dysplasia, cystic, susceptibility to, 601331, Autosomal dominant; CYSRD (BICC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BICC1	BICC1, BICC, CYSRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal dysplasia, cystic (CYSRD) [MIM:601331]: An anomaly of the kidney characterized by numerous renal cysts and apparent disorder of differentiation of the renal parenchyma. Kidney of affected individuals lack the normal renal bean shape, and the collection drainage system. The cystic, dysplastic kidney contains undifferentiated mesenchyme, cartilaginous tissue, and immature collecting ducts. {ECO:0000269	PubMed:21922595}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal Displazi, Kistik, Yatkınlık, 601331, Otozomal Dominant; CYSRD (Prenatal) (BICC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renal dysplasia, cystic, susceptibility to, 601331, Autosomal dominant; CYSRD (BICC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BICC1	BICC1, BICC, CYSRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Renal dysplasia, cystic (CYSRD) [MIM:601331]: An anomaly of the kidney characterized by numerous renal cysts and apparent disorder of differentiation of the renal parenchyma. Kidney of affected individuals lack the normal renal bean shape, and the collection drainage system. The cystic, dysplastic kidney contains undifferentiated mesenchyme, cartilaginous tissue, and immature collecting ducts. {ECO:0000269	PubMed:21922595}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal Glikozüri, 233100, Otozomal Resesif, Otozomal Dominant; GLYS (Renal Glukozüri) (SLC5A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal glucosuria, 233100, Autosomal recessive, Autosomal dominant; GLYS (Renal glucosuria) (SLC5A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC5A2	SLC5A2, SGLT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal glucosuria (GLYS) [MIM:233100]: A disorder characterized by persistent isolated glucosuria, normal fasting serum glucose concentration, decreased renal tubular resorption of glucose from the urine, and absence of any other signs of tubular dysfunction. {ECO:0000269	PubMed:14614622}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal Hipodisplazi / Aplazi 1, 191830, Otozomal Resesif; RHDA1 (Renal Agenez) (ITGA8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal hypodysplasia/aplasia 1, 191830, Autosomal recessive; RHDA1 (Renal agenesis) (ITGA8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA8	ITGA8, RHDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renal Hipodisplazi / Aplazi 1, 191830, Otozomal Resesif; RHDA1 (Renal Agenez) (Prenatal) (ITGA8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renal hypodysplasia/aplasia 1, 191830, Autosomal recessive; RHDA1 (Renal agenesis) (ITGA8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITGA8	ITGA8, RHDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal Hipodisplazi / Aplazi 2, 615721, Otozomal Resesif; RHDA2 (Renal Agenez) (FGF20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal hypodysplasia/aplasia 2, 615721, Autosomal recessive; RHDA2 (Renal agenesis) (FGF20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF20	FGF20, RHDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renal Hipodisplazi / Aplazi 2, 615721, Otozomal Resesif; RHDA2 (Renal Agenez) (Prenatal) (FGF20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renal hypodysplasia/aplasia 2, 615721, Autosomal recessive; RHDA2 (Renal agenesis) (FGF20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGF20	FGF20, RHDA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Renal Tübüler Asidoz, Sağırlık 267300, Otozomal Resesif (Distal Renal Tübüler Asidoz) (ATP6V1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular acidosis with deafness, 267300, Autosomal recessive (Distal renal tubular acidosis) (ATP6V1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V1B1	ATP6B1, VPP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular acidosis, distal, with progressive nerve deafness (dRTA-D) [MIM:267300]: An autosomal recessive disease characterized by the association of renal distal tubular acidosis with sensorineural hearing loss. Distal renal tubular acidosis is characterized by reduced ability to acidify urine, variable hyperchloremic hypokalemic metabolic acidosis, nephrocalcinosis, and nephrolithiasis. It is due to functional failure of alpha- intercalated cells of the cortical collecting duct of the distal nephron, where vectorial proton transport is required for urinary acidification. {ECO:0000269|PubMed:12414817, ECO:0000269|PubMed:12579397, ECO:0000269|PubMed:9916796}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal Tübüler Asidoz, Distal, AD, 179800, Otozomal Dominant (Distal Renal Tübüler Asidoz) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular acidosis, distal, AD, 179800, Autosomal dominant (Distal renal tubular acidosis) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Asidoz, Distal, AD, 179800, Otozomal Dominant (Distal Renal Tübüler Asidoz) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular acidosis, distal, AR, 611590, autosomal recessive (Distal renal tubular acidosis) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Asidoz, Distal, Otozomal Resesif, Beraber 602722 RTA0R (Distal Renal Tübüler Asidoz) (ATP6V0A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular acidosis, distal, autosomal recessive, 602722, RTA0R (Distal renal tubular acidosis) (ATP6V0A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V0A4	ATP6N1B, VPP2, RTA1C, RTA0R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Renal Tübüler Asidoz, Distal, Hemolitik Anemi (Distal Renal Tübüler Asidoz) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	RENAL TUBULAR ACIDOSIS, DISTAL, WITH HEMOLYTIC ANEMIA (Distal renal tubular acidosis) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Asidoz, Proksimal, Oküler Anormallikler, 604278, Otozomal Resesif (Proksimal Renal Tübüler Asidoz) (SLC4A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular acidosis, proximal, with ocular abnormalities, 604278, Autosomal recessive (Proximal renal tubular acidosis) (SLC4A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A4	SLC4A4, NBC1, KNBC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular acidosis, proximal, with ocular abnormalities and mental retardation (pRTA-OA) [MIM:604278]: An extremely rare autosomal recessive syndrome characterized by short stature, profound proximal renal tubular acidosis, mental retardation, bilateral glaucoma, cataracts and bandkeratopathy. pRTA is due to a failure of the proximal tubular cells to reabsorb filtered bicarbonate from the urine, leading to urinary bicarbonate wasting and subsequent acidemia. {ECO:0000269|PubMed:10545938, ECO:0000269|PubMed:15471865, ECO:0000269|PubMed:15713912, ECO:0000269|PubMed:15930088, ECO:0000269|PubMed:16636648, ECO:0000269|PubMed:17661077}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Disgenezis, 267430, Otozomal Resesif (Renal Tübüler Disgenezis) (AGTR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular dysgenesis, 267430, Autosomal recessive (Renal tubular dysgenesis) (AGTR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGTR1	AGTR1, AGTR1A, AT2R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal Tübüler Disgenezis, 267430, Otozomal Resesif (Renal Tübüler Disgenezis) (AGT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular dysgenesis, 267430, Autosomal recessive (Renal tubular dysgenesis) (AGT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGT	AGT, SERPINA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Essential hypertension (EHT) [MIM:145500]: A condition in which blood pressure is consistently higher than normal with no identifiable cause. {ECO:0000269|PubMed:1394429, ECO:0000269|PubMed:8513325}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Disgenezis, 267430, Otozomal Resesif (Renal Tübüler Disgenezis) (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular dysgenesis, 267430, Autosomal recessive (Renal tubular dysgenesis) (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal Tübüler Disgenezis, 267430, Otozomal Resesif, RTD (Renal Tübüler Disgenezis) (REN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal tubular dysgenesis, 267430, Autosomal recessive; RTD (Renal tubular dysgenesis) (REN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REN	REN, HNFJ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal, Hepatik, Pankreatik Displazi 1, 208540, Otozomal Resesif, RHPD1 (Renal-Karaciğer-Pankreas Displazi) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal-hepatic-pancreatic dysplasia 1, 208540, Autosomal recessive; RHPD1 (Renal-hepatic-pancreatic dysplasia) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. {ECO:0000269	PubMed:18371931}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Renal, Hepatik, Pankreatik Displazi 1, 208540, Otozomal Resesif, RHPD1 (Renal-Karaciğer-Pankreas Displazi) (Prenatal) (NPHP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renal-hepatic-pancreatic dysplasia 1, 208540, Autosomal recessive; RHPD1 (Renal-hepatic-pancreatic dysplasia) (NPHP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP3	NPHP3, NPH3, RHPD1, MKS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Renal-hepatic-pancreatic dysplasia 1 (RHPD1) [MIM:208540]: A disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. {ECO:0000269	PubMed:18371931}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renal-Hepatik-Pankreatik Displazi 2, 615415, Otozomal Resesif (Renal-Karaciğer-Pankreas Displazi) (NEK8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renal-hepatic-pancreatic dysplasia 2, 615415, Autosomal recessive (Renal-hepatic-pancreatic dysplasia) (NEK8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK8	NEK8, JCK, NPHP9, RHPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal-hepatic-pancreatic dysplasia 2 (RHPD2) [MIM:615415]: A form of renal-hepatic-pancreatic dysplasia, a disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. {ECO:0000269	PubMed:23418306}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renal-Hepatik-Pankreatik Displazi 2, 615415, Otozomal Resesif (Renal-Karaciğer-Pankreas Displazi) (Prenatal) (NEK8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renal-hepatic-pancreatic dysplasia 2, 615415, Autosomal recessive (Renal-hepatic-pancreatic dysplasia) (NEK8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEK8	NEK8, JCK, NPHP9, RHPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Renal-hepatic-pancreatic dysplasia 2 (RHPD2) [MIM:615415]: A form of renal-hepatic-pancreatic dysplasia, a disease characterized by cystic malformations of the kidneys, liver, and pancreas. The pathological findings consist of multicystic dysplastic kidneys, dilated and dysgenetic bile ducts, a dysplastic pancreas with dilated ducts, cysts, fibrosis and inflammatory infiltrates. {ECO:0000269	PubMed:23418306}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Renpenning Sendromu, 309500, X'e Bağlı Resesif, RENS1 (Renpenning Sendromu) (PQBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Renpenning syndrome, 309500, X-linked recessive; RENS1 (Renpenning syndrome) (PQBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PQBP1	PQBP1, NPW38, SHS, MRX55, MRXS3, RENS1, MRXS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renpenning syndrome 1 (RENS1) [MIM:309500]: A X-linked mental retardation syndrome characterized by mental retardation, microcephaly, short stature, and small testes. The craniofacies tends to be narrow and tall with upslanting palpebral fissures, abnormal nasal configuration, cupped ears, and short philtrum. The nose may appear long or bulbous, with overhanging columella. Less consistent manifestations include ocular colobomas, cardiac malformations, cleft palate, and anal anomalies. {ECO:0000269|PubMed:14634649, ECO:0000269|PubMed:16740914}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Renpenning Sendromu, 309500, X'e Bağlı Resesif, RENS1 (Renpenning Sendromu) (Prenatal) (PQBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Renpenning syndrome, 309500, X-linked recessive; RENS1 (Renpenning syndrome) (PQBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PQBP1	PQBP1, NPW38, SHS, MRX55, MRXS3, RENS1, MRXS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		15	30	20	Prenatal	DISEASE: Renpenning syndrome 1 (RENS1) [MIM:309500]: A X-linked mental retardation syndrome characterized by mental retardation, microcephaly, short stature, and small testes. The craniofacies tends to be narrow and tall with upslanting palpebral fissures, abnormal nasal configuration, cupped ears, and short philtrum. The nose may appear long or bulbous, with overhanging columella. Less consistent manifestations include ocular colobomas, cardiac malformations, cleft palate, and anal anomalies. {ECO:0000269	PubMed:14634649, ECO:0000269	PubMed:16740914}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
G6PD Eksikliğine Bağlı Sıtmaya Direnç, 611162 (Sıtma) (G6PD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Resistance to malaria due to G6PD deficiency, 611162 (Malaria) (G6PD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	G6PD	G6PD, G6PD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, non-spherocytic hemolytic, due to G6PD deficiency (NSHA) [MIM:300908]: A disease characterized by G6PD deficiency, acute hemolytic anemia, fatigue, back pain, and jaundice. In most patients, the disease is triggered by an exogenous agent, such as some drugs, food, or infection. Increased unconjugated bilirubin, lactate dehydrogenase, and reticulocytosis are markers of the disorder. Although G6PD deficiency can be life-threatening, most patients are asymptomatic throughout their life. {ECO:0000269	PubMed:1611091}. Note=The disease is caused by mutations affecting the gene represented in this entry. Deficiency of G6PD is associated with hemolytic anemia in two different situations. First, in areas in which malaria has been endemic, G6PD-deficiency alleles have reached high frequencies (1% to 50%) and deficient individuals, though essentially asymptomatic in the steady state, have a high risk of acute hemolytic attacks. Secondly, sporadic cases of G6PD deficiency occur at a very low frequencies, and they usually present a more severe phenotype. Several types of NSHA are recognized. Class-I variants are associated with severe NSHA; class-II have an activity <10% of normal; class-III have an activity of 10% to 60% of normal; class- IV have near normal activity.;	EDTA Blood Sample

G6PD Eksikliğine Bağlı Sıtmaya Direnç, 611162 (Sıtma) (Prenatal) (G6PD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Resistance to malaria due to G6PD deficiency, 611162 (Malaria) (G6PD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	G6PD	G6PD, G6PD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Anemia, non-spherocytic hemolytic, due to G6PD deficiency (NSHA) [MIM:300908]: A disease characterized by G6PD deficiency, acute hemolytic anemia, fatigue, back pain, and jaundice. In most patients, the disease is triggered by an exogenous agent, such as some drugs, food, or infection. Increased unconjugated bilirubin, lactate dehydrogenase, and reticulocytosis are markers of the disorder. Although G6PD deficiency can be life-threatening, most patients are asymptomatic throughout their life. {ECO:0000269	PubMed:1611091}. Note=The disease is caused by mutations affecting the gene represented in this entry. Deficiency of G6PD is associated with hemolytic anemia in two different situations. First, in areas in which malaria has been endemic, G6PD-deficiency alleles have reached high frequencies (1% to 50%) and deficient individuals, though essentially asymptomatic in the steady state, have a high risk of acute hemolytic attacks. Secondly, sporadic cases of G6PD deficiency occur at a very low frequencies, and thoy usually present a more severe phenotype. Several types of NSHA are recognized. Class-I variants are associated with severe NSHA; class-II have an activity <10% of normal; class-III have an activity of 10% to 60% of normal; class- IV have near normal activity.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İmatinib (Gleevec) Direnci (ABL1 geni) (G250E, Y253H, E255K, T315I, F317L, M315T dahil) (Ekzon 4 - 10) (ABL1 geni) (Dizi Analizi) (Postnatal)	Resistant or Intolerant to Imatinib (Gleevec) (ABL1 gene) (G250E, Y253H, E255K, T315I, F317L, M315T) (Exon 4-10) (ABL1 gene) (Sequence Analysis) (Postnatal)	ABL1	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, chronic myeloid (CML) [MIM:608232]: A clonal myeloproliferative disorder of a pluripotent stem cell with a specific cytogenetic abnormality, the Philadelphia chromosome (Ph), involving myeloid, erythroid, megakaryocytic, B-lymphoid, and sometimes T-lymphoid cells, but not marrow fibroblasts. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample / EDTA'lı kemik iliği
İmatinib (Gleevec) Direnci (ABL1 geni) (G250E, Y253H, E255K, T315I, F317L, M315T dahil) (Ekzon 4 - 10) (ABL1 geni) (Dizi Analizi) (Postnatal)	Resistant or Intolerant to Imatinib (Gleevec) (ABL1 gene) (G250E, Y253H, E255K, T315I, F317L, M315T) (Exon 4-10) (ABL1 gene) (Sequence Analysis) (Postnatal)	ABL1	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, chronic myeloid (CML) [MIM:608232]: A clonal myeloproliferative disorder of a pluripotent stem cell with a specific cytogenetic abnormality, the Philadelphia chromosome (Ph), involving myeloid, erythroid, megakaryocytic, B-lymphoid, and sometimes T-lymphoid cells, but not marrow fibroblasts. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample / EDTA'lı kemik iliği
Kısıtlayıcı Dermopati, Letal, 275210, Otozomal Resesif (Restriktif Dermopati) (ZMPSTE24 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Restrictive dermopathy, lethal, 275210, Autosomal recessive (Restrictive dermopathy) (ZMPSTE24 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısıtlayıcı Dermopati, Letal, 275210, Otozomal Resesif (Restriktif Dermopati) (MLPA)	Restrictive dermopathy, lethal, 275210, Autosomal recessive (Restrictive dermopathy) (MLPA)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısıtlayıcı Dermopati, Letal, 275210, Otozomal Resesif (Restriktif Dermopati) (Prenatal) (ZMPSTE24 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Restrictive dermopathy, lethal, 275210, Autosomal recessive (Restrictive dermopathy) (ZMPSTE24 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısıtlayıcı Dermopati, Letal, 275210, Otozomal Resesif (Restriktif Dermopati) (Prenatal) (MLPA)	Restrictive dermopathy, lethal, 275210, Autosomal recessive (Restrictive dermopathy) (Prenatal) (MLPA)	ZMPSTE24	ZMPSTE24, FACE1, STE24, MADB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lethal tight skin contracture syndrome (LTSCS) [MIM:275210]: Rare disorder mainly characterized by intrauterine growth retardation, tight and rigid skin with erosions, prominent superficial vasculature and epidermal hyperkeratosis, facial features (small mouth, small pinched nose and micrognathia), sparse/absent eyelashes and eyebrows, mineralization defects of the skull, thin dysplastic clavicles, pulmonary hypoplasia, multiple joint contractures and an early neonatal lethal course. Liveborn children usually die within the first week of life. The overall prevalence of consanguineous cases suggested an autosomal recessive inheritance. {ECO:0000269	PubMed:15317753}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
RET Onkogeni (Ekzon 10 - 11 - 13 - 14 - 15 ve 16) (RET geni) (Dizi Analizi) (Postnatal)	RET protooncogene (Exon 10-11-13-14-15 and 16) (RET gene) (Sequence Analysis) (Postnatal)	RET	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Retiküler Disgenezis, 267500, Otozomal Resesif (Retiküler Disgenezis) (AK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Reticular dysgenesis, 267500, Autosomal recessive (Reticular dysgenesis) (AK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AK2	AK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Reticular dysgenesis (RDYS) [MIM:267500]: Most severe form of inborn severe combined immunodeficiencies (SCID) and is characterized by absence of granulocytes and almost complete deficiency of lymphocytes in peripheral blood, hypoplasia of the thymus and secondary lymphoid organs, and lack of innate and adaptive humoral and cellular immune functions, leading to fatal septicemia within days after birth. In bone marrow of individuals with reticular dysgenesis, myeloid differentiation is blocked at the promyelocytic stage, whereas erythro- and megakaryocytic maturation is generally normal. In addition, affected newborns have bilateral sensorineural deafness. Defects may be due to its absence in leukocytes and inner ear, in which its absence can not be compensated by AK1. {ECO:0000269|PubMed:19043418, ECO:0000269|PubMed:19043417}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kitamura'nın Retiküler Akropigmentasyonu, 615537, Otozomal Dominant; RAK (Retiküle Akropigmentasyon, Kitamura Tipi) (ADAM10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Reticulate acropigmentation of Kitamura, 615537, Autosomal dominant; RAK (Reticulate acropigmentation of Kitamura) (ADAM10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAM10	ADAM10, MADM, RAK, AD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Reticulate acropigmentation of Kitamura (RAK) [MIM:615537]: A rare cutaneous pigmentation disorder characterized by reticulate, slightly depressed, sharply demarcated brown macules without hypopigmentation, affecting the dorsa of the hands and feet and appearing in the first or second decade of life. The macules gradually darken and extend to the proximal regions of the extremities. The manifestations tend to progress until middle age, after which progression of the eruptions stops. The pigmentary augmentation is found on the flexor aspects of the wrists, neck, patella and olecranon. Other features include breaks in the epidermal ridges on the palms and fingers, palmoplantar pits, occasionally plantar keratoderma, and partial alopecia. {ECO:0000269	PubMed:23666529}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retina Arter Anevrizması-Supravalvüler Pulmoner Stenoz, 614224, Otozomal Resesif; RAMSVPS (Ailesel Retinal Arter Makroanevrizması) (IGFBP7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal arterial macroaneurysm with supravalvular pulmonic stenosis, 614224, Autosomal recessive; RAMSVPS (Familial retinal arterial macroaneurysm) (IGFBP7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IGFBP7	IGFBP7, MAC25, RAMSVPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal arterial macroaneurysm with supravalvular pulmonic stenosis (RAMSVPS) [MIM:614224]: An autosomal recessive condition characterized by the bilateral appearance of 'beading' along the major retinal arterial trunks, with the subsequent formation of macroaneurysms. Affected individuals also have supravalvular pulmonic stenosis, often requiring surgical correction. {ECO:0000269	PubMed:21835307}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retina Arteler, Tortiosite, 180000, Otozomal Dominant; RATOR (Retinal Arter Tortiositesi) (COL4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal arteries, tortuosity of, 180000, Autosomal dominant; RATOR (Retinal arterial tortuosity) (COL4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL4A1	COL4A1, PNREN1, HANAC, ICH, BSVD, RATOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Porencephaly 1 (PNREN1) [MIM:175780]: A neurologic disorder characterized by a fluid-filled cysts or cavities within the cerebral hemispheres, neurologic manifestations, facial paresis, and visual defects. Affected individuals typically have hemiplegia, seizures, and intellectual disability. Porencephaly type 1 is usually unilateral and results from focal destructive lesions such as fetal vascular occlusion or birth trauma. {ECO:0000269|PubMed:15905400, ECO:0000269|PubMed:16107487, ECO:0000269|PubMed:19194877}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinal Kon Distrofisi 3, 610024, Otozomal Resesif, Otozomal Dominant; RCD3A (Akromatopsi) (PDE6H geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal cone dystrophy 3, 610024, Autosomal recessive, Autosomal dominant; RCD3A (Achromatopsia) (PDE6H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6H	PDE6H, RCD3, ACHM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone dystrophy, retinal 3A (RCD3A) [MIM:610024]: A rare form of cone dystrophy associated with supernormal rod responses. The disorder is characterized by reduced visual acuity, photoaversion, night blindness, and abnormal color vision. At an early age, the retina shows subtle depigmentation at the macula and, later, more obvious areas of atrophy. {ECO:0000269	PubMed:15629837}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinal Kon Distrofisi 3B, 610356, Otozomal Resesif; RCD3B (Kon Distrofisi-Süpernormal Rod Yanıtı) (KCNV2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal cone dystrophy 3B, 610356, Autosomal recessive; RCD3B (Cone dystrophy with supernormal rod response) (KCNV2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNV2	KCNV2, KV11.1, RCD3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone dystrophy retinal 3B (RCD3B) [MIM:610356]: A rare form of cone dystrophy associated with supernormal rod responses. The disorder is characterized by reduced visual acuity, photoaversion, night blindness, and abnormal color vision. At an early age, the retina shows subtle depigmentation at the macula and, later, more obvious areas of atrophy. {ECO:0000269	PubMed:16909397}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Retinal Kon Distrofisi 4, 610478, Otozomal Resesif; RCD4 (Progresif Kon Distrofisi) (CACNA2D4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal cone dystrophy 4, 610478, Autosomal recessive; RCD4 (Cone rod dystrophy) (CACNA2D4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA2D4	CACNA2D4, RCD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal cone dystrophy 4 (RCD4) [MIM:610478]: Characterized by minimal symptoms except for slowly progressive reduction in visual acuity. (ECO:0000269	PubMed:17033974). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retina Dejenerasyonu, Otozomal Resesif, Kıvırcık Pigment Tipi (NRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal degeneration, autosomal recessive, clumped pigment type (NRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRL	NRL, D14S46E, RP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal degeneration autosomal recessive clumped pigment type (RDCP) [MIM:162080]: A retinopathy characterized by night blindness since early childhood, consistent with a severe reduction in rod function. Color vision is normal although there is a relatively enhanced function of short-wavelength-sensitive cones in the macula. Signs of retinal degeneration and clusters of clumped pigment deposits in the peripheral fundus at the level of the retinal pigment epithelium are present. (ECO:0000269	PubMed:15591106). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinal Dejenerasyon, Geç Başlangıçlı, Otozomal Dominant, 605670, Otozomal Dominant; LORD (Geç Başlangıçlı Retinal Dejenerasyon) (C1QTNF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal degeneration, late-onset, autosomal dominant, 605670, Autosomal dominant; LORD (Late-onset retinal degeneration) (C1QTNF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1QTNF5	C1QTNF5, CTRP5, LORD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Late-onset retinal degeneration (LORD) [MIM:605670]: Autosomal dominant disorder characterized by onset in the fifth to sixth decade with night blindness and punctate yellow-white deposits in the retinal fundus, progressing to severe central and peripheral degeneration, with choroidal neovascularization and chorioretinal atrophy. (ECO:0000269|PubMed:12944416, ECO:0000269|PubMed:22892318). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Usher Sendromu Tip 2A, Retina Hastalığı, Modifiye Edici, 276901, Otozomal Resesif; USH2A (Usher Sendromu) (PDZD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal disease in Usher syndrome type IIA, modifier of, 276901, Autosomal recessive; USH2A (Usher syndrome) (PDZD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDZD7	PDZD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 2C (USH2C) [MIM:605472]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. (ECO:0000269|PubMed:20440071). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. PDZD7 mutations have been found in combination with mutations in USH2A and GPR98 in patients affected by Usher syndrome, suggesting a role as contributor to digenic Usher syndrome or a modifier of retinal disease expression. (ECO:0000269|PubMed:20440071).;	EDTA Blood Sample
Retina Distrofi- Iris Kolobomu ± Katarakt, 616722, Otozomal Dominant; RDICC (MIR204 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy and iris coloboma with or without cataract, 616722, Autosomal dominant; RDICC (MIR204 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIR204	MIR204, MIRN204, RDICC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retina Distrofisi ve Obezite, 616188, Otozomal Resesif; RDOB (Retinitis Pigmentozaa) (TUB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy and obesity, 616188, Autosomal recessive; RDOB (Retinitis pigmentosa) (TUB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUB	TUB, RDOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal dystrophy and obesity (RDOB) [MIM:616188]: A disease characterized by obesity, night blindness, decreased visual acuity, and electrophysiological features of a rod cone dystrophy. (ECO:0000269	PubMed:24375934). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retina Distrofisi, İnner Retina Disfonksiyonu ve Gangliyon Hücresi Anormallikleri, 616079, Otozomal Dominant; RDGCA (Retinal Distrofi-İç Retinal Disfonksiyon ve Ganglion Hücre Anomalileri) (ITM2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079, Autosomal dominant; RDGCA (Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. (ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362). Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. (ECO:0000269|PubMed:10781099).	EDTA Blood Sample
Retina Distrofisi, İnner Retina Disfonksiyonu ve Gangliyon Hücresi Anormallikleri, 616079, Otozomal Dominant; RDGCA (Retinal Distrofi-İç Retinal Disfonksiyon ve Ganglion Hücre Anomalileri) (Prenatal) (ITM2B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Retinal dystrophy with inner retinal dysfunction and ganglion cell abnormalities, 616079, Autosomal dominant; RDGCA (Retinal dystrophy with inner retinal dysfunction and ganglion cell anomalies) (ITM2B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ITM2B	ITM2B, BRI, ABRI, FBD, RDGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cerebral amyloid angiopathy, ITM2B-related 2 (CAA-ITM2B2) [MIM:117300]: A disorder characterized by amyloid deposition in the walls of the blood vessels of the cerebrum, choroid plexus, cerebellum, spinal cord and retina. Plaques and neurofibrillary tangles are observed in the hippocampus. Clinical features include progressive ataxia, dementia, cataracts and deafness. (ECO:0000269|PubMed:10781099, ECO:0000269|PubMed:14656991, ECO:0000269|PubMed:16091362). Note=The disease is caused by mutations affecting the gene represented in this entry. A decamer duplication in the 3' region of ITM2B results in the production of the ADan amyloidogenic peptide (PubMed:10781099). ADan is generated by cleavage of the mutated precursor at the normal furin processing site. ADan accumulates in the brain and produces amyloid fibrils responsible for neuronal dysfunction and dementia. (ECO:0000269|PubMed:10781099).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Retina Distrofisi, ± Ekstraoküler Anomaliler, 617175, Otozomal Resesif; RDEOA (Retina Pigment Epitelinin Retiküler Distrofisi) (RCBTB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy with or without extraocular anomalies, 617175, Autosomal recessive; RDEOA (Reticular dystrophy of the retinal pigment epithelium) (RCBTB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RCBTB1	RCBTB1, CLLD7, GLP, RDEOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retina Distrofisi, ± Ekstraoküler Anomaliler, 617175, Otozomal Resesif; RDEOA (Retina Pigment Epitelinin Retiküler Distrofisi) (Prenatal) (RCBTB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Retinal dystrophy with or without extraocular anomalies, 617175, Autosomal recessive; RDEOA (Reticular dystrophy of the retinal pigment epithelium) (RCBTB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RCBTB1	RCBTB1, CLLD7, GLP, RDEOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Retina Distrofisi, Şiddetli Erken Başlangıçlı, 248200, Otozomal Resesif (Stargardt Hastalığı) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy, early-onset severe, 248200, Autosomal recessive (Stargardt disease) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 2 (ARMD2) [MIM:153800]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. (ECO:0000269|PubMed:19028736, ECO:0000269|PubMed:9295268). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Retina Distrofisi, Şiddetli Erken Başlangıçlı, 613341, Otozomal Resesif (LRAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy, early-onset severe, 613341, Autosomal recessive (LRAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRAT	LRAT, LCA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 14 (LCA14) [MIM:613341]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. (ECO:0000269|PubMed:11381255, ECO:0000269|PubMed:17011878, ECO:0000269|PubMed:18055821). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retina Distrofisi, Erken Başlangıç, ± Pitüiter Disfonksiyon, 610125, Otozomal Dominant (Kelebek Şeklinde Pigment Distrofisi) (OTX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125, Autosomal dominant (Butterfly-shaped pigment dystrophy) (OTX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OTX2	OTX2, MCOPS5, CPHD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microphthalmia, syndromic, 5 (MCOPS5) [MIM:610125]: Patients manifest unilateral or bilateral microphthalmia/clinical anophthalmia and variable additional features including pituitary dysfunction, coloboma, microcornea, cataract, retinal dystrophy, hypoplasia or agenesis of the optic nerve, agenesis of the corpus callosum, developmental delay, joint laxity, hypotonia, and seizures. Microphthalmia is a disorder of eye formation, ranging from small size of a single eye to complete bilateral absence of ocular tissues (anophthalmia). In many cases, microphthalmia/anophthalmia occurs in association with syndromes that include non-ocular abnormalities. (ECO:0000269|PubMed:15846561, ECO:0000269|PubMed:20396904, ECO:0000269|PubMed:22577225, ECO:0000269|PubMed:24167467). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retina Distrofisi, İris Kolobomu ve Komedojen Akne Sendromu, 615147, Otozomal Resesif; RDCCAS (Progresif Retina Distrofisi, Retinol Transport Defektine Bağlı) (RBP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147, Autosomal recessive; RDCCAS (Progressive retinal dystrophy due to retinol transport defect) (RBP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBP4	RBP4, RDCCAS, MCOPCB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal dystrophy, iris coloboma, and comedogenic acne syndrome (RDCCAS) [MIM:615147]: A disease characterized by retinal degeneration, ocular colobomas involving both the anterior and posterior segment, impaired night vision and loss of visual acuity. Additional characteristic features include developmental abnormalities and severe acne. (ECO:0000269|PubMed:23189188, ECO:0000269|PubMed:9888420). Note=The disease is caused by mutations affecting the gene represented in this entry. Loss of functional RBP4 protein results in serum retinol deficiency. Lack of normal levels of retinol impairs the visual cycle leading to night blindness at early stages; prolonged deficiency may lead to retinal degeneration. Additionally, retinol deficiency may result in dry skin, increased susceptibility to infection and acne (PubMed:23189188). {ECO:0000269	PubMed:23189188}.	EDTA Blood Sample
Retina Distrofisi, İris Kolobomu ve Komedojen Akne Sendromu, 615147, Otozomal Resesif; RDCCAS (Progresif Retina Distrofisi, Retinol Transport Defektine Bağlı) (Prenatal) (RBP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147, Autosomal recessive; RDCCAS (Progressive retinal dystrophy due to retinol transport defect) (RBP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBP4	RBP4, RDCCAS, MCOPCB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Retinal dystrophy, iris coloboma, and comedogenic acne syndrome (RDCCAS) [MIM:615147]: A disease characterized by retinal degeneration, ocular colobomas involving both the anterior and posterior segment, impaired night vision and loss of visual acuity. Additional characteristic features include developmental abnormalities and severe acne. (ECO:0000269|PubMed:23189188, ECO:0000269|PubMed:9888420). Note=The disease is caused by mutations affecting the gene represented in this entry. Loss of functional RBP4 protein results in serum retinol deficiency. Lack of normal levels of retinol impairs the visual cycle leading to night blindness at early stages; prolonged deficiency may lead to retinal degeneration. Additionally, retinol deficiency may result in dry skin, increased susceptibility to infection and acne (PubMed:23189188). {ECO:0000269	PubMed:23189188}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Retina Distrofisi, Jüvenil Katarakt ve Kısa Boy Sendromu, 616108, Otozomal Resesif; RDJCSS (Retinitis Pigmentoza-Jüvenil Katarakt-Boy Kısalığı-Entelektüel Yetersizlik Sendromu) (RDH11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinal dystrophy, juvenile cataracts, and short stature syndrome, 616108, Autosomal recessive; RDJCSS (Retinitis pigmentosa-juvenile cataract-short stature-intellectual disability syndrome) (RDH11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RDH11	RDH11, PSDR1, RALR1, RDJCSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal dystrophy, juvenile cataracts, and short stature syndrome (RDJCSS) [MIM:616108]: A disorder characterized by retinal dystrophy resulting in progressive decrease in visual acuity and difficulties with night vision in the first decade of life, development of juvenile cataracts, facial dysmorphism, psychomotor developmental delays, learning disabilities and short stature. Ophthalmological findings include salt-and-pepper retinopathy, attenuation of the arterioles, generalized rod-cone dysfunction, mottled macula at an early age, and peripapillary sparing of the retinal pigment epithelium. {ECO:0000269	PubMed:24916380}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 1, 180100, Otozomal Resesif, Otozomal Dominant; RP1 (Retinitis Pigmentoza) (RP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 1, 180100, Autosomal recessive, Autosomal dominant; RP1 (Retinitis pigmentosa) (RP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RP1	RP1, ORP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 1 (RP1) [MIM:180100]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:10391211, ECO:0000269|PubMed:10484783, ECO:0000269|PubMed:11095597, ECO:0000269|PubMed:15863674, ECO:0000269|PubMed:15933747, ECO:0000269|PubMed:19956407, ECO:0000269|PubMed:20664799, ECO:0000269|PubMed:22052604, ECO:0000269|PubMed:22334370}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 10, 180105, Otozomal Dominant; RP10 (Retinitis Pigmentoza) (IMPDH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 10, 180105, Autosomal dominant; RP10 (Retinitis pigmentosa) (IMPDH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPDH1	IMPDH1, RP10, LCA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 10 (RP10) [MIM:180105]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:11875049, ECO:0000269|PubMed:11875050, ECO:0000269|PubMed:16384941}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 11, 600138, Otozomal Dominant; RP11 (Retinitis Pigmentoza) (PRPF31 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 11, 600138, Autosomal dominant; RP11 (Retinitis pigmentosa) (PRPF31 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPF31	PRPF31, PRP31, RP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 11 (RP11) [MIM:600138]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:11545739, ECO:0000269|PubMed:12923864, ECO:0000269|PubMed:8808602}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 13, 600059, Otozomal Dominant; RP13 (Retinitis Pigmentoza) (PRPF8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 13, 600059, Autosomal dominant; RP13 (Retinitis pigmentosa) (PRPF8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPF8	PRPF8, PRPC8, RP13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 13 (RP13) [MIM:600059]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:11468273, ECO:0000269|PubMed:11910563, ECO:0000269|PubMed:12714658, ECO:0000269|Ref.33}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 14, 600132, Otozomal Resesif; RP14 (Retinitis Pigmentoza) (TULP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 14, 600132, Autosomal recessive; RP14 (Retinitis pigmentosa) (TULP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TULP1	TULP1, RP14, LCA15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 15 (LCA15) [MIM:613843]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:17962469}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 17, 600852, Otozomal Dominant; RP17 (Retinitis Pigmentoza) (CA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 17, 600852, Autosomal dominant; RP17 (Retinitis pigmentosa) (CA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CA4	CA4, RP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 18, 601414, Otozomal Dominant; RP18 (Retinitis Pigmentoza) (PRPF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 18, 601414, Autosomal dominant; RP18 (Retinitis pigmentosa) (PRPF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPF3	PRPF3, HPRP3, RP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 18 (RP18) [MIM:601414]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:11773002, ECO:0000269|PubMed:12714658}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 19, 601718; RP19 (Retinitis Pigmentoza) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 19, 601718; RP19 (Retinitis pigmentosa) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 2 (ARMD2) [MIM:153800]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269|PubMed:19028736, ECO:0000269|PubMed:9295268}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 2, 312600, X'e Bağlı; RP2 (Retinitis Pigmentoza) (RP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 2, 312600, X-linked; RP2 (Retinitis pigmentosa) (RP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RP2	RP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 2 (RP2) [MIM:312600]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:10090907, ECO:0000269|PubMed:10520237, ECO:0000269|PubMed:10634633, ECO:0000269|PubMed:10937588, ECO:0000269|PubMed:11462235, ECO:0000269|PubMed:11992260, ECO:0000269|PubMed:12657579, ECO:0000269|PubMed:14564670, ECO:0000269|PubMed:22334370, ECO:0000269|PubMed:9697692}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 20, 613794, Otozomal Resesif; RP20 (Retinitis Pigmentoza) (RPE65 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 20, 613794, Autosomal recessive; RP20 (Retinitis pigmentosa) (RPE65 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPE65	RPE65, RP20, LCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 2 (LCA2) [MIM:204100]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:10090910, ECO:0000269|PubMed:10766140, ECO:0000269|PubMed:11462243, ECO:0000269|PubMed:14611946, ECO:0000269|PubMed:14962443, ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:16205573, ECO:0000269|PubMed:17297704, ECO:0000269|PubMed:17724218, ECO:0000269|PubMed:18682808, ECO:0000269|PubMed:9326941, ECO:0000269|PubMed:9801879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 20, 613794, Otozomal Resesif; RP20 (Retinitis Pigmentoza) (MLPA)	Retinitis pigmentosa 20, 613794, Autosomal recessive; RP20 (Retinitis pigmentosa) (MLPA)	RPE65	RPE65, RP20, LCA2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leber congenital amaurosis 2 (LCA2) [MIM:204100]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:10090910, ECO:0000269|PubMed:10766140, ECO:0000269|PubMed:11462243, ECO:0000269|PubMed:14611946, ECO:0000269|PubMed:14962443, ECO:0000269|PubMed:15024725, ECO:0000269|PubMed:16205573, ECO:0000269|PubMed:17297704, ECO:0000269|PubMed:17724218, ECO:0000269|PubMed:18682808, ECO:0000269|PubMed:9326941, ECO:0000269|PubMed:9801879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 23, 300424, X'e Bağlı Resesif; RP23 (Retinitis Pigmentoza) (OFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 23, 300424, X-linked recessive; RP23 (Retinitis pigmentosa) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. {ECO:0000269	PubMed:16783569}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Retinitis Pigmentoza 25, 602772; RP25 (Retinitis Pigmentoza) (EYS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 25, 602772; RP25 (Retinitis pigmentosa) (EYS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EYS	EYS, RP25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 26, 608380; RP26 (Retinitis Pigmentoza) (CERKL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 26, 608380; RP26 (Retinitis pigmentosa) (CERKL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CERKL	CERKL, RP26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 26 (RP26) [MIM:608380]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269|PubMed:14681825, ECO:0000269|PubMed:18978954). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 27 613750, Otozomal Dominant; RP27 (Retinitis Pigmentoza) (NRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 27, 613750, Autosomal dominant; RP27 (Retinitis pigmentosa) (NRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRL	NRL, D14S46E, RP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal degeneration autosomal recessive clumped pigment type (RDCP) [MIM:162080]: A retinopathy characterized by night blindness since early childhood, consistent with a severe reduction in rod function. Color vision is normal although there is a relatively enhanced function of short-wavelength-sensitive cones in the macula. Signs of retinal degeneration and clusters of clumped pigment deposits in the peripheral fundus at the level of the retinal pigment epithelium are present. (ECO:0000269	PubMed:15591106). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 28 606068; RP28 (Retinitis Pigmentoza) (FAM161A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 28, 606068; RP28 (Retinitis pigmentosa) (FAM161A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM161A	FAM161A, RP28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 3, 300029; RP3 (Retinitis Pigmentoza) (RPGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 3, 300029; RP3 (Retinitis pigmentosa) (RPGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGR	RPGR, RP3, CRD, RP15, COD1, CORDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 3 (RP3) [MIM:300029]: A X-linked retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. In RP3, affected males have a severe phenotype, and carrier females show a wide spectrum of clinical features ranging from completely asymptomatic to severe retinitis pigmentosa. Heterozygous women can manifest a form of choroideretinal degeneration which is distinguished from other types by the absence of visual defects in the presence of a brilliant, scintillating, golden-hued, patchy appearance most striking around the macula, called a tapetal-like retinal reflex. (ECO:0000269|PubMed:10482958, ECO:0000269|PubMed:10737996, ECO:0000269|PubMed:10932196, ECO:0000269|PubMed:10937588, ECO:0000269|PubMed:10970770, ECO:0000269|PubMed:11180598, ECO:0000269|PubMed:11992260, ECO:0000269|PubMed:12657579, ECO:0000269|PubMed:14564670, ECO:0000269|PubMed:8671101, ECO:0000269|PubMed:8817343, ECO:0000269|PubMed:9399904, ECO:0000269|PubMed:9851162). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 30 607921; RP30 (Retinitis Pigmentoza) (FSCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 30, 607921; RP30 (Retinitis pigmentosa) (FSCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FSCN2	FSCN2, RFSN, RP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 31 609923; RP31 (Retinitis Pigmentoza) (TOPORS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 31, 609923; RP31 (Retinitis pigmentosa) (TOPORS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TOPORS	TOPORS, PS3BP3, LUN, RP31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 31 (RP31) [MIM:609923]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269	PubMed:17924349). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 33, 610359, Otozomal Dominant; RP33 (Retinitis Pigmentoza) (SNRNP200 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 33, 610359, Autosomal dominant; RP33 (Retinitis pigmentosa) (SNRNP200 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNRNP200	SNRNP200, ASCC3L1, KIAA0788, RP33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 33 (RP33) [MIM:610359]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269|PubMed:16723661, ECO:0000269|PubMed:19710410, ECO:0000269|PubMed:19878916, ECO:0000269|PubMed:21618346, ECO:0000269|PubMed:23029027, ECO:0000269|PubMed:23887765, ECO:0000269|PubMed:24318334). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 35, 610282, Otozomal Resesif, Otozomal Dominant; RP35 (Retinitis Pigmentoza) (SEMA4A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 35, 610282, Autosomal recessive, Autosomal dominant; RP35 (Retinitis pigmentosa) (SEMA4A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEMA4A	SEMA4A, SEMB, RP35, CORD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 35 (RP35) [MIM:610282]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269	PubMed:16199541). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 36, 610599; RP36 (Retinitis Pigmentoza) (PRCD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 36, 610599; RP36 (Retinitis pigmentosa) (PRCD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRCD	PRCD, RP36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 36 (RP36) [MIM:610599]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269	PubMed:16938425). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 37, 611131, Otozomal Resesif, Otozomal Dominant; RP37 (Retinitis Pigmentoza) (NR2E3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 37, 611131, Autosomal recessive, Autosomal dominant; RP37 (Retinitis pigmentosa) (NR2E3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR2E3	NR2E3, PNR, ESCS, RP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Enhanced S cone syndrome (ESCS) [MIM:268100]: Autosomal recessive retinopathy in which patients have increased sensitivity to blue light perception of blue light is mediated by what is normally the least populous cone photoreceptor subtype, the S (short wavelength, blue) cones. ESCS is also associated with visual loss, with night blindness occurring from early in life, varying degrees of L (long, red)- and M (middle, green)-cone vision, and retinal degeneration. (ECO:0000269|PubMed:10655056, ECO:0000269|PubMed:11071390, ECO:0000269|PubMed:12963616, ECO:0000269|PubMed:15459973, ECO:0000269|PubMed:16225923, ECO:0000269|PubMed:18294254, ECO:0000269|PubMed:19006237). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 38, 613862, Otozomal Resesif, RP38 (Retinitis Pigmentoza) (MERTK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 38, 613862, Autosomal recessive; RP38 (Retinitis pigmentosa) (MERTK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MERTK	MERTK, RP38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 38 (RP38) [MIM:613862]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269	PubMed:21062461). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 39, 613809; RP39 (Retinitis Pigmentoza) (USH2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 39, 613809; RP39 (Retinitis pigmentosa) (USH2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USH2A	USH2A, RP39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 2A (USH2A) [MIM:276901]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. (ECO:0000269|PubMed:10729113, ECO:0000269|PubMed:10738000, ECO:0000269|PubMed:10909849, ECO:0000269|PubMed:11311042, ECO:0000269|PubMed:12112664, ECO:0000269|PubMed:12525556, ECO:0000269|PubMed:14970843, ECO:0000269|PubMed:15015129, ECO:0000269|PubMed:15025721, ECO:0000269|PubMed:15241801, ECO:0000269|PubMed:15325563, ECO:0000269|PubMed:17085681, ECO:0000269|PubMed:17406132, ECO:0000269|PubMed:18273898, ECO:0000269|PubMed:18452394, ECO:0000269|PubMed:19683999, ECO:0000269|PubMed:19737284, ECO:0000269|PubMed:20309401, ECO:0000269|PubMed:20440071, ECO:0000269|PubMed:20507924, ECO:0000269|PubMed:21593743, ECO:0000269|PubMed:21686329, ECO:0000269|PubMed:22004887, ECO:0000269|PubMed:23737954, ECO:0000269|PubMed:9624953). Note=The disease is caused	EDTA Blood Sample

Retinitis Pigmentoza 4, Otozomal Dominant Veya Resesif, 613731, Otozomal Resesif, Otozomal Dominant; RP4 (Retinitis Pigmentoza) (RHO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 4, autosomal dominant or recessive, 613731, Autosomal recessive, Autosomal dominant; RP4 (Retinitis pigmentosa) (RHO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHO	RHO, RP4, OPN2, CSNBAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 1 (CSNBAD1) [MIM:610445]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. {ECO:0000269|PubMed:7846071, ECO:0000269|PubMed:8358437, ECO:0000269|PubMed:9888392}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 41, 612095, Otozomal Resesif, RP41 (Retinitis Pigmentoza) (PROM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 41, 612095, Autosomal recessive; RP41 (Retinitis pigmentosa) (PROM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROM1	PROM1, PROML1, AC133, RP41, CORD12, CD133, MCDR2, STGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stargardt disease 4 (STGD4) [MIM:603786]: A common hereditary macular degeneration. It is characterized by decreased central vision, atrophy of the macula and underlying retinal pigment epithelium, and frequent presence of prominent flecks in the posterior pole of the retina. {ECO:0000269	PubMed:18654668}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 42, 612943, Otozomal Dominant; RP42 (Retinitis Pigmentoza) (KLHL7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 42, 612943, Autosomal dominant; RP42 (Retinitis pigmentosa) (KLHL7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL7	KLHL7, RP42, CIS53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 43, 613810; RP43 (Retinitis Pigmentoza) (PDE6A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 43, 613810; RP43 (Retinitis pigmentosa) (PDE6A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6A	PDE6A, PDEA, RP43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 43 (RP43) [MIM:613810]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:10393062, ECO:0000269|PubMed:7493036}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 44, 613769; RP44 (Retinitis Pigmentoza) (RGR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 44, 613769; RP44 (Retinitis pigmentosa) (RGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RGR	RGR, RP44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 44 (RP44) [MIM:613769]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:10581022}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 45, 613767, Otozomal Resesif, RP45 (Retinitis Pigmentoza) (CNGB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 45, 613767, Autosomal recessive; RP45 (Retinitis pigmentosa) (CNGB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNGB1	CNGB1, CNCG3L, CNCG2, RP45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 45 (RP45) [MIM:613767]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:11379879}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 46, 612572; RP46 (Retinitis Pigmentoza) (IDH3B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 46, 612572; RP46 (Retinitis pigmentosa) (IDH3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IDH3B	IDH3B, RP46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 46 (RP46) [MIM:612572]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:18806796}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 47, 613758; RP47 (Retinitis Pigmentoza) (SAG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 47, 613758; RP47 (Retinitis pigmentosa) (SAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAG	SAG, RP47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 47 (RP47) [MIM:613758]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:9565049}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 48, 613827; RP48 (Retinitis Pigmentoza) (GUCA1B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 48, 613827; RP48 (Retinitis pigmentosa) (GUCA1B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GUCA1B	GUCA1B, GCAP2, GUCA2, RP48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Retinitis Pigmentoza 49, 613756; Rp49 (Retinitis Pigmentoza) (CNGA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 49, 613756; Rp49 (Retinitis pigmentosa) (CNGA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CNGA1	CNGA1, CNCG1, RP49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 49 (RP49) [MIM:613756]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:15570217, ECO:0000269|PubMed:7479749}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 51, 613464, Otozomal Resesif, RP51 (Retinitis Pigmentoza) (TTC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 51, 613464, Autosomal recessive; RP51 (Retinitis pigmentosa) (TTC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC8	TTC8, BBS8, RP51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 8 (BBS8) [MIM:615985]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:14520415, ECO:0000269|PubMed:16308600}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 54, 613428; RP54 (Retinitis Pigmentoza) (C2orf71 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 54, 613428; RP54 (Retinitis pigmentosa) (C2orf71 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C2orf71	C2orf71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 55, 613575; RP55 (Retinitis Pigmentoza) (ARL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 55, 613575; RP55 (Retinitis pigmentosa) (ARL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL6	ARL6, BBS3, RP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 3 (BBS3) [MIM:600151]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:15258860, ECO:0000269|PubMed:15314642, ECO:0000269|PubMed:23219996}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 56, 613581, Otozomal Resesif, RP56 (Retinitis Pigmentoza) (IMPG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 56, 613581, Autosomal recessive; RP56 (Retinitis pigmentosa) (IMPG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IMPG2	IMPG2, IPM200, RP56, VMDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 56 (RP56) [MIM:613581]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:20673862}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 57, 613582, Otozomal Resesif, RP57 (Retinitis Pigmentoza) (PDE6G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 57, 613582, Autosomal recessive; RP57 (Retinitis pigmentosa) (PDE6G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6G	PDE6G, PDEG, RP57	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 57 (RP57) [MIM:613582]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:20655036}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Retinitis Pigmentoza 58, 613617, Otozomal Resesif, RP58 (Retinitis Pigmentoza) (ZNF513 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 58, 613617, Autosomal recessive; RP58 (Retinitis pigmentosa) (ZNF513 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF513	ZNF513, RP58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 59, 613861, Otozomal Resesif, RP59 (Retinitis Pigmentoza) (DHDDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 59, 613861, Autosomal recessive; RP59 (Retinitis pigmentosa) (DHDDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHDDS	DHDDS, HDS, RP59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 60, 613983, Otozomal Dominant, RP60 (Retinitis Pigmentoza) (PRPF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 60, 613983, Autosomal dominant; RP60 (Retinitis pigmentosa) (PRPF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPF6	PRPF6, PRP6, ANT1, TOM, C20orf14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 61, 614180; RP61 (Retinitis Pigmentoza) (CLRN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 61, 614180; RP61 (Retinitis pigmentosa) (CLRN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLRN1	CLRN1, USH3A, USH3, RP61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 61 (RP61) [MIM:614180]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269	PubMed:21310491}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Retinitis Pigmentoza 62, 614181, Otozomal Resesif, RP62 (Retinitis Pigmentoza) (MAK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 62, 614181, Autosomal recessive; RP62 (Retinitis pigmentosa) (MAK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAK	MAK, RP62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 62 (RP62) [MIM:614181]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:21825139, ECO:0000269|PubMed:21835304}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 64, 614500, Otozomal Resesif (Retinitis Pigmentoza) (C8orf37 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 64, 614500, Autosomal recessive (Retinitis pigmentosa) (C8orf37 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C8orf37	C8orf37, CORD16, RP64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 64 (RP64) [MIM:614500]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:22177090}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 65, 613660, Otozomal Resesif (Retinitis Pigmentoza) (CDHR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 65, 613660, Autosomal recessive (Retinitis pigmentosa) (CDHR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDHR1	CDHR1, PCDH21, PRCAD, CORD15, RP65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cone-rod dystrophy 15 (CORD15) [MIM:613660]: An inherited retinal dystrophy characterized by retinal pigment deposits visible on fundus examination, predominantly in the macular region, and initial loss of cone photoreceptors followed by rod degeneration. This leads to decreased visual acuity and sensitivity in the central visual field, followed by loss of peripheral vision. Severe loss of vision occurs earlier than in retinitis pigmentosa. {ECO:0000269|PubMed:20805371}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 66, 615233, Otozomal Resesif, RP66 (Retinitis Pigmentoza) (RBP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 66, 615233, Autosomal recessive; RP66 (Retinitis pigmentosa) (RBP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBP3	RBP3, RP66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 66 (RP66) [MIM:615233]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:19074801}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 67, 615565, Otozomal Resesif, RP67 (Retinitis Pigmentoza) (NEK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 67, 615565, Autosomal recessive; RP67 (Retinitis pigmentosa) (NEK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK2	NEK2, RP67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 67 (RP67) [MIM:615565]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:24043777}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 68, 615725, Otozomal Resesif, RP68 (Retinitis Pigmentoza) (SLC7A14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 68, 615725, Autosomal recessive; RP68 (Retinitis pigmentosa) (SLC7A14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC7A14	SLC7A14, KIAA1613, RP68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 69, 615780, Otozomal Resesif, RP69 (Retinitis Pigmentoza) (KIZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 69, 615780, Autosomal recessive; RP69 (Retinitis pigmentosa) (KIZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIZ	KIZ, KIZUNA, C20orf19, RP69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 69 (RP69) [MIM:615780]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:24680887}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 7 ve Dijenik, 608133, Otozomal Resesif, Otozomal Dominant; RP7 (Retinitis Pigmentoza) (PRPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 7 and digenic, 608133, Autosomal recessive, Autosomal dominant; RP7 (Retinitis pigmentosa) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, ADFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 7, Dijenik, 608133, Otozomal Resesif, Otozomal Dominant (Retinitis Pigmentoza) (ROM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 7, digenic, 608133, Autosomal recessive, Autosomal dominant (Retinitis pigmentosa) (ROM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROM1	ROM1, ROSP1, RP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 70, 615922, Otozomal Dominant; RP70 (Retinitis Pigmentoza) (PRPF4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 70, 615922, Autosomal dominant; RP70 (Retinitis pigmentosa) (PRPF4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPF4	PRPF4, PRP4, HPRP4, RP70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 70 (RP70) [MIM:615922]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:24419317, ECO:0000269|PubMed:25383878}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 71, 616394, Otozomal Resesif, RP71 (Retinitis Pigmentoza) (IFT172 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 71, 616394, Autosomal recessive; RP71 (Retinitis pigmentosa) (IFT172 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT172	IFT172, SLB, KIAA1179, SRTD10, RP71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 10 with or without polydactyly (SRTD10) [MIM:615630]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269|PubMed:24140113}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 72, 616469, Otozomal Resesif, RP72 (Retinitis Pigmentoza) (ZNF408 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 72, 616469, Autosomal recessive; RP72 (Retinitis pigmentosa) (ZNF408 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF408	ZNF408, EVR6, RP72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 72 (RP72) [MIM:616469]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:25882705}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 73, 616544, Otozomal Resesif, RP73 (Retinitis Pigmentoza) (HGSNAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 73, 616544, Autosomal recessive; RP73 (Retinitis pigmentosa) (HGSNAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HGSNAT	HGSNAT, TMEM76, MPS3C, RP73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mucopolysaccharidosis 3C (MPS3C) [MIM:252930]: A form of mucopolysaccharidosis type 3, an autosomal recessive lysosomal storage disease due to impaired degradation of heparan sulfate. MPS3 is characterized by severe central nervous system degeneration, but only mild somatic disease. Onset of clinical features usually occurs between 2 and 6 years; severe neurologic degeneration occurs in most patients between 6 and 10 years of age, and death occurs typically during the second or third decade of life. {ECO:0000269|PubMed:16960811, ECO:0000269|PubMed:17033958, ECO:0000269|PubMed:17397050, ECO:0000269|PubMed:18024218, ECO:0000269|PubMed:19479962}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza 74, 616562, Otozomal Resesif, RP74 (Retinitis Pigmentoza) (BBS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 74, 616562, Autosomal recessive; RP74 (Retinitis pigmentosa) (BBS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BBS2	BBS2, RP74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 75, 617023, Otozomal Resesif, RP75 (Retinitis Pigmentoza) (AGBL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 75, 617023, Autosomal recessive; RP75 (Retinitis pigmentosa) (AGBL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGBL5	AGBL5, CCP5, RP75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 76, 617123, Otozomal Resesif, RP76 (Retinitis Pigmentoza) (POMGNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 76, 617123, Autosomal recessive; RP76 (Retinitis pigmentosa) (POMGNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Retinitis Pigmentoza 76, 617123, Otozomal Resesif, RP76 (Retinitis Pigmentoza) (MLPA)	Retinitis pigmentosa 76, 617123, Autosomal recessive; RP76 (Retinitis pigmentosa) (MLPA)	POMGNT1	POMGNT1, MEB, MDDGA3, MDDGB3, MDDGC3, RP76	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Muscular dystrophy-dystroglycanopathy congenital with brain and eye anomalies A3 (MDDGA3) [MIM:253280]: An autosomal recessive disorder characterized by congenital muscular dystrophy, ocular abnormalities, cobblestone lissencephaly, and cerebellar and pontine hypoplasia. Patients present severe congenital myopia, congenital glaucoma, pallor of the optic disks, retinal hypoplasia, mental retardation, hydrocephalus, abnormal electroencephalograms, generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease. {ECO:0000269|PubMed:11709191, ECO:0000269|PubMed:12588800, ECO:0000269|PubMed:15236414, ECO:0000269|PubMed:15466003, ECO:0000269|PubMed:17030669, ECO:0000269|PubMed:19067344}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza 77, 617304, Otozomal Resesif, RP77 (Retinitis Pigmentoza) (REEP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 77, 617304, Autosomal recessive; RP77 (Retinitis pigmentosa) (REEP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REEP6	REEP6, DP1L1, TB2L1, C19orf32, RP77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza 9, 180104, Otozomal Dominant; RP9 (Retinitis Pigmentoza) (RP9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa 9, 180104, Autosomal dominant; RP9 (Retinitis pigmentosa) (RP9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RP9	RP9, PAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza ve Eritrosit Mikrositozu, 616959, Otozomal Resesif; RPEM (TRNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa and erythrocytic microcytosis, 616959, Autosomal recessive; RPEM (TRNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRNT1	TRNT1, SIFD, RPEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD) [MIM:616084]: An autosomal recessive disease characterized by severe sideroblastic anemia with onset in the neonatal period or infancy, recurrent periodic fevers without an infectious etiology, B-cell lymphopenia and hypogammaglobulinemia. Affected individuals show delayed psychomotor development with variable neurodegeneration. Additional variable features include sensorineural hearing loss, retinitis pigmentosa, nephrocalcinosis, and cardiomyopathy. {ECO:0000269|PubMed:25193871}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Retinitis Pigmentoza, ± Situs İnversus, 615434, Otozomal Resesif (Retinitis Pigmentoza) (ARL2BP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa with or without situs inversus, 615434, Autosomal recessive (Retinitis pigmentosa) (ARL2BP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL2BP	ARL2BP, BART	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza-12, Otozomal Resesif, 600105, Otozomal Resesif; RP12 (Retinitis Pigmentoza) (CRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa-12, autosomal recessive, 600105, Autosomal recessive; RP12 (Retinitis pigmentosa) (CRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRB1	CRB1, RP12, LCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269|PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Retinitis Pigmentoza-12, Otozomal Resesif, 600105, Otozomal Resesif; RP12 (Retinitis Pigmentoza) (MLPA)	Retinitis pigmentosa-12, autosomal recessive, 600105, Autosomal recessive; RP12 (Retinitis pigmentosa) (MLPA)	CRB1	CRB1, RP12, LCA8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pigmented paravenous chorioretinal atrophy (PPCRA) [MIM:172870]: Unusual retinal degeneration characterized by accumulation of pigmentation along retinal veins. PPCRA is dominantly inherited, but exhibited variable expressivity. Males are more likely to exhibit a severe phenotype, whereas females may remain virtually asymptomatic even in later years. The PPCRA phenotype is associated with a mutation in CRB1 gene which is likely to affect the structure of the CRB1 protein. {ECO:0000269|PubMed:15623792}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Retinitis Pigmentoza-40, 613801, Otozomal Resesif; RP40 (Retinitis Pigmentoza) (PDE6B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa-40, 613801, Autosomal recessive; RP40 (Retinitis pigmentosa) (PDE6B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE6B	PDE6B, PDEB, RP40, CSNBAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 2 (CSNBAD2) [MIM:163500]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. {ECO:0000269|PubMed:8075643}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza-50, 613194 (Retinitis Pigmentoza) (BEST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa-50, 613194 (Retinitis pigmentosa) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy, vitelliform, 2 (VMD2) [MIM:153700]: An autosomal dominant form of macular degeneration that usually begins in childhood or adolescence. VMD2 is characterized by typical 'egg-yolk' macular lesions due to abnormal accumulation of lipofuscin within and beneath the retinal pigment epithelium cells. Progression of the disease leads to destruction of the retinal pigment epithelium and vision loss. {ECO:0000269|PubMed:10331951, ECO:0000269|PubMed:10394929, ECO:0000269|PubMed:10453731, ECO:0000269|PubMed:10682987, ECO:0000269|PubMed:10798642, ECO:0000269|PubMed:11241846, ECO:0000269|PubMed:11449320, ECO:0000269|PubMed:12187431, ECO:0000269|PubMed:12324875, ECO:0000269|PubMed:13129869, ECO:0000269|PubMed:14517959, ECO:0000269|PubMed:15176385, ECO:0000269|PubMed:18766995, ECO:0000269|PubMed:19357557, ECO:0000269|PubMed:9662395, ECO:0000269|PubMed:9700209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza, Konsantrik 613194 (Retinitis Pigmentoza) (BEST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa, concentric, 613194 (Retinitis pigmentosa) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular dystrophy, vitelliform, 2 (VMD2) [MIM:153700]: An autosomal dominant form of macular degeneration that usually begins in childhood or adolescence. VMD2 is characterized by typical 'egg-yolk' macular lesions due to abnormal accumulation of lipofuscin within and beneath the retinal pigment epithelium cells. Progression of the disease leads to destruction of the retinal pigment epithelium and vision loss. {ECO:0000269|PubMed:10331951, ECO:0000269|PubMed:10394929, ECO:0000269|PubMed:10453731, ECO:0000269|PubMed:10682987, ECO:0000269|PubMed:10798642, ECO:0000269|PubMed:11241846, ECO:0000269|PubMed:11449320, ECO:0000269|PubMed:12187431, ECO:0000269|PubMed:12324875, ECO:0000269|PubMed:13129869, ECO:0000269|PubMed:14517959, ECO:0000269|PubMed:15176385, ECO:0000269|PubMed:18766995, ECO:0000269|PubMed:19357557, ECO:0000269|PubMed:9662395, ECO:0000269|PubMed:9700209}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Pigmentoza, Jüvenil, 604393, Otozomal Resesif (Retinitis Pigmentoza) (AIPL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa, juvenile, 604393, Autosomal recessive (Retinitis pigmentosa) (AIPL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AIPL1	AIPL1, LCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza, Jüvenil, 604393, Otozomal Resesif (Retinitis Pigmentoza) (MLPA)	Retinitis pigmentosa, juvenile, 604393, Autosomal recessive (Retinitis pigmentosa) (MLPA)	AIPL1	AIPL1, LCA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Retinitis Pigmentoza, Jüvenil, 613341, Otozomal Resesif (Retinitis Pigmentoza) (LRAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa, juvenile, 613341, Autosomal recessive (Retinitis pigmentosa) (LRAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRAT	LRAT, LCA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leber congenital amaurosis 14 (LCA14) [MIM:613341]: A severe dystrophy of the retina, typically becoming evident in the first years of life. Visual function is usually poor and often accompanied by nystagmus, sluggish or near-absent pupillary responses, photophobia, high hyperopia and keratoconus. {ECO:0000269|PubMed:11381255, ECO:0000269|PubMed:17011878, ECO:0000269|PubMed:18055821}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Pigmentoza, Jüvenil Otozomal Resesif, 604232 (Retinitis Pigmentoza) (SPATA7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa, juvenile, autosomal recessive, 604232 (Retinitis pigmentosa) (SPATA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPATA7	SPATA7, HSD3, LCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa autosomal recessive (ARRP) [MIM:268000]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. {ECO:0000269|PubMed:19268277}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample

Retinitis Pigmentoza X'e Bağlı, Sinorespiratuar Enfeksiyonlar, ± Sağırlık, 300455 (Primer Silkyer Diskinezi-Retinit Pigmentosa Sendromu) (RPGR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis pigmentosa, X-linked, and sinorespiratory infections, with or without deafness, 300455 (Primary ciliary dyskinesia-retinitis pigmentosa syndrome) (RPGR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPGR	RPGR, RP3, CRD, RP15, COD1, CORDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 3 (RP3) [MIM:300029]: A X-linked retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. In RP3, affected males have a severe phenotype, and carrier females show a wide spectrum of clinical features ranging from completely asymptomatic to severe retinitis pigmentosa. Heterozygous women can manifest a form of choroideretinal degeneration which is distinguished from other types by the absence of visual defects in the presence of a brilliant, scintillating, golden-hued, patchy appearance most striking around the macula, called a tapetal-like retinal reflex. {ECO:0000269|PubMed:10482958, ECO:0000269|PubMed:10737996, ECO:0000269|PubMed:10932196, ECO:0000269|PubMed:10937588, ECO:0000269|PubMed:10970770, ECO:0000269|PubMed:11180598, ECO:0000269|PubMed:11992260, ECO:0000269|PubMed:12657579, ECO:0000269|PubMed:14564670, ECO:0000269|PubMed:8673101, ECO:0000269|PubMed:8817343, ECO:0000269|PubMed:9399904, ECO:0000269|PubMed:9851162}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinitis Punctata Albescens, 136880, Otozomal Resesif, Otozomal Dominant (Fundus Albipunctatus) (RLBP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis punctata albescens, 136880, Autosomal recessive, Autosomal dominant (Fundus albipunctatus) (RLBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RLBP1	RLBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. {ECO:0000269	PubMed:10102299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Punctata Albescens, 136880, Otozomal Resesif, Otozomal Dominant (Fundus Albipunctatus) (RHO gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis punctata albescens, 136880, Autosomal recessive, Autosomal dominant (Fundus albipunctatus) (RHO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHO	RHO, RP4, OPN2, CSNBAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Night blindness, congenital stationary, autosomal dominant 1 (CSNBAD1) [MIM:610445]: A non-progressive retinal disorder characterized by impaired night vision, often associated with nystagmus and myopia. {ECO:0000269|PubMed:7846071, ECO:0000269|PubMed:8358437, ECO:0000269|PubMed:9886392}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinitis Punctata Albescens, 136880, Otozomal Resesif, Otozomal Dominant (Fundus Albipunctatus) (PRPH2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinitis punctata albescens, 136880, Autosomal recessive, Autosomal dominant (Fundus albipunctatus) (PRPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRPH2	PRPH2, DS, RP7, PRPH, AVMD, AOFMD, CACD2, MDBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis punctata albescens (RPA) [MIM:136880]: Rare form of stationary night blindness characterized by a delay in the regeneration of cone and rod photopigments. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Retinoblastoma (13q14) (FISH)	Retinoblastoma (13q14) (FISH)	13q14	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Retinoblastom (RB1 13q14) (MLPA)	Retinoblastoma (RB1 13q14) (MLPA)	RB1 13q14	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Retinoblastom (RB1 13q14) (MLPA) (Prenatal)	Retinoblastoma (RB1 13q14) (MLPA) (Prenatal)	RB1 13q14	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Retinoblastom, 180200, Otozomal Dominant, Somatik Mutasyon; RB1 (Retinoblastom) (RB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinoblastoma, 180200, Autosomal dominant, Somatic mutation; RB1 (Retinoblastoma) (RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1	RB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Retinoblastom, Trilateral, 180200, Otozomal Dominant, Somatik Mutasyon (Retinoblastom) (RB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinoblastoma, trilateral, 180200, Autosomal dominant, Somatic mutation; RB1, RB1 (Retinoblastoma) (RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1	RB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Prematüre Retinopatisi, 133780, Otozomal Dominant (Ailesel Eksudatif Vitreoretinopati) (FZD4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinopathy of prematurity, 133780, Autosomal dominant (Familial exudative vitreoretinopathy) (FZD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FZD4	FZD4, EVR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Vitreoretinopathy, exudative 1 (EVR1) [MIM:133780]: A disorder of the retinal vasculature characterized by an abrupt cessation of growth of peripheral capillaries, leading to an avascular peripheral retina. This may lead to compensatory retinal neovascularization, which is thought to be induced by hypoxia from the initial avascular insult. New vessels are prone to leakage and rupture causing exudates and bleeding, followed by scarring, retinal detachment and blindness. Clinical features can be highly variable, even within the same family. Patients with mild forms of the disease are asymptomatic, and their only disease related abnormality is an arc of avascular retina in the extreme temporal periphery. In many ways the disease resembles retinopathy of prematurity but there is no evidence of prematurity or small birth weight in the patient history. {ECO:0000269|PubMed:12172548, ECO:0000269|PubMed:14507768, ECO:0000269|PubMed:15035869, ECO:0000269|PubMed:15223780, ECO:0000269|PubMed:15370539, ECO:0000269|PubMed:15488808, ECO:0000269|PubMed:15733276, ECO:0000269|PubMed:15981244, ECO:0000269|PubMed:17093393, ECO:0000269|PubMed:19172507, ECO:0000269|PubMed:19324841, ECO:0000269|PubMed:20340138}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Retinoşizis, 312700, X'e Bağlı Dominant; RS1 (Retinoşizis, X'e Bağlı) RS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Retinoschisis, 312700, X-linked dominant; RS1 (X-linked retinoschisis) (RS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RS1	RS1, XLRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinoschisis juvenile X-linked 1 (XLRS1) [MIM:312700]: A vitreo-retinal dystrophy characterized by macular pathology and by splitting of the superficial layer of the retina. Macular changes are present in almost all cases. In the fundi, radially oriented intraretinal foveomacular cysts are seen in a spoke-wheel configuration, with the absence of foveal reflex in most cases. In addition, approximately half of cases have bilateral peripheral retinoschisis in the inferotemporal part of the retina. Aside from the typical fundus appearance, strabismus, nystagmus, axial hyperopia, defective color vision and foveal ectopy can be present. The most important complications are vitreous hemorrhage, retinal detachment, and neovascular glaucoma. {ECO:0000269|PubMed:10079181, ECO:0000269|PubMed:10220153, ECO:0000269|PubMed:10234514, ECO:0000269|PubMed:10450864, ECO:0000269|PubMed:10533068, ECO:0000269|PubMed:17304551, ECO:0000269|PubMed:17615541, ECO:0000269|PubMed:19093009, ECO:0000269|PubMed:9326935, ECO:0000269|PubMed:9760195, ECO:0000269|Ref.10}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rett Sendromu, 312750, X'e Bağlı Dominant (Rett Sendromu) (MECP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rett syndrome, 312750, X-linked dominant (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, 312750, X'e Bağlı Dominant (Rett Sendromu) (MLPA)	Rett syndrome, 312750, X-linked dominant (Rett syndrome) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, 312750, X'e Bağlı Dominant (Rett Sendromu) (Prenatal) (MECP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rett syndrome, 312750, X-linked dominant (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rett Sendromu, 312750, X'e Bağlı Dominant (Rett Sendromu) (Prenatal) (MLPA)	Rett syndrome, 312750, X-linked dominant (Rett syndrome) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rett Sendromu, Tipik Olmayan 312750, X'e Bağlı Dominant (Atipik Rett Sendromu) (MECP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rett syndrome, atypical, 312750, X-linked dominant (Atypical Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, Tipik Olmayan 312750, X'e Bağlı Dominant (Atipik Rett Sendromu) (MLPA)	Rett syndrome, atypical, 312750, X-linked dominant (Atypical Rett syndrome) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, Tipik Olmayan 312750, X'e Bağlı Dominant (Atipik Rett Sendromu) (Prenatal) (MECP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rett syndrome, atypical, 312750, X-linked dominant (Atypical Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269	PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Rett Sendromu, Tipik Olmayan 312750, X'e Bağlı Dominant (Atipik Rett Sendromu) (Prenatal) (MLPA)	Rett syndrome, atypical, 312750, X-linked dominant (Atypical Rett syndrome) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rett Sendromu, Konjenital Varyant, 613454, Sporadik Vakalar (Atipik Rett Sendromu) (FOXG1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rett syndrome, congenital variant, 613454, Isolated cases (Atypical Rett syndrome) (FOXG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXG1	FOXG1, FOXG1B, FKHL1, FKH2, QIN, BF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Rett Sendromu, Konjenital Varyant, 613454, Sporadik Vakalar (Atipik Rett Sendromu) (Prenatal) (FOXG1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rett syndrome, congenital variant, 613454, Isolated cases (Atypical Rett syndrome) (FOXG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXG1	FOXG1, FOXG1B, FKHL1, FKH2, QIN, BF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rett Sendromu, Korunmuş Konuşma Varyantı, 312750, X'e Bağlı Dominant; RTT (Rett Sendromu) (MECP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rett syndrome, preserved speech variant, 312750, X-linked dominant; RTT (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, Korunmuş Konuşma Varyantı, 312750, X'e Bağlı Dominant; RTT (Rett Sendromu) (MLPA)	Rett syndrome, preserved speech variant, 312750, X-linked dominant; RTT (Rett syndrome) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rett Sendromu, Korunmuş Konuşma Varyantı, 312750, X'e Bağlı Dominant; RTT (Rett Sendromu) (Prenatal) (MECP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rett syndrome, preserved speech variant, 312750, X-linked dominant; RTT (Rett syndrome) (MECP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rett Sendromu, Korunmuş Konuşma Varyantı, 312750, X'e Bağlı Dominant; RTT (Rett Sendromu) (MECP2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rett syndrome, preserved speech variant, 312750, X-linked dominant; RTT (Rett syndrome) (Prenatal) (MLPA)	MECP2	MECP2, RTT, PPMX, MRX16, MRX79, AUTSX3, MRXSL, MRXS13, MRX79, MRX16	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Angelman syndrome (AS) [MIM:105830]: A neurodevelopmental disorder characterized by severe motor and intellectual retardation, ataxia, frequent jerky limb movements and flapping of the arms and hands, hypotonia, seizures, absence of speech, frequent smiling and episodes of paroxysmal laughter, open-mouthed expression revealing the tongue. {ECO:0000269|PubMed:11283202}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Revesz Sendromu, 268130, Otozomal Dominant (Revesz Sendromu) (TINF2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Revesz syndrome, 268130, Autosomal dominant (Revesz syndrome) (TINF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TINF2	TINF2, TIN2, DKCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyskeratosis congenita, autosomal dominant, 5 (DKCA5) [MIM:268130]: A disease characterized by bone marrow hypoplasia, nail dystrophy, fine sparse hair, fine reticulate skin pigmentation, oral leukoplakia, bilateral exudative retinopathy, cerebellar hypoplasia, and growth retardation. {ECO:0000269|PubMed:18252230}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Revesz Sendromu, 268130, Otozomal Dominant (Revesz Sendromu) (Prenatal) (TINF2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Revesz syndrome, 268130, Autosomal dominant (Revesz syndrome) (TINF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TINF2	TINF2, TIN2, DKCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyskeratosis congenita, autosomal dominant, 5 (DKCA5) [MIM:268130]: A disease characterized by bone marrow hypoplasia, nail dystrophy, fine sparse hair, fine reticulate skin pigmentation, oral leukoplakia, bilateral exudative retinopathy, cerebellar hypoplasia, and growth retardation. {ECO:0000269|PubMed:18252230}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Reynolds Sendromu, 613471, Otozomal Dominant (Reynolds Sendromu) (LBR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Reynolds syndrome, 613471, Autosomal dominant (Reynolds syndrome) (LBR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pelger-Huet anomaly (PHA) [MIM:169400]: An autosomal dominant inherited abnormality of granulocytes, characterized by abnormal ovoid shape, reduced nuclear segmentation and an apparently looser chromatin structure. Some individuals occasionally have skeletal anomalies, developmental delay, and seizures. {ECO:0000269|PubMed:14617022}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Reynolds Sendromu, 613471, Otozomal Dominant (Reynolds Sendromu) (Prenatal) (LBR gen) (Tüm Gen Dizi Analizi) (Prenatal)	Reynolds syndrome, 613471, Autosomal dominant (Reynolds syndrome) (LBR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LBR	LBR, PHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pelger-Huet anomaly (PHA) [MIM:169400]: An autosomal dominant inherited abnormality of granulocytes, characterized by abnormal ovoid shape, reduced nuclear segmentation and an apparently looser chromatin structure. Some individuals occasionally have skeletal anomalies, developmental delay, and seizures. {ECO:0000269|PubMed:14617022}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rh Mod Sendromu (Aşırı Hidrate Kalıtsal Stomatositoz) (RHAG gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rh-mod syndrome (Overhydrated hereditary stomatocytosis) (RHAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHAG	RHAG, RH50A, OHST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Rh-Null Hastalık, Amorf Tip# (RHCE gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rh-null disease, amorph type (RHCE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHCE	RHCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Rh-Null, Regülatör Tip; Rhn (Rh Eksikliği Sendromu) (RHAG gen) (Tüm Gen Dizi Analizi) (Postnatal)	RH-NULL, REGULATOR TYPE; RHN (Rh deficiency syndrome) (RHAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHAG	RHAG, RH50A, OHST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Rabdoid Yatkınlık Sendromu 1, 609322, Otozomal Dominant; RTPS1 (Ailesel Rabdoid Tümör) (SMARCB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdoid predisposition syndrome 1, 609322, Autosomal dominant; RTPS1 (Familial rhabdoid tumor) (SMARCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCB1	SMARCB1, SNF5, IN1, RDT, RTPS1, MRD15, SWNTS1, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schwannomatosis 1 (SWNTS1) [MIM:162091]: A cancer syndrome in which patients develop multiple non-vestibular schwannomas, benign neoplasms that arise from Schwann cells of the cranial, peripheral, and autonomic nerves. {ECO:0000269|PubMed:17357086, ECO:0000269|PubMed:18072270}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Rabdoid Tümör Yatkınlık Sendromu 2, 613325, Otozomal Dominant; RTPS2 (Ailesel Rabdoid Tümör) (SMARCA4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdoid tumor predisposition syndrome 2, 613325, Autosomal dominant; RTPS2 (Familial rhabdoid tumor) (SMARCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCA4	SMARCA4, BRG1, RTPS2, MRD16, CSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 16 (MRD16) [MIM:614609]: A disease characterized by multiple congenital anomalies and mental retardation. Mental retardation is defined by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD16 patients manifest developmental delay, absent or hypoplastic fifth fingernails or toenails, thick eyebrows and long eyelashes, hirsutism. Additional findings include hypotonia, microcephaly, seizures, a Dandy-Walker malformation, and vision and hearing problems. {ECO:0000269|PubMed:22426308}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Rabdoid Tümörler, Somatik, 609322 (SMARCB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdoid tumors, somatic, 609322 (SMARCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCB1	SMARCB1, SNF5, IN1, RDT, RTPS1, MRD15, SWNTS1, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Rabdomiyoliz, Serivastatin İle Uyarılan (CYP2C8 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyolysis, cerivastatin-induced (CYP2C8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C8	CYP2C8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Rabdomiyosarkom 2 Alveolar, 268220, Otozomal Resesif (Rabdomyosarkom) (PAX3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyosarcoma 2, alveolar, 268220, Autosomal recessive (Rhabdomyosarcoma) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. {ECO:0000269|PubMed:8664898}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rabdomiyosarkom 2 Alveolar, 268220, Otozomal Resesif (Rabdomyosarkom) (MLPA)	Rhabdomyosarcoma 2, alveolar, 268220, Autosomal recessive (Rhabdomyosarcoma) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniofacial-deafness-hand syndrome (CDHS) [MIM:122880]: Thought to be an autosomal dominant disease which comprises absence or hypoplasia of the nasal bones, hypoplastic maxilla, small and short nose with thin nares, limited movement of the wrist, short palpebral fissures, ulnar deviation of the fingers, hypertelorism and profound sensory-neural deafness. {ECO:0000269|PubMed:8664898}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rabdomiyosarkom 2 Alveolar, 268220, Otozomal Resesif; RMS2 (Rabdomyosarkom) (PAX7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyosarcoma 2, alveolar, 268220, Autosomal recessive; RMS2 (Rhabdomyosarcoma) (PAX7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX7	PAX7, RMS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhabdomyosarcoma 2 (RMS2) [MIM:268220]: A form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines. Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. {ECO:0000269|PubMed:8187070}. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration involving PAX7 is found in rhabdomyosarcoma. Translocation t(1;13)(p36;q14) with FOXO1. The resulting protein is a transcriptional activator. {ECO:0000269|PubMed:8187070}..	EDTA Blood Sample
Rabdomiyosarkom, Alveolar, 268220, Otozomal Resesif; Rabdomyosarkom) (FOXO1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyosarcoma, alveolar, 268220, Autosomal recessive (Rhabdomyosarcoma) (FOXO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXO1	FOXO1A, FKHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhabdomyosarcoma 2 (RMS2) [MIM:268220]: A form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines. Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. Note=The gene represented in this entry may be involved in disease pathogenesis. Chromosomal aberrations involving FOXO1 are found in rhabdomyosarcoma. Translocation (2:13)(q35;q14) with PAX3 and translocation t(1;13)(p36;q14) with PAX7. The resulting protein is a transcriptional activator.;	EDTA Blood Sample
Rabdomiyosarkom, Embriyonal, 2, 180295; RMSE2 (Pleviropulmoner Blastom-Ailesel Tümör Yatkınlık Sendromu) (DICER1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyosarcoma, embryonal, 2, 180295; RMSE2 (Pleuropulmonary blastoma family tumor susceptibility syndrome) (DICER1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DICER1	DICER1, HERNA, KIAA0928, MNG1, RMSE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Goiter multinodular 1, with or without Sertoli-Leydig cell tumors (MNG1) [MIM:138800]: A common disorder characterized by nodular overgrowth of the thyroid gland. Some individuals may also develop Sertoli-Leydig cell tumors, usually of the ovary. {ECO:0000269|PubMed:21205668}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Rabdomiyosarkom, Somatik, 268210; RMSE1 (Rabdomyosarkom) (SLC22A18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rhabdomyosarcoma, somatic, 268210; RMSE1 (Rhabdomyosarcoma) (SLC22A18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A18	SLC22A1L, BWSCR1A, IMPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Rhabdomyosarcoma, embryonal, 1 (RMSE1) [MIM:268210]: A form of rhabdomyosarcoma, a highly malignant tumor of striated muscle derived from primitive mesenchymal cells and exhibiting differentiation along rhabdomyoblastic lines. Rhabdomyosarcoma is one of the most frequently occurring soft tissue sarcomas and the most common in children. It occurs in four forms: alveolar, pleomorphic, embryonal and botryoidal rhabdomyosarcomas. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Romatoid Artrit, İlerlemesi, 180300 (IL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, progression of, 180300 (IL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL10	IL10, CSIF, GVHDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (SLC22A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (SLC22A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC22A4	SLC22A4, OCTN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis (RA) [MIM:180300]: An inflammatory disease with autoimmune features and a complex genetic component. It primarily affects the joints and is characterized by inflammatory changes in the synovial membranes and articular structures, widespread fibrinoid degeneration of the collagen fibers in mesenchymal tissues, and by atrophy and rarefaction of bony structures. {ECO:0000269	PubMed:14608356}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (PTPN22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (PTPN22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN22	PTPN22, PEP, PTPN8, LYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269	PubMed:15273934}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (PADI4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (PADI4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PADI4	PADI4, PADI5, PAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis (RA) [MIM:180300]: An inflammatory disease with autoimmune features and a complex genetic component. It primarily affects the joints and is characterized by inflammatory changes in the synovial membranes and articular structures, widespread fibrinoid degeneration of the collagen fibers in mesenchymal tissues, and by atrophy and rarefaction of bony structures. {ECO:0000269|PubMed:12833157}. Note=The gene represented in this entry may be involved in disease pathogenesis. The association to rheumatoid arthritis was initially thought to result from increased citrullination of target proteins (PubMed:12833157). However, variants that have been associated to rheumatoid arthritis (Ser-55, Ala-82 and Ala-112) do not affect the catalytic activity or the citrullination activity of PADI4, suggesting that these variants may affect the mRNA stability rather than the protein (PubMed:21245532). {ECO:0000269|PubMed:12833157, ECO:0000269|PubMed:21245532}.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (NFKBIL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (NFKBIL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFKBIL1	NFKBIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (HLA-DRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (HLA-DRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DRB1	HLA-DRB1, SS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (CIITA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (CIITA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIITA	CIITA, MHC2TA, C2TA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bare lymphocyte syndrome 2 (BLS2) [MIM:209920]: A severe combined immunodeficiency disease with early onset. It is characterized by a profound defect in constitutive and interferon- gamma induced MHC II expression, absence of cellular and humoral T-cell response to antigen challenge, hypogammaglobulinemia and impaired antibody production. The consequence include extreme susceptibility to viral, bacterial and fungal infections. {ECO:0000269|PubMed:10501838, ECO:0000269|PubMed:11466404, ECO:0000269|PubMed:11862382, ECO:0000269|PubMed:7749984, ECO:0000269|PubMed:8402893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Romatoid Artrit, Yatkınlık, 180300 (CD244 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, susceptibility to, 180300 (CD244 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CD244	CD244, NAIL, NKR2B4, SLAMF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Romatoid Artrit, Sistemik Jüvenil, 604302 (Sistemik Başlangıçlı Jüvenil İdiopatik Artrit) (IL6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, systemic juvenile, 604302 (Systemic-onset juvenile idiopathic arthritis) (IL6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL6	IL6, IFNB2, BSF2, HSF, HGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis systemic juvenile (RASJ) [MIM:604302]: An inflammatory articular disorder with systemic-onset beginning before the age of 16. It represents a subgroup of juvenile arthritis associated with severe extraarticular features and occasionally fatal complications. During active phases of the disorder, patients display a typical daily spiking fever, an evanescent macular rash, lymphadenopathy, hepatosplenomegaly, serositis, myalgia and arthritis. {ECO:0000269	PubMed:9769329}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Romatoid Artrit, Sistemik Jüvenil, Yatkınlık, 604302 (Sistemik Başlangıçlı Jüvenil İdiopatik Artrit) (MIF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rheumatoid arthritis, systemic juvenile, susceptibility to, 604302 (Systemic-onset juvenile idiopathic arthritis) (MIF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MIF	MIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis systemic juvenile (RASJ) [MIM:604302]: An inflammatory articular disorder with systemic-onset beginning before the age of 16. It represents a subgroup of juvenile arthritis associated with severe extraarticular features and occasionally fatal complications. During active phases of the disorder, patients display a typical daily spiking fever, an evanescent macular rash, lymphadenopathy, hepatosplenomegaly, serositis, myalgia and arthritis. {ECO:0000269	PubMed:11508429}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. {ECO:0000269	EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 1, 215100, Otozomal Resesif; RCDP1 (Rizomelik Kondrodisplazia Punktata) (PEX7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rhizomelic chondrodysplasia punctata, type 1, 215100, Autosomal recessive; RCDP1 (Rhizomelic chondrodysplasia punctata) (PEX7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX7	PEX7, RCDP1, PBD9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhizomelic chondrodysplasia punctata 1 (RCDP1) [MIM:215100]: A peroxisome biogenesis disorder. It is characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269	PubMed:9090381}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 1, 215100, Otozomal Resesif; RCDP1 (Rizomelik Kondrodisplazia Punktata) (Prenatal) (PEX7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rhizomelic chondrodysplasia punctata, type 1, 215100, Autosomal recessive; RCDP1 (Rhizomelic chondrodysplasia punctata) (PEX7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEX7	PEX7, RCDP1, PBD9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rhizomelic chondrodysplasia punctata 1 (RCDP1) [MIM:215100]: A peroxisome biogenesis disorder. It is characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269	PubMed:9090381}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 2, 222765, Otozomal Resesif; RCDP2 (Rizomelik Kondrodisplazia Punktata) (GNPAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rhizomelic chondrodysplasia punctata, type 2, 222765, Autosomal recessive; RCDP2 (Rhizomelic chondrodysplasia punctata) (GNPAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNPAT	GNPAT, DHAPAT, RCDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhizomelic chondrodysplasia punctata 2 (RCDP2) [MIM:222765]: A disease characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269|PubMed:11152660, ECO:0000269|PubMed:21990100, ECO:0000269|PubMed:9536089}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 2, 222765, Otozomal Resesif; RCDP2 (Rizomelik Kondrodisplazia Punktata) (Prenatal) (GNPAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rhizomelic chondrodysplasia punctata, type 2, 222765, Autosomal recessive; RCDP2 (Rhizomelic chondrodysplasia punctata) (GNPAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNPAT	GNPAT, DHAPAT, RCDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rhizomelic chondrodysplasia punctata 2 (RCDP2) [MIM:222765]: A disease characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269|PubMed:11152660, ECO:0000269|PubMed:21990100, ECO:0000269|PubMed:9536089}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 3, 600121, Otozomal Resesif; RCDP3 (Rizomelik Kondrodisplazia Punktata) (AGPS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rhizomelic chondrodysplasia punctata, type 3, 600121, Autosomal recessive; RCDP3 (Rhizomelic chondrodysplasia punctata) (AGPS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGPS	AGPS, ADHAPS, RCDP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rhizomelic chondrodysplasia punctata 3 (RCDP3) [MIM:600121]: A disease characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269|PubMed:11152660, ECO:0000269|PubMed:21990100, ECO:0000269|PubMed:9553082}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata Tip 3, 600121, Otozomal Resesif; RCDP3 (Rizomelik Kondrodisplazia Punktata) (Prenatal) (AGPS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rhizomelic chondrodysplasia punctata, type 3, 600121, Autosomal recessive; RCDP3 (Rhizomelic chondrodysplasia punctata) (AGPS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGPS	AGPS, ADHAPS, RCDP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rhizomelic chondrodysplasia punctata 3 (RCDP3) [MIM:600121]: A disease characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation. {ECO:0000269|PubMed:11152660, ECO:0000269|PubMed:21990100, ECO:0000269|PubMed:9553082}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rizomelik Kondrodisplazia Punktata, Tip 5, 616716, Otozomal Resesif (Rizomelik Kondrodisplazia Punktata) (PEX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rhizomelic chondrodysplasia punctata, type 5, 616716, Autosomal recessive (Rhizomelic chondrodysplasia punctata) (PEX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PEX5	PEX5, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10462504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Rizomelik Kondrodisplazia Punktata, Tip 5, 616716, Otozomal Resesif (Rizomelik Kondrodisplazia Punktata) (Prenatal) (PEXS gen) (Tüm Gen Dizi Analizi) (Prenatal)	Rhizomelic chondrodysplasia punctata, type 5, 616716, Autosomal recessive (Rhizomelic chondrodysplasia punctata) (PEXS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PEXS	PEXS, PXR1, PTS1R, PBD2A, PBD2B, RCDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Peroxisome biogenesis disorder 2B (PBD2B) [MIM:202370]: A peroxisome biogenesis disorder that includes neonatal adrenoleukodystrophy (NALD) and infantile Refsum disease (IRD), two milder manifestations of the Zellweger disease spectrum. The clinical course of patients with the NALD and IRD presentation is variable and may include developmental delay, hypotonia, liver dysfunction, sensorineural hearing loss, retinal dystrophy and vision impairment. Children with the NALD presentation may reach their teens, while patients with the IRD presentation may reach adulthood. The clinical conditions are often slowly progressive in particular with respect to loss of hearing and vision. The biochemical abnormalities include accumulation of phytanic acid, very long chain fatty acids (VLCFA), di- and trihydroxycholestanoic acid and pipecolic acid. {ECO:0000269|PubMed:10482504, ECO:0000269|PubMed:7719337}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Riboflavin Eksikliği, 615026, Otozomal Dominant; RBFVD (Maternal Riboflavin Eksikliği) (SLC52A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Riboflavin deficiency, 615026, Autosomal dominant; RBFVD (Maternal riboflavin deficiency) (SLC52A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC52A1	SLC52A1, GPR172B, GPCR42, PAR2, FLJ10060, RBFVD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Riboflavin Eksikliği, 615026, Otozomal Dominant; RBFVD (Maternal Riboflavin Eksikliği) (Prenatal) (SLC52A1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Riboflavin deficiency, 615026, Autosomal dominant; RBFVD (Maternal riboflavin deficiency) (SLC52A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC52A1	SLC52A1, GPR172B, GPCR42, PAR2, FLJ10060, RBFVD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Riboz 5-Fosfat İzomeraz Eksikliği, 608611, Otozomal Resesif (Riboz-5-P İzomeraz Eksikliği) (RPIA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ribose 5-phosphate isomerase deficiency, 608611, Autosomal recessive (Ribose-5-P isomerase deficiency) (RPIA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RPIA	RPIA, RPI, RPIAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ribose 5-phosphate isomerase deficiency (RPID) [MIM:608611]: A patient has been described with a deficiency of ribose 5-phosphate isomerase who presented with leukoencephalopathy and peripheral neuropathy. Proton magnetic resonance spectroscopy of the brain revealed a highly elevated level of the polyols ribitol and D-arabitol, which were subsequently also found in high concentrations in body fluids. Deficient activity of RPIA, one of the pentose phosphate pathway enzymes, has been demonstrated in fibroblasts. {ECO:0000269	PubMed:14988808}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Riboz 5-Fosfat İzomeraz Eksikliği, 608611, Otozomal Resesif (Riboz-5-P İzomeraz Eksikliği) (Prenatal) (RPIA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ribose 5-phosphate isomerase deficiency, 608611, Autosomal recessive (Ribose-5-P isomerase deficiency) (RPIA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RPIA	RPIA, RPI, RPIAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ribose 5-phosphate isomerase deficiency (RPID) [MIM:608611]: A patient has been described with a deficiency of ribose 5-phosphate isomerase who presented with leukoencephalopathy and peripheral neuropathy. Proton magnetic resonance spectroscopy of the brain revealed a highly elevated level of the polyols ribitol and D-arabitol, which were subsequently also found in high concentrations in body fluids. Deficient activity of RPIA, one of the pentose phosphate pathway enzymes, has been demonstrated in fibroblasts. {ECO:0000269	PubMed:14988808}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Raşitizm, Vitamin D 25 Hidroksilasyon Defektine Bağlı, 600081, Otozomal Resesif (Hipokalsemik D Vitamini Bağımlı Raşitizm) (CYP2R1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rickets due to defect in vitamin D 25-hydroxylation, 600081, Autosomal recessive (Hypocalcemic vitamin D-dependent rickets) (CYP2R1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2R1	CYP2R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rickets vitamin D-dependent 1B (VDDR1B) [MIM:600081]: A disorder caused by a selective deficiency of the active form of vitamin D (1,25-dihydroxyvitamin D3) and resulting in defective bone mineralization and clinical features of rickets. The patients sera have low calcium concentrations, low phosphate concentrations, elevated alkaline phosphatase activity and low levels of 25-hydroxyvitamin D. {ECO:0000269	PubMed:15128933}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Raşitizm, Vitamin D Dirençli, Tip 2A, 277440, Otozomal Resesif; VDDR2A (Hipokalsemik Vitamin D Dirençli Raşitizm) (VDR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rickets, vitamin D-resistant, type IIA, 277440, Autosomal recessive; VDDR2A (Hypocalcemic vitamin D-resistant rickets) (VDR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VDR	VDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rickets vitamin D-dependent 2A (VDDR2A) [MIM:277440]: A disorder of vitamin D metabolism resulting in severe rickets, hypocalcemia and secondary hyperparathyroidism. Most patients have total alopecia in addition to rickets. {ECO:0000269|PubMed:1652893, ECO:0000269|PubMed:2177843, ECO:0000269|PubMed:2849209, ECO:0000269|PubMed:7828346, ECO:0000269|PubMed:8106618, ECO:0000269|PubMed:8381803, ECO:0000269|PubMed:8392085, ECO:0000269|PubMed:8675579, ECO:0000269|PubMed:8961271, ECO:0000269|PubMed:9005998}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
RIDDLE Sendromu, 611943, Otozomal Resesif (RIDDLE Sendromu) (RNF168 geni) (Tüm Gen Dizi Analizi) (Postnatal)	RIDDLE syndrome, 611943, Autosomal recessive (RIDDLE syndrome) (RNF168 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF168	RNF168	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Riddle syndrome (RIDDLES) [MIM:611943]: Characterized by increased radiosensitivity, immunodeficiency, mild motor control and learning difficulties, facial dysmorphism, and short stature. Defects are probably due to impaired localization of TP53BP1 and BRCA1 at DNA lesions. {ECO:0000269	PubMed:19203578}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
RIDDLE Sendromu, 611943, Otozomal Resesif (RIDDLE Sendromu) (Prenatal) (RNF168 geni) (Tüm Gen Dizi Analizi) (Prenatal)	RIDDLE syndrome, 611943, Autosomal recessive (RIDDLE syndrome) (RNF168 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNF168	RNF168	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Riddle syndrome (RIDDLES) [MIM:611943]: Characterized by increased radiosensitivity, immunodeficiency, mild motor control and learning difficulties, facial dysmorphism, and short stature. Defects are probably due to impaired localization of TP53BP1 and BRCA1 at DNA lesions. {ECO:0000269	PubMed:19203578}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sağ Atriyal İzomerizm, 208530, Otozomal Resesif; RAI (Ivemark Sendromu (Sağ Atriyal İzomerizm)) (GDF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Right atrial isomerism, 208530, Autosomal recessive; RAI (Ivemark syndrome) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269	PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sağ Atriyal İzomerizm, 208530, Otozomal Resesif; RAI (Ivemark Sendromu (Sağ Atriyal İzomerizm)) (Prenatal) (GDF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Right atrial isomerism, 208530, Autosomal recessive; RAI (Ivemark syndrome) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269	PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rijidite ve Multifokal Nöbet Sendromu, Letal Neonatal, 614498, Otozomal Resesif; RMFSL (Letal Neonatal Spastisite-Epileptik Ensefalopati Sendromu) (BRAT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498, Autosomal recessive; RMFSL (Lethal neonatal spasticity-epileptic encephalopathy syndrome) (BRAT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRAT1	BRAT1, BAAT1, C7orf27, RMFSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rigidity and multifocal seizure syndrome, lethal neonatal (RMFSL) [MIM:614498]: A lethal, neonatal, neurologic disorder characterized by episodic jerking that is apparent in utero, lack of psychomotor development, axial and limb rigidity, frequent multifocal seizures, and dysautonomia. At birth, affected individuals have small heads, overlapping cranial sutures, small or absent fontanels, and depressed frontal bones. Infants show poorly responsive focal jerks of the tongue, face and arms in a nearly continuous sequence throughout life. {ECO:0000269	PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Rijidite ve Multifokal Nöbet Sendromu, Letal Neonatal, 614498, Otozomal Resesif; RMFSL (Letal Neonatal Spastisite-Epileptik Ensefalopati Sendromu) (Prenatal) (BRAT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rigidity and multifocal seizure syndrome, lethal neonatal, 614498, Autosomal recessive; RMFSL (Lethal neonatal spasticity-epileptic encephalopathy syndrome) (BRAT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BRAT1	BRAT1, BAAT1, C7orf27, RMFSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rigidity and multifocal seizure syndrome, lethal neonatal (RMFSL) [MIM:614498]: A lethal, neonatal, neurologic disorder characterized by episodic jerking that is apparent in utero, lack of psychomotor development, axial and limb rigidity, frequent multifocal seizures, and dysautonomia. At birth, affected individuals have small heads, overlapping cranial sutures, small or absent fontanels, and depressed frontal bones. Infants show poorly responsive focal jerks of the tongue, face and arms in a nearly continuous sequence throughout life. {ECO:0000269	PubMed:22279524}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Korneanın Halka Dermoidi, 180550, Otozomal Dominant; RDC (Korneanın Halka Dermoidi) (PITX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ring dermoid of cornea, 180550, Autosomal dominant; RDC (Ring dermoid of cornea) (PITX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Iridogoniodysgenesis 2 (IRID2) [MIM:137600]: Autosomal dominant inherited disease. {ECO:0000269|PubMed:9437321, ECO:0000269|PubMed:9618168}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Korneanın Halka Dermoidi, 180550, Otozomal Dominant; RDC (Korneanın Halka Dermoidi) (MLPA)	Ring dermoid of cornea, 180550, Autosomal dominant; RDC (Ring dermoid of cornea) (MLPA)	PITX2	PITX2, IDG2, RIEG1, RGS, IGDS2, ASGD4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Iridogoniodysgenesis 2 (IRID2) [MIM:137600]: Autosomal dominant inherited disease. {ECO:0000269|PubMed:9437321, ECO:0000269|PubMed:9618168}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dalgalanan (Ripling) Kas Hastalığı, 606072, Otozomal Dominant; RMD (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (CAV3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rippling muscle disease, 606072, Autosomal dominant; RMD (Rippling muscle disease) (CAV3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAV3	CAV3, LGMD1C, LQT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Dalgalanan (Ripling) Kas Hastalığı, 606072, Otozomal Dominant; RMD (Dalgalanan Kas Hastalığı (Rippling Muscle Disease)) (MLPA)	Rippling muscle disease, 606072, Autosomal dominant; RMD (Rippling muscle disease) (MLPA)	CAV3	CAV3, LGMD1C, LQT9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: HyperCKmia (HYPCK) [MIM:123320]: Characterized by persistent elevated levels of serum creatine kinase without muscle weakness. {ECO:0000269|PubMed:10746614, ECO:0000269|PubMed:12082049, ECO:0000269|PubMed:14663034, ECO:0000269|PubMed:15099591, ECO:0000269|PubMed:15580566}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ritscher-Schinzel Sendromu 1, 220210, Otozomal Resesif; RTSC1 (3C Sendromu Tip (Ritscher-Schinzel Sendromu)) (WASHC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ritscher-Schinzel syndrome 1, 220210, Autosomal recessive; RTSC1 (3C syndrome) (WASHC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WASHC5	KIAA0196, SPG8, RTSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ritscher-Schinzel Sendromu 1, 220210, Otozomal Resesif; RTSC1 (3C Sendromu Tip (Ritscher-Schinzel Sendromu)) (Prenatal) (WASHC5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ritscher-Schinzel syndrome 1, 220210, Autosomal recessive; RTSC1 (3C syndrome) (WASHC5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WASHC5	KIAA0196, SPG8, RTSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Ritscher-Schinzel Sendromu 2, 300963, X'e Bağlı: RTSC2 (3C Sendromu Tip (Ritscher-Schinzel Sendromu)) (CCDC22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ritscher-Schinzel syndrome 2, 300963, X-linked recessive; RTSC2 (3C syndrome) (CCDC22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC22	CCDC22, RTSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ritscher-Schinzel Sendromu 2, 300963, X'e Bağlı: RTSC2 (3C Sendromu Tip (Ritscher-Schinzel Sendromu)) (Prenatal) (CCDC22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ritscher-Schinzel syndrome 2, 300963, X-linked recessive; RTSC2 (3C syndrome) (CCDC22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCDC22	CCDC22, RTSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Roberts Sendromu, 268300, Otozomal Resesif; RBS (Roberts Sendromu (SC Fokomel Sendromu)) (ESCO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Roberts syndrome, 268300, Autosomal recessive; RBS (Roberts syndrome) (ESCO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESCO2	ESCO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: SC phocomelia syndrome (SCPS) [MIM:269000]: Has a milder phenotype than RBS, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common. {ECO:0000269|PubMed:16380922}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Roberts Sendromu, 268300, Otozomal Resesif; RBS (Roberts Sendromu (SC Fokomel Sendromu)) (Prenatal) (ESCO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Roberts syndrome, 268300, Autosomal recessive; RBS (Roberts syndrome) (ESCO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ESCO2	ESCO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: SC phocomelia syndrome (SCPS) [MIM:269000]: Has a milder phenotype than RBS, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common. {ECO:0000269|PubMed:16380922}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robin Sekans, Yarık mandıbula ve Ekstremite Anomalileri, 268305, Otozomal Resesif (Richieri Costa-Pereira Sendromu) (EIF4A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robin sequence with cleft mandible and limb anomalies, 268305, Autosomal recessive (Richieri Costa-Pereira syndrome) (EIF4A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF4A3	DDX4B, EIF4A3, MUK34, NMP265, KIAA0111, RCPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Robin Sekans, Yarık mandıbula ve Ekstremite Anomalileri, 268305, Otozomal Resesif (Richieri Costa-Pereira Sendromu) (Prenatal) (EIF4A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robin sequence with cleft mandible and limb anomalies, 268305, Autosomal recessive (Richieri Costa-Pereira syndrome) (EIF4A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EIF4A3	DDX4B, EIF4A3, MUK34, NMP265, KIAA0111, RCPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 1, 180700, Otozomal Dominant; DRS1 (Otozomal Dominant Robinow Sendromu) (WNT5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow syndrome, autosomal dominant 1, 180700, Autosomal dominant; DRS1 (Autosomal dominant Robinow syndrome) (WNT5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT5A	WNT5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Robinow syndrome, autosomal dominant 1 (DRS1) [MIM:180700]: A disease characterized by short-limb dwarfism, costovertebral segmentation defects and abnormalities of the head, face and external genitalia. The clinical signs are generally milder in dominant cases. {ECO:0000269|PubMed:19918918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 1, 180700, Otozomal Dominant; DRS1 (Otozomal Dominant Robinow Sendromu) (Prenatal) (WNT5A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robinow syndrome, autosomal dominant 1, 180700, Autosomal dominant; DRS1 (Autosomal dominant Robinow syndrome) (WNT5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT5A	WNT5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Robinow syndrome, autosomal dominant 1 (DRS1) [MIM:180700]: A disease characterized by short-limb dwarfism, costovertebral segmentation defects and abnormalities of the head, face and external genitalia. The clinical signs are generally milder in dominant cases. {ECO:0000269|PubMed:19918918}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 2, 616331, Otozomal Dominant; DRS2 (Otozomal Dominant Robinow Sendromu) (DVL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow syndrome, autosomal dominant 2, 616331, Autosomal dominant; DRS2 (Autosomal dominant Robinow syndrome) (DVL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DVL1	DVL1, DRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Robinow syndrome, autosomal dominant 2 (DRS2) [MIM:616331]: A rare skeletal dysplasia syndrome characterized by dysmorphic features resembling a fetal face, mesomelic limb shortening, hypoplastic external genitalia in males, costovertebral segmentation defects, and renal anomalies. {ECO:0000269|PubMed:25817014, ECO:0000269|PubMed:25817016}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 2, 616331, Otozomal Dominant; DRS2 (Otozomal Dominant Robinow Sendromu) (Prenatal) (DVL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robinow syndrome, autosomal dominant 2, 616331, Autosomal dominant; DRS2 (Autosomal dominant Robinow syndrome) (DVL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DVL1	DVL1, DRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Robinow syndrome, autosomal dominant 2 (DRS2) [MIM:616331]: A rare skeletal dysplasia syndrome characterized by dysmorphic features resembling a fetal face, mesomelic limb shortening, hypoplastic external genitalia in males, costovertebral segmentation defects, and renal anomalies. {ECO:0000269|PubMed:25817014, ECO:0000269|PubMed:25817016}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 3, 616894, Otozomal Dominant; DRS3 (Otozomal Dominant Robinow Sendromu) (DVL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow syndrome, autosomal dominant 3, 616894, Autosomal dominant; DRS3 (Autosomal dominant Robinow syndrome) (DVL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DVL3	DVL3, DRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Robinow Sendromu, Otozomal Dominant 3, 616894, Otozomal Dominant; DRS3 (Otozomal Dominant Robinow Sendromu) (Prenatal) (DVL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robinow syndrome, autosomal dominant 3, 616894, Autosomal dominant; DRS3 (Autosomal dominant Robinow syndrome) (DVL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DVL3	DVL3, DRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow Sendromu, Otozomal Resesif, 268310, Otozomal Resesif; RRS (Robinow Sendromu, Otozomal Resesif) (ROR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow syndrome, autosomal recessive, 268310, Autosomal recessive; RRS (Autosomal recessive Robinow syndrome) (ROR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROR2	ROR2, BDB1, BDB, NTRKR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Robinow Sendromu, Otozomal Resesif, 268310, Otozomal Resesif; RRS (Robinow Sendromu, Otozomal Resesif) (MLPA)	Robinow syndrome, autosomal recessive, 268310, Autosomal recessive; RRS (Autosomal recessive Robinow syndrome) (MLPA)	ROR2	ROR2, BDB1, BDB, NTRKR2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Robinow Sendromu, Otozomal Resesif, 268310, Otozomal Resesif; RRS (Robinow Sendromu, Otozomal Resesif) (Prenatal) (ROR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robinow syndrome, autosomal recessive, 268310, Autosomal recessive; RRS (Autosomal recessive Robinow syndrome) (ROR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ROR2	ROR2, BDB1, BDB, NTRKR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow Sendromu, Otozomal Resesif, 268310, Otozomal Resesif; RRS (Robinow Sendromu, Otozomal Resesif) (MLPA)	Robinow syndrome, autosomal recessive, 268310, Autosomal recessive; RRS (Autosomal recessive Robinow syndrome) (Prenatal) (MLPA)	ROR2	ROR2, BDB1, BDB, NTRKR2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brachydactyly B1 (BDB1) [MIM:113000]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type B1 the middle phalanges are short but in addition the terminal phalanges are rudimentary or absent. Both fingers and toes are affected. The thumbs and big toes are usually deformed. Symphalangism is also a feature. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow-Sorauf Sendromu, 180750, Otozomal Dominant (Robinow-Sorauf Sendromu) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow-Sorauf syndrome, 180750, Autosomal dominant (Robinow-Sorauf syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow-Sorauf Sendromu, 180750, Otozomal Dominant (Robinow-Sorauf Sendromu) (MLPA)	Robinow-Sorauf syndrome, 180750, Autosomal dominant (Robinow-Sorauf syndrome) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow-Sorauf Sendromu, 180750, Otozomal Dominant (Robinow-Sorauf Sendromu) (Prenatal) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Robinow-Sorauf syndrome, 180750, Autosomal dominant (Robinow-Sorauf syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Robinow-Sorauf Sendromu, 180750, Otozomal Dominant (Robinow-Sorauf Sendromu) (Prenatal) (MLPA)	Robinow-Sorauf syndrome, 180750, Autosomal dominant (Robinow-Sorauf syndrome) (Prenatal) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rolandik Epilepsi, Mental Retardasyon ve Konuşma Dispraksi, 300643 (Rolandik Epilepsi-Konuşma Dispraksi Sendromu) (SRPX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (Rolandic epilepsy-speech dyspraxia syndrome) (SRPX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRPX2	SRPX2, SRPUL, RESDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rolandic epilepsy with speech dyspraxia and mental retardation X-linked (RESDX) [MIM:300643]: A condition characterized by the association of rolandic seizures with oral and speech dyspraxia, and mental retardation. Rolandic seizures occur during a period of significant brain maturation. During this time, dysfunction of neural network activities such as focal discharges may be associated with specific developmental disabilities resulting in specific cognitive impairments of language, visuo-spatial abilities or attention. {ECO:0000269|PubMed:16497722}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Rolandik Epilepsi, Mental Retardasyon ve Konuşma Dispraksi, 300643 (Rolandik Epilepsi-Konuşma Dispraksi Sendromu) (Prenatal) (SRPX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rolandic epilepsy, mental retardation, and speech dyspraxia, 300643 (Rolandic epilepsy-speech dyspraxia syndrome) (SRPX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SRPX2	SRPX2, SRPUL, RESDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rolandic epilepsy with speech dyspraxia and mental retardation X-linked (RESDX) [MIM:300643]: A condition characterized by the association of rolandic seizures with oral and speech dyspraxia, and mental retardation. Rolandic seizures occur during a period of significant brain maturation. During this time, dysfunction of neural network activities such as focal discharges may be associated with specific developmental disabilities resulting in specific cognitive impairments of language, visuo-spatial abilities or attention. {ECO:0000269|PubMed:16497722}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ROS1 (6q22) (FISH)	ROS1 (FISH)	6q22	.	FISH	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinle örtülü bloğu		10	30	.	.	20	Postnatal	.	.	Tumor tissue (Fresh or etanol fixed) /Paraffinized tissue sample
Rothmund-Thomson Sendromu, 268400, Otozomal Resesif; RTS (Rothmund-Thomson Sendromu) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rothmund-Thomson syndrome, 268400, Autosomal recessive; RTS (Rothmund-Thomson syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. {ECO:0000269|PubMed:15964893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rothmund-Thomson Sendromu, 268400, Otozomal Resesif; RTS (Rothmund-Thomson Sendromu) (Prenatal) (RECQL4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rothmund-Thomson syndrome, 268400, Autosomal recessive; RTS (Rothmund-Thomson syndrome) (RECQL4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RECQL4	RECQL4, RTS, RECQ4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Baller-Gerold syndrome (BGS) [MIM:218600]: An autosomal recessive syndrome characterized by short stature, craniosynostosis, absent or hypoplastic radii, short and curved ulna, fused carpal bones and absent carpals, metacarpals and phalanges. Some patients manifest poikiloderma. Cases reported as Baller-Gerold syndrome have phenotypic overlap with several other disorders, including Saethre-Chotzen syndrome. {ECO:0000269|PubMed:15964893}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Roussy-Levy Sendromu, 180800, (Roussy-Levy Sendromu) (MPZ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Roussy-Levy syndrome, 180800, (Roussy-Lévy syndrome) (MPZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adie pupil (ADIEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269|PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Roussy-Levy Sendromu, 180800, (Roussy-Levy Sendromu) (MLPA)	Roussy-Levy syndrome, 180800, (Roussy-Lévy syndrome) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adie pupil (ADIEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269|PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Roussy-Levy Sendromu, 180800, (Roussy-Levy Sendromu) (Prenatal) (MPZ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Roussy-Levy syndrome, 180800, (Roussy-Lévy syndrome) (MPZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adie pupil (ADIEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269|PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Roussy-Levy Sendromu, 180800, (Roussy-Levy Sendromu) (Prenatal) (MLPA)	Roussy-Levy syndrome, 180800, (Roussy-Lévy syndrome) (Prenatal) (MLPA)	MPZ	MPZ, CMT1B, CMTDID, CHM, DSS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Adie pupil (ADIEP) [MIM:103100]: A stationary, benign disorder characterized by tonic, sluggishly reacting pupil and hypoactive or absent tendon reflexes. Adie pupil is a characteristic of Charcot-Marie-Tooth disease type 2J. {ECO:0000269|PubMed:16775239}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Roussy-Levy Sendromu, 180800, Otozomal Dominant (Roussy-Levy Sendromu) (PMP22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Roussy-Levy syndrome, 180800, Autosomal dominant (Roussy-Lévy syndrome) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Roussy-Levy Sendromu, 180800, Otozomal Dominant (Roussy-Levy Sendromu) (MLPA)	Roussy-Levy syndrome, 180800, Autosomal dominant (Roussy-Lévy syndrome) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Roussy-Levy Sendromu, 180800, Otozomal Dominant (Roussy-Levy Sendromu) (Prenatal) (PMP22 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Roussy-Levy syndrome, 180800, Autosomal dominant (Roussy-Lévy syndrome) (PMP22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140841, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Roussy-Levy Sendromu, 180800, Otozomal Dominant (Roussy-Levy Sendromu) (Prenatal) (MLPA)	Roussy-Levy syndrome, 180800, Autosomal dominant (Roussy-Levy syndrome) (Prenatal) (MLPA)	PMP22	PMP22, CMT1A, CMT1E, DSS, CIDP	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Charcot-Marie-Tooth disease 1A (CMT1A) [MIM:118220]: A dominant demyelinating form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot- Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Demyelinating neuropathies are characterized by severely reduced nerve conduction velocities (less than 38 m/sec), segmental demyelination and remyelination with onion bulb formations on nerve biopsy, slowly progressive distal muscle atrophy and weakness, absent deep tendon reflexes, and hollow feet. {ECO:0000269|PubMed:10489052, ECO:0000269|PubMed:10737979, ECO:0000269|PubMed:11140941, ECO:0000269|PubMed:11835375, ECO:0000269|PubMed:12402337, ECO:0000269|PubMed:12497641, ECO:0000269|PubMed:1303281, ECO:0000269|PubMed:15205993, ECO:0000269|PubMed:8252046, ECO:0000269|PubMed:8510709, ECO:0000269|PubMed:8615087, ECO:0000269|PubMed:8655153, ECO:0000269|PubMed:8777804, ECO:0000269|PubMed:9040744}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rubinstein-Taybi Sendromu 1, 180849, Otozomal Dominant; RSTS1 (Rubinstein-Taybi Sendromu) (CREBBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rubinstein-Taybi syndrome 1, 180849, Autosomal dominant; RSTS1 (Rubinstein-Taybi syndrome) (CREBBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CREBBP	CREBBP, CBP, RSTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rubinstein-Taybi syndrome 1 (RSTS1) [MIM:180849]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. {ECO:0000269|PubMed:11331617, ECO:0000269|PubMed:12114483, ECO:0000269|PubMed:12566391, ECO:0000269|PubMed:15706485, ECO:0000269|PubMed:20684013, ECO:0000269|PubMed:25388907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rubinstein-Taybi Sendromu 1, 180849, Otozomal Dominant; RSTS1 (Rubinstein-Taybi Sendromu) (MLPA)	Rubinstein-Taybi syndrome 1, 180849, Autosomal dominant; RSTS1 (Rubinstein-Taybi syndrome) (MLPA)	CREBBP	CREBBP, CBP, RSTS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Rubinstein-Taybi syndrome 1 (RSTS1) [MIM:180849]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. {ECO:0000269|PubMed:11331617, ECO:0000269|PubMed:12114483, ECO:0000269|PubMed:12566391, ECO:0000269|PubMed:15706485, ECO:0000269|PubMed:20684013, ECO:0000269|PubMed:25388907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rubinstein-Taybi Sendromu 1, 180849, Otozomal Dominant; RSTS1 (Rubinstein-Taybi Sendromu) (Prenatal) (CREBBP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rubinstein-Taybi syndrome 1, 180849, Autosomal dominant; RSTS1 (Rubinstein-Taybi syndrome) (CREBBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CREBBP	CREBBP, CBP, RSTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rubinstein-Taybi syndrome 1 (RSTS1) [MIM:180849]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. {ECO:0000269|PubMed:11331617, ECO:0000269|PubMed:12114483, ECO:0000269|PubMed:12566391, ECO:0000269|PubMed:15706485, ECO:0000269|PubMed:20684013, ECO:0000269|PubMed:25388907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rubinstein-Taybi Sendromu 1, 180849, Otozomal Dominant; RSTS1 (Rubinstein-Taybi Sendromu) (Prenatal) (MLPA)	Rubinstein-Taybi syndrome 1, 180849, Autosomal dominant; RSTS1 (Rubinstein-Taybi syndrome) (Prenatal) (MLPA)	CREBBP	CREBBP, CBP, RSTS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rubinstein-Taybi syndrome 1 (RSTS1) [MIM:180849]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. {ECO:0000269|PubMed:11331617, ECO:0000269|PubMed:12114483, ECO:0000269|PubMed:12566391, ECO:0000269|PubMed:15706485, ECO:0000269|PubMed:20684013, ECO:0000269|PubMed:25388907}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Rubinstein-Taybi Sendromu, 2, 613684, Otozomal Dominant; RSTS2 (Rubinstein-Taybi Sendromu) (EP300 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Rubinstein-Taybi syndrome 2, 613684, Autosomal dominant; RSTS2 (Rubinstein-Taybi syndrome) (EP300 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EP300	EP300, RSTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rubinstein-Taybi syndrome 2 (RSTS2) [MIM:613684]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. Some individuals with RSTS2 have less severe mental impairment, more severe microcephaly, and a greater degree of changes in facial bone structure than RSTS1 patients. {ECO:0000269|PubMed:15706485}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Rubinstein-Taybi Sendromu, 2, 613684, Otozomal Dominant; RSTS2 (Rubinstein-Taybi Sendromu) (Prenatal) (EP300 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Rubinstein-Taybi syndrome 2, 613684, Autosomal dominant; RSTS2 (Rubinstein-Taybi syndrome) (EP300 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EP300	EP300, RSTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Rubinstein-Taybi syndrome 2 (RSTS2) [MIM:613684]: A disorder characterized by craniofacial abnormalities, postnatal growth deficiency, broad thumbs, broad big toes, mental retardation and a propensity for development of malignancies. Some individuals with RSTS2 have less severe mental impairment, more severe microcephaly, and a greater degree of changes in facial bone structure than RSTS1 patients. {ECO:0000269|PubMed:15706485}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ruijs-Aalfs Sendromu, 616200, Otozomal Resesif; RJALS (Progeroid Bulgular-Hepatoselüler Karsinom Yatkınlık Sendromu (Ruijs-Aalfs Sendromu)) (SPRTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ruijs-Aalfs syndrome, 616200, Autosomal recessive; RJALS (Progeroid features-hepatocellular carcinoma predisposition syndrome) (SPRTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPRTN	SPRTN, DVC1, C1orf124, RJALS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ruijs-Aalfs syndrome (RJALS) [MIM:616200]: A syndrome characterized by genomic instability, progeroid features, and susceptibility toward early onset hepatocellular carcinoma. {ECO:0000269|PubMed:25261934}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ruijs-Aalfs Sendromu, 616200, Otozomal Resesif; RJALS (Progeroid Bulgular-Hepatoselüler Karsinom Yatkınlık Sendromu (Ruijs-Aalfs Sendromu)) (Prenatal) (SPRTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ruijs-Aalfs syndrome, 616200, Autosomal recessive; RJALS (Progeroid features-hepatocellular carcinoma predisposition syndrome) (SPRTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SPRTN	SPRTN, DVC1, C1orf124, RJALS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ruijs-Aalfs syndrome (RJALS) [MIM:616200]: A syndrome characterized by genomic instability, progeroid features, and susceptibility toward early onset hepatocellular carcinoma. {ECO:0000269|PubMed:25261934}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Russel -Silver Sendromu (MEST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Russel-Silver syndrome (MEST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MEST	MEST, PEG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Russel -Silver Sendromu (Prenatal) (MEST geni) (Tüm Gen Dizi Analizi) (Prenatal)	Russel-Silver syndrome (MEST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MEST	MEST, PEG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sakkaropinüri, 268700, Otozomal Resesif (Sakkaropinüri) (AASS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Saccharopinuria, 268700, Autosomal recessive (Saccharopinuria) (AASS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AASS	AASS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperlysinemia, 1 (HYPLYS1) [MIM:238700]: An autosomal recessive metabolic condition with variable clinical features. Some patients present with non-specific seizures, hypotonia, or mildly delayed psychomotor development, and increased serum lysine and pipecolic acid on laboratory analysis. However, about half of the probands are reported to be asymptomatic, and hyperlysinemia is generally considered to be a benign metabolic variant. {ECO:0000269|PubMed:10775527}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sakral Agenezis-Vertebral Anomaliler, 615709, Otozomal Resesif; SAVA (Sakral Agenezisi-Vertebralarda Anormal Ossifikasyon-Kalıcı Notokord Kanal Sendromu) (T geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sacral agenesis with vertebral anomalies, 615709, Autosomal recessive; SAVA (Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome) (T gene) (Sequence Analysis-All Coding Exons) (Postnatal)	T	T, TFT, SAVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chordoma (CHDM) [MIM:215400]: Rare, clinically malignant tumors derived from notochordal remnants. They occur along the length of the spinal axis, predominantly in the sphenooccipital, vertebral and sacrococcygeal regions. They are characterized by slow growth, local destruction of bone, extension into adjacent soft tissues and rarely, distant metastatic spread. {ECO:0000269|PubMed:19801981}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to development of chordomas is due to a T gene duplication.	EDTA Blood Sample
Sakral Agenezis-Vertebral Anomaliler, 615709, Otozomal Resesif; SAVA (Sakral Agenezisi-Vertebralarda Anormal Ossifikasyon-Kalıcı Notokord Kanal Sendromu) (Prenatal) (T geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sacral agenesis with vertebral anomalies, 615709, Autosomal recessive; SAVA (Sacral agenesis-abnormal ossification of the vertebral bodies-persistent notochordal canal syndrome) (T gene) (Sequence Analysis-All Coding Exons) (Prenatal)	T	T, TFT, SAVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Chordoma (CHDM) [MIM:215400]: Rare, clinically malignant tumors derived from notochordal remnants. They occur along the length of the spinal axis, predominantly in the sphenooccipital, vertebral and sacrococcygeal regions. They are characterized by slow growth, local destruction of bone, extension into adjacent soft tissues and rarely, distant metastatic spread. {ECO:0000269|PubMed:19801981}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Susceptibility to development of chordomas is due to a T gene duplication.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sakral Defekt-Anterior Meningosel (Sirenomeli) (VANGL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SACRAL DEFECT WITH ANTERIOR MENINGOCELE (Sirenomelia) (VANGL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VANGL1	VANGL1, STBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sacral defect with anterior meningocele (SDAM) [MIM:600145]: Form of caudal dysgenesis. It is present at birth and becomes symptomatic later in life, usually because of obstructive labor in females, chronic constipation, or meningitis. Inheritance is autosomal dominant. {ECO:0000269|PubMed:17403244}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sakral Defekt-Anterior Meningosel (Sirenomeli) (Prenatal) (VANGL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SACRAL DEFECT WITH ANTERIOR MENINGOCELE (Sirenomelia) (VANGL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VANGL1	VANGL1, STBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sacral defect with anterior meningocele (SDAM) [MIM:600145]: Form of caudal dysgenesis. It is present at birth and becomes symptomatic later in life, usually because of obstructive labor in females, chronic constipation, or meningitis. Inheritance is autosomal dominant. {ECO:0000269|PubMed:17403244}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Saethre-Chotzen Sendromu- Göz Kapağı Anomalileri, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Saethre-Chotzen syndrome with eyelid anomalies, 101400, Autosomal dominant (Saethre-Chotzen syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Saethre-Chotzen Sendromu- Göz Kapağı Anomalileri, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (MLPA)	Saethre-Chotzen syndrome with eyelid anomalies, 101400, Autosomal dominant (Saethre-Chotzen syndrome) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Saethre-Chotzen Sendromu- Göz Kapağı Anomalileri, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (Prenatal) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Saethre-Chotzen syndrome with eyelid anomalies, 101400, Autosomal dominant (Saethre-Chotzen syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Saethre-Chotzen Sendromu- Göz Kapağı Anomalileri, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (Prenatal) (MLPA)	Saethre-Chotzen syndrome with eyelid anomalies, 101400, Autosomal dominant (Saethre-Chotzen syndrome) (Prenatal) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Saethre-Chotzen syndrome, 101400, autosomal dominant (Saethre-Chotzen syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant (Saethre-Chotzen Sendromu) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Saethre-Chotzen syndrome, 101400, autosomal dominant (Saethre-Chotzen syndrome) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant; SCS (Saethre-Chotzen Sendromu) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Saethre-Chotzen syndrome, 101400, Autosomal dominant; SCS (Saethre-Chotzen syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant; SCS (Saethre-Chotzen Sendromu) (MLPA)	Saethre-Chotzen syndrome, 101400, Autosomal dominant; SCS (Saethre-Chotzen syndrome) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant; SCS (Saethre-Chotzen Sendromu) (Prenatal) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Saethre-Chotzen syndrome, 101400, Autosomal dominant; SCS (Saethre-Chotzen syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Saethre-Chotzen Sendromu, 101400, Otozomal Dominant; SCS (Saethre-Chotzen Sendromu) (Prenatal) (MLPA)	Saethre-Chotzen syndrome, 101400, Autosomal dominant; SCS (Saethre-Chotzen syndrome) (Prenatal) (MLPA)	TWIST1	TWIST1, ACS3, SCS, CRS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269	PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Salla Hastalığı, 604369, Otozomal Resesif; SD (Serbest Sialik Asit Depo Hastalığı) (SLC17A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Salla disease, 604369, Autosomal recessive; SD (Free sialic acid storage disease) (SLC17A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A5	SLC17A5, SIASD, SLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile sialic acid storage disorder (ISSD) [MIM:269920]: Severe form of sialic acid storage disease. Affected newborns exhibit visceromegaly, coarse features and failure to thrive immediately after birth. These patients have a shortened life span, usually less than 2 years. {ECO:0000269|PubMed:10581036}. ECO:0000269|PubMed:10947946}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Salla Hastalığı, 604369, Otozomal Resesif; SD (Serbest Sialik Asit Depo Hastalığı) (Prenatal) (SLC17A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Salla disease, 604369, Autosomal recessive; SD (Free sialic acid storage disease) (SLC17A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC17A5	SLC17A5, SIASD, SLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile sialic acid storage disorder (ISSD) [MIM:269920]: Severe form of sialic acid storage disease. Affected newborns exhibit visceromegaly, coarse features and failure to thrive immediately after birth. These patients have a shortened life span, usually less than 2 years. {ECO:0000269|PubMed:10581036}. ECO:0000269|PubMed:10947946}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tuz ve Biber Gelişim Regresyonu Sendromu, 609056, Otozomal Resesif; SPDRS (Amish İnfantil Epilepsi Sendromu) (ST3GAL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Salt and pepper developmental regression syndrome, 609056, Autosomal recessive; SPDRS (Amish infantile epilepsy syndrome) (ST3GAL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ST3GAL5	SIAT9, ST3GALV, SPDRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amish infantile epilepsy syndrome (AIES) [MIM:609056]: An autosomal recessive, infantile-onset symptomatic epilepsy associated with developmental stagnation and blindness. {ECO:0000269	PubMed:15502825}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sandhoff Hastalığı, İnfantil, Jüvenil ve Erişkin Formlar, 268800, Otozomal Resesif (Sandhoff Hastalığı) (HEXB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sandhoff disease, infantile, juvenile, and adult forms, 268800, Autosomal recessive (Sandhoff disease) (HEXB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEXB	HEXB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM2-gangliosidosis 2 (GM2G2) [MIM:268800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. Clinically indistinguishable from GM2-gangliosidosis type 1, presenting startle reactions, early blindness, progressive motor and mental deterioration, macrocephaly and cherry-red spots on the macula. {ECO:0000269|PubMed:1531140, ECO:0000269|PubMed:1720305, ECO:0000269|PubMed:7557963, ECO:0000269|PubMed:7626071, ECO:0000269|PubMed:7633435, ECO:0000269|PubMed:8357844, ECO:0000269|PubMed:8950198, ECO:0000269|PubMed:9401004, ECO:0000269|PubMed:9694901, ECO:0000269|PubMed:9856491}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sandhoff Hastalığı, İnfantil, Jüvenil ve Erişkin Formlar, 268800, Otozomal Resesif (Sandhoff Hastalığı) (Prenatal) (HEXB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sandhoff disease, infantile, juvenile, and adult forms, 268800, Autosomal recessive (Sandhoff disease) (HEXB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HEXB	HEXB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis 2 (GM2G2) [MIM:268800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 gangliosides in the neuronal cells. Clinically indistinguishable from GM2-gangliosidosis type 1, presenting startle reactions, early blindness, progressive motor and mental deterioration, macrocephaly and cherry-red spots on the macula. {ECO:0000269|PubMed:1531140, ECO:0000269|PubMed:1720305, ECO:0000269|PubMed:7557963, ECO:0000269|PubMed:7626071, ECO:0000269|PubMed:7633435, ECO:0000269|PubMed:8357844, ECO:0000269|PubMed:8950198, ECO:0000269|PubMed:9401004, ECO:0000269|PubMed:9694901, ECO:0000269|PubMed:9856491}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sarkoidoz, Yatlenik 1, 181000, Sporadik Vakalar; SS1 (Sarkoidoz) (HLA-DRB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sarcoidosis, susceptibility to, 1, 181000, Isolated cases; SS1 (Sarcoidosis) (HLA-DRB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-DRB1	HLA-DRB1, SS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Sarkoidoz, Yatlenik, 2, 612387; SS2 (Sarkoidoz) (BTNL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sarcoidosis, susceptibility to, 2, 612387; SS2 (Sarcoidosis) (BTNL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BTNL2	BTNL2, SS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sarcoidosis 2 (SS2) [MIM:612387]: An idiopathic, systemic, inflammatory disease characterized by the formation of immune granulomas in involved organs. Granulomas predominantly invade the lungs and the lymphatic system, but also skin, liver, spleen, eyes and other organs may be involved. {ECO:0000269	PubMed:15735647}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. A nucleotide transition affecting a splice donor site results in the use of an alternative splice site and the production of isoform 3. Individuals expressing isoform 3 have a higher risk for sarcoidosis.;	EDTA Blood Sample

Sarkom, Sinoviyal (SS18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sarcoma, synovial (SS18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SS18	SS18, SSXT, SYT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sarkom, Sinoviyal, 300813 (Sinoviyal Sarkom) (SSX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sarcoma, synovial, 300813 (Synovial sarcoma) (SSX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SSX2	SSX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sarkom, Sinoviyal, 300813 (Sinoviyal Sarkom) (SSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sarcoma, synovial, 300813 (Synovial sarcoma) (SSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SSX1	SSX1, SSRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SARS Enfeksiyonu, Korunma, 605872 (CLEC4M geni) (Tüm Gen Dizi Analizi) (Postnatal)	SARS infection, protection against, 605872 (CLEC4M gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLEC4M	CLEC4M, CD209L, L-SIGN, DCSIGNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
SARS İlerlemesi (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	SARS, progression of (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SBBYSS Sendromu, 603736 (Blefarofimozis-Entelektüel Yetersizlik Sendromu, SBBYS Tipi (SBBYSS Sendromu)) (KAT6B geni) (Tüm Gen Dizi Analizi) (Postnatal)	SBBYSS syndrome, 603736 (Blepharophimosis-intellectual disability syndrome, SBBYS type) (KAT6B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KAT6B	KAT6B, MYST4, MORF, GTPTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ohdo syndrome, SBBYS variant (SBBYSS) [MIM:603736]: A syndrome characterized by distinctive facial appearance with severe blepharophimosis, an immobile mask-like face, a bulbous nasal tip, and a small mouth with a thin upper lip. The condition presents in infancy with severe hypotonia and feeding problems. Associated skeletal problems include joint laxity, abnormally long thumbs and great toes, and dislocated or hypoplastic patellae. Structural cardiac defects are present in around 50% of cases, and dental anomalies, including small and pointed teeth, are common. Optic atrophy and conductive or sensorineural deafness are repeatedly reported. Many affected individuals have abnormalities of thyroid structure or function. SBBYSS is usually associated with severe mental retardation, delayed motor milestones, and significantly impaired speech. {ECO:0000269	PubMed:22077973}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SBBYSS Sendromu, 603736 (Blefarofimozis-Entelektüel Yetersizlik Sendromu, SBBYS Tipi (SBBYSS Sendromu)) (Prenatal) (KAT6B geni) (Tüm Gen Dizi Analizi) (Prenatal)	SBBYSS syndrome, 603736 (Blepharophimosis-intellectual disability syndrome, SBBYS type) (KAT6B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KAT6B	KAT6B, MYST4, MORF, GTPTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ohdo syndrome, SBBYS variant (SBBYSS) [MIM:603736]: A syndrome characterized by distinctive facial appearance with severe blepharophimosis, an immobile mask-like face, a bulbous nasal tip, and a small mouth with a thin upper lip. The condition presents in infancy with severe hypotonia and feeding problems. Associated skeletal problems include joint laxity, abnormally long thumbs and great toes, and dislocated or hypoplastic patellae. Structural cardiac defects are present in around 50% of cases, and dental anomalies, including small and pointed teeth, are common. Optic atrophy and conductive or sensorineural deafness are repeatedly reported. Many affected individuals have abnormalities of thyroid structure or function. SBBYSS is usually associated with severe mental retardation, delayed motor milestones, and significantly impaired speech. {ECO:0000269	PubMed:22077973}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SC Fokomeli Sendromu, 269000, Otozomal Resesif (Roberts Sendromu (SC Fokomeli Sendromu)) (ESCO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SC phocomelia syndrome, 269000, Autosomal recessive (Roberts syndrome) (ESCO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ESCO2	ESCO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: SC phocomelia syndrome (SCPS) [MIM:269000]: Has a milder phenotype than RBS, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common. {ECO:0000269	PubMed:16380922}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
SC Fokomeli Sendromu, 269000, Otozomal Resesif (Roberts Sendromu (SC Fokomeli Sendromu)) (Prenatal) (ESCO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SC phocomelia syndrome, 269000, Autosomal recessive (Roberts syndrome) (ESCO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ESCO2	ESCO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: SC phocomelia syndrome (SCPS) [MIM:269000]: Has a milder phenotype than RBS, with a lesser degree of symmetric limb reduction and additionally includes flexion contractures of various joints, midfacial hemangioma, hypoplastic cartilage of ears and nose, scant silvery-blond hair, and cloudy corneae. Although microcephaly is present, mental retardation may be mild and survival into adulthood is common. {ECO:0000269	PubMed:16380922}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Skalp-Kulak-Meme Başı Sendromu, 181270, Otozomal Dominant; SENS (Skalp-Kulak-Meme Başı Sendromu) (KCTD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scalp-ear-nipple syndrome, 181270, Autosomal dominant; SENS (Scalp-ear-nipple syndrome) (KCTD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCTD1	KCTD1, SENS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scalp-ear-nipple syndrome (SENS) [MIM:181270]: A disease characterized by aplasia cutis congenita of the scalp, breast anomalies that range from hypothelia or athelia to amastia, and minor anomalies of the external ears. Less frequent clinical characteristics include nail dystrophy, dental anomalies, cutaneous syndactyly of the digits, and renal malformations. Penetrance appears to be high, although there is substantial variable expressivity within families. {ECO:0000269	PubMed:23541344}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Scafosefali ve Axenfeld-Rieger Anomalisi (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scaphocephaly and Axenfeld-Rieger anomaly (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Scafosefali ve Axenfeld-Rieger Anomalisi (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Scaphocephaly and Axenfeld-Rieger anomaly (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Skafosefali, Maksiller Retrüzyon ve Mental Retardasyon, 609579 (Aleksei Skafosefali Sendromu, Mcgillivray Tipi) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (Aleksei Skafosefali Sendromu, McGillivray type) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Skafosefali, Maksiller Retrüzyon ve Mental Retardasyon, 609579 (Aleksei Skafosefali Sendromu, Mcgillivray Tipi) (Prenatal) (FGFR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (Aleksei Skafosefali Sendromu, McGillivray type) (FGFR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR2	FGFR2, BEK, CFD1, JWS, TK14, BBDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Apert syndrome (APRS) [MIM:101200]: A syndrome characterized by facio-cranio-synostosis, osseous and membranous syndactyly of the four extremities, and midface hypoplasia. The craniosynostosis is bicoronal and results in acrocephaly or brachysphenocephalic type. Syndactyly of the fingers and toes may be total (mitten hands and sock feet) or partial affecting the second, third, and fourth digits. Intellectual deficit is frequent and often severe, usually being associated with cerebral malformations. {ECO:0000269|PubMed:11781872, ECO:0000269|PubMed:7668257, ECO:0000269|PubMed:7719344, ECO:0000269|PubMed:9002682, ECO:0000269|PubMed:9452027, ECO:0000269|PubMed:9677057}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Skapuloperoneal Miyopati, X'e Bağlı Dominant, 300695, X'e Bağlı Dominant; SPM (X'e Bağlı Skapuloperoneal Müsküler Distrofi) (FHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scapuloperoneal myopathy, X-linked dominant, 300695, X-linked dominant; SPM (X-linked scapuloperoneal muscular dystrophy) (FHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FHL1	FHL1, SLIM1, XMPMA, KYOT, FHL1A, FHL1B, RBMX1A, RBMX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Scapuloperoneal myopathy, X-linked dominant (SPM) [MIM:300695]: A disease characterized by progressive muscle weakness and wasting, upper and lower limbs weakness, foot drop, scapular winging, and myopathic changes on muscle biopsy. Most affected individuals become wheelchair-bound. {ECO:0000269|PubMed:18179901}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Skapuloperoneal Spinal Müsküler Atrofi, 18; SPSMA (Skapuloperoneal spinal müsküler atrofi) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scapuloperoneal spinal muscular atrophy, 18; SPSMA (Scapuloperoneal spinal muscular atrophy) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, SCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269|PubMed:18587394}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Skapuloperoneal Sendromu, Miyopatik Türü, 181430, Otozomal Dominant; SPMM (MYH7 İlişkili Geç Başlangıçlı Skapuloperoneal Müsküler Distrofi) (MYH7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scapuloperoneal syndrome, myopathic type, 181430, Autosomal dominant; SPMM (MYH7-related late-onset scapuloperoneal muscular dystrophy) (MYH7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH7	MYH7, CMH1, MPD1, CMD1S, SPMM, SPMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, distal, 1 (MPD1) [MIM:160500]: A muscular disorder characterized by early-onset selective weakness of the great toe and ankle dorsiflexors, followed by weakness of the finger extensors. Mild proximal weakness occasionally develops years later after the onset of the disease. {ECO:0000269|PubMed:15322983, ECO:0000269|PubMed:17548557}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Skapuloperoneal Sendromu, Nörojenik, Kaeser Tipi, 181400, Otozomal Dominant, SCPNK (Skapuloperoneal Sendrom, Nörojenik, Kaeser Tipi) (DES geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scapuloperoneal syndrome, neurogenic, Kaeser type, 181400, Autosomal dominant; SCPNK (Neurogenic scapuloperoneal syndrome, Kaeser type) (DES gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DES	DES, CMD1I, MFM1, SCPNK, ARVD7, ARVC7, LGMD2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurogenic scapuloperoneal syndrome Kaeser type (Kaeser syndrome) [MIM:181400]: Autosomal dominant disorder with a peculiar scapuloperoneal distribution of weakness and atrophy. A large clinical variability is observed ranging from scapuloperoneal, limb girdle and distal phenotypes with variable cardiac or respiratory involvement. Facial weakness, dysphagia and gynaecomastia are frequent additional symptoms. Affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Histological and immunohistochemical examination of muscle biopsy specimens reveal a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin. {ECO:0000269|PubMed:17439987, ECO:0000269|PubMed:25394388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schaaf Yang Sendromu, 615547, Otozomal Dominant; SHFYNG (Prader-Willi sendromu, nokta mutasyonu ile) (MAGEL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schaaf-Yang syndrome, 615547, Autosomal dominant; SHFYNG (Prader-Willi syndrome due to point mutation) (MAGEL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAGEL2	MAGEL2, NDNL1, SHFYNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Schaaf Yang Sendromu, 615547, Otozomal Dominant; SHFYNG (Prader-Willi sendromu, nokta mutasyonu ile) (MLPA)	Schaaf-Yang syndrome, 615547, Autosomal dominant; SHFYNG (Prader-Willi syndrome due to point mutation) (MLPA)	MAGEL2	MAGEL2, NDNL1, SHFYNG	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Schaaf Yang Sendromu, 615547, Otozomal Dominant; SHFYNG (Prader-Willi sendromu, nokta mutasyonu ile) (Prenatal) (MAGEL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schaaf-Yang syndrome, 615547, Autosomal dominant; SHFYNG (Prader-Willi syndrome due to point mutation) (MAGEL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAGEL2	MAGEL2, NDNL1, SHFYNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schaaf Yang Sendromu, 615547, Otozomal Dominant; SHFYNG (Prader-Willi sendromu, nokta mutasyonu ile) (Prenatal) (MLPA)	Schaaf-Yang syndrome, 615547, Autosomal dominant; SHFYNG (Prader-Willi syndrome due to point mutation) (Prenatal) (MLPA)	MAGEL2	MAGEL2, NDNL1, SHFYNG	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schimke İmmünoosseöz Displazi, 242900, Otozomal Resesif; SIOD (Schimke İmmüno-Osseöz Displazi) (SMARCAL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schimke immunoosseous dysplasia, 242900, Autosomal recessive; SIOD (Schimke immuno-osseous dysplasia) (SMARCAL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCAL1	SMARCAL1, HARP, SIOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schimke immuno-osseous dysplasia (SIOD) [MIM:242900]: Causes spondyloepiphyseal dysplasia, renal dysfunction and T-cell immunodeficiency. Approximately half of all patients also exhibit hyperthyroidism, while around half also exhibit episodal cerebral ischemia. {ECO:0000269	PubMed:11799392}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schimke İmmünoosseöz Displazi, 242900, Otozomal Resesif; SIOD (Schimke İmmüno-Osseöz Displazi) (Prenatal) (SMARCAL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schimke immunoosseous dysplasia, 242900, Autosomal recessive; SIOD (Schimke immuno-osseous dysplasia) (SMARCAL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMARCAL1	SMARCAL1, HARP, SIOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schimke immuno-osseous dysplasia (SIOD) [MIM:242900]: Causes spondyloepiphyseal dysplasia, renal dysfunction and T-cell immunodeficiency. Approximately half of all patients also exhibit hyperthyroidism, while around half also exhibit episodal cerebral ischemia. {ECO:0000269	PubMed:11799392}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schimmelpenning-Feuerstein-Mims Sendromu, Somatik Mozaik, 163200 (Lineer Sebase Nevus Sendromu) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (Linear nevus sebaceus syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Schimmelpenning-Feuerstein-Mims Sendromu, Somatik Mozaik, 163200 (Lineer Sebase Nevus Sendromu) (Prenatal) (HRAS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200 (Linear nevus sebaceus syndrome) (HRAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bladder cancer (BLC) [MIM:109800]: A malignancy originating in tissues of the urinary bladder. It often presents with multiple tumors appearing at different times and at different sites in the bladder. Most bladder cancers are transitional cell carcinomas that begin in cells that normally make up the inner lining of the bladder. Other types of bladder cancer include squamous cell carcinoma (cancer that begins in thin, flat cells) and adenocarcinoma (cancer that begins in cells that make and release mucus and other fluids). Bladder cancer is a complex disorder with both genetic and environmental influences. {ECO:0000269|PubMed:6298635, ECO:0000269|PubMed:6844927}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schimmelpenning-Feuerstein-Mims Syndrome, Somatic Mosaic, 163200, Somatic Mosaic (Linear Nevus Sebaceus Syndrome) (KRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200, somatic mosaic (Linear nevus sebaceus syndrome) (KRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schimmelpenning-Feuerstein-Mims Syndrome, Somatic Mosaic, 163200, Somatic Mosaic (Linear Nevus Sebaceus Syndrome) (MLPA)	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200, somatic mosaic (Linear nevus sebaceus syndrome) (MLPA)	KRAS	KRAS, KRAS2, RASK2, NS, CFC2, RALD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schimmelpenning-Feuerstein-Mims Sendromu, Somatik Mozaik, 163200; SFM (Lineer Sebase Nevus Sendromu) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200; SFM (Linear nevus sebaceus syndrome) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Schindler Hastalığı, Tip 1, 609241, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (NAGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schindler disease, type I, 609241, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schindler disease (SCHIND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schindler Hastalığı, Tip 1, 609241, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (Prenatal) (NAGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schindler disease, type I, 609241, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schindler disease (SCHIND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schindler Hastalığı, Tip 3, 609241, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (NAGA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schindler disease, type III, 609241, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schindler disease (SCHIND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schindler Hastalığı, Tip 3, 609241, Otozomal Resesif (Alfa-N-Asetil Galaktosaminidaz Eksikliği) (Prenatal) (NAGA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schindler disease, type III, 609241, Autosomal recessive (Alpha-N-acetylgalactosaminidase deficiency) (NAGA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NAGA	NAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Schindler disease (SCHIND) [MIM:609241]: Form of NAGA deficiency characterized by early-onset neuroaxonal dystrophy and neurological signs (convulsion during fever, epilepsy, psychomotor retardation and hypotonia). NAGA deficiency is typically classified in three main phenotypes: NAGA deficiency type I (Schindler disease or Schindler disease type I) with severe manifestations; NAGA deficiency type II (Kanzaxi disease or Schindler disease type II) which is mild; NAGA deficiency type III (Schindler disease type III) characterized by mild-to-moderate neurologic manifestations. NAGA deficiency results in the increased urinary excretion of glycopeptides and oligosaccharides containing alpha-N-acetylgalactosaminyl moieties. Inheritance is autosomal recessive. {ECO:0000269|PubMed:2243144, ECO:0000269|PubMed:8782044}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schinzel-Giedion Orta Yüz Retraksiyon Sendromu, 269150, Otozomal Dominant (Schinzel-Giedion Sendromu) (SETBP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schinzel-Giedion midface retraction syndrome, 269150, Autosomal dominant (Schinzel-Giedion syndrome) (SETBP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETBP1	SETBP1, KIAA0437, SEB, MRD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:23689083}. Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample

Schinzel-Giedion Orta Yüz Retraksiyon Sendromu, 269150, Otozomal Dominant (Schinzel-Giedion Sendromu) (Prenatal) (SETBP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schinzel-Giedion midface retraction syndrome, 269150, Autosomal dominant (Schinzel-Giedion syndrome) (SETBP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SETBP1	SETBP1, KIAA0437, SEB, MRD29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269|PubMed:23648668, ECO:0000269|PubMed:23889083). Note=The gene represented in this entry is involved in disease pathogenesis.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (SIX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizencephaly, 269160 (Schizencephaly) (SIX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIX3	SIX3, HPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. (ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (SHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizencephaly, 269160 (Schizencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (EMX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizencephaly, 269160 (Schizencephaly) (EMX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EMX2	EMX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (MLPA)	Schizencephaly, 269160 (Schizencephaly) (MLPA)	SIX3	SIX3, HPE2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. (ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (MLPA)	Schizencephaly, 269160 (Schizencephaly) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (Prenatal) (SIX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schizencephaly, 269160 (Schizencephaly) (SIX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIX3	SIX3, HPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. (ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (Prenatal) (SHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schizencephaly, 269160 (Schizencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (Prenatal) (EMX2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schizencephaly, 269160 (Schizencephaly) (EMX2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EMX2	EMX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (Prenatal) (MLPA)	Schizencephaly, 269160 (Schizencephaly) (Prenatal) (MLPA)	SIX3	SIX3, HPE2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Holoprosencephaly 2 (HPE2) [MIM:157170]: A structural anomaly of the brain, in which the developing forebrain fails to correctly separate into right and left hemispheres. Holoprosencephaly is genetically heterogeneous and associated with several distinct facies and phenotypic variability. (ECO:0000269|PubMed:10369266, ECO:0000269|PubMed:15221788, ECO:0000269|PubMed:15523651, ECO:0000269|PubMed:17001667, ECO:0000269|PubMed:18791198, ECO:0000269|PubMed:20531442). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizensefali, 269160 (Şizensefali) (Prenatal) (MLPA)	Schizencephaly, 269160 (Schizencephaly) (Prenatal) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şizoaffektif Bozukluk, Yatkınlık, 181500, Otozomal Dominant (DISC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizoaffective disorder, susceptibility to, 181500, Autosomal dominant (DISC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DISC1	DISC1, SCZD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schizophrenia 9 (SCZD9) [MIM:604906]: A complex, multifactorial psychotic disorder or group of disorders characterized by disturbances in the form and content of thought (e.g. delusions, hallucinations), in mood (e.g. inappropriate affect), in sense of self and relationship to the external world (e.g. loss of ego boundaries, withdrawal), and in behavior (e.g. bizarre or apparently purposeless behavior). Although it affects emotions, it is distinguished from mood disorders in which such disturbances are primary. Similarly, there may be mild impairment of cognitive function, and it is distinguished from the dementias in which disturbed cognitive function is considered primary. Some patients manifest schizophrenic as well as bipolar disorder symptoms and are often given the diagnosis of schizoaffective disorder. (ECO:0000269|PubMed:11468279, ECO:0000269|PubMed:14532331, ECO:0000269|PubMed:15939883). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Şizofreni 15, 613950 (Monozomi 22q13; SCZD15 (Phelan-Mcdermid Sendromu)) (2200)	Schizophrenia 15, 613950; SCZD15 (Monosomy 22q13) (440)	SHANK3	SHANK3, PSAP2, PROSAP2, KIAA1650, DEL22q13.3, SCZD15	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	DISEASE: Phelan-McDermid syndrome (PHMDS) [MIM:606232]: A developmental disorder with variable features. Common features include neonatal hypotonia, global developmental delay, normal to accelerated growth, absent to severely delayed speech, autistic behavior, and minor dysmorphic features. (ECO:0000269|PubMed:22892527, ECO:0000269|PubMed:23758760, ECO:0000269|PubMed:24132240). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample and Heparinized blood sample
Şizofreni Yatkınlık 18, 615232 (SLC1A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia susceptibility 18, 615232 (SLC1A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC1A1	SLC1A1, EAAC1, SCZD18, DCBXA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dicarboxylic aminoaciduria (DCBXA) [MIM:222730]: An autosomal recessive disorder characterized by abnormal excretion of urinary glutamate and aspartate, resulting from the incomplete reabsorption of anionic amino acids from the glomerular filtrate in the kidney. It can be associated with mental retardation. (ECO:0000269	PubMed:21123949). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Şizofreni, 181500, Otozomal Dominant (DAOA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, 181500, Autosomal dominant (DAOA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAOA	DAOA, G72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, 181500, Otozomal Dominant (DAO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, 181500, Autosomal dominant (DAO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAO	DAO, DAMOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, 181500, Otozomal Dominant (APOL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, 181500, Autosomal dominant (APOL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOL4	APOL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, 181500, Otozomal Dominant (APOL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, 181500, Autosomal dominant (APOL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOL2	APOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Nörofizyolojik Defekt, 118511 (CHRNA7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, neurophysiologic defect in, 118511 (CHRNA7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHRNA7	CHRNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Yatkınlık, 17, 614332 (Pitt-Hopkins Benzeri Sendrom) (NRXN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 17, 614332 (Pitt-Hopkins-like syndrome) (NRXN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRXN1	NRXN1, PTHSL2, SCZD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Şizofreni, Yatkınlık, 181500, Otozomal Dominant (SYN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (SYN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYN2	SYN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (RTN4R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (RTN4R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTN4R	RTN4R, NOGOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (MTHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (MTHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (DRD3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (DRD3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DRD3	DRD3, ETM1, FET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tremor, hereditary essential 1 (ETM1) [MIM:190300]: A common movement disorder mainly characterized by postural tremor of the arms. Head, legs, trunk, voice, jaw, and facial muscles also may be involved. The condition can be aggravated by emotions, hunger, fatigue and temperature extremes, and may cause a functional disability or even incapacitation. Inheritance is autosomal dominant. {ECO:0000269|PubMed:16650084, ECO:0000269|PubMed:16809426}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (COMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (COMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COMT	COMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (CHI3L1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (CHI3L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHI3L1	CHI3L1, GP39, YKL40, ASRT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Asthma-related traits 7 (ASRT7) [MIM:611960]: Asthma- related traits include clinical symptoms of asthma, such as coughing, wheezing, dyspnea, bronchial hyperresponsiveness as assessed by methacholine challenge test, serum IgE levels, atopy and atopic dermatitis. {ECO:0000269	PubMed:18403759}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Şizofreni, Yatkınlık, 181500, Otozomal Dominant (AKT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 181500, Autosomal dominant (AKT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AKT1	AKT1, CWS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Breast cancer (BC) [MIM:114480]: A common malignancy originating from breast epithelial tissue. Breast neoplasms can be distinguished by their histologic pattern. Invasive ductal carcinoma is by far the most common type. Breast cancer is etiologically and genetically heterogeneous. Important genetic factors have been indicated by familial occurrence and bilateral involvement. Mutations at more than one locus can be involved in different families or even in the same case. {ECO:0000269	PubMed:17611497}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Şizofreni, Yatkınlık, 4, 600850, Otozomal Dominant (Hiperprolinemi Tip 1) (PRODH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 4, 600850, Autosomal dominant (Hyperprolinemia type 1) (PRODH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRODH	PRODH, PRODH2, SCZD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperprolinemia 1 (HYRPRO1) [MIM:239500]: An inborn error of proline metabolism resulting in elevated levels of proline in the plasma and urine. The disorder is generally benign and most affected individuals are clinically asymptomatic. Some patients, however, have neurologic manifestations, including epilepsy and mental retardation. Association with certain forms of schizophrenia have been reported. {ECO:0000269|PubMed:12217952, ECO:0000269|PubMed:15662599, ECO:0000269|PubMed:17135275}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Şizofreni, Yatkınlık, 603013 (NRG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 603013 (NRG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRG1	NRG1, HGL, HRGA, ARIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving NRG1 produces gamma-heregulin. Translocation t(8;11) with TENM4. The translocation fuses the 5'-end of TENM4 to NRG1 (isoform 8). The product of this translocation was first thought to be an alternatively spliced isoform. Gamma-heregulin is a soluble activating ligand for the ERBB2-ERBB3 receptor complex and acts as an autocrine growth factor in a specific breast cancer cell line (MDA-MB-175). Not detected in breast carcinoma samples, including ductal, lobular, medullary, and mucinous histological types, neither in other breast cancer cell lines.;	EDTA Blood Sample
Şizofreni, Yatkınlık, 604906 (DISC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schizophrenia, susceptibility to, 604906 (DISC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DISC1	DISC1, SCZD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schizophrenia 9 (SCZD9) [MIM:604906]: A complex, multifactorial psychotic disorder or group of disorders characterized by disturbances in the form and content of thought (e.g. delusions, hallucinations), in mood (e.g. inappropriate affect), in sense of self and relationship to the external world (e.g. loss of ego boundaries, withdrawal), and in behavior (e.g. bizarre or apparently purposeless behavior). Although it affects emotions, it is distinguished from mood disorders in which such disturbances are primary. Similarly, there may be mild impairment of cognitive function, and it is distinguished from the dementias in which disturbed cognitive function is considered primary. Some patients manifest schizophrenic as well as bipolar disorder symptoms and are often given the diagnosis of schizoaffective disorder. {ECO:0000269|PubMed:11468279, ECO:0000269|PubMed:14532331, ECO:0000269|PubMed:15939883}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Schneckenbecken Displazi, 269250, Otozomal Resesif (Schneckenbecken Displazi) (SLC35D1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schneckenbecken dysplasia, 269250, Autosomal recessive (Schneckenbecken dysplasia) (SLC35D1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC35D1	SLC35D1, UGTREL7, KIAA0260	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Schneckenbecken Displazi, 269250, Otozomal Resesif (Schneckenbecken Displazi) (Prenatal) (SLC35D1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schneckenbecken dysplasia, 269250, Autosomal recessive (Schneckenbecken dysplasia) (SLC35D1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC35D1	SLC35D1, UGTREL7, KIAA0260	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schopf-Schulz-Passarge Sendromu, 224750, Otozomal Resesif; SSPS (Schopf-Schulz-Passarge Sendromu) (WNT10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schopf-Schulz-Passarge syndrome, 224750, Autosomal recessive; SSPS (Schopf-Schulz-Passarge syndrome) (WNT10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT10A	WNT10A, SSPS, STHAG4, OODD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 4 (STHAG4) [MIM:150400]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). In STHAG4, the upper lateral incisors are absent or peg-shaped. {ECO:0000269|PubMed:21484994, ECO:0000269|PubMed:22581971}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Schopf-Schulz-Passarge Sendromu, 224750, Otozomal Resesif; SSPS (Schopf-Schulz-Passarge Sendromu) (Prenatal) (WNT10A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schopf-Schulz-Passarge syndrome, 224750, Autosomal recessive; SSPS (Schopf-Schulz-Passarge syndrome) (WNT10A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT10A	WNT10A, SSPS, STHAG4, OODD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tooth agenesis selective 4 (STHAG4) [MIM:150400]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). In STHAG4, the upper lateral incisors are absent or peg-shaped. {ECO:0000269|PubMed:21484994, ECO:0000269|PubMed:22581971}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schuurs-Hoeijmakers Sendromu, 615009, Otozomal Dominant; SHMS (Mental Retardasyon-Kraniofasiyal Dismorfizm-Kriptorşidizm Sendromu) (PACS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schuurs-Hoeijmakers syndrome, 615009, Autosomal dominant; SHMS (Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome) (PACS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PACS1	PACS1, SHMS, MRD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mental retardation, autosomal dominant 17 (MRD17) [MIM:615009]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD17 patients have intellectual disability in combination with distinct craniofacial features and genital abnormalities. {ECO:0000269	PubMed:23159249}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Schuurs-Hoeijmakers Sendromu, 615009, Otozomal Dominant; SHMS (Mental Retardasyon-Kraniofasiyal Dismorfizm-Kriptorşidizm Sendromu) (Prenatal) (PACS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schuurs-Hoeijmakers syndrome, 615009, Autosomal dominant; SHMS (Intellectual disability-craniofacial dysmorphism-cryptorchidism syndrome) (PACS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PACS1	PACS1, SHMS, MRD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mental retardation, autosomal dominant 17 (MRD17) [MIM:615009]: A disorder characterized by significantly below average general intellectual functioning associated with impairments in adaptive behavior and manifested during the developmental period. MRD17 patients have intellectual disability in combination with distinct craniofacial features and genital abnormalities. {ECO:0000269	PubMed:23159249}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Schwannomatosis-1, Yatkınlık, 162091, Otozomal Dominant (Nörofibromatoz Tip 3) (SMARCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schwannomatosis-1, susceptibility to, 162091, Autosomal dominant (Neurofibromatosis type 3) (SMARCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMARCB1	SMARCB1, SNF5, INI1, RDT, RTPS1, MRD15, SWNTS1, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schwannomatosis 1 (SWNTS1) [MIM:162091]: A cancer syndrome in which patients develop multiple non-vestibular schwannomas, benign neoplasms that arise from Schwann cells of the cranial, peripheral, and autonomic nerves. {ECO:0000269|PubMed:17357086, ECO:0000269|PubMed:18072270}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Schwannomatosis-2, Yatkınlık, 615670, Otozomal Dominant; SWNTS2 (Nörofibromatoz Tip 5) (LZTR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schwannomatosis-2, susceptibility to, 615670, Autosomal dominant; SWNTS2 (Neurofibromatosis type 3) (LZTR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LZTR1	LZTR1, SWNTS2, NS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Schwannomatosis 2 (SWNTS2) [MIM:615670]: A cancer predisposition syndrome in which patients develop multiple non- vestibular schwannomas, benign neoplasms that arise from Schwann cells of the cranial, peripheral, and autonomic nerves. {ECO:0000269	PubMed:24362817}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample

Schwannomatosis, 162091, Otozomal Dominant; SWNTS1 (Nörofibromatoz Tip 3) (NF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schwannomatosis, 162091, Autosomal dominant; SWNTS1 (Neurofibromatosis type 3) (NF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF2	NF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schwannomatosis, 162091, Otozomal Dominant; SWNTS1 (Nörofibromatoz Tip 3) (MLPA)	Schwannomatosis, 162091, Autosomal dominant; SWNTS1 (Neurofibromatosis type 3) (MLPA)	NF2	NF2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Neurofibromatosis 2 (NF2) [MIM:101000]: Genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. {ECO:0000269|PubMed:10090912, ECO:0000269|PubMed:10790209, ECO:0000269|PubMed:12709270, ECO:0000269|PubMed:20445339, ECO:0000269|PubMed:7666400, ECO:0000269|PubMed:7759081, ECO:0000269|PubMed:7913580, ECO:0000269|PubMed:8081368, ECO:0000269|PubMed:8230593, ECO:0000269|PubMed:8566958, ECO:0000269|PubMed:8698340, ECO:0000269|PubMed:9643284}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Schwartz-Jampel Sendromu, Tip 1, 255800, Otozomal Resesif; SJS1 (Schwartz-Jampel Sendromu Tip 1) (HSPG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Schwartz-Jampel syndrome, type 1, 255800, Autosomal recessive; SJS1 (Schwartz-Jampel syndrome) (HSPG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPG2	HSPG2, PLC, SJS, SJA, SJS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyssegmental dysplasia Silverman-Handmaker type (DDSH) [MIM:224410]: The dyssegmental dysplasias are rare, autosomal recessive skeletal dysplasias with anisospondyly and micromelia. There are two recognized types: the severe, lethal DDSH and the milder Rolland-Desbuquois form. Individuals with DDSH also have a flat face, micrognathia, cleft palate and reduced joint mobility, and frequently have an encephalocoele. The endochondral growth plate is short, the calcospherites (which are spherical calcium- phosphorus crystals produced by hypertrophic chondrocytes) are unfused, and there is mucoid degeneration of the resting cartilage. {ECO:0000269|PubMed:11279527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Schwartz-Jampel Sendromu, Tip 1, 255800, Otozomal Resesif; SJS1 (Schwartz-Jampel Sendromu Tip 1) (Prenatal) (HSPG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Schwartz-Jampel syndrome, type 1, 255800, Autosomal recessive; SJS1 (Schwartz-Jampel syndrome) (HSPG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HSPG2	HSPG2, PLC, SJS, SJA, SJS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyssegmental dysplasia Silverman-Handmaker type (DDSH) [MIM:224410]: The dyssegmental dysplasias are rare, autosomal recessive skeletal dysplasias with anisospondyly and micromelia. There are two recognized types: the severe, lethal DDSH and the milder Rolland-Desbuquois form. Individuals with DDSH also have a flat face, micrognathia, cleft palate and reduced joint mobility, and frequently have an encephalocoele. The endochondral growth plate is short, the calcospherites (which are spherical calcium- phosphorus crystals produced by hypertrophic chondrocytes) are unfused, and there is mucoid degeneration of the resting cartilage. {ECO:0000269|PubMed:11279527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SCID, Otozomal Resesif, T-Negatif / B-Pozitif Tip, 600802, Otozomal Resesif (T-B + Ağır Kombine Immun Yetmezlik, JAK3 Eksikliğine Bağlı) (JAK3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SCID, autosomal recessive, T-negative/B-positive type, 600802, Autosomal recessive (T-B+ severe combined immunodeficiency due to JAK3 deficiency) (JAK3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAK3	JAK3, JAKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell positive/NK-cell-negative (T-)B(+)NK(-) SCID) [MIM:600802]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269|PubMed:10982185, ECO:0000269|PubMed:14615376, ECO:0000269|PubMed:7659163, ECO:0000269|PubMed:9354668, ECO:0000269|PubMed:9753072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
SCID, Otozomal Resesif, T-Negatif / B-Pozitif Tip, 600802, Otozomal Resesif (T-B + Ağır Kombine Immun Yetmezlik, JAK3 Eksikliğine Bağlı) (Prenatal) (JAK3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SCID, autosomal recessive, T-negative/B-positive type, 600802, Autosomal recessive (T-B+ severe combined immunodeficiency due to JAK3 deficiency) (JAK3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JAK3	JAK3, JAKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-positive/NK-cell-negative (T-)B(+)NK(-) SCID) [MIM:600802]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269|PubMed:10982185, ECO:0000269|PubMed:14615376, ECO:0000269|PubMed:7659163, ECO:0000269|PubMed:9354668, ECO:0000269|PubMed:9753072}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sklerosteozis 1, 269500, Otozomal Resesif; SOST1 (Sklerosteozis) (SOST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sclerosteosis 1, 269500, Autosomal recessive; SOST1 (Sclerosteosis) (SOST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of cranial facial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269|PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	EDTA Blood Sample
Sklerosteozis 2, 614305, Otozomal Resesif, Otozomal Dominant; SOST2 (Sklerosteozis) (LRP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sclerosteosis 2, 614305, Autosomal recessive, Autosomal dominant; SOST2 (Sclerosteosis) (LRP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP4	LRP4, MEGF7, CLSS, SOST2, CMS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sclerosteosis 2 (SOST2) [MIM:614305]: A sclerosing bone dysplasia characterized by a generalized hyperostosis and sclerosis leading to a markedly thickened skull, with mandible, ribs, clavicles and all long bones also being affected. Due to narrowing of the foramina of the cranial nerves, facial nerve palsy, hearing loss and atrophy of the optic nerves can occur. Sclerosteosis is clinically and radiologically very similar to van Buchem disease, mainly differentiated by hand malformations and a large stature in sclerosteosis patients. {ECO:0000269|PubMed:21471202, ECO:0000269|PubMed:24234652}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Scott Sendromu, 262890, Otozomal Resesif; SCTS (Scott Sendromu) (ANO6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Scott syndrome, 262890, Autosomal recessive; SCTS (Scott syndrome) (ANO6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO6	ANO6, TMEM16F, SCTS, BDPLT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Scott Sendromu, 262890, Otozomal Resesif; SCTS (Scott Sendromu) (Prenatal) (ANO6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Scott syndrome, 262890, Autosomal recessive; SCTS (Scott syndrome) (ANO6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ANO6	ANO6, TMEM16F, SCTS, BDPLT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Deniz Mavisi Histiosit Hastalığı, 269600, Otozomal Resesif (Deniz Mavisi Histiositoz) (APOE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sea-blue histiocyte disease, 269600, Autosomal recessive (Sea-blue histiocytosis) (APOE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APOE	APOE, AD2, LPG, LDLCQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alzheimer disease 2 (AD2) [MIM:104310]: A late-onset neurodegenerative disorder characterized by progressive dementia, loss of cognitive abilities, and deposition of fibrillar amyloid proteins as intraneuronal neurofibrillary tangles, extracellular amyloid plaques and vascular amyloid deposits. The major constituent of these plaques is the neurotoxic amyloid-beta-APP 40-42 peptide (s), derived proteolytically from the transmembrane precursor protein APP by sequential secretase processing. The cytotoxic C-terminal fragments (CTFs) and the caspase-cleaved products such as C31 derived from APP, are also implicated in neuronal death. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. The APOE*4 allele is genetically associated with the common late onset familial and sporadic forms of Alzheimer disease. Risk for AD increased from 20% to 90% and mean age at onset decreased from 84 to 68 years with increasing number of APOE*4 alleles in 42 families with late onset AD. Thus APOE*4 gene dose is a major risk factor for late onset AD and, in these families, homozygosity for APOE*4 was virtually sufficient to cause AD by age 80. The mechanism by which APOE*4 participates in pathogenesis is not known.	EDTA Blood Sample
Mevsimsel Afektif Bozukluk, Yatkınlık, 608516 (HTR2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seasonal affective disorder, susceptibility to, 608516 (HTR2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mevsimsel Afektif Bozukluk, Yatkınlık, 608516 (Prenatal) (HTR2A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seasonal affective disorder, susceptibility to, 608516 (HTR2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HTR2A	HTR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sebase Tümörler, Somatik (LEF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sebaceous tumors, somatic (LEF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LEF1	LEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media

Sebastian Sendromu, 605249, Otozomal Dominant; SBS (Sebastian Sendromu) (MYH9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sebastian syndrome, 605249, Autosomal dominant; SBS (Sebastian syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sebastian Sendromu, 605249, Otozomal Dominant; SBS (Sebastian Sendromu) (Prenatal) (MYH9 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sebastian syndrome, 605249, Autosomal dominant; SBS (Sebastian syndrome) (MYH9 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MYH9	MYH9, MHA, FTNS, DFNA17, BDPLT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fechtner syndrome (FTNS) [MIM:153640]: Autosomal dominant macrothrombocytopenia characterized by thrombocytopenia, giant platelets and leukocyte inclusions that are small and poorly organized. Additionally, FTNS is distinguished by Alport-like clinical features of sensorineural deafness, cataracts and nephritis. {ECO:0000269|PubMed:10973259, ECO:0000269|PubMed:11776386, ECO:0000269|PubMed:12533692, ECO:0000269|PubMed:12792306}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sebore Benzeri Dermatit, Psoriasiform Elementler, 610227 (Sebore-Benzeri Dermatit-Psöriazis Benzeri Elementler) (ZNF750 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seborrhea-like dermatitis with psoriasiform elements, 610227 (Seborrhea-like dermatitis with psoriasiform elements) (ZNF750 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZNF750	ZNF750, FLJ13841	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seborrhea-like dermatitis with psoriasiform elements (SLDP) [MIM:610227]: Characterized by a chronic fine diffuse scaly erythematous rash on the face, particularly on the chin, nasolabial folds and eyebrows, around earlobes and over the scalp. The rash exacerbate in the winter, with emotional stress and after strenuous physical activity. Hyperkeratosis of skin over the elbows, knees, palms, soles and metacarpophalangeal joints is evident. There is no arthralgia, arthritis or neurological disorders. {ECO:0000269|PubMed:16751772}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seckel Sendromu 1, 210600, Otozomal Resesif; SCKL1 (Seckel Sendromu) (ATR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 1, 210600, Autosomal recessive; SCKL1 (Seckel syndrome) (ATR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATR	ATR, FRP1, SCKL1, FCTCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seckel syndrome 1 (SCKL1) [MIM:210600]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:12640452}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seckel Sendromu 1, 210600, Otozomal Resesif; SCKL1 (Seckel Sendromu) (MLPA)	Seckel syndrome 1, 210600, Autosomal recessive; SCKL1 (Seckel syndrome) (MLPA)	ATR	ATR, FRP1, SCKL1, FCTCS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Seckel syndrome 1 (SCKL1) [MIM:210600]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:12640452}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seckel Sendromu 1, 210600, Otozomal Resesif; SCKL1 (Seckel Sendromu) (Prenatal) (ATR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 1, 210600, Autosomal recessive; SCKL1 (Seckel syndrome) (ATR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATR	ATR, FRP1, SCKL1, FCTCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 1 (SCKL1) [MIM:210600]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:12640452}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 1, 210600, Otozomal Resesif; SCKL1 (Seckel Sendromu) (Prenatal) (MLPA)	Seckel syndrome 1, 210600, Autosomal recessive; SCKL1 (Seckel syndrome) (Prenatal) (MLPA)	ATR	ATR, FRP1, SCKL1, FCTCS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 1 (SCKL1) [MIM:210600]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:12640452}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 10, 617253, Otozomal Resesif; SCKL10 (Mikrosefalik Primordial Dwarfizm-İnsülin Direnci Sendromu) (NSMCE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 10, 617253, Autosomal recessive; SCKL10 (Microcephalic primordial dwarfism-insulin resistance syndrome) (NSMCE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSMCE2	NSMCE2, NSE2, MMS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seckel Sendromu 10, 617253, Otozomal Resesif; SCKL10 (Mikrosefalik Primordial Dwarfizm-İnsülin Direnci Sendromu) (Prenatal) (NSMCE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 10, 617253, Autosomal recessive; SCKL10 (Microcephalic primordial dwarfism-insulin resistance syndrome) (NSMCE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSMCE2	NSMCE2, NSE2, MMS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 2, 606744, Otozomal Resesif; SCKL2 (Seckel Sendromu) (RBBP8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 2, 606744, Autosomal recessive; SCKL2 (Seckel syndrome) (RBBP8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBBP8	RBBP8, RIM, SCKL2, JWDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Jawad syndrome (JWDS) [MIM:251255]: A syndrome characterized by congenital microcephaly, moderately severe mental retardation, and symmetrical digital anomalies. Digital malformations of variable degree include hallux valgus, syndactyly of toes 4 and 5, short fifth fingers, single flexion crease of fifth fingers, polydactyly and syndolydactyly. {ECO:0000269|PubMed:21998596}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seckel Sendromu 2, 606744, Otozomal Resesif; SCKL2 (Seckel Sendromu) (Prenatal) (RBBP8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 2, 606744, Autosomal recessive; SCKL2 (Seckel syndrome) (RBBP8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBBP8	RBBP8, RIM, SCKL2, JWDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Jawad syndrome (JWDS) [MIM:251255]: A syndrome characterized by congenital microcephaly, moderately severe mental retardation, and symmetrical digital anomalies. Digital malformations of variable degree include hallux valgus, syndactyly of toes 4 and 5, short fifth fingers, single flexion crease of fifth fingers, polydactyly and syndolydactyly. {ECO:0000269|PubMed:21998596}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 4, 613676, Otozomal Resesif; SCKL4 (Seckel Sendromu) (CENPJ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 4, 613676, Autosomal recessive; SCKL4 (Seckel syndrome) (CENPJ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CENPJ	CENPJ, CPAP, MCPH6, SCKL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Microcephaly 6, primary, autosomal recessive (MCPH6) [MIM:608393]: A disease defined as a head circumference more than 3 standard deviations below the age, related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Seckel Sendromu 4, 613676, Otozomal Resesif; SCKL4 (Seckel Sendromu) (Prenatal) (CENPJ geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 4, 613676, Autosomal recessive; SCKL4 (Seckel syndrome) (CENPJ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CENPJ	CENPJ, CPAP, MCPH6, SCKL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Microcephaly 6, primary, autosomal recessive (MCPH6) [MIM:608393]: A disease defined as a head circumference more than 3 standard deviations below the age-related mean. Brain weight is markedly reduced and the cerebral cortex is disproportionately small. Despite this marked reduction in size, the gyral pattern is relatively well preserved, with no major abnormality in cortical architecture. Affected individuals are mentally retarded. Primary microcephaly is further defined by the absence of other syndromic features or significant neurological deficits due to degenerative brain disorder. {ECO:0000269|PubMed:15793586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 5, 613823, Otozomal Resesif; SCKL5 (Seckel Sendromu) (CEP152 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 5, 613823, Autosomal recessive; SCKL5 (Seckel syndrome) (CEP152 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP152	CEP152, KIAA0912, MCPH9, SCKL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seckel syndrome 5 (SCKL5) [MIM:613823]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21131973}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seckel Sendromu 5, 613823, Otozomal Resesif; SCKL5 (Seckel Sendromu) (Prenatal) (CEP152 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 5, 613823, Autosomal recessive; SCKL5 (Seckel syndrome) (CEP152 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP152	CEP152, KIAA0912, MCPH9, SCKL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 5 (SCKL5) [MIM:613823]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21131973}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 6, 614728, Otozomal Resesif; SCKL6 (CEP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 6, 614728, Autosomal recessive; SCKL6 (CEP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP63	CEP63, SCKL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seckel syndrome 6 (SCKL6) [MIM:614728]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21983783}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seckel Sendromu 6, 614728, Otozomal Resesif; SCKL6 (Prenatal) (CEP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 6, 614728, Autosomal recessive; SCKL6 (CEP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP63	CEP63, SCKL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 6 (SCKL6) [MIM:614728]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:21983783}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 7, 614851, Otozomal Resesif; SCKL7 (Mikrosefalik Primordial Dwarfizm, Dauber Tipi) (NIN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 7, 614851, Autosomal recessive; SCKL7 (Microcephalic primordial dwarfism, Dauber type) (NIN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NIN	NIN, KIAA1565, SCKL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seckel syndrome 7 (SCKL7) [MIM:614851]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:22933543}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Seckel Sendromu 7, 614851, Otozomal Resesif; SCKL7 (Mikrosefalik Primordial Dwarfizm, Dauber Tipi) (Prenatal) (NIN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 7, 614851, Autosomal recessive; SCKL7 (Microcephalic primordial dwarfism, Dauber type) (NIN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NIN	NIN, KIAA1565, SCKL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Seckel syndrome 7 (SCKL7) [MIM:614851]: A rare autosomal recessive disorder characterized by proportionate dwarfism of prenatal onset associated with low birth weight, growth retardation, severe microcephaly with a bird-headed like appearance, and mental retardation. {ECO:0000269|PubMed:22933543}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 8, 615807, Otozomal Resesif; SCKL8 (Seckel Sendromu) (DNA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 8, 615807, Autosomal recessive; SCKL8 (Seckel syndrome) (DNA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNA2	DNA2, DNA2L, KIAA0083, PEOA6, SCKL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 (PEOA6) [MIM:615156]: A disorder characterized by muscle weakness, mainly affecting the lower limbs, external ophthalmoplegia, exercise intolerance, and mitochondrial DNA deletions on muscle biopsy. Symptoms may appear in childhood or adulthood and show slow progression. {ECO:0000269|PubMed:23252259}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Seckel Sendromu 8, 615807, Otozomal Resesif; SCKL8 (Seckel Sendromu) (Prenatal) (DNA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 8, 615807, Autosomal recessive; SCKL8 (Seckel syndrome) (DNA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNA2	DNA2, DNA2L, KIAA0083, PEO46, SCKL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 6 (PEOA6) [MIM:615156]: A disorder characterized by muscle weakness, mainly affecting the lower limbs, external ophthalmoplegia, exercise intolerance, and mitochondrial DNA deletions on muscle biopsy. Symptoms may appear in childhood or adulthood and show slow progression. {ECO:0000269	PubMed:23352259}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Seckel Sendromu 9, 616777, Otozomal Resesif; SCKL9 (Seckel Sendromu) (TRAIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seckel syndrome 9, 616777, Autosomal recessive; SCKL9 (Seckel syndrome) (TRAIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAIP	TRAIP, TRIP, RNF206, SCKL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Seckel Sendromu 9, 616777, Otozomal Resesif; SCKL9 (Seckel Sendromu) (Prenatal) (TRAIP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seckel syndrome 9, 616777, Autosomal recessive; SCKL9 (Seckel syndrome) (TRAIP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAIP	TRAIP, TRIP, RNF206, SCKL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SED Konjenita, 183900, Otozomal Dominant (Spondiloepifizyal Displazi Konjenita) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SED congenita, 183900, autosomal dominant (Spondyloepiphyseal dysplasia congenita) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SED Konjenita, 183900, Otozomal Dominant (Spondiloepifizyal Displazi Konjenita) (MLPA)	SED congenita, 183900, autosomal dominant (Spondyloepiphyseal dysplasia congenita) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SED Konjenita, 183900, Otozomal Dominant (Spondiloepifizyal Displazi Konjenita) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SED congenita, 183900, autosomal dominant (Spondyloepiphyseal dysplasia congenita) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SED Konjenita, 183900, Otozomal Dominant (Spondiloepifizyal Displazi Konjenita) (Prenatal) (MLPA)	SED congenita, 183900, autosomal dominant (Spondyloepiphyseal dysplasia congenita) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SED, Maroteaux Tipi, 184095, Otozomal Dominant (Spondiloepifizyal Displazi, Maroteaux Tip) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SED, Maroteaux type, 184095, Autosomal dominant (Spondyloepiphyseal dysplasia, Maroteaux type) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269	PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SED, Maroteaux Tipi, 184095, Otozomal Dominant (Spondiloepifizyal Displazi, Maroteaux Tip) (Prenatal) (TRPV4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SED, Maroteaux type, 184095, Autosomal dominant (Spondyloepiphyseal dysplasia, Maroteaux type) (TRPV4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPV4	TRPV4, VROAC, HMSN2C, CMT2C, SPSMA, SSQTL1, SMAL, BCYM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Brachyolmia 3 (BCYM3) [MIM:113500]: A form of brachyolmia, a clinically and genetically heterogeneous skeletal dysplasia primarily affecting the spine and characterized by a short trunk, short stature, and platyspondyly. BCYM3 is an autosomal dominant form with severe scoliosis with or without kyphosis, and flattened irregular cervical vertebrae. {ECO:0000269	PubMed:18587396}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Segawa Sendromu, Resesif, 605407, Otozomal Resesif (Otozomal Resesif Dopa-Yanıtlı Distoni) (TH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Segawa syndrome, recessive, 605407, Autosomal recessive (Autosomal recessive dopa-responsive dystonia) (TH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TH	TH, TYH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=May play a role in the pathogenesis of Parkinson disease (PD). A genome-wide copy number variation analysis has identified a 34 kilobase deletion over the TH gene in a PD patient but not in any controls. {ECO:0000269	PubMed:20809526}.	EDTA Blood Sample
Segawa Sendromu, Resesif, 605407, Otozomal Resesif (Otozomal Resesif Dopa-Yanıtlı Distoni) (MLPA)	Segawa syndrome, recessive, 605407, Autosomal recessive (Autosomal recessive dopa-responsive dystonia) (MLPA)	TH	TH, TYH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Note=May play a role in the pathogenesis of Parkinson disease (PD). A genome-wide copy number variation analysis has identified a 34 kilobase deletion over the TH gene in a PD patient but not in any controls. {ECO:0000269	PubMed:20809526}.	EDTA Blood Sample
Segawa Sendromu, Resesif, 605407, Otozomal Resesif (Otozomal Resesif Dopa-Yanıtlı Distoni) (Prenatal) (TH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Segawa syndrome, recessive, 605407, Autosomal recessive (Autosomal recessive dopa-responsive dystonia) (TH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TH	TH, TYH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=May play a role in the pathogenesis of Parkinson disease (PD). A genome-wide copy number variation analysis has identified a 34 kilobase deletion over the TH gene in a PD patient but not in any controls. {ECO:0000269	PubMed:20809526}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Segawa Sendromu, Resesif, 605407, Otozomal Resesif (Otozomal Resesif Dopa-Yanıtlı Distoni) (Prenatal) (MLPA)	Segawa syndrome, recessive, 605407, Autosomal recessive (Autosomal recessive dopa-responsive dystonia) (Prenatal) (MLPA)	TH	TH, TYH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=May play a role in the pathogenesis of Parkinson disease (PD). A genome-wide copy number variation analysis has identified a 34 kilobase deletion over the TH gene in a PD patient but not in any controls. {ECO:0000269	PubMed:20809526}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöbetler, Benign Ailesel İnfantil, 2, 605751, Otozomal Dominant; BFIS2 (Benign Ailesel İnfantil Epilepsi) (PRRT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, benign familial infantile, 2, 605751, Autosomal dominant; BFIS2 (Benign familial infantile epilepsy) (PRRT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRRT2	PRRT2, PKC, DYT10, EKD1, BFIS2, BFIC2, ICCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Episodic kinesigenic dyskinesia 1 (EKD1) [MIM:128200]: An autosomal dominant neurologic condition characterized by recurrent and brief attacks of abnormal involuntary movements, triggered by sudden voluntary movement. These attacks usually have onset during childhood or early adulthood and can involve dystonic postures, chorea, or athetosis. {ECO:0000269|PubMed:22101681, ECO:0000269|PubMed:22120146, ECO:0000269|PubMed:22131361, ECO:0000269|PubMed:22209761}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease- causing mutations that produce truncation of the C-terminus of the protein alter subcellular location, from plasma membrane to cytoplasm {PubMed:22101681}. {ECO:0000269|PubMed:22101681}.	EDTA Blood Sample
Nöbetler, Benign Ailesel İnfantil, 3, 607745, Otozomal Dominant; BFIS3 (Benign Ailesel Neonatal-İnfantil Nöbetler 3b) (SCN2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, benign familial infantile, 3, 607745, Autosomal dominant; BFIS3 (Benign familial neonatal-infantile seizures) (SCN2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN2A	SCN2A, SCN2A1, BFIC3, EIEE11, BFIS3, BFNIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seizures, benign familial infantile 3 (BFIS3) [MIM:607745]: An autosomal dominant disorder in which afebrile seizures occur in clusters during the first year of life, without neurologic sequelae. {ECO:0000269|PubMed:11371648, ECO:0000269|PubMed:12243921, ECO:0000269|PubMed:15048894, ECO:0000269|PubMed:20371507, ECO:0000269|PubMed:22612257}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöbetler, Benign Ailesel İnfantil, 5, 617080, Otozomal Dominant; BFIS5 (Benign Ailesel İnfantil Epilepsi) (SCN8A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, benign familial infantile, 5, 617080, Autosomal dominant; BFIS5 (Benign familial infantile epilepsy) (SCN8A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN8A	SCN8A, CIAT, EIEE13, BFIS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cognitive impairment with or without cerebellar ataxia (CIAT) [MIM:614306]: A disorder characterized by markedly delayed cognitive and motor development, attention deficit disorder, and cerebellar ataxia. Features include bilateral esophoria, strabismatic amblyopia, unsustained gaze evoked nystagmus on horizontal gaze, ataxic gait, dysmetria in the upper limbs and dysarthria, with normal strength, tone, and reflexes. {ECO:0000269	PubMed:16236819}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Nöbetler, Benign Neonatal, 1, 121200, Otozomal Dominant (Benign Ailesel Neonatal Epilepsi) (KCNQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, benign neonatal, 1, 121200, Autosomal dominant (Benign familial neonatal epilepsy) (KCNQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ2	KCNQ2, EBN1, EIEE7, BFNS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seizures, benign familial neonatal 1 (BFNS1) [MIM:121200]: A disorder characterized by clusters of seizures occurring in the first days of life. Most patients have spontaneous remission by 12 months of age and show normal psychomotor development. Some rare cases manifest an atypical severe phenotype associated with epileptic encephalopathy and psychomotor retardation. The disorder is distinguished from benign familial infantile seizures by an earlier age at onset. In some patients, neonatal convulsions are followed later in life by myokymia, a benign condition characterized by spontaneous involuntary contractions of skeletal muscles fiber groups that can be observed as vermiform movement of the overlying skin. Electromyography typically shows continuous motor unit activity with spontaneous oligo- and multiplet-discharges of high intraburst frequency (myokymic discharges). Some patients may have isolated myokymia. {ECO:0000269|PubMed:11175290, ECO:0000269|PubMed:11572947, ECO:0000269|PubMed:14534157, ECO:0000269|PubMed:15249611, ECO:0000269|PubMed:9425895}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöbetler, Benign Neonatal, Tip 2, 121201, Otozomal Dominant; BFNS2 (Benign Ailesel Neonatal Epilepsi) (KCNQ3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, benign neonatal, type 2, 121201, Autosomal dominant; BFNS2 (Benign familial neonatal epilepsy) (KCNQ3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ3	KCNQ3, EBN2, BFNC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seizures, benign familial neonatal 2 (BFNS2) [MIM:121201]: A disorder characterized by clusters of seizures occurring in the first days of life. Most patients have spontaneous remission by 12 months of age and show normal psychomotor development. The disorder is distinguished from benign familial infantile seizures by an earlier age at onset. {ECO:0000269|PubMed:10852552, ECO:0000269|PubMed:14534157, ECO:0000269|PubMed:9425900}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöbetler, Kortikal Körlük, Mikrosefali Sendromu, 616632, Otozomal Resesif; SCBMS (Progresif Mikrosefali-Nöbet-Kortikal Körlük-Gelişimsel Gecikme Sendromu) (DIAPH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, cortical blindness, microcephaly syndrome, 616632, Autosomal recessive; SCBMS (Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome) (DIAPH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DIAPH1	DIAPH1, DFNA1, LFHL1, SCBMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Deafness, autosomal dominant, 1 (DFNA1) [MIM:124900]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:9360932}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Nöbetler, Kortikal Körlük, Mikrosefali Sendromu, 616632, Otozomal Resesif; SCBMS (Progresif Mikrosefali-Nöbet-Kortikal Körlük-Gelişimsel Gecikme Sendromu) (Prenatal) (DIAPH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seizures, cortical blindness, microcephaly syndrome, 616632, Autosomal recessive; SCBMS (Progressive microcephaly-seizures-cortical blindness-developmental delay syndrome) (DIAPH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DIAPH1	DIAPH1, DFNA1, LFHL1, SCBMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Deafness, autosomal dominant, 1 (DFNA1) [MIM:124900]: A form of non-syndromic sensorineural hearing loss. Sensorineural deafness results from damage to the neural receptors of the inner ear, the nerve pathways to the brain, or the area of the brain that receives sound information. {ECO:0000269|PubMed:9360932}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöbetler, Skolyoz ve Makrosefalisi Sendromu, 616682, Otozomal Resesif; SSMS (Nöbetler-Skolyoz-Makrosefali Sendromu) (EXT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Seizures, scoliosis, and macrocephaly syndrome, 616682, Autosomal recessive; SSMS (Seizures-scoliosis-macrocephaly syndrome) (EXT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EXT2	EXT2, SSMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. Note=The gene represented in this entry is involved in disease pathogenesis.;	EDTA Blood Sample
Nöbetler, Skolyoz ve Makrosefalisi Sendromu, 616682, Otozomal Resesif; SSMS (Nöbetler-Skolyoz-Makrosefali Sendromu) (MLPA)	Seizures, scoliosis, and macrocephaly syndrome, 616682, Autosomal recessive; SSMS (Seizures-scoliosis-macrocephaly syndrome) (MLPA)	EXT2	EXT2, SSMS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. Note=The gene represented in this entry is involved in disease pathogenesis.;	EDTA Blood Sample
Nöbetler, Skolyoz ve Makrosefalisi Sendromu, 616682, Otozomal Resesif; SSMS (Nöbetler-Skolyoz-Makrosefali Sendromu) (Prenatal) (EXT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Seizures, scoliosis, and macrocephaly syndrome, 616682, Autosomal recessive; SSMS (Seizures-scoliosis-macrocephaly syndrome) (EXT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EXT2	EXT2, SSMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. Note=The gene represented in this entry is involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Nöbetler, Skolyoz ve Makrosefalisi Sendromu, 616682, Otozomal Resesif; SSMS (Nöbetler-Skolyoz-Makrosefali Sendromu) (Prenatal) (MLPA)	Seizures, scoliosis, and macrocephaly syndrome, 616682, Autosomal recessive; SSMS (Seizures-scoliosis-macrocephaly syndrome) (Prenatal) (MLPA)	EXT2	EXT2, SSMS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Potocki-Shaffer syndrome (POSHS) [MIM:601224]: A syndrome characterized by foramina parietalia permagna, multiple exostoses, and craniofacial dysostosis and mental retardation in some cases. Note=The gene represented in this entry is involved in disease pathogenesis.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sengers Sendromu, 212350, Otozomal Resesif (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (AGK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sengers syndrome, 212350, Autosomal recessive (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (AGK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AGK	AGK, MULK, MTDPS10, CATCS, CTRCT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cataract 38 (CTRCT38) [MIM:614691]: An opacification of the crystalline lens of the eye becoming evident at birth. It frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269|PubMed:22415731}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sengers Sendromu, 212350, Otozomal Resesif (Konjenital Katarakt-Hipertrofik Kardiyomiyopati-Mitokondriyal Miyopati Sendromu) (Prenatal) (AGK geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sengers syndrome, 212350, Autosomal recessive (Congenital cataract-hypertrophic cardiomyopathy-mitochondrial myopathy syndrome) (AGK gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AGK	AGK, MULK, MTDPS10, CATCS, CTRCT38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cataract 38 (CTRCT38) [MIM:614691]: An opacification of the crystalline lens of the eye becoming evident at birth. It frequently results in visual impairment or blindness. Opacities vary in morphology, are often confined to a portion of the lens, and may be static or progressive. In general, the more posteriorly located and dense an opacity, the greater the impact on visual function. {ECO:0000269|PubMed:22415731}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 4, 606996, Otozomal Resesif; SLSN4 (Senior-Loken Sendromu) (NPHP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 4, 606996, Autosomal recessive; SLSN4 (Senior-Loken syndrome) (NPHP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP4	NPHP4, SLSN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Senior-Loken syndrome 4 (SLSN4) [MIM:606996]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. {ECO:0000269|PubMed:12244321, ECO:0000269|PubMed:15776426}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Senior-Loken Sendromu 4, 606996, Otozomal Resesif; SLSN4 (Senior-Loken Sendromu) (Prenatal) (NPHP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 4, 606996, Autosomal recessive; SLSN4 (Senior-Loken syndrome) (NPHP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP4	NPHP4, SLSN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Senior-Loken syndrome 4 (SLSN4) [MIM:606996]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. {ECO:0000269|PubMed:12244321, ECO:0000269|PubMed:15776426}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 5, 609254, Otozomal Resesif; SLSN5 (Senior-Loken Sendromu) (IQCB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 5, 609254, Autosomal recessive; SLSN5 (Senior-Loken syndrome) (IQCB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IQCB1	IQCB1, NPHP5, KIAA0036	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Senior-Loken syndrome 5 (SLSN5) [MIM:609254]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. {ECO:0000269|PubMed:15723066}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Senior-Loken Sendromu 5, 609254, Otozomal Resesif; SLSN5 (Senior-Loken Sendromu) (Prenatal) (IQCB1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 5, 609254, Autosomal recessive; SLSN5 (Senior-Loken syndrome) (IQCB1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IQCB1	IQCB1, NPHP5, KIAA0036	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Senior-Loken syndrome 5 (SLSN5) [MIM:609254]: A renal- retinal disorder characterized by progressive wasting of the filtering unit of the kidney (nephronophthisis), with or without medullary cystic renal disease, and progressive eye disease. Typically this disorder becomes apparent during the first year of life. {ECO:0000269|PubMed:15723066}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 6, 610189, Otozomal Resesif; SLSN6 (Senior-Loken Sendromu) (CEP290 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 6, 610189, Autosomal recessive; SLSN6 (Senior-Loken syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Senior-Loken Sendromu 6, 610189, Otozomal Resesif; SLSN6 (Senior-Loken Sendromu) (Prenatal) (CEP290 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 6, 610189, Autosomal recessive; SLSN6 (Senior-Loken syndrome) (CEP290 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP290	CEP290, KIAA0373, 3H1AG, JBTS5, SLSN6, LCA10, BBS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 5 (JBTS5) [MIM:610188]: A disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. Joubert syndrome type 5 shares the neurologic and neuroradiologic features of Joubert syndrome together with severe retinal dystrophy and/or progressive renal failure characterized by nephronophthisis. {ECO:0000269|PubMed:16682970, ECO:0000269|PubMed:16682973, ECO:0000269|PubMed:22425360}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 7, 613615; SLSN7 (Senior-Loken Sendromu) (SDCCAG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 7, 613615; SLSN7 (Senior-Loken syndrome) (SDCCAG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SDCCAG8	SDCCAG8, CCCAP, SLSN7, BBS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome 16 (BBS16) [MIM:615993]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:20835237, ECO:0000269|PubMed:22626039}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Senior-Loken Sendromu 7, 613615; SLSN7 (Senior-Loken Sendromu) (Prenatal) (SDCCAG8 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 7, 613615; SLSN7 (Senior-Loken syndrome) (SDCCAG8 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SDCCAG8	SDCCAG8, CCCAP, SLSN7, BBS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome 16 (BBS16) [MIM:615993]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. {ECO:0000269|PubMed:20835237, ECO:0000269|PubMed:22626039}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 8, 616307, Otozomal Resesif; SLSN8 (Senior-Loken Sendromu) (WDR19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 8, 616307, Autosomal recessive; SLSN8 (Senior-Loken syndrome) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269	PubMed:22019273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Senior-Loken Sendromu 8, 616307, Otozomal Resesif; SLSN8 (Senior-Loken Sendromu) (Prenatal) (WDR19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 8, 616307, Autosomal recessive; SLSN8 (Senior-Loken syndrome) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269	PubMed:22019273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu 9, 616629, Otozomal Resesif; SLSN9 (Senior-Loken Sendromu) (TRAF3IP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome 9, 616629, Autosomal recessive; SLSN9 (Senior-Loken syndrome) (TRAF3IP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAF3IP1	TRAF3IP1, MIP73, SLSN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Senior-Loken Sendromu 9, 616629, Otozomal Resesif; SLSN9 (Senior-Loken Sendromu) (Prenatal) (TRAF3IP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome 9, 616629, Autosomal recessive; SLSN9 (Senior-Loken syndrome) (TRAF3IP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAF3IP1	TRAF3IP1, MIP73, SLSN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu-1, 266900, Otozomal Resesif; SLSN1 (Senior-Loken Sendromu) (NPHP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Senior-Loken syndrome-1, 266900, Autosomal recessive; SLSN1 (Senior-Loken syndrome) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Senior-Loken Sendromu-1, 266900, Otozomal Resesif; SLSN1 (Senior-Loken Sendromu) (MLPA)	Senior-Loken syndrome-1, 266900, Autosomal recessive; SLSN1 (Senior-Loken syndrome) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Senior-Loken Sendromu-1, 266900, Otozomal Resesif; SLSN1 (Senior-Loken Sendromu) (Prenatal) (NPHP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Senior-Loken syndrome-1, 266900, Autosomal recessive; SLSN1 (Senior-Loken syndrome) (NPHP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Senior-Loken Sendromu-1, 266900, Otozomal Resesif; SLSN1 (Senior-Loken Sendromu) (Prenatal) (MLPA)	Senior-Loken syndrome-1, 266900, Autosomal recessive; SLSN1 (Senior-Loken syndrome) (Prenatal) (MLPA)	NPHP1	NPHP1, NPH1, SLSN1, JBTS4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Nephronophthisis 1 (NPHP1) [MIM:256100]: An autosomal recessive inherited disease characterized by anemia, polyuria, polydipsia, isosthenuria and death in uremia. Symmetrical destruction of the kidneys involving both tubules and glomeruli occurs. The underlying pathology is a chronic tubulo-interstitial nephropathy with characteristic tubular basement membrane thickening and medullary cyst formation. Associations with extrarenal symptoms, especially ocular lesions, are frequent. The age at death ranges from about 4 to 15 years. {ECO:0000269	PubMed:10839884}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sensorinöral Sağırlık-Hafif Renal Disfonksiyon, 602522, Otozomal Resesif (İnfantil Bartter Sendromu-Sensorinöral İşitme Kaybı) (BSND geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sensorineural deafness with mild renal dysfunction, 602522, Autosomal recessive (Infantile Bartter syndrome with sensorineural deafness) (BSND gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSND	BSND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bartter syndrome 4A (BS4A) [MIM:602522]: An autosomal recessive disorder characterized by impaired salt reabsorption in the thick ascending loop of Henle with pronounced salt wasting, hypokalemic metabolic alkalosis, and varying degrees of hypercalciuria. Bartter syndrome type 4 is associated with sensorineural deafness. {ECO:0000269|PubMed:11687798, ECO:0000269|PubMed:12574213}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Duysal Ataksik Nöropati, Dizartri ve Oftalmoparezi; Sando (Duysal Ataksik Nöropati-Disartri-Oftalmoparezi Sendromu) (POLG geni) (Tüm Gen Dizi Analizi) (Postnatal)	SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS; SANDO (Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome) (POLG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Duysal Ataksik Nöropati, Dizartri ve Oftalmoparezi; Sando (Duysal Ataksik Nöropati-Disartri-Oftalmoparezi Sendromu) (MLPA)	SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS; SANDO (Sensory ataxic neuropathy-dysarthria-ophthalmoparesis syndrome) (MLPA)	POLG	POLG, POLG1, POLGA, PEO, SANDO, SCAE, MTDPS4A, MTDPS4B, MIRAS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant, 1 (PEOA1) [MIM:157640]: A disorder characterized by progressive weakness of ocular muscles and levator muscle of the upper eyelid. In a minority of cases, it is associated with skeletal myopathy, which predominantly involves axial or proximal muscles and which causes abnormal fatigability and even permanent muscle weakness. Ragged-red fibers and atrophy are found on muscle biopsy. A large proportion of chronic ophthalmoplegias are associated with other symptoms, leading to a multisystemic pattern of this disease. Additional symptoms are variable, and may include cataracts, hearing loss, sensory axonal neuropathy, ataxia, depression, hypogonadism, and parkinsonism. {ECO:0000269|PubMed:12210792, ECO:0000269|PubMed:15351195, ECO:0000269|PubMed:15534189, ECO:0000269|PubMed:17420318, ECO:0000269|PubMed:18575922}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sepsis, Yatkınlık (CASP12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sepsis, susceptibility to (CASP12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASP12	CASP12, CASP12P1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Septik Şok, Yatkınlık (TNF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Septic shock, susceptibility to (TNF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNF	TNF, TNFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Psoriatic arthritis (PSORAS) [MIM:607507]: An inflammatory, seronegative arthritis associated with psoriasis. It is a heterogeneous disorder ranging from a mild, non-destructive disease to a severe, progressive, erosive arthropathy. Five types of psoriatic arthritis have been defined: asymmetrical oligoarthritis characterized by primary involvement of the small joints of the fingers or toes; asymmetrical arthritis which involves the joints of the extremities; symmetrical polyarthritis characterized by a rheumatoid like pattern that can involve hands, wrists, ankles, and feet;arthritis mutilans, which is a rare but deforming and destructive condition; arthritis of the sacroiliac joints and spine (psoriatic spondylitis). {ECO:0000269	PubMed:12746914}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample

Septooptik Displazi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (HESX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Septooptic dysplasia, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (HESX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HESX1	HESX1, RPX, CPHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. (ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Septooptik Displazi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (MLPA)	Septooptic dysplasia, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (MLPA)	HESX1	HESX1, RPX, CPHD5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. (ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Septooptik Displazi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (Prenatal) (HESX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Septooptic dysplasia, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (HESX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HESX1	HESX1, RPX, CPHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. (ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Septooptik Displazi, 182230, Otozomal Resesif, Otozomal Dominant (Septo-Optik Displazi Spektrumu) (Prenatal) (MLPA)	Septooptic dysplasia, 182230, Autosomal recessive, Autosomal dominant (Septo-optic dysplasia spectrum) (Prenatal) (MLPA)	HESX1	HESX1, RPX, CPHD5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Septooptic dysplasia (SOD) [MIM:182230]: A clinically heterogeneous disorder defined by any combination of optic nerve hypoplasia, pituitary gland hypoplasia with panhypopituitarism, and midline abnormalities of the brain, including absence of the corpus callosum and septum pellucidum. (ECO:0000269|PubMed:11136712, ECO:0000269|PubMed:9620767). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SERKAL Sendromu, 611812, Otozomal Resesif (SERKAL Sendromu) (WNT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SERKAL syndrome, 611812, Autosomal recessive (SERKAL syndrome) (WNT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT4	WNT4, SERKAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
SERKAL Sendromu, 611812, Otozomal Resesif (SERKAL Sendromu) (MLPA)	SERKAL syndrome, 611812, Autosomal recessive (SERKAL syndrome) (MLPA)	WNT4	WNT4, SERKAL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
SERKAL Sendromu, 611812, Otozomal Resesif (SERKAL Sendromu) (Prenatal) (WNT4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SERKAL syndrome, 611812, Autosomal recessive (SERKAL syndrome) (WNT4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT4	WNT4, SERKAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SERKAL Sendromu, 611812, Otozomal Resesif (SERKAL Sendromu) (Prenatal) (MLPA)	SERKAL syndrome, 611812, Autosomal recessive (SERKAL syndrome) (Prenatal) (MLPA)	WNT4	WNT4, SERKAL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mullerian aplasia and hyperandrogenism (MULLAPL) [MIM:158330]: A disorder of sex development. Affected females manifest dysgenesis of Mullerian duct derivatives absent or rudimentary uterus and vagina, functional ovaries, primary amenorrhea, hyperandrogenism and hirsutism. (ECO:0000269|PubMed:15317892, ECO:0000269|PubMed:16959810, ECO:0000269|PubMed:18182450). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Serpin Peptidaz İnhibitörü, Clade A, Üye 2, Psödogen; Serpina2p (SERPINA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SERPIN PEPTIDASE INHIBITOR, CLADE A, MEMBER 2, PSEUDOGENE; SERPINA2P (SERPINA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sertoli-Cell-Only Sendromu (DAZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sertoli-cell-only syndrome (DAZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAZ1	DAZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure Y-linked 2 (SPGFY2) [MIM:415000]: A disorder resulting in the absence (azoospermia) or reduction (oligozoospermia) of sperm in the semen, leading to male infertility. (ECO:0000269|PubMed:11095434, ECO:0000269|PubMed:11870237, ECO:0000269|PubMed:12801575). Note=The disease may be caused by mutations affecting the gene represented in this entry. AZFc deletions in the Yq11.23 region including the DAZ genes are the most common known genetic cause of human male infertility.;	EDTA Blood Sample
SESAME Sendromu, 612780, Otozomal Resesif (EAST Sendromu) (KCNJ10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SESAME syndrome, 612780, Autosomal recessive (EAST syndrome) (KCNJ10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ10	KCNJ10, SESAME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Seizures, sensorineural deafness, ataxia, mental retardation, and electrolyte imbalance (SESAMES) [MIM:612780]: A complex disorder characterized by generalized seizures with onset in infancy, delayed psychomotor development, ataxia, sensorineural hearing loss, hypokalemia, metabolic alkalosis, and hypomagnesemia. (ECO:0000269|PubMed:19289823, ECO:0000269|PubMed:19420365, ECO:0000269|PubMed:22612257). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sesil Digi Polipozis Kanser Sendromu, 617108, Otozomal Dominant (Hiperplastik Polipozis Sendromu) (RNF43 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sessile serrated polyposis cancer syndrome, 617108, Autosomal dominant (Hyperplastic polyposis syndrome) (RNF43 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF43	RNF43, RNF124, SSPCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, ADA Eksikliğine Bağlı, 102700, Otozomal Resesif, Somatik Mozaisizm (Ağır Kombine İmmun Yetmezlik, Adenozin Deaminaz Eksikliğine Bağlı) (ADA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency due to ADA deficiency, 102700, Autosomal recessive, Somatic mosaicism (Severe combined immunodeficiency due to adenosine deaminase deficiency) (ADA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADA	ADA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, ADA Eksikliğine Bağlı, 102700, Otozomal Resesif, Somatik Mozaisizm (Ağır Kombine İmmun Yetmezlik, Adenozin Deaminaz Eksikliğine Bağlı) (Prenatal) (ADA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency due to ADA deficiency, 102700, Autosomal recessive, Somatic mosaicism (Severe combined immunodeficiency due to adenosine deaminase deficiency) (ADA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADA	ADA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-negative due to adenosine deaminase deficiency (ADASCID) [MIM:102700]: An autosomal recessive disorder accounting for about 50% of non-X-linked SCIDs. SCID refers to a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients with SCID present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell- mediated cellular immunity due to a defect in T-cell development. ADA deficiency has been diagnosed in chronically ill teenagers and adults (late or adult onset). Population and newborn screening programs have also identified several healthy individuals with normal immunity who have partial ADA deficiency. (ECO:0000269|PubMed:10200056, ECO:0000269|PubMed:1284479, ECO:0000269|PubMed:2166947, ECO:0000269|PubMed:2783588, ECO:0000269|PubMed:3182793, ECO:0000269|PubMed:3839802, ECO:0000269|PubMed:6208479, ECO:0000269|PubMed:7599635, ECO:0000269|PubMed:8227344, ECO:0000269|PubMed:8299233). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, IL2 Eksikliğine Bağlı (IL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency due to IL2 deficiency (IL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL2	IL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving IL2 is found in a form of T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(4;16)(q26;p13) with involves TNFRSF17. (ECO:0000269	PubMed:11965833).	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, IL2 Eksikliğine Bağlı (Prenatal) (IL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency due to IL2 deficiency (IL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL2	IL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving IL2 is found in a form of T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(4;16)(q26;p13) with involves TNFRSF17. (ECO:0000269	PubMed:11965833).	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, Mikrosefali, Büyüme Geriliği, ve İyonize Radyasyona Hassasiyet, 611291 (Cernunnos-XLF Eksikliği) (NHEJ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (Cernunnos-XLF deficiency) (NHEJ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NHEJ1	NHEJ1, XLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency due to NHEJ1 deficiency (NHEJ1-SCID) [MIM:611291]: SCID refers to a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia and low or absent antibody levels. Patients with SCID present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. NHEJ1-SCID is characterized by a profound T- and B-lymphocytopenia associated with increased cellular sensitivity to ionizing radiation, microcephaly and growth retardation. Some patients may manifest SCID with sensitivity to ionizing radiation without microcephaly and mild growth retardation, probably due to hypomorphic NHEJ1 mutations. (ECO:0000269|PubMed:16439204, ECO:0000269|PubMed:16439205). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ağır Kombine İmmün Yetmezlik, Mikrosefali, Büyüme Geriliği, ve İyonize Radyasyona Hassasiyet, 611291 (Cernunnos-XLF Eksikliği) (Prenatal) (NHEJ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency with microcephaly, growth retardation, and sensitivity to ionizing radiation, 611291 (Cernunnos-XLF deficiency) (NHEJ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NHEJ1	NHEJ1, XLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency due to NHEJ1 deficiency (NHEJ1-SCID) [MIM:611291]: SCID refers to a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia and low or absent antibody levels. Patients with SCID present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. NHEJ1-SCID is characterized by a profound T- and B-lymphocytopenia associated with increased cellular sensitivity to ionizing radiation, microcephaly and growth retardation. Some patients may manifest SCID with sensitivity to ionizing radiation without microcephaly and mild growth retardation, probably due to hypomorphic NHEJ1 mutations. {ECO:0000269|PubMed:16439204, ECO:0000269|PubMed:16435055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, Athabascan Türü, 602450, Otozomal Resesif (Ağır Kombine İmmun Yetmezlik, DCLRE1C Eksikliğine Bağlı) (DCLRE1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, Athabascan type, 602450, Autosomal recessive (Severe combined immunodeficiency due to DCLRE1C deficiency) (DCLRE1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, Athabascan Türü, 602450, Otozomal Resesif (Ağır Kombine İmmun Yetmezlik, DCLRE1C Eksikliğine Bağlı) (MLPA)	Severe combined immunodeficiency, Athabascan type, 602450, Autosomal recessive (Severe combined immunodeficiency due to DCLRE1C deficiency) (MLPA)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, Athabascan Türü, 602450, Otozomal Resesif (Ağır Kombine İmmun Yetmezlik, DCLRE1C Eksikliğine Bağlı) (Prenatal) (DCLRE1C geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, Athabascan type, 602450, Autosomal recessive (Severe combined immunodeficiency due to DCLRE1C deficiency) (DCLRE1C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, Athabascan Türü, 602450, Otozomal Resesif (Ağır Kombine İmmun Yetmezlik, DCLRE1C Eksikliğine Bağlı) (Prenatal) (MLPA)	Severe combined immunodeficiency, Athabascan type, 602450, Autosomal recessive (Severe combined immunodeficiency due to DCLRE1C deficiency) (Prenatal) (MLPA)	DCLRE1C	DCLRE1C, ARTEMIS, SCIDA	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-negative/NK-cell-positive with sensitivity to ionizing radiation (RSSCID) [MIM:602450]: A form of severe combined immunodeficiency, a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy with recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T- cell-mediated cellular immunity due to a defect in T-cell development. Individuals affected by RS-SCID show defects in the DNA repair machinery necessary for coding joint formation and the completion of V(D)J recombination. A subset of cells from such patients show increased radiosensitivity. {ECO:0000269|PubMed:11336668, ECO:0000269|PubMed:12406895, ECO:0000269|PubMed:12569164, ECO:0000269|PubMed:12592555, ECO:0000269|PubMed:12921762}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, B Hücre-Negatif, 601457, Otozomal Resesif (Ağır Kombine İmmün Yetmezlik,Komplet RAG1/2 Eksikliğinebağlı) (RAG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, B cell-negative, 601457, Autosomal recessive (Severe combined immunodeficiency due to complete RAG1/2 deficiency) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. {ECO:0000269|PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, B Hücre-Negatif, 601457, Otozomal Resesif (Ağır Kombine İmmün Yetmezlik,Komplet RAG1/2 Eksikliğinebağlı) (RAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, B cell-negative, 601457, Autosomal recessive (Severe combined immunodeficiency due to complete RAG1/2 deficiency) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. {ECO:0000269|PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, B Hücre-Negatif, 601457, Otozomal Resesif (Ağır Kombine İmmün Yetmezlik,Komplet RAG1/2 Eksikliğinebağlı) (Prenatal) (RAG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, B cell-negative, 601457, Autosomal recessive (Severe combined immunodeficiency due to complete RAG1/2 deficiency) (RAG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG2	RAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. {ECO:0000269|PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, B Hücre-Negatif, 601457, Otozomal Resesif (Ağır Kombine İmmün Yetmezlik,Komplet RAG1/2 Eksikliğinebağlı) (Prenatal) (RAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, B cell-negative, 601457, Autosomal recessive (Severe combined immunodeficiency due to complete RAG1/2 deficiency) (RAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAG1	RAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Combined cellular and humoral immune defects with granulomas (CHDG) [MIM:233650]: Immunodeficiency disease with granulomas in the skin, mucous membranes, and internal organs. Other characteristics include hypogammaglobulinemia, a diminished number of T and B-cells, and sparse thymic tissue on ultrasonography. {ECO:0000269|PubMed:18463379}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, T Hücresi-Negatif, B-Hücresi / NK Hücresi Pozitif, 608971, Otozomal Resesif (T-B + IL-7Ralfa Eksikliğine Bağlı Ağır Kombine İmmun Yetmelik) (PTPRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971, Autosomal recessive (T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency) (PTPRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPRC	PTPRC, CD45, LCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple sclerosis (MS) [MIM:126200]: A multifactorial, inflammatory, demyelinating disease of the central nervous system. Sclerotic lesions are characterized by perivascular infiltration of monocytes and lymphocytes and appear as indurated areas in pathologic specimens (sclerosis in plaques). The pathological mechanism is regarded as an autoimmune attack of the myelin sheath, mediated by both cellular and humoral immunity. Clinical manifestations include visual loss, extra-ocular movement disorders, paresthesias, loss of sensation, weakness, dysarthria, spasticity, ataxia and bladder dysfunction. Genetic and environmental factors influence susceptibility to the disease. {ECO:0000269|PubMed:11101853}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, T Hücresi-Negatif, B-Hücresi / NK Hücresi Pozitif, 608971, Otozomal Resesif (T-B + IL-7Ralfa Eksikliğine Bağlı Ağır Kombine İmmun Yetmelik) (Prenatal) (PTPRC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, T cell-negative, B-cell/natural killer-cell positive, 608971, Autosomal recessive (T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency) (PTPRC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTPRC	PTPRC, CD45, LCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple sclerosis (MS) [MIM:126200]: A multifactorial, inflammatory, demyelinating disease of the central nervous system. Sclerotic lesions are characterized by perivascular infiltration of monocytes and lymphocytes and appear as indurated areas in pathologic specimens (sclerosis in plaques). The pathological mechanism is regarded as an autoimmune attack of the myelin sheath, mediated by both cellular and humoral immunity. Clinical manifestations include visual loss, extra-ocular movement disorders, paresthesias, loss of sensation, weakness, dysarthria, spasticity, ataxia and bladder dysfunction. Genetic and environmental factors influence susceptibility to the disease. {ECO:0000269|PubMed:11101853}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Ağır Kombine İmmün Yetmezlik, T-Hücresi Negatif, B-Hücresi / NK Hücresi Pozitif Tip, 608971, Otozomal Resesif (T-B + IL-7Ralfa Eksikliğine Bağlı Ağır Kombine İmmun Yetmezlik (IL7R geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971, Autosomal recessive (T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency) (IL7R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL7R	IL7R, IL7RA, CD127	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-positive/NK-cell-positive (T(-)B(+)NK(+) SCID) [MIM:608971]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, T-Hücresi Negatif, B-Hücresi / NK Hücresi Pozitif Tip, 608971, Otozomal Resesif (T-B + IL-7Ralfa Eksikliğine Bağlı Ağır Kombine İmmun Yetmezlik) (Prenatal) (IL7R geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 608971, Autosomal recessive (T-B+ severe combined immunodeficiency due to IL-7Ralpha deficiency) (IL7R gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL7R	IL7R, IL7RA, CD127	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency autosomal recessive T- cell-negative/B-cell-positive/NK-cell-positive (T(-)B(+)NK(+) SCID) [MIM:608971]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, X'e Bağlı, 300-400, X'e Bağlı; SCIDX1 (İmmun Yetmezlik, Ağır Kombine, T-B + Gama Zinciri Eksikliğine Bağlı) (IL2RG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Severe combined immunodeficiency, X-linked, 300400, X-linked recessive; SCIDX1 (T-B+ severe combined immunodeficiency due to gamma chain deficiency) (IL2RG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IL2RG	IL2RG, SCIDX1, SCIDX, IMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Severe combined immunodeficiency X-linked T-cell-negative/B-cell-positive/NK-cell-negative (XSCID) [MIM:300400]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269|PubMed:7557965, ECO:0000269|PubMed:7668284, ECO:0000269|PubMed:7860773, ECO:0000269|PubMed:7937790, ECO:0000269|PubMed:8027558, ECO:0000269|PubMed:8088810, ECO:0000269|PubMed:8296698, ECO:0000269|PubMed:8401490, ECO:0000269|PubMed:8900989, ECO:0000269|PubMed:9049783, ECO:0000269|PubMed:9150740}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ağır Kombine İmmün Yetmezlik, X'e Bağlı, 300-400, X'e Bağlı; SCIDX1 (İmmun Yetmezlik, Ağır Kombine, T-B + Gama Zinciri Eksikliğine Bağlı) (Prenatal) (IL2RG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Severe combined immunodeficiency, X-linked, 300400, X-linked recessive; SCIDX1 (T-B+ severe combined immunodeficiency due to gamma chain deficiency) (IL2RG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IL2RG	IL2RG, SCIDX1, SCIDX, IMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Severe combined immunodeficiency X-linked T-cell-negative/B-cell-positive/NK-cell-negative (XSCID) [MIM:300400]: A form of severe combined immunodeficiency (SCID), a genetically and clinically heterogeneous group of rare congenital disorders characterized by impairment of both humoral and cell-mediated immunity, leukopenia, and low or absent antibody levels. Patients present in infancy recurrent, persistent infections by opportunistic organisms. The common characteristic of all types of SCID is absence of T-cell-mediated cellular immunity due to a defect in T-cell development. {ECO:0000269|PubMed:7557965, ECO:0000269|PubMed:7668284, ECO:0000269|PubMed:7860773, ECO:0000269|PubMed:7937790, ECO:0000269|PubMed:8027558, ECO:0000269|PubMed:8088810, ECO:0000269|PubMed:8296698, ECO:0000269|PubMed:8401490, ECO:0000269|PubMed:8900989, ECO:0000269|PubMed:9049783, ECO:0000269|PubMed:9150740}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Şiddetli Kütanöz Yan Etki, Yatkınlık (Stevens-Johnson Sendromu) (HLA-A geni) (Tüm Gen Dizi Analizi) (Postnatal)	SEVERE CUTANEOUS ADVERSE REACTION, SUSCEPTIBILITY TO (Stevens-Johnson syndrome) (HLA-A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-A	HLA-A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cinsiyet farklılaşma bozuklukları paneli (SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2) (MLPA)	Sex determination (SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2) (MLPA)	SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Cinsiyet farklılaşma bozuklukları paneli (SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2) (MLPA) (Prenatal)	Sex determination (SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2) (MLPA) (Prenatal)	SOX9 17q24.3; NR5A1 9q33; WNT4 1p36.12; CXorf21 & NROB1 Xp21.2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sezary Sendromu, Somatik (BCL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sezary syndrome, somatic (BCL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BCL10	BCL10, IMD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 37 (IMD37) [MIM:616098]: A form of primary combined immunodeficiency, a group of disorders characterized by severe recurrent infections, with normal numbers or an absence of T and B lymphocytes, and impaired cellular and humoral immunity. IMD37 is characterized by hypogammaglobulinemia without lymphopenia, but with profoundly reduced memory B cells and memory T cells, and increased numbers of circulating naive lymphocytes. Inheritance is autosomal recessive. {ECO:0000269	PubMed:25365219}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA blood sample / EDTA bone marrow
Shaheen Sendromu, 615328, Otozomal Resesif; SHNS (Hipohidroz-Mine Hipoplazisi-Palmoplantar Keratoderma-Mental Retardasyon Sendromu) (COG6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Shaheen syndrome, 615328, Autosomal recessive; SHNS (Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome) (COG6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COG6	COG6, COD2, KIAA1134, CDG2L, SHNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital disorder of glycosylation 2L (CDG2L) [MIM:614576]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2L include neonatal intractable focal seizures, vomiting, loss of consciousness, intracranial bleeding due to vitamin K deficiency, and death in infancy. {ECO:0000269	PubMed:20605848}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Shaheen Sendromu, 615328, Otozomal Resesif; SHNS (Hipohidroz-Mine Hipoplazisi-Palmoplantar Keratoderma-Mental Retardasyon Sendromu) (Prenatal) (COG6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Shaheen syndrome, 615328, Autosomal recessive; SHNS (Hypohidrosis-enamel hypoplasia-palmoplantar keratoderma-intellectual disability syndrome) (COG6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COG6	COG6, COD2, KIAA1134, CDG2L, SHNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital disorder of glycosylation 2L (CDG2L) [MIM:614576]: A multisystem disorder caused by a defect in glycoprotein biosynthesis and characterized by under-glycosylated serum glycoproteins. Congenital disorders of glycosylation result in a wide variety of clinical features, such as defects in the nervous system development, psychomotor retardation, dysmorphic features, hypotonia, coagulation disorders, and immunodeficiency. The broad spectrum of features reflects the critical role of N- glycoproteins during embryonic development, differentiation, and maintenance of cell functions. Clinical features of CDG2L include neonatal intractable focal seizures, vomiting, loss of consciousness, intracranial bleeding due to vitamin K deficiency, and death in infancy. {ECO:0000269	PubMed:20605848}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Shashi-Pena Sendromu, 617190, Otozomal Dominant; SHAPNS (ASXL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Shashi-Pena syndrome, 617190, Autosomal dominant; SHAPNS (ASXL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASXL2	ASXL2, KIAA1685, SHAPNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving ASXL2 is a cause of therapy-related myelodysplastic syndrome. Translocation t(2;8)(q23;p11.2) with KAT6A generates a KAT6A-ASXL2 fusion protein.;	EDTA Blood Sample
Shashi-Pena Sendromu, 617190, Otozomal Dominant; SHAPNS (Prenatal) (ASXL2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Shashi-Pena syndrome, 617190, Autosomal dominant; SHAPNS (ASXL2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASXL2	ASXL2, KIAA1685, SHAPNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving ASXL2 is a cause of therapy-related myelodysplastic syndrome. Translocation t(2;8)(q23;p11.2) with KAT6A generates a KAT6A-ASXL2 fusion protein.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa QT Sendromu 1, 609620; SQT1 (Ailesel Kısa QT Sendromu) (KCNH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short QT syndrome 1, 609620; SQT1 (Familial short QT syndrome) (KCNH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNH2	KCNH2, LQT2, HERG, SQT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. {ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısa QT Sendromu 1, 609620; SQT1 (Ailesel Kısa QT Sendromu) (MLPA)	Short QT syndrome 1, 609620; SQT1 (Familial short QT syndrome) (MLPA)	KCNH2	KCNH2, LQT2, HERG, SQT1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Short QT syndrome 1 (SQT1) [MIM:609620]: A heart disorder characterized by idiopathic persistently and uniformly short QT interval on ECG in the absence of structural heart disease in affected individuals. It causes syncope and sudden death. {ECO:0000269|PubMed:14676148, ECO:0000269|PubMed:15828882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısa QT Sendromu 2, 609621, Otozomal Dominant; SQT2 (Ailesel Kısa QT Sendromu) (KCNQ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short QT syndrome 2, 609621, Autosomal dominant; SQT2 (Familial short QT syndrome) (KCNQ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNQ1	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB3, SQT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Diabetes mellitus, non-insulin-dependent (NIDDM) [MIM:125853]: A multifactorial disorder of glucose homeostasis caused by a lack of sensitivity to the body's own insulin. Affected individuals usually have an obese body habitus and manifestations of a metabolic syndrome characterized by diabetes, insulin resistance, hypertension and hypertriglyceridemia. The disease results in long-term complications that affect the eyes, kidneys, nerves, and blood vessels. {ECO:0000269|PubMed:18711366, ECO:0000269|PubMed:18711367, ECO:0000269|PubMed:24390345}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa QT Sendromu 3, 609622; SQT3 (Ailesel Kısa QT Sendromu) (KCNJ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short QT syndrome 3, 609622; SQT3 (Familial short QT syndrome) (KCNJ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. {ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324964}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Kısa QT Sendromu 3, 609622; SQT3 (Ailesel Kısa QT Sendromu) (MLPA)	Short QT syndrome 3, 609622; SQT3 (Familial short QT syndrome) (MLPA)	KCNJ2	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7, SQT3, ATFB9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Long QT syndrome 7 (LQT7) [MIM:170390]: A heart disorder characterized by a prolonged QT interval on the ECG and polymorphic ventricular arrhythmias. They cause syncope and sudden death in response to exercise or emotional stress, and can present with a sentinel event of sudden cardiac death in infancy. Long QT syndrome type 7 manifests itself as a clinical triad consisting of potassium-sensitive periodic paralysis, ventricular ectopy and dysmorphic features. [ECO:0000269|PubMed:11371347, ECO:0000269|PubMed:12148092, ECO:0000269|PubMed:12163457, ECO:0000269|PubMed:16571646, ECO:0000269|PubMed:17324064). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Mikrosefali, Kısa Boy, Belirgin Yüz Bulguları, 615789, Otozomal Resesif (CRIPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature with microcephaly and distinctive facies, 615789, Autosomal recessive (CRIPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRIPT	CRIPT, SSMDF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short stature with microcephaly and distinctive facies (SSMF) [MIM:615789]: A disease characterized by dwarfism, microcephaly, and distinctive facial dysmorphism involving frontal bossing, high forehead, sparse hair and eyebrows, telecanthus, mild proptosis, anteverted nares, and flat nasal bridge. [ECO:0000269	PubMed:24389050). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Mikrosefali, Kısa Boy, Belirgin Yüz Bulguları, 615789, Otozomal Resesif (Prenatal) (CRIPT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature with microcephaly and distinctive facies, 615789, Autosomal recessive (CRIPT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRIPT	CRIPT, SSMDF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short stature with microcephaly and distinctive facies (SSMF) [MIM:615789]: A disease characterized by dwarfism, microcephaly, and distinctive facial dysmorphism involving frontal bossing, high forehead, sparse hair and eyebrows, telecanthus,, mild proptosis, anteverted nares, and flat nasal bridge. [ECO:0000269	PubMed:24389050). Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy-Spesifik Olmayan İskelet Anomalileri, 616255, Otozomal Dominant; SNSK (NPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature with nonspecific skeletal abnormalities, 616255, Autosomal dominant; SNSK (NPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acromesomelic dysplasia, Maroteaux type (AMDM) [MIM:602875]: An autosomal recessive acromesomelic chondrodysplasia. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs and handfoot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knob-like fingers). AMDM is characterized by axial skeletal involvement with wedging of vertebral bodies. In AMDM all skeletal elements are present but show abnormal rates of linear growth. [ECO:0000269	PubMed:15146390). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Boy-Spesifik Olmayan İskelet Anomalileri, 616255, Otozomal Dominant; SNSK (Prenatal) (NPR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature with nonspecific skeletal abnormalities, 616255, Autosomal dominant; SNSK (NPR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NPR2	NPR2, ANPRB, AMDM, ECDM, SNSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acromesomelic dysplasia, Maroteaux type (AMDM) [MIM:602875]: An autosomal recessive acromesomelic chondrodysplasia. Acromesomelic chondrodysplasias are rare hereditary skeletal disorders characterized by short stature, very short limbs and handfoot malformations. The severity of limb abnormalities increases from proximal to distal with profoundly affected hands and feet showing brachydactyly and/or rudimentary fingers (knob-like fingers). AMDM is characterized by axial skeletal involvement with wedging of vertebral bodies. In AMDM all skeletal elements are present but show abnormal rates of linear growth. [ECO:0000269	PubMed:15146390). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy, Kulak Yolu Atrezisi, Mandibuler Hipoplazi, İskelet Anormallikleri, 602471, Otozomal Resesif; SAMS (Kısa Boy-Dış Kulak Yolu Kanal Atrezisi-Mandibular Hipoplazi-İskelet Anormalleri Sendromu) (GSC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471, Autosomal recessive; SAMS (Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome) (GSC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSC	GSC, SAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities (SAMS) [MIM:602471]: An autosomal recessive developmental disorder with features of a first and second branchial arch syndrome, and with unique rhizomelic skeletal anomalies. Craniofacial abnormalities can lead to conductive hearing loss, respiratory insufficiency, and feeding difficulties. Skeletal features include bilateral humeral hypoplasia, humeroscapular synostosis, pelvic abnormalities, and proximal defects of the femora. Affected individuals may also have some features of a neurocristopathy or abnormal mesoderm development, such as urogenital anomalies, that are distinct from other branchial arch syndromes. [ECO:0000269	PubMed:24290375). Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Kısa Boy, Kulak Yolu Atrezisi, Mandibuler Hipoplazi, İskelet Anormallikleri, 602471, Otozomal Resesif; SAMS (Kısa Boy-Dış Kulak Yolu Kanal Atrezisi-Mandibular Hipoplazi-İskelet Anormalleri Sendromu) (Prenatal) (GSC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471, Autosomal recessive; SAMS (Short stature-auditory canal atresia-mandibular hypoplasia-skeletal anomalies syndrome) (GSC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GSC	GSC, SAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities (SAMS) [MIM:602471]: An autosomal recessive developmental disorder with features of a first and second branchial arch syndrome, and with unique rhizomelic skeletal anomalies. Craniofacial abnormalities can lead to conductive hearing loss, respiratory insufficiency, and feeding difficulties. Skeletal features include bilateral humeral hypoplasia, humeroscapular synostosis, pelvic abnormalities, and proximal defects of the femora. Affected individuals may also have some features of a neurocristopathy or abnormal mesoderm development, such as urogenital anomalies, that are distinct from other branchial arch syndromes. [ECO:0000269	PubMed:24290375). Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy, Brakidaktili, Entelektüel Gelişim Geriliği, ve Paralizi, 617157, Otozomal Resesif; SBIDDS (Kısa Boy-Brakidaktili-Obezite-Global Gelişimsel Gecikme Sendromu) (PRMT7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157, Autosomal recessive; SBIDDS (Short stature-brachydactyly-obesity-global developmental delay syndrome) (PRMT7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRMT7	PRMT7, KIAA1933, SBIDDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Boy, Brakidaktili, Entelektüel Gelişim Geriliği, ve Paralizi, 617157, Otozomal Resesif; SBIDDS (Kısa Boy-Brakidaktili-Obezite-Global Gelişimsel Gecikme Sendromu) (Prenatal) (PRMT7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature, brachydactyly, intellectual developmental disability, and seizures, 617157, Autosomal recessive; SBIDDS (Short stature-brachydactyly-obesity-global developmental delay syndrome) (PRMT7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PRMT7	PRMT7, KIAA1933, SBIDDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy, Gelişimsel Gecikme ve Konjenital Kalp Kusurları, 617044, Otozomal Resesif (TKT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, developmental delay, and congenital heart defects, 617044, Autosomal recessive (TKT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TKT	TKT, SDDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Boy, Gelişimsel Gecikme ve Konjenital Kalp Kusurları, 617044, Otozomal Resesif (Prenatal) (TKT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature, developmental delay, and congenital heart defects, 617044, Autosomal recessive (TKT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TKT	TKT, SDDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy, İdyopatik Ailesel, 300582; SDDHD (SHOX İlgili Kısa Boy) (SHOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, idiopathic familial, 300582; SDDHD (SHOX-related short stature) (SHOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHOX	SHOX, GCFX, SS, PHOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. [ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Boy, İdyopatik Ailesel, 300582; SDDHD (SHOX İlgili Kısa Boy) (MLPA)	Short stature, idiopathic familial, 300582; SDDHD (SHOX-related short stature) (MLPA)	SHOX	SHOX, GCFX, SS, PHOG	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Leri-Weill dyschondrosteosis (LWD) [MIM:127300]: Dominantly inherited skeletal dysplasia characterized by moderate short stature predominantly because of short mesomelic limb segments. It is often associated with the Madelung deformity of the wrist, comprising bowing of the radius and dorsal dislocation of the distal ulna. [ECO:0000269|PubMed:11030412, ECO:0000269|PubMed:11403039). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Boy, Mikrosefali, ve Endokrin Bozukluklar, 616541, Otozomal Resesif; SSMED (Mikrosefalik Primordial Dwarfizm-İnsülin Direnci Sendromu) (XRCC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, microcephaly, and endocrine dysfunction, 616541, Autosomal recessive; SSMED (Microcephalic primordial dwarfism-insulin resistance syndrome) (XRCC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XRCC4	XRCC4, SSMED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Boy, Mikrosefali, ve Endokrin Bozukluklar, 616541, Otozomal Resesif; SSMED (Mikrosefalik Primordial Dwarfizm-İnsülin Direnci Sendromu) (Prenatal) (XRCC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature, microcephaly, and endocrine dysfunction, 616541, Autosomal recessive; SSMED (Microcephalic primordial dwarfism-insulin resistance syndrome) (XRCC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XRCC4	XRCC4, SSMED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Boy, Onikodisplazisi, Yüzde Dismorfizm ve Hipotrikozis, 614813, Otozomal Resesif; SOFT (Kısa Boy-Onikodisplazisi-Fasiyal Dismorfizm-Hipotrikoz Sendromu) (POC1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813, Autosomal recessive; SOFT (Short stature-onychodysplasia-facial dysmorphism-hypotrichosis syndrome) (POC1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POC1A	POC1A, PIX2, SOFT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis (SOFT) [MIM:614813]: A syndrome characterized by severely short long bones, peculiar facies associated with paucity of hair, and nail anomalies. Growth retardation is evident on prenatal ultrasound as early as the second trimester of pregnancy, and affected individuals reach a final stature consistent with a height age of 6 years to 8 years. Relative macrocephaly is present during early childhood but head circumference is markedly low by adulthood. Psychomotor development is normal. Facial dysmorphism includes a long, triangular face with prominent nose and small ears, and affected individuals have an unusual high-pitched voice. Clinodactyly, brachydactyly, and hypoplastic distal phalanges and fingernails are present in association with postpubertal sparse and short hair. Typical skeletal findings include short and thick long bones with mild irregular metaphyseal changes, short femoral necks, and hypoplastic pelvis and sacrum. All long bones of the hand are short, with major delay of carpal ossification and cone- shaped epiphyses. Vertebral body ossification is also delayed. [ECO:0000269|PubMed:22840363, ECO:0000269|PubMed:22840364). Note=The disease is caused by mutations affecting the gene represented in this entry. Cells derived from affected individuals have abnormal mitotic mechanics with multipolar spindles, in addition to clearly impaired ciliogenesis.;	EDTA Blood Sample
Kısa Boy, Optik Sinir Atrofisi ve Pelger-Huet Anomalisi, 614800, Otozomal Resesif; SOPH (Kısa Boy-Optik Atrofi-Pelger-Huet Anomali Sendromu) (NBAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800, Autosomal recessive; SOPH (Short stature-optic atrophy-Pelger-Huet anomaly syndrome) (NBAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NBAS	NBAS, NAG, SOPH, ILFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile liver failure syndrome 2 (ILFS2) [MIM:616483]: A form of infantile liver failure syndrome, a life-threatening disorder of hepatic function that manifests with acute liver failure in the first few months of life. Clinical features include anemia, renal tubulopathy, developmental delay, seizures, failure to thrive, and liver steatosis and fibrosis. [ECO:0000269	PubMed:26073778). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Boy Kısalığı, Rizomelik, Mikrosefali, Mikrognati ve Gelişme Geriliği, 617164, Otozomal Dominant; SRMMD (ARCN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164, Autosomal dominant; SRMMD (ARCN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARCN1	ARCN1, SRMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Boy Kısalığı, Rizomelik, Mikrosefali, Mikrognati ve Gelişme Geriliği, 617164, Otozomal Dominant; SRNMD (Prenatal) (ARCN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short stature, rhizomelic, with microcephaly, micrognathia, and developmental delay, 617164, Autosomal dominant; SRNMD (ARCN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARCN1	ARCN1, SRNMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SHORT Sendromu, 269880, Otozomal Dominant (SHORT Sendromu) (PIK3R1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	SHORT syndrome, 269880, Autosomal dominant (SHORT syndrome) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: SHORT syndrome (SHORTS) [MIM:269880]: A rare, multisystem disease characterized by short stature, anomalies of the anterior chamber of the eye, characteristic facial features such as triangular facies, lack of facial fat, and hypoplastic nasal alae with overhanging columella, partial lipodystrophy, hernias, hyperextensibility, and delayed dentition. The clinical phenotype can include insulin resistance, nephrocalcinosis, and hearing deficits. Developmental milestones and cognition are normal. {ECO:0000269|PubMed:23810378, ECO:0000269|PubMed:23810379, ECO:0000269|PubMed:23810382}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SHORT Sendromu, 269880, Otozomal Dominant (SHORT Sendromu) (Prenatal) (PIK3R1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	SHORT syndrome, 269880, Autosomal dominant (SHORT syndrome) (PIK3R1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PIK3R1	PIK3R1, GRB1, AGM7, SHORT, IMD36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: SHORT syndrome (SHORTS) [MIM:269880]: A rare, multisystem disease characterized by short stature, anomalies of the anterior chamber of the eye, characteristic facial features such as triangular facies, lack of facial fat, and hypoplastic nasal alae with overhanging columella, partial lipodystrophy, hernias, hyperextensibility, and delayed dentition. The clinical phenotype can include insulin resistance, nephrocalcinosis, and hearing deficits. Developmental milestones and cognition are normal. {ECO:0000269|PubMed:23810378, ECO:0000269|PubMed:23810379, ECO:0000269|PubMed:23810382}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi 10 ± Polidaktili, 615630, Otozomal Resesif; SRTD10 (Jeune Sendromu) (IFT172 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630, Autosomal recessive; SRTD10 (Jeune syndrome) (IFT172 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT172	IFT172, SLB, KIAA1179, SRTD10, RP71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 10 with or without polydactyly (SRTD10) [MIM:615630]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269|PubMed:24140113}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Kosta Torasik Displazi 10 ± Polidaktili, 615630, Otozomal Resesif; SRTD10 (Jeune Sendromu) (Prenatal) (IFT172 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 10 with or without polydactyly, 615630, Autosomal recessive; SRTD10 (Jeune syndrome) (IFT172 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT172	IFT172, SLB, KIAA1179, SRTD10, RP71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 10 with or without polydactyly (SRTD10) [MIM:615630]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269|PubMed:24140113}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 11, 615633, Otozomal Resesif; SRTD11 (Jeune Sendromu) (WDR34 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633, Autosomal recessive; SRTD11 (Jeune syndrome) (WDR34 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR34	WDR34, SRTD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 11, 615633, Otozomal Resesif; SRTD11 (Jeune Sendromu) (Prenatal) (WDR34 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 11 with or without polydactyly, 615633, Autosomal recessive; SRTD11 (Jeune syndrome) (WDR34 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR34	WDR34, SRTD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 13, 616300, Otozomal Resesif (Jeune Sendromu) (CEP120 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300, Autosomal recessive (Jeune syndrome) (CEP120 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEP120	CEP120, CCDC100, SRTD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 13 with or without polydactyly (SRTD13) [MIM:616300]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269|PubMed:25361962}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 13, 616300, Otozomal Resesif (Jeune Sendromu) (Prenatal) (CEP120 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 13 with or without polydactyly, 616300, Autosomal recessive (Jeune syndrome) (CEP120 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CEP120	CEP120, CCDC100, SRTD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 13 with or without polydactyly (SRTD13) [MIM:616300]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. {ECO:0000269|PubMed:25361962}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi 14-Polidaktili, 616546, Otozomal Resesif (Joubert Sendromu-Jeune Asfiktik Torasik Distrofi) (KIAA0586 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 14 with polydactyly, 616546, Autosomal recessive (Joubert syndrome with Jeune asphyxiating thoracic dystrophy) (KIAA0586 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIAA0586	KIAA0586, TALPID3, JBTS23, SRTD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Joubert syndrome 23 (JBTS23) [MIM:616490]: A mild form of Joubert syndrome, a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269|PubMed:26096313}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Kısa Kosta Torasik Displazi 14-Polidaktili, 616546, Otozomal Resesif (Joubert Sendromu-Jeune Asfiktik Torasik Distrofi) (Prenatal) (KIAA0586 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 14 with polydactyly, 616546, Autosomal recessive (Joubert syndrome with Jeune asphyxiating thoracic dystrophy) (KIAA0586 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIAA0586	KIAA0586, TALPID3, JBTS23, SRTD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Joubert syndrome 23 (JBTS23) [MIM:616490]: A mild form of Joubert syndrome, a disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy, renal disease, liver fibrosis, and polydactyly. {ECO:0000269|PubMed:26096313}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 16, 617102, Otozomal Resesif (Kraniyoektodermal Displazi 4) (IFT52 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102, Autosomal recessive (Cranioectodermal dysplasia) (IFT52 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT52	IFT52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 16, 617102, Otozomal Resesif (Kraniyoektodermal Displazi 4) (Prenatal) (IFT52 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 16 with or without polydactyly, 617102, Autosomal recessive (Cranioectodermal dysplasia) (IFT52 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT52	IFT52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 2, 611263, Otozomal Resesif; SRTD2 (Jeune Sendromu) (IFT80 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263, Autosomal recessive; SRTD2 (Jeune syndrome) (IFT80 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT80	IFT80, KIAA1374, WDR56, SRTD2, ATD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 2, 611263, Otozomal Resesif; SRTD2 (Jeune Sendromu) (Prenatal) (IFT80 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263, Autosomal recessive; SRTD2 (Jeune syndrome) (IFT80 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT80	IFT80, KIAA1374, WDR56, SRTD2, ATD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Kısa Kosta Torasik Displazi ± Polidaktili 3, 613091, Otozomal Resesif, Dijenik Resesif; SRTD3 (Jeune Sendromu) (DYNC2H1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091, Autosomal recessive, Digenic recessive; SRTD3 (Jeune syndrome) (DYNC2H1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYNC2H1	DYNC2H1, DNCH2, DHC2, ATD3, SRPS2B, SRTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 3 with or without polydactyly (SRTD3) [MIM:613091]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269|PubMed:19361615, ECO:0000269|PubMed:19442771, ECO:0000269|PubMed:22499340, ECO:0000269|PubMed:23456818). Note=The disease is caused by mutations affecting the gene represented in this entry. In some cases DYNC2H1 mutations result in disease phenotype in the presence of mutations in NEK1 indicating digenic inheritance (digenic short rib-polydactyly syndrome 3/6 with polydactyly) (PubMed:21211617). (ECO:0000269|PubMed:21211617).;	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 3, 613091, Otozomal Resesif, Dijenik Resesif; SRTD3 (Jeune Sendromu) (Prenatal) (DYNC2H1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091, Autosomal recessive, Digenic recessive; SRTD3 (Jeune syndrome) (DYNC2H1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYNC2H1	DYNC2H1, DNCH2, DHC2, ATD3, SRPS2B, SRTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 3 with or without polydactyly (SRTD3) [MIM:613091]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269|PubMed:19361615, ECO:0000269|PubMed:19442771, ECO:0000269|PubMed:22499340, ECO:0000269|PubMed:23456818). Note=The disease is caused by mutations affecting the gene represented in this entry. In some cases DYNC2H1 mutations result in disease phenotype in the presence of mutations in NEK1 indicating digenic inheritance (digenic short rib-polydactyly syndrome 3/6 with polydactyly) (PubMed:21211617). (ECO:0000269|PubMed:21211617).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 4 ± Polidaktili, 613819, Otozomal Resesif; SRTD4 (Jeune Sendromu) (TTC21B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819, Autosomal recessive; SRTD4 (Jeune syndrome) (TTC21B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC21B	TTC21B, THM1, NPHP12, SRTD4, ATD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:21258341). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 4 ± Polidaktili, 613819, Otozomal Resesif; SRTD4 (Jeune Sendromu) (Prenatal) (TTC21B gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 4 with or without polydactyly, 613819, Autosomal recessive; SRTD4 (Jeune syndrome) (TTC21B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTC21B	TTC21B, THM1, NPHP12, SRTD4, ATD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bardet-Biedl syndrome (BBS) [MIM:209900]: A syndrome characterized by usually severe pigmentary retinopathy, early- onset obesity, polydactyly, hypogenitalism, renal malformation and mental retardation. Secondary features include diabetes mellitus, hypertension and congenital heart disease. Bardet-Biedl syndrome inheritance is autosomal recessive, but three mutated alleles (two at one locus, and a third at a second locus) may be required for clinical manifestation of some forms of the disease. (ECO:0000269	PubMed:21258341). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi 5 ± Polidaktili, 614376, Otozomal Resesif; SRTD5 (Jeune Sendromu) (WDR19 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 5 with or without polydactyly, 614376, Autosomal recessive; SRTD5 (Jeune syndrome) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269	PubMed:22019273). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Kosta Torasik Displazi 5 ± Polidaktili, 614376, Otozomal Resesif; SRTD5 (Jeune Sendromu) (Prenatal) (WDR19 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 5 with or without polydactyly, 614376, Autosomal recessive; SRTD5 (Jeune syndrome) (WDR19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR19	WDR19, SRTD5, ATD5, NPHP13, CED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 5 with or without polydactyly (SRTD5) [MIM:614376]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. (ECO:0000269	PubMed:22019273). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 6, 263520, Otozomal Resesif, Dijenik Resesif; SRTD6 (Kısa Kosta Polidaktili Sendromu, Majewski Tipi) (NEK1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520, Autosomal recessive, Digenic recessive; SRTD6 (Short rib-polydactyly syndrome, Majewski type) (NEK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEK1	NEK1, SRTD6, SRPS2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 6, 263520, Otozomal Resesif, Dijenik Resesif; SRTD6 (Kısa Kosta Polidaktili Sendromu, Majewski Tipi) (Prenatal) (NEK1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520, Autosomal recessive, Digenic recessive; SRTD6 (Short rib-polydactyly syndrome, Majewski type) (NEK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEK1	NEK1, SRTD6, SRPS2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 7 ± Polidaktili, 614091, Otozomal Resesif; SRTD7 (Kısa Kosta Polidaktili Sendromu, Verma-Naumoff Tipi) (WDR35 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091, Autosomal recessive; SRTD7 (Short rib-polydactyly syndrome, Verma-Naumoff type) (WDR35 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR35	WDR35, NAOFEN, KIAA1336, CED2, SRTD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cranioectodermal dysplasia 2 (CED2) [MIM:613610]: A disorder characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include short stature, dolichocephaly, craniosynostosis, narrow thorax with pectus excavatum, short limbs, brachydactyly, joint laxity, narrow palpebral fissures, telecanthus with hypertelorism, low-set simple ears, everted lower lip, and short neck. Teeth abnormalities include widely spaced, hypoplastic and fused teeth. (ECO:0000269	PubMed:20817137). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kısa Kosta Torasik Displazi 7 ± Polidaktili, 614091, Otozomal Resesif; SRTD7 (Kısa Kosta Polidaktili Sendromu, Verma-Naumoff Tipi) (Prenatal) (WDR35 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091, Autosomal recessive; SRTD7 (Short rib-polydactyly syndrome, Verma-Naumoff type) (WDR35 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR35	WDR35, NAOFEN, KIAA1336, CED2, SRTD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cranioectodermal dysplasia 2 (CED2) [MIM:613610]: A disorder characterized by craniofacial, skeletal and ectodermal abnormalities. Clinical features include short stature, dolichocephaly, craniosynostosis, narrow thorax with pectus excavatum, short limbs, brachydactyly, joint laxity, narrow palpebral fissures, telecanthus with hypertelorism, low-set simple ears, everted lower lip, and short neck. Teeth abnormalities include widely spaced, hypoplastic and fused teeth. (ECO:0000269	PubMed:20817137). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi 8 ± Polidaktili, 615503, Otozomal Resesif; SRTD8 (Kısa Kosta Polidaktili Sendromu, Verma-Naumoff Tipi) (WDR60 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503, Autosomal recessive; SRTD8 (Short rib-polydactyly syndrome, Verma-Naumoff type) (WDR60 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WDR60	WDR60, SRTD8, SRPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi 8 ± Polidaktili, 615503, Otozomal Resesif; SRTD8 (Kısa Kosta Polidaktili Sendromu, Verma-Naumoff Tipi) (Prenatal) (WDR60 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503, Autosomal recessive; SRTD8 (Short rib-polydactyly syndrome, Verma-Naumoff type) (WDR60 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WDR60	WDR60, SRTD8, SRPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi ± Polidaktili 9, 266920, Otozomal Resesif; SRTD9 (Saldino-Mainzer Sendromu) (IFT140 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920, Autosomal recessive; SRTD9 (Saldino-Mainzer syndrome) (IFT140 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFT140	IFT140, KIAA0590, SRTD9, MZSDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Short-rib thoracic dysplasia 9 with or without polydactyly (SRTD9) [MIM:266920]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. SRTD9 is characterized by phalangeal cone-shaped epiphyses, chronic renal disease, nearly constant retinal dystrophy, and mild radiographic abnormality of the proximal femur. Occasional features include short stature, cerebellar ataxia, and hepatic fibrosis. (ECO:0000269|PubMed:22503633, ECO:0000269|PubMed:23418020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Kısa Kosta Torasik Displazi ± Polidaktili 9, 266920, Otozomal Resesif; SRTD9 (Saldino-Mainzer Sendromu) (Prenatal) (IFT140 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920, Autosomal recessive; SRTD9 (Saldino-Mainzer syndrome) (IFT140 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFT140	IFT140, KIAA0590, SRTD9, MZSDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Short-rib thoracic dysplasia 9 with or without polydactyly (SRTD9) [MIM:266920]: A form of short-rib thoracic dysplasia, a group of autosomal recessive ciliopathies that are characterized by a constricted thoracic cage, short ribs, shortened tubular bones, and a 'trident' appearance of the acetabular roof. Polydactyly is variably present. Non-skeletal involvement can include cleft lip/palate as well as anomalies of major organs such as the brain, eye, heart, kidneys, liver, pancreas, intestines, and genitalia. Some forms of the disease are lethal in the neonatal period due to respiratory insufficiency/ secondary to a severely restricted thoracic cage, whereas others are compatible with life. Disease spectrum encompasses Ellis-van Creveld syndrome, asphyxiating thoracic dystrophy (Jeune syndrome), Mainzer-Saldino syndrome, and short rib-polydactyly syndrome. SRTD9 is characterized by phalangeal cone-shaped epiphyses, chronic renal disease, nearly constant retinal dystrophy, and mild radiographic abnormality of the proximal femur. Occasional features include short stature, cerebellar ataxia, and hepatic fibrosis. (ECO:0000269|PubMed:22503633, ECO:0000269|PubMed:23418020). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kısa Kosta Torasik Displazi- Polidaktili 15, 617088, Otozomal Resesif (Jeune Sendromu) (DYNC2LI1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Short-rib thoracic dysplasia 15 with polydactyly, 617088, Autosomal recessive (Jeune syndrome) (DYNC2LI1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYNC2LI1	DYNC2LI1, D2LIC, LIC3, SRTD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kısa Kosta Torasik Displazi- Polidaktili 15, 617088, Otozomal Resesif (Jeune Sendromu) (Prenatal) (DYNC2LI1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Short-rib thoracic dysplasia 15 with polydactyly, 617088, Autosomal recessive (Jeune syndrome) (DYNC2LI1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYNC2LI1	DYNC2LI1, D2LIC, LIC3, SRTD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SHOX Xp22.33/ Yp11.32 (FISH)	SHOX Xp22.33/ Yp11.32 (FISH)	Xp22.33	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Shprintzen-Goldberg Sendromu, 182212, Otozomal Dominant; SGS (Shprintzen-Goldberg Sendromu) (SKI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Shprintzen-Goldberg syndrome, 182212, Autosomal dominant; SGS (Shprintzen-Goldberg syndrome) (SKI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SKI	SKI, SGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Shprintzen-Goldberg craniosynostosis syndrome (SGS) [MIM:182212]: A very rare syndrome characterized by a marfanoid habitus, craniosynostosis, characteristic dysmorphic facial features, skeletal and cardiovascular abnormalities, mental retardation, developmental delay and learning disabilities. (ECO:0000269|PubMed:23023332, ECO:0000269|PubMed:23103230, ECO:0000269|PubMed:24357594, ECO:0000269|PubMed:24736733). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Shprintzen-Goldberg Sendromu, 182212, Otozomal Dominant; SGS (Shprintzen-Goldberg Sendromu) (SKI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Shprintzen-Goldberg syndrome, 182212, Autosomal dominant; SGS (Shprintzen-Goldberg syndrome) (SKI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SKI	SKI, SGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Shprintzen-Goldberg craniosynostosis syndrome (SGS) [MIM:182212]: A very rare syndrome characterized by a marfanoid habitus, craniosynostosis, characteristic dysmorphic facial features, skeletal and cardiovascular abnormalities, mental retardation, developmental delay and learning disabilities. (ECO:0000269|PubMed:23023332, ECO:0000269|PubMed:23103230, ECO:0000269|PubMed:24357594, ECO:0000269|PubMed:24736733). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Shwachman-Diamond Sendromu, 260400, Otozomal Resesif; SDS (Shwachman-Diamond Sendromu) (SBDS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Shwachman-Diamond syndrome, 260400, Autosomal recessive; SDS (Shwachman-Diamond syndrome) (SBDS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Shwachman-Diamond Sendromu, 260400, Otozomal Resesif; SDS (Shwachman-Diamond Sendromu) (Prenatal) (SBDS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Shwachman-Diamond syndrome, 260400, Autosomal recessive; SDS (Shwachman-Diamond syndrome) (SBDS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SBDS	SBDS, SDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sialik Asit Depo Hastalığı, İnfantil, 269920, Otozomal Resesif (Serbest Sialik Asit Depo Hastalığı) (SLC17A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sialic acid storage disorder, infantile, 269920, Autosomal recessive (Free sialic acid storage disease) (SLC17A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC17A5	SLC17A5, SIASD, SLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile sialic acid storage disorder (ISSD) [MIM:269920]: Severe form of sialic acid storage disease. Affected newborns exhibit visceromegaly, coarse features and failure to thrive immediately after birth. These patients have a shortened life span, usually less than 2 years. (ECO:0000269|PubMed:10581036, ECO:0000269|PubMed:10947946). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sialik Asit Depo Hastalığı, İnfantil, 269920, Otozomal Resesif (Serbest Sialik Asit Depo Hastalığı) (Prenatal) (SLC17A5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sialic acid storage disorder, infantile, 269920, Autosomal recessive (Free sialic acid storage disease) (SLC17A5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC17A5	SLC17A5, SIASD, SLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile sialic acid storage disorder (ISSD) [MIM:269920]: Severe form of sialic acid storage disease. Affected newborns exhibit visceromegaly, coarse features and failure to thrive immediately after birth. These patients have a shortened life span, usually less than 2 years. (ECO:0000269|PubMed:10581036, ECO:0000269|PubMed:10947946). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sialidosis, Tip 1, 256550, Otozomal Resesif (Sialidoz Tip 1) (NEU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sialidosis, type I, 256550, Autosomal recessive (Sialidosis type 1) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insidious visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. (ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sialidosis, Tip 1, 256550, Otozomal Resesif (Sialidoz Tip 1) (Prenatal) (NEU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sialidosis, type I, 256550, Autosomal recessive (Sialidosis type 1) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insidious visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. (ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sialidosis, Tip 2, 256550, Otozomal Resesif (Sialidozis Tip 2) (NEU1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sialidosis, type II, 256550, Autosomal recessive (Sialidosis type 2) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insidious visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. (ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sialidosis, Tip 2, 256550, Otozomal Resesif (Sialidozis Tip 2) (Prenatal) (NEU1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sialidosis, type II, 256550, Autosomal recessive (Sialidosis type 2) (NEU1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NEU1	NEU1, NEU, SIAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sialidosis (SIALIDOSIS) [MIM:256550]: Lysosomal storage disease occurring as two types with various manifestations. Type 1 sialidosis (cherry red spot-myoclonus syndrome or normosomatic type) is late-onset and it is characterized by the formation of cherry red macular spots in childhood, progressive debilitating myoclonus, insidious visual loss and rarely ataxia. The diagnosis can be confirmed by the screening of the urine for sialyloligosaccharides. Type 2 sialidosis (also known as dysmorphic type) occurs as several variants of increasing severity with earlier age of onset. It is characterized by the presence of abnormal somatic features including coarse facies and dysostosis multiplex, vertebral deformities, mental retardation, cherry-red spot/myoclonus, sialuria, cytoplasmic vacuolation of peripheral lymphocytes, bone marrow cells and conjunctival epithelial cells. (ECO:0000269|PubMed:10944856, ECO:0000269|PubMed:11063730, ECO:0000269|PubMed:11279074, ECO:0000269|PubMed:11829139, ECO:0000269|PubMed:14695530, ECO:0000269|PubMed:8985184, ECO:0000269|PubMed:9054950). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Sialüri, 269921, Otozomal Dominant (Sialüri) (GNE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sialuria, 269921, Autosomal dominant (Sialuria) (GNE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNE	GNE, GLCNE, IBM2, DMRV, NM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sialuria (SIALURIA) [MIM:269921]: In sialuria, free sialic acid accumulates in the cytoplasm and gram quantities of neuraminic acid are secreted in the urine. The metabolic defect involves lack of feedback inhibition of UDP-GlcNAc 2-epimerase by CMP-NeuSAc, resulting in constitutive overproduction of free NeuSAc. Clinical features include variable degrees of developmental delay, coarse facial features and hepatomegaly. Sialuria inheritance is autosomal dominant. {ECO:0000269|PubMed:10330343, ECO:0000269|PubMed:10356312, ECO:0000269|PubMed:2808337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sialüri, 269921, Otozomal Dominant (Sialüri) (Prenatal) (GNE geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sialuria, 269921, Autosomal dominant (Sialuria) (GNE gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GNE	GNE, GLCNE, IBM2, DMRV, NM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Sialuria (SIALURIA) [MIM:269921]: In sialuria, free sialic acid accumulates in the cytoplasm and gram quantities of neuraminic acid are secreted in the urine. The metabolic defect involves lack of feedback inhibition of UDP-GlcNAc 2-epimerase by CMP-NeuSAc, resulting in constitutive overproduction of free NeuSAc. Clinical features include variable degrees of developmental delay, coarse facial features and hepatomegaly. Sialuria inheritance is autosomal dominant. {ECO:0000269|PubMed:10330343, ECO:0000269|PubMed:10356312, ECO:0000269|PubMed:2808337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Hasta Sinüs Sendromu 1, 608567, Otozomal Resesif; SSS1 (Ailesel Hasta Sinüs Sendromu) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sick sinus syndrome 1, 608567, Autosomal recessive; SSS1 (Familial sick sinus syndrome) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial standstill 1 (ATRST1) [MIM:108770]: A rare arrhythmia characterized by the absence of electrical and mechanical activity in the atria. Electrocardiographically, it is characterized by bradycardia, the absence of P waves, and a junctional narrow complex escape rhythm. {ECO:0000269|PubMed:12522116}. Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry. A mutation in SCN5A has been detected in combination with a rare GJA5 genotype in a large family with atrial standstill.	EDTA Blood Sample
Hasta Sinüs Sendromu 2, 163800, Otozomal Dominant (Ailesel Hasta Sinüs Sendromu) (HCN4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sick sinus syndrome 2, 163800, Autosomal dominant (Familial sick sinus syndrome) (HCN4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HCN4	HCN4, SSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sick sinus syndrome 2 (SSS2) [MIM:163800]: The term 'sick sinus syndrome' encompasses a variety of conditions caused by sinus node dysfunction. The most common clinical manifestations are syncope, presyncope, dizziness, and fatigue. Electrocardiogram typically shows sinus bradycardia, sinus arrest, and/or sinoatrial block. Episodes of atrial tachycardias coexisting with sinus bradycardia (tachycardia-bradycardia syndrome) are also common in this disorder. SSS occurs most often in the elderly associated with underlying heart disease or previous cardiac surgery, but can also occur in the fetus, infant, or child without heart disease or other contributing factors. SSS2 onset is in utero or at birth. {ECO:0000269|PubMed:15123648, ECO:0000269|PubMed:16407510, ECO:0000269|PubMed:20662977}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Hasta Sinüs Sendromu 3, 614090; SSS3 (Ailesel Hasta Sinüs Sendromu) (MYH6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sick sinus syndrome 3, 614090; SSS3 (Familial sick sinus syndrome) (MYH6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYH6	MYH6, ASD3, MYHCA, CMD1EE, CMH14, SSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, familial hypertrophic 14 (CMH14) [MIM:613251]: A hereditary heart disorder characterized by ventricular hypertrophy, which is usually asymmetric and often involves the interventricular septum. The symptoms include dyspnea, syncope, collapse, palpitations, and chest pain. They can be readily provoked by exercise. The disorder has inter- and intrafamilial variability ranging from benign to malignant forms with high risk of cardiac failure and sudden cardiac death. {ECO:0000269|PubMed:11815426, ECO:0000269|PubMed:15998695}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sickle Cell Anemia, Orak Hücreli Anemi (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sickle cell anemia (GLU6VAL) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sickle Cell Anemia, Orak Hücreli Anemi (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sickle cell anemia (GLU6VAL) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Orak Hücreli Anemi, 603903, Otozomal Resesif (Orak Hücre Anemisi) (HBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sickle cell anemia, 603903, Autosomal recessive (Sickle cell anemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Orak Hücreli Anemi, 603903, Otozomal Resesif (Orak Hücre Anemisi) (Prenatal) (HBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sickle cell anemia, 603903, Autosomal recessive (Sickle cell anemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sideroblastik Anemi-B Hücre İmmün Yetmezlik, Periyodik Ateşler ve Gelişimsel Gecikme, 616084, Otozomal Resesif; SIFD (Konjenital Sideroblastik Anemi-B-Hücre İmmün Yetmezliği-Periyodik Ateş-Gelişimsel Gecikme Sendromu) (TRNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084, Autosomal recessive; SIFD (Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome) (TRNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRNT1	TRNT1, SIFD, RPEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD) [MIM:616084]: An autosomal recessive disease characterized by severe sideroblastic anemia with onset in the neonatal period or infancy, recurrent periodic fevers without an infectious etiology, B-cell lymphopenia and hypogammaglobulinemia. Affected individuals show delayed psychomotor development with variable neurodegeneration. Additional variable features include sensorineural hearing loss, retinitis pigmentosa, nephrocalcinosis, and cardiomyopathy. {ECO:0000269|PubMed:25193871}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Sideroblastik Anemi-B Hücre İmmün Yetmezlik, Periyodik Ateşler ve Gelişimsel Gecikme, 616084, Otozomal Resesif; SIFD (Konjenital Sideroblastik Anemi-B-Hücre İmmün Yetmezliği-Periyodik Ateş-Gelişimsel Gecikme Sendromu) (Prenatal) (TRNT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay, 616084, Autosomal recessive; SIFD (Congenital sideroblastic anemia-B-cell immunodeficiency-periodic fever-developmental delay syndrome) (TRNT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRNT1	TRNT1, SIFD, RPEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD) [MIM:616084]: An autosomal recessive disease characterized by severe sideroblastic anemia with onset in the neonatal period or infancy, recurrent periodic fevers without an infectious etiology, B-cell lymphopenia and hypogammaglobulinemia. Affected individuals show delayed psychomotor development with variable neurodegeneration. Additional variable features include sensorineural hearing loss, retinitis pigmentosa, nephrocalcinosis, and cardiomyopathy. {ECO:0000269|PubMed:25193871}. Note=The disease is caused by mutations affecting the gene represented in this entry..	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sifrim-Hitz-Weiss Sendromu, 617159, Otozomal Dominant; SIHIWES (CHD4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sifrim-Hitz-Weiss syndrome, 617159, Autosomal dominant; SIHIWES (CHD4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHD4	CHD4, SIHIWES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Sifrim-Hitz-Weiss Sendromu, 617159, Otozomal Dominant; SIHIWES (Prenatal) (CHD4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sifrim-Hitz-Weiss syndrome, 617159, Autosomal dominant; SIHIWES (CHD4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHD4	CHD4, SIHIWES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Silver Spastik Paraplejі Sendromu, 270685, Otozomal Dominant; SPG17 (Otozomal Dominant Spastik Paraplejі Tip 17) (BSCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Silver spastic paraplegia syndrome, 270685, Autosomal dominant; SPG17 (Autosomal dominant spastic paraplegia type 17) (BSCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BSCL2	BSCL2, SPG17, HMN5, PELD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 17, autosomal dominant (SPG17) [MIM:270685]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG17 is characterized by prominent amyotrophy of the hand muscles, the presence of mild to severe pyramidal tract signs and spastic paraplegia. SPG17 is a motor neuron disease overlapping with distal spinal muscular atrophy type 5. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Silver-Russell Sendromu, 180860, Sporadik Vakalar; SRS (Silver-Russell Sendromu) (H19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Silver-Russell syndrome, 180860, Isolated cases; SRS (Silver-Russell syndrome) (H19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Silver-Russell Sendromu, 180860, Sporadik Vakalar; SRS (Silver-Russell Sendromu) (MLPA)	Silver-Russell syndrome, 180860, Isolated cases; SRS (Silver-Russell syndrome) (MLPA)	H19	H19, D11S813E, ASM1, BWS, WT2	MLPA (CNV + Metilasyon)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Silver-Russell Sendromu, 180860, Sporadik Vakalar; SRS (Silver-Russell Sendromu) (Prenatal) (H19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Silver-Russell syndrome, 180860, Isolated cases; SRS (Silver-Russell syndrome) (H19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Silver-Russell Sendromu, 180860, Sporadik Vakalar; SRS (Silver-Russell Sendromu) (Prenatal) (MLPA)	Silver-Russell syndrome, 180860, Isolated cases; SRS (Silver-Russell syndrome) (Prenatal) (MLPA)	H19	H19, D11S813E, ASM1, BWS, WT2	MLPA (CNV + Metilasyon)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Simpson-Golabi-Behmel sendromu (GPC3, GPC4, Xq26) (MLPA)	Simpson-Golabi-Behmel syndrome (GPC3, GPC4, Xq26) (MLPA)	GPC3, GPC4, Xq26	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Simpson-Golabi-Behmel sendromu (GPC3, GPC4, Xq26) (MLPA) (Prenatal)	Simpson-Golabi-Behmel syndrome (GPC3, GPC4, Xq26) (MLPA) (Prenatal)	GPC3, GPC4, Xq26	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 1, 312870, X'e Bağlı Resesif; SGBS1 (Simpson-Golabi-Behmel Sendromu) (GPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Simpson-Golabi-Behmel syndrome, type 1, 312870, X-linked recessive; SGBS1 (Simpson-Golabi-Behmel syndrome) (GPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPC3	GPC3, SDYS, SGBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 1, 312870, X'e Bağlı Resesif; SGBS1 (Simpson-Golabi-Behmel Sendromu) (MLPA)	Simpson-Golabi-Behmel syndrome, type 1, 312870, X-linked recessive; SGBS1 (Simpson-Golabi-Behmel syndrome) (MLPA)	GPC3	GPC3, SDYS, SGBS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 1, 312870, X'e Bağlı Resesif; SGBS1 (Simpson-Golabi-Behmel Sendromu) (Prenatal) (GPC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Simpson-Golabi-Behmel syndrome, type 1, 312870, X-linked recessive; SGBS1 (Simpson-Golabi-Behmel syndrome) (GPC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPC3	GPC3, SDYS, SGBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 1, 312870, X'e Bağlı Resesif; SGBS1 (Simpson-Golabi-Behmel Sendromu) (Prenatal) (MLPA)	Simpson-Golabi-Behmel syndrome, type 1, 312870, X-linked recessive; SGBS1 (Simpson-Golabi-Behmel syndrome) (Prenatal) (MLPA)	GPC3	GPC3, SDYS, SGBS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 2, 300209, X'e Bağlı Resesif; SGBS2 (Simpson Golabi-Behmel Sendromu Tip 2) (OFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Simpson-Golabi-Behmel syndrome, type 2, 300209, X-linked recessive; SGBS2 (Simpson-Golabi-Behmel syndrome type 2) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. (ECO:0000269	PubMed:16783569). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Simpson Golabi-Behmel Sendromu, Tip 2, 300209, X'e Bağlı Resesif; SGBS2 (Simpson Golabi-Behmel Sendromu Tip 2) (Prenatal) (OFD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Simpson-Golabi-Behmel syndrome, type 2, 300209, X-linked recessive; SGBS2 (Simpson-Golabi-Behmel syndrome type 2) (OFD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OFD1	OFD1, CXorf5, SGBS2, JBTS10, RP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Simpson-Golabi-Behmel syndrome 2 (SGBS2) [MIM:300209]: A severe variant of Simpson-Golabi-Behmel syndrome, a condition characterized by pre- and postnatal overgrowth (gigantism), facial dysmorphism and a variety of inconstant visceral and skeletal malformations. (ECO:0000269	PubMed:16783569). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tek Orta Maksiller Kesici, 147250, Otozomal Dominant; SMMCI (Holoprozensefali) (SHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Single median maxillary central incisor, 147250, Autosomal dominant; SMMCI (Holoprosencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Tek Orta Maksiller Kesici, 147250, Otozomal Dominant; SMMCI (Holoprozensefali) (MLPA)	Single median maxillary central incisor, 147250, Autosomal dominant; SMMCI (Holoprosencephaly) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	EDTA Blood Sample
Tek Orta Maksiller Kesici, 147250, Otozomal Dominant; SMMCI (Holoprozensefali) (Prenatal) (SHH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Single median maxillary central incisor, 147250, Autosomal dominant; SMMCI (Holoprosencephaly) (SHH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tek Orta Maksiller Kesici, 147250, Otozomal Dominant; SMMCI (Holoprozensefali) (Prenatal) (MLPA)	Single median maxillary central incisor, 147250, Autosomal dominant; SMMCI (Holoprosencephaly) (Prenatal) (MLPA)	SHH	SHH, HPE3, HLP3, SMMCI, MCOPCB5	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269	PubMed:24456159). Note=The gene represented in this entry is involved in disease pathogenesis. Abnormal SHH limb expression with pathological consequences is caused by duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis-regulatory element, located in LMBR1 intron 5 (PubMed:24456159).;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Singleton-Merten Sendromu 1, 182250, Otozomal Dominant; SGMRT1 (Singleton-Merten Displazi) (IFIH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Singleton-Merten syndrome 1, 182250, Autosomal dominant; SGMRT1 (Singleton-Merten dysplasia) (IFIH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFIH1	IFIH1, MDA5, AGS7, SGMRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Singleton-Merten syndrome 1 (SGMRT1) [MIM:182250]: An autosomal dominant disorder with variable expression. Core features are marked aortic calcification, dental anomalies, osteopenia, acro-osteolysis, and to a lesser extend glaucoma, psoriasis, muscle weakness, and joint laxity. Dental anomalies include delayed eruption and immature root formation of anterior permanent teeth, early loss of permanent teeth due to short roots, acute root resorption, high caries, and aggressive alveolar bone loss. Additional clinical manifestations include particular facial characteristics and abnormal joint and muscle ligaments. (ECO:0000269	PubMed:25620204). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Singleton-Merten Sendromu 1, 182250, Otozomal Dominant; SGMRT1 (Singleton-Merten Displazi) (Prenatal) (IFIH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Singleton-Merten syndrome 1, 182250, Autosomal dominant; SGMRT1 (Singleton-Merten dysplasia) (IFIH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IFIH1	IFIH1, MDA5, AGS7, SGMRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Singleton-Merten syndrome 1 (SGMRT1) [MIM:182250]: An autosomal dominant disorder with variable expression. Core features are marked aortic calcification, dental anomalies, osteopenia, acro-osteolysis, and to a lesser extend glaucoma, psoriasis, muscle weakness, and joint laxity. Dental anomalies include delayed eruption and immature root formation of anterior permanent teeth, early loss of permanent teeth due to short roots, acute root resorption, high caries, and aggressive alveolar bone loss. Additional clinical manifestations include particular facial characteristics and abnormal joint and muscle ligaments. (ECO:0000269	PubMed:25620204). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Singleton-Merten Sendromu 2, 616298, Otozomal Dominant; SGMRT2 (Singleton-Merten Displazi) (DDX58 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Singleton-Merten syndrome 2, 616298, Autosomal dominant; SGMRT2 (Singleton-Merten dysplasia) (DDX58 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDX58	DDX58, RIGI, SGMRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Singleton-Merten syndrome 2 (SGMRT2) [MIM:616298]: A form of Singleton-Merten syndrome, an autosomal dominant disorder characterized by marked aortic calcification, dental anomalies, osteopenia, acro-osteolysis, and to a lesser extend glaucoma, psoriasis, muscle weakness, and joint laxity. Additional clinical manifestations include particular facial characteristics and abnormal joint and muscle ligaments. SGMRT2 is an atypical form characterized by variable expression of glaucoma, aortic calcification, and skeletal abnormalities, without dental anomalies. (ECO:0000269	PubMed:25620203). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Singleton-Merten Sendromu 2, 616298, Otozomal Dominant; SGMRT2 (Singleton-Merten Displazi) (Prenatal) (DDX58 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Singleton-Merten syndrome 2, 616298, Autosomal dominant; SGMRT2 (Singleton-Merten dysplasia) (DDX58 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDX58	DDX58, RIGI, SGMRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Singleton-Merten syndrome 2 (SGMRT2) [MIM:616298]: A form of Singleton-Merten syndrome, an autosomal dominant disorder characterized by marked aortic calcification, dental anomalies, osteopenia, acro-osteolysis, and to a lesser extend glaucoma, psoriasis, muscle weakness, and joint laxity. Additional clinical manifestations include particular facial characteristics and abnormal joint and muscle ligaments. SGMRT2 is an atypical form characterized by variable expression of glaucoma, aortic calcification, and skeletal abnormalities, without dental anomalies. (ECO:0000269	PubMed:25620203). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sinoatriyal Nodu Disfonksiyonu ve Sağırlık, 614896, Otozomal Resesif; SANDD (Sinoatriyal Düğüm Disfonksiyonu ve Sağırlık) (CACNA1D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sinoatrial node dysfunction and deafness, 614896, Autosomal recessive; SANDD (Sinoatrial node dysfunction and deafness) (CACNA1D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1D	CACNA1D, CACNL1A2, CCHL1A2, SANDD, PASNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sinoatrial node dysfunction and deafness (SANDD) [MIM:614896]: A disease characterized by congenital severe to profound deafness without vestibular dysfunction, associated with episodic syncope due to intermittent pronounced bradycardia. (ECO:0000269	PubMed:21319563). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sitosterolemi, 210250, Otozomal Resesif (Sitosterolemi) (ABCG8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sitosterolemia, 210250, Autosomal recessive (Sitosterolemia) (ABCG8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCG8	ABCG8, GBD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sitosterolemia (STSL) [MIM:210250]: Rare autosomal recessive disorder characterized by increased intestinal absorption of all sterols including cholesterol, plant and shellfish sterols, and decreased biliary excretion of dietary sterols into bile. Sitosterolemia patients have hypercholesterolemia, very high levels of plant sterols in the plasma, and frequently develop tendon and tuberous xanthomas, accelerated atherosclerosis and premature coronary artery disease. (ECO:0000269|PubMed:11099417, ECO:0000269|PubMed:11452259). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sitosterolemi, 210250, Otozomal Resesif (Sitosterolemi) (ABCG5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sitosterolemia, 210250, Autosomal recessive (Sitosterolemia) (ABCG5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCG5	ABCG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sjogren-Larsson Sendromu, 270200, Otozomal Resesif; SLS (Sjögren-Larsson Sendromu) (ALDH3A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sjogren-Larsson syndrome, 270200, Autosomal recessive; SLS (Sjögren-Larsson syndrome) (ALDH3A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH3A2	ALDH3A2, ALDH10, SLS, FALDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spegren-Larsson syndrome (SLS) [MIM:270200]: An autosomal recessive neurocutaneous disorder characterized by a combination of severe mental retardation, spastic di- or tetraplegia and congenital ichthyosis. Ichthyosis is usually evident at birth with varying degrees of erythema and scaling, neurologic symptoms appear in the first or second year of life. Most patients have an IQ of less than 60. Additional clinical features include glistening white spots on the retina, seizures, short stature and speech defects. {ECO:0000269|PubMed:10577908, ECO:0000269|PubMed:10792573, ECO:0000269|PubMed:8528251, ECO:0000269|PubMed:9254849, ECO:0000269|PubMed:9829906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sjogren-Larsson Sendromu, 270200, Otozomal Resesif; SLS (Sjögren-Larsson Sendromu) (Prenatal) (ALDH3A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sjogren-Larsson syndrome, 270200, Autosomal recessive; SLS (Sjögren-Larsson syndrome) (ALDH3A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH3A2	ALDH3A2, ALDH10, SLS, FALDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Spegren-Larsson syndrome (SLS) [MIM:270200]: An autosomal recessive neurocutaneous disorder characterized by a combination of severe mental retardation, spastic di- or tetraplegia and congenital ichthyosis. Ichthyosis is usually evident at birth with varying degrees of erythema and scaling, neurologic symptoms appear in the first or second year of life. Most patients have an IQ of less than 60. Additional clinical features include glistening white spots on the retina, seizures, short stature and speech defects. {ECO:0000269|PubMed:10577908, ECO:0000269|PubMed:10792573, ECO:0000269|PubMed:8528251, ECO:0000269|PubMed:9254849, ECO:0000269|PubMed:9829906}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İskelet Kusurları, Genital Hipoplazi ve Mental Retardasyon, 612447, Otozomal Resesif (ZBTB16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Skeletal defects, genital hypoplasia, and mental retardation, 612447, Autosomal recessive (ZBTB16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZBTB16	ZBTB16, ZNF145, PLZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Skeletal defects, genital hypoplasia, and mental retardation (SGYMR) [MIM:612447]: A disorder characterized by mental retardation, craniofacial dysmorphism, microcephaly and short stature. Additional features include absence of the thumbs, hypoplasia of the radii and ulnae, additional vertebrae and ribs, retarded bone age and genital hypoplasia. {ECO:0000269|PubMed:18611983}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
İskelet Kusurları, Genital Hipoplazi ve Mental Retardasyon, 612447, Otozomal Resesif (Prenatal) (ZBTB16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Skeletal defects, genital hypoplasia, and mental retardation, 612447, Autosomal recessive (ZBTB16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZBTB16	ZBTB16, ZNF145, PLZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Skeletal defects, genital hypoplasia, and mental retardation (SGYMR) [MIM:612447]: A disorder characterized by mental retardation, craniofacial dysmorphism, microcephaly and short stature. Additional features include absence of the thumbs, hypoplasia of the radii and ulnae, additional vertebrae and ribs, retarded bone age and genital hypoplasia. {ECO:0000269|PubMed:18611983}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Cilt Biyopsisi Kromozom Analizi	Skin biopsy- Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Merkezimizden temin edilen transport besi yeri içinde/ Acil durumlarda steril serum fizyolojik içinde		15	45	.	.	21	Postnatal	.	Transport Medium suppliedfrom our center/ In urgency in Sterile Serum Physiologic
Cilt Kırışıklıkları, Konjenital Simetrik Dairesel, 2; Cscs2 (Multipl Benign Çembersel Ekstremite Deri Kattanları 2) (MAPRE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SKIN CREASES, CONGENITAL SYMMETRIC CIRCUMFERENTIAL, 2; CSCS2 (Multiple benign circumferential skin creases on limbs) (MAPRE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPRE2	MAPRE2, EB2, RP1, CSCSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Cilt Kırışıklıkları, Konjenital Simetrik Dairesel, 2; Cscs2 (Multipl Benign Çembersel Ekstremite Deri Kattanları 2) (Prenatal) (MAPRE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SKIN CREASES, CONGENITAL SYMMETRIC CIRCUMFERENTIAL, 2; CSCS2 (Multiple benign circumferential skin creases on limbs) (MAPRE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAPRE2	MAPRE2, EB2, RP1, CSCSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Deri Frajilitesi-Yünsü Saç Sendromu, 607655, Otozomal Resesif; SFWHS (Cilt Kırılganlık-Yünsü Saç-Palmoplantar Keratoderma Sendromu) (DSP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Skin fragility-woolly hair syndrome, 607655, Autosomal recessive; SFWHS (Skin fragility-woolly hair-palmoplantar keratoderma syndrome) (DSP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cardiomyopathy, dilated, with woolly hair and keratoderma (DCWHK) [MIM:605676]: An autosomal recessive cardiocutaneous syndrome characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. {ECO:0000269|PubMed:11063735}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Deri Frajilitesi-Yünsü Saç Sendromu, 607655, Otozomal Resesif; SFWHS (Cilt Kırılganlık-Yünsü Saç-Palmoplantar Keratoderma Sendromu) (Prenatal) (DSP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Skin fragility-woolly hair syndrome, 607655, Autosomal recessive; SFWHS (Skin fragility-woolly hair-palmoplantar keratoderma syndrome) (DSP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DSP	DSP, KPPS2, PPKS2, DCWHKTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cardiomyopathy, dilated, with woolly hair and keratoderma (DCWHK) [MIM:605676]: An autosomal recessive cardiocutaneous syndrome characterized by a generalized striate keratoderma particularly affecting the palmoplantar epidermis, woolly hair, and dilated left ventricular cardiomyopathy. {ECO:0000269|PubMed:11063735}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SLE Yatkınlık (CR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SLE susceptibility (CR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR1	CR1, C3BR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yavaşlamış Sinir İletim Hızı, OD, 608236, Otozomal Dominant; SNCV (Otozomal Dominant Sinir İletim Hızı Yavaşlaması) (ARHGEF10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Slowed nerve conduction velocity, AD, 608236, Autosomal dominant; SNCV (Autosomal dominant slowed nerve conduction velocity) (ARHGEF10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARHGEF10	ARHGEF10, KIAA0294, SNCV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Slowed nerve conduction velocity (SNCV) [MIM:608236]: Affected individuals present a reduction in nerve conduction velocities without any clinical signs of peripheral or central nervous system dysfunction. SNCV inheritance is autosomal dominant. {ECO:0000269|PubMed:14508709}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Küçük Hücreli Kanser, Akciğer, Somatik 182280 (Küçük Hücreli Akciğer Kanseri) (RB1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Small cell cancer of the lung, somatic, 182280 (Small cell lung cancer) (RB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RB1	RB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Küçük Lif Nöropati, 133020 (Sodyum Kanalopatisi İlişkili Small Fiber Nöropati) (SCN9A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Small fiber neuropathy, 133020 (Sodium channelopathy-related small fiber neuropathy) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Küçük Lif Nöropati, 133020 (Sodyum Kanalopatisi İlişkili Small Fiber Nöropati) (Prenatal) (SCN9A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Small fiber neuropathy, 133020 (Sodium channelopathy-related small fiber neuropathy) (SCN9A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN9A	SCN9A, NENA, PN1, FEB3B, GEFSP7, SFNP, HSAN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	15	30	20	Prenatal	DISEASE: Primary erythermalgia (PERYTHM) [MIM:133020]: Autosomal dominant disease characterized by recurrent episodes of severe pain associated with redness and warmth in the feet or hands. {ECO:0000269|PubMed:14985375, ECO:0000269|PubMed:15955112, ECO:0000269|PubMed:15958509, ECO:0000269|PubMed:16216943, ECO:0000269|PubMed:16392115, ECO:0000269|PubMed:18945915, ECO:0000269|PubMed:19369487, ECO:0000269|PubMed:24311784}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SMED Strudwick Tipi, 184250, Otozomal Dominant; SEMDSTWK (Spondiloepimetafizyal Displazia Konjenita, Strudwick Tipi) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SMED Strudwick type, 184250, autosomal dominant; SEMDSTWK (Spondyloepimetaphyseal dysplasia congenita, Strudwick type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
SMED Strudwick Tipi, 184250, Otozomal Dominant; SEMDSTWK (Spondiloepimetafizyal Displazia Konjenita, Strudwick Tipi) (MLPA)	SMED Strudwick type, 184250, autosomal dominant; SEMDSTWK (Spondyloepimetaphyseal dysplasia congenita, Strudwick type) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

SMED Strudwick Tipi, 184250, Otozomal Dominant; SEMDSTWK (Spondiloepimetafizyal Displazia Konjenita, Strudwick Tipi) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SMED Strudwick type, 184250, autosomal dominant; SEMDSTWK (Spondyloepimetaphyseal dysplasia congenita, Strudwick type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
SMED Strudwick Tipi, 184250, Otozomal Dominant; SEMDSTWK (Spondiloepimetafizyal Displazia Konjenita, Strudwick Tipi) (Prenatal) (MLPA)	SMED Strudwick type, 184250, autosomal dominant; SEMDSTWK (Spondyloepimetaphyseal dysplasia congenita, Strudwick type) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith Magenis Sendromu (17p11.2 Mikrodelesyonu) (FISH)	Smith Magenis syndrome (17p11.2 microdeletion) (FISH)	17p11.2	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Smith Magenis Sendromu (17p11.2 Mikrodelesyonu) (FISH) (Prenatal)	Smith Magenis syndrome (17p11.2 microdeletion) (Prenatal) (FISH)	17p11.2	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Kingsmore Sendromu, 616638, Otozomal Dominant; SKS (Makrosefali-Mental Retardasyon-Nörogelişimsel Bozukluk-Küçük Toraks Sendromu) (MTOR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Smith-Kingsmore syndrome, 616638, Autosomal dominant; SKS (Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome) (MTOR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTOR	MTOR, FRAP1, SKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Smith-Kingsmore syndrome (SKS) [MIM:616638]: An autosomal dominant syndrome characterized by intellectual disability, macrocephaly, seizures, umbilical hernia, and facial dysmorphic features. {ECO:0000269|PubMed:25851998, ECO:0000269|PubMed:26542245}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Smith-Kingsmore Sendromu, 616638, Otozomal Dominant; SKS (Makrosefali-Mental Retardasyon-Nörogelişimsel Bozukluk-Küçük Toraks Sendromu) (Prenatal) (MTOR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Smith-Kingsmore syndrome, 616638, Autosomal dominant; SKS (Macrocephaly-intellectual disability-neurodevelopmental disorder-small thorax syndrome) (MTOR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MTOR	MTOR, FRAP1, SKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Smith-Kingsmore syndrome (SKS) [MIM:616638]: An autosomal dominant syndrome characterized by intellectual disability, macrocephaly, seizures, umbilical hernia, and facial dysmorphic features. {ECO:0000269|PubMed:25851998, ECO:0000269|PubMed:26542245}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Lemli Opitz Sendromu, 270400, Otozomal Resesif; SLOS (Smith-Lemli-Opitz Sendromu) (DHCR7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Smith-Lemli-Opitz syndrome, 270400, Autosomal recessive; SLOS (Smith-Lemli-Opitz syndrome) (DHCR7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DHCR7	DHCR7, SLOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Smith-Lemli Opitz Sendromu, 270400, Otozomal Resesif; SLOS (Smith-Lemli-Opitz Sendromu) (MLPA)	Smith-Lemli-Opitz syndrome, 270400, Autosomal recessive; SLOS (Smith-Lemli-Opitz syndrome) (MLPA)	DHCR7	DHCR7, SLOS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Smith-Lemli Opitz Sendromu, 270400, Otozomal Resesif; SLOS (Smith-Lemli-Opitz Sendromu) (Prenatal) (DHCR7 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Smith-Lemli-Opitz syndrome, 270400, Autosomal recessive; SLOS (Smith-Lemli-Opitz syndrome) (DHCR7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DHCR7	DHCR7, SLOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Lemli Opitz Sendromu, 270400, Otozomal Resesif; SLOS (Smith-Lemli-Opitz Sendromu) (Prenatal) (MLPA)	Smith-Lemli-Opitz syndrome, 270400, Autosomal recessive; SLOS (Smith-Lemli-Opitz syndrome) (Prenatal) (MLPA)	DHCR7	DHCR7, SLOS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Magenis sendromu (17p11.2) (MLPA)	Smith-Magenis syndrome (17p11.2) (MLPA)	17p11.2	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Smith-Magenis sendromu (17p11.2) (MLPA) (Prenatal)	Smith-Magenis syndrome (17p11.2) (MLPA) (Prenatal)	17p11.2	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Magenis Sendromu, 182290, Otozomal Dominant, Sporadik Vakalar; SMS (Smith-Magenis Sendromu) (MLPA)	Smith-Magenis syndrome, 182290, Autosomal dominant, Isolated cases; SMS (Smith-Magenis syndrome) (MLPA)	RAI1	RAI1, SMCR, SMS	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Smith-Magenis syndrome (SMS) [MIM:182290]: Characterized by congenital mental retardation associated with development and growth delays. Affected persons have characteristic behavioral abnormalities, including self-injurious behaviors and sleep disturbance, and distinct craniofacial and skeletal anomalies. {ECO:0000269|PubMed:11404004, ECO:0000269|PubMed:12652298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Smith-Magenis Sendromu, 182290, Otozomal Dominant, Sporadik Vakalar; SMS (Smith-Magenis Sendromu) (Prenatal) (MLPA)	Smith-Magenis syndrome, 182290, Autosomal dominant, Isolated cases; SMS (Smith-Magenis syndrome) (Prenatal) (MLPA)	RAI1	RAI1, SMCR, SMS	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Smith-Magenis syndrome (SMS) [MIM:182290]: Characterized by congenital mental retardation associated with development and growth delays. Affected persons have characteristic behavioral abnormalities, including self-injurious behaviors and sleep disturbance, and distinct craniofacial and skeletal anomalies. {ECO:0000269|PubMed:11404004, ECO:0000269|PubMed:12652298}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Mccort Displazi 2, 615222, Otozomal Resesif; SMC2 (Smith-Mccort Displazisi) (RAB33B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Smith-McCort dysplasia 2, 615222, Autosomal recessive; SMC2 (Smith-McCort dysplasia) (RAB33B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB33B	RAB33B, SMC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Smith-McCort dysplasia 2 (SMC2) [MIM:615222]: A rare autosomal recessive osteochondrodysplasia with skeletal features identical to those of Dyggve-Melchior-Clausen syndrome, but with normal intelligence and no microcephaly. It is characterized by short limbs and trunk with barrel-shaped chest. The radiographic phenotype includes platyspondyly, generalized abnormalities of the epiphyses and metaphyses, and a distinctive lacy appearance of the iliac crest. {ECO:0000269|PubMed:22652534, ECO:0000269|PubMed:23042644}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Smith-Mccort Displazi 2, 615222, Otozomal Resesif; SMC2 (Smith-Mccort Displazisi) (Prenatal) (RAB33B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Smith-McCort dysplasia 2, 615222, Autosomal recessive; SMC2 (Smith-McCort dysplasia) (RAB33B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB33B	RAB33B, SMC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Smith-McCort dysplasia 2 (SMC2) [MIM:615222]: A rare autosomal recessive osteochondrodysplasia with skeletal features identical to those of Dyggve-Melchior-Clausen syndrome, but with normal intelligence and no microcephaly. It is characterized by short limbs and trunk with barrel-shaped chest. The radiographic phenotype includes platyspondyly, generalized abnormalities of the epiphyses and metaphyses, and a distinctive lacy appearance of the iliac crest. {ECO:0000269|PubMed:22652534, ECO:0000269|PubMed:23042644}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Smith-Mccort Displazi, 607326, Otozomal Resesif (Smith-Mccort Displazisi) (DYM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Smith-McCort dysplasia, 607326, Autosomal recessive (Smith-McCort dysplasia) (DYM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYM	DYM, FLJ90130, DMC, SMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dyggve-Melchior-Clausen syndrome (DMC) [MIM:223800]: A rare autosomal recessive disorder belonging to the group of spondyloepimetaphyseal dysplasias. DMC is characterized by progressive short stature with short trunk dwarfism, microcephaly, protruding sternum, and psychomotor retardation. Radiological features include a platyspondyly with double vertebral humps, an epiphyso-metaphyseal dysplasia and lacy pelvis iliac crests. {ECO:0000269|PubMed:12491225, ECO:0000269|PubMed:12554689}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Smith-Mccort Displazi, 607326, Otozomal Resesif (Smith-Mccort Displazisi) (Prenatal) (DYM geni) (Tüm Gen Dizi Analizi) (Prenatal)	Smith-McCort dysplasia, 607326, Autosomal recessive (Smith-McCort dysplasia) (DYM gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYM	DYM, FLJ90130, DMC, SMC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Dyggve-Melchior-Clausen syndrome (DMC) [MIM:223800]: A rare autosomal recessive disorder belonging to the group of spondyloepimetaphyseal dysplasias. DMC is characterized by progressive short stature with short trunk dwarfism, microcephaly, protruding sternum, and psychomotor retardation. Radiological features include a platyspondyly with double vertebral humps, an epiphyso-metaphyseal dysplasia and lacy pelvis iliac crests. {ECO:0000269|PubMed:12491225, ECO:0000269|PubMed:12554689}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sneddon Sendromu, 182410, Otozomal Resesif (Sneddon Sendromu) (ADA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sneddon syndrome, 182410, Autosomal recessive (Sneddon syndrome) (ADA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADA2	CECR1, PAN, SNEDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sneddon Sendromu, 182410, Otozomal Resesif (Sneddon Sendromu) (Prenatal) (ADA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sneddon syndrome, 182410, Autosomal recessive (Sneddon syndrome) (ADA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADA2	CECR1, PAN, SNEDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kar Tanesi Vitreoretinal Dejenerasyon, 193230, Otozomal Dominant (Kar Tanesi Vitreoretinal Dejenerasyonu) (KCNJ13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Snowflake vitreoretinal degeneration, 193230, Autosomal dominant (Snowflake vitreoretinal degeneration) (KCNJ13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ13	KCNJ13, SVD, LCA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Snowflake vitreoretinal degeneration (SVD) [MIM:193230]: Developmental and progressive hereditary eye disorder that affects multiple tissues within the eye. Diagnostic features of SVD include fibrillar degeneration of the vitreous humor, early-onset cataract, minute crystalline deposits in the neurosensory retina, and retinal detachment. {ECO:0000269|PubMed:18178896}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Solid Doku Biyopsi Kromozom Analizi	Solid Tissue biopsy- Chromosome analysis	.	.	Kromozom analizi/ Karyotype analysis	Merkezimizden temin edilen transport besi yeri içinde/ Acil durumlarda steril serum fizyolojik içinde		15	45	.	.	21	Postnatal	.	Transport Medium suppliedfrom our center/ in urgency in Sterile Serum Physiologic
Somatostatin Analoğu Rezistansı, Otozomal Dominant, Somatik Mutasyon (SSTR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Somatostatin analog, resistance to, Autosomal dominant, Somatic mutation (SSTR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SSTR5	SSTR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etkilenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Somatostatin Reseptörü 5; Sstr5 (SSTR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SOMATOSTATIN RECEPTOR 5; SSTR5 (SSTR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SSTR5	SSTR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Sorsby Fundus Distrofisi, 136900, Otozomal Dominant; SFD (Sorsby Fundus Distrofisi) (TIMP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sorsby fundus dystrophy, 136900, Autosomal dominant; SFD (Sorsby's fundus dystrophy) (TIMP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIMP3	TIMP3, SFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sorsby fundus dystrophy (SFD) [MIM:136900]: Rare autosomal dominant macular disorder with an age of onset in the fourth decade. It is characterized by loss of central vision from subretinal neovascularization and atrophy of the ocular tissues. Generally, macular discform degeneration develops in the patients eye within 6 months to 6 years. {ECO:0000269|PubMed:7503009, ECO:0000269|PubMed:7894485, ECO:0000269|PubMed:8634721, ECO:0000269|PubMed:8728699, ECO:0000269|PubMed:8981947}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sotos Sendromu (Sq35 Mikrodelesyonu) (FISH)	Sotos syndrome (Sq35 microdeletion) (FISH)	5q35	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Sotos Sendromu (Sq35 Mikrodelesyonu) (FISH) (Prenatal)	Sotos syndrome (Sq35 microdeletion) (Prenatal) (FISH)	5q35	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sotos Sendromu 1, 117550, Otozomal Dominant; SOTOS1 (Sotos Sendromu) (NSD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sotos syndrome 1, 117550, Autosomal dominant; SOTOS1 (Sotos syndrome) (NSD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NSD1	NSD1, ARA267, STO, SOTOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:14997421}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sotos Sendromu 1, 117550, Otozomal Dominant; SOTOS1 (Sotos Sendromu) (MLPA)	Sotos syndrome 1, 117550, Autosomal dominant; SOTOS1 (Sotos syndrome) (MLPA)	NSD1	NSD1, ARA267, STO, SOTOS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:14997421}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sotos Sendromu 1, 117550, Otozomal Dominant; SOTOS1 (Sotos Sendromu) (Prenatal) (NSD1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sotos syndrome 1, 117550, Autosomal dominant; SOTOS1 (Sotos syndrome) (NSD1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NSD1	NSD1, ARA267, STO, SOTOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:14997421}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sotos Sendromu 1, 117550, Otozomal Dominant; SOTOS1 (Sotos Sendromu) (Prenatal) (MLPA)	Sotos syndrome 1, 117550, Autosomal dominant; SOTOS1 (Sotos syndrome) (Prenatal) (MLPA)	NSD1	NSD1, ARA267, STO, SOTOS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Beckwith-Wiedemann syndrome (BWS) [MIM:130650]: A disorder characterized by anterior abdominal wall defects including exomphalos (omphalocele), pre- and postnatal overgrowth, and macroglossia. Additional less frequent complications include specific developmental defects and a predisposition to embryonal tumors. {ECO:0000269|PubMed:14997421}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sotos Sendromu 2, 614753, Otozomal Dominant; SOTOS2 (Malan Aşırı Büyüme Sendromu) (NFIX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sotos syndrome 2, 614753, Autosomal dominant; SOTOS2 (Malan overgrowth syndrome) (NFIX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NFIX	NFIX, NF1A, SOTOS2, MRSHSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marshall-Smith syndrome (MRSHSS) [MIM:602535]: A distinct malformation syndrome characterized by accelerated skeletal maturation, relative failure to thrive, respiratory difficulties, mental retardation, and unusual facies, including prominent forehead, shallow orbits, blue sclerae, depressed nasal bridge, and micrognathia. Additional skeletal findings include long and thin tubular bones, broad middle phalanges with relatively narrow distal phalanges, and scoliosis. {ECO:0000269|PubMed:20673863}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sotos Sendromu 2, 614753, Otozomal Dominant; SOTOS2 (Malan Aşırı Büyüme Sendromu) (Prenatal) (NFIX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sotos syndrome 2, 614753, Autosomal dominant; SOTOS2 (Malan overgrowth syndrome) (NFIX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NFIX	NFIX, NF1A, SOTOS2, MRSHSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Marshall-Smith syndrome (MRSHSS) [MIM:602535]: A distinct malformation syndrome characterized by accelerated skeletal maturation, relative failure to thrive, respiratory difficulties, mental retardation, and unusual facies, including prominent forehead, shallow orbits, blue sclerae, depressed nasal bridge, and micrognathia. Additional skeletal findings include long and thin tubular bones, broad middle phalanges with relatively narrow distal phalanges, and scoliosis. {ECO:0000269|PubMed:20673863}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sotos Sendromu 3, 617169, Otozomal Resesif; SOTOS3 (Sotos Sendromu) (APC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sotos syndrome 3, 617169, Autosomal recessive; SOTOS3 (Sotos syndrome) (APC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APC2	APC2, APCL, SOTOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sotos Sendromu 3, 617169, Otozomal Resesif; SOTOS3 (Sotos Sendromu) (Prenatal) (APC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sotos syndrome 3, 617169, Autosomal recessive; SOTOS3 (Sotos syndrome) (APC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	APC2	APC2, APCL, SOTOS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spastik Ataksi 1, Otozomal Dominant, 108600, Otozomal Dominant; SPAX1 (Otozomal Dominant Spastik Ataksi Tip 1) (VAMP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia 1, autosomal dominant, 108600, Autosomal dominant; SPAX1 (Autosomal dominant spastic ataxia type 1) (VAMP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VAMP1	VAMP1, SYB1, SPAX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Ataksi 2, Otozomal Resesif, 611302, Otozomal Resesif; SPAX2 (Otozomal Resesif Spastik Paraplegi Tip 58) (KIF1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia 2, autosomal recessive, 611302, Autosomal recessive; SPAX2 (Autosomal recessive spastic paraplegia type 58) (KIF1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1C	KIF1C, LTXS1, KIAA0706, SPAX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic ataxia 2, autosomal recessive (SPAX2) [MIM:611302]: A neurologic disorder characterized by cerebellar ataxia, dysarthria, and variable spasticity of the lower limbs. Cognition is not affected. {ECO:0000269|PubMed:24319291}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Ataksi 3, Otozomal Resesif, 611390, Otozomal Resesif; SPAX3 (Otozomal Resesif Spastik Ataksi-Lökoensefalopati) (MARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia 3, autosomal recessive, 611390, Autosomal recessive; SPAX3 (Autosomal recessive spastic ataxia with leukoencephalopathy) (MARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MARS2	MARS2, SPAX3, COXPD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic ataxia 3, autosomal recessive (SPAX3) [MIM:611390]: A neurologic disorder characterized by cerebellar ataxia, ataxic gait, spasticity, and hyperreflexia. Other variable features include dysarthria, dysmetria, mild cognitive impairment, urinary urgency and dystonic positioning. {ECO:0000269|PubMed:22448145}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Ataksi 4, Otozomal Resesif, 613672, Otozomal Resesif; SPAX4 (Otozomal Resesif Spastik Ataksi-Optik Atrofi-Konuşma Bozukluğu Sendromu) (MTPAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia 4, autosomal recessive, 613672, Autosomal recessive; SPAX4 (Autosomal recessive spastic ataxia-optic atrophy-dysarthria syndrome) (MTPAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTPAP	MTPAP, PAPD1, SPAX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Ataksi 5, Otozomal Resesif, 614487, Otozomal Resesif; SPAX5 (Erken Başlangıçlı Spastik Ataksi-Nöropati Sendromu) (AFG3L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia 5, autosomal recessive, 614487, Autosomal recessive; SPAX5 (Early-onset spastic ataxia-neuropathy syndrome) (AFG3L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFG3L2	AFG3L2, SCA28, SPAX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic ataxia 5, autosomal recessive (SPAX5) [MIM:614487]: A neurodegenerative disorder characterized by early onset spasticity, peripheral neuropathy, ptosis, oculomotor apraxia, dystonia, cerebellar atrophy, and progressive myoclonic epilepsy. {ECO:0000269|PubMed:22022284}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Ataksi, Charlevoix-Saguenay Tipi, 270550, Otozomal Resesif; SACS (Otozomal Resesif Spastik Ataksi, Charlevoix-Saguenay Tipi) (SACS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic ataxia, Charlevoix-Saguenay type, 270550, Autosomal recessive; SACS (Autosomal recessive spastic ataxia of Charlevoix-Saguenay) (SACS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SACS	SACS, ARSACS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic ataxia Charlevoix-Saguenay type (SACS) [MIM:270550]: A neurodegenerative disease characterized by early- onset cerebellar ataxia, spasticity, retinal hypermyelination, pyramidal signs, and both axonal and demyelinating neuropathy with loss of sensory nerve conduction and reduced motor conduction velocities. Other features include dysarthria, distal muscle wasting, nystagmus, defect in conjugate pursuit ocular movements, retinal striation (from prominent retinal nerves) obscuring the retinal blood vessels in places, and the frequent presence of mitral valve prolapse. {ECO:0000269|PubMed:10655055, ECO:0000269|PubMed:12873855, ECO:0000269|PubMed:14718708, ECO:0000269|PubMed:15156359, ECO:0000269|PubMed:15985586, ECO:0000269|PubMed:16007637, ECO:0000269|PubMed:17290461, ECO:0000269|PubMed:17716690, ECO:0000269|PubMed:18398442, ECO:0000269|PubMed:18465152, ECO:0000269|PubMed:18484239, ECO:0000269|PubMed:19529988, ECO:0000269|PubMed:20876471}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Paralizi, İnfantil Başlangıçlı Yükselen, 607225, Otozomal Resesif; IAHSP (Infantil Başlangıçlı Asendan Kalıtsal Spastik Paralizi) (ALS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paralysis, infantile onset ascending, 607225, Autosomal recessive; IAHSP (Infantile-onset ascending hereditary spastic paralysis) (ALS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALS2	ALS2, ALSJ, PLSJ, IAHSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 2 (ALS2) [MIM:205100]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:11586298}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 10, Otozomal Dominant, 604187, Otozomal Dominant; SPG10 (Otozomal Dominant Spastik Parapleji Tip 10) (KIF5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 10, autosomal dominant, 604187, Autosomal dominant; SPG10 (Autosomal dominant spastic paraplegia type 10) (KIF5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF5A	KIF5A, NKHC, SPG10, NEIMY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 10, autosomal dominant (SPG10) [MIM:604187]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:12355402, ECO:0000269|PubMed:15452312, ECO:0000269|PubMed:16476820, ECO:0000269|PubMed:16489470, ECO:0000269|PubMed:18245137, ECO:0000269|PubMed:18853458, ECO:0000269|PubMed:21107874}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 11, Otozomal Resesif, 604360, Otozomal Resesif; SPG11 (Otozomal Resesif Spastik Parapleji Tip 11) (SPG11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 11, autosomal recessive, 604360, Autosomal recessive; SPG11 (Autosomal recessive spastic paraplegia type 11) (SPG11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG11	SPG11, KIAA1840, FLJ21439, ALS5, CMT2X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Spastik Parapleji 12, Otozomal Dominant, 604805, Otozomal Dominant; SPG12 (Otozomal Dominant Spastik Parapleji Tip 12) (RTN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 12, autosomal dominant, 604805, Autosomal dominant; SPG12 (Autosomal dominant spastic paraplegia type 12) (RTN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RTN2	RTN2, NSPL1, SPG12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 13, Otozomal Dominant, 605280, Otozomal Dominant; SPG13 (Otozomal Dominant Spastik Parapleji Tip 13) (HSPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 13, autosomal dominant, 605280, Autosomal dominant; SPG13 (Autosomal dominant spastic paraplegia type 13) (HSPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HSPD1	HSPD1, SPG13, HSP60, HLD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 13, autosomal dominant (SPG13) [MIM:605280]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:11898127}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 15, Otozomal Resesif, 270700, Otozomal Resesif; SPG15 (Otozomal Resesif Spastik Parapleji Tip 15) (ZFYVE26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 15, autosomal recessive, 270700, Autosomal recessive; SPG15 (Autosomal recessive spastic paraplegia type 15) (ZFYVE26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFYVE26	ZFYVE26, KIAA0321, SPG15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 15, autosomal recessive (SPG15) [MIM:270700]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG15 is a complex form associated with additional neurological symptoms such as cognitive deterioration or mental retardation, axonal neuropathy, mild cerebellar signs, and, less frequently, a central hearing deficit, decreased visual acuity, or retinal degeneration. {ECO:0000269|PubMed:18394578, ECO:0000269|PubMed:19084844, ECO:0000269|PubMed:19805727}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 15, Otozomal Resesif, 270700, Otozomal Resesif; SPG15 (Otozomal Resesif Spastik Parapleji Tip 15) (Prenatal) (ZFYVE26 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spastic paraplegia 15, autosomal recessive, 270700, Autosomal recessive; SPG15 (Autosomal recessive spastic paraplegia type 15) (ZFYVE26 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZFYVE26	ZFYVE26, KIAA0321, SPG15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (iKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 15, autosomal recessive (SPG15) [MIM:270700]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG15 is a complex form associated with additional neurological symptoms such as cognitive deterioration or mental retardation, axonal neuropathy, mild cerebellar signs, and, less frequently, a central hearing deficit, decreased visual acuity, or retinal degeneration. {ECO:0000269|PubMed:18394578, ECO:0000269|PubMed:19084844, ECO:0000269|PubMed:19805727}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spastik Parapleji 18, Otozomal Resesif, 611225, Otozomal Resesif; SPG18 (Otozomal Resesif Spastik Parapleji Tip 18) (ERLIN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 18, autosomal recessive, 611225, Autosomal recessive; SPG18 (Autosomal recessive spastic paraplegia type 18) (ERLIN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERLIN2	ERLIN2, SPFH2, C8orf2, SPG18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 2, X'e Bağlı, 312920, X'e Bağlı Resesif; SPG2 (Spastik Parapleji Tip 2) (PLP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 2, X-linked, 312920, X-linked recessive; SPG2 (Spastic paraplegia type 2) (PLP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLP1	PLP1, PMD, HLD1, SPG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukodystrophy, hypomyelinating, 1 (HLD1) [MIM:312080]: A X-linked recessive disorder of the central nervous system in which myelin is not formed properly. Clinically characterized by nystagmus, spastic quadriplegia, ataxia, and developmental delay. {ECO:0000269|PubMed:10417279, ECO:0000269|PubMed:10425042, ECO:0000269|PubMed:11093273, ECO:0000269|PubMed:11786921, ECO:0000269|PubMed:1376966, ECO:0000269|PubMed:1384324, ECO:0000269|PubMed:15712223, ECO:0000269|PubMed:1707231, ECO:0000269|PubMed:1708672, ECO:0000269|PubMed:1715570, ECO:0000269|PubMed:2479017, ECO:0000269|PubMed:2480601, ECO:0000269|PubMed:2773936, ECO:0000269|PubMed:7531827, ECO:0000269|PubMed:7539213, ECO:0000269|PubMed:7541731, ECO:0000269|PubMed:7573159, ECO:0000269|PubMed:7679906, ECO:0000269|PubMed:7683951, ECO:0000269|PubMed:7684886, ECO:0000269|PubMed:8037216, ECO:0000269|PubMed:8909455, ECO:0000269|PubMed:9008538, ECO:0000269|PubMed:9143933, ECO:0000269|PubMed:9482656, ECO:0000269|PubMed:9633722, ECO:0000269|PubMed:9747038, ECO:0000269|PubMed:9788732	EDTA Blood Sample
Spastik Parapleji 26, Otozomal Resesif, 609195, Otozomal Resesif; SPG26 (Otozomal Resesif Spastik Parapleji Tip 26) (B4GALNT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 26, autosomal recessive, 609195, Autosomal recessive; SPG26 (Autosomal recessive spastic paraplegia type 26) (B4GALNT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B4GALNT1	B4GALNT1, GALGT, GALNACT, SPG26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 26, autosomal recessive (SPG26) [MIM:609195]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG26 is a complicated form characterized by onset in the first 2 decades of life of gait abnormalities due to lower limb spasticity and muscle weakness. Some patients have upper limb involvement. Additional features include intellectual disability, peripheral neuropathy, dysarthria, cerebellar signs, extrapyramidal signs, and cortical atrophy. The disorder is slowly progressive. {ECO:0000269	PubMed:23746551}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 28 Otozomal Resesif, 609340, Otozomal Resesif; SPG28 (Otozomal Resesif Spastik Parapleji Tip 28) (DDHD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 28, autosomal recessive, 609340, Autosomal recessive; SPG28 (Autosomal recessive spastic paraplegia type 28) (DDHD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDHD1	DDHD1, PAPLA1, KIAA1705, SPG28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 3, Otozomal Dominant; Spg3a (Otozomal Dominant Spastik Parapleji Tip 3) (ATL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SPASTIC PARAPLEGIA 3, AUTOSOMAL DOMINANT; SPG3A (Autosomal dominant spastic paraplegia type 3) (ATL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATL1	ATL1, SPG3A, HSN1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 1D (HSN1D) [MIM:613708]: A disease characterized by adult-onset distal axonal sensory neuropathy leading to mutilating ulcerations as well as hyporeflexia. Some patients may show features suggesting upper neuron involvement. {ECO:0000269	PubMed:21194679}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 30, Otozomal Resesif, 610357, Otozomal Resesif; SPG30 (Otozomal Spastik Parapleji Tip 30) (KIF1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 30, autosomal recessive, 610357, Autosomal recessive; SPG30 (Autosomal spastic paraplegia type 30) (KIF1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF1A	KIF1A, ATSV, UNC104, SPG30, HSN2C, MRD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 2C (HSN2C) [MIM:614213]: A neurodegenerative disorder characterized by onset in the first decade of progressive distal sensory loss leading to ulceration and amputation of the fingers and toes. Affected individuals also develop distal muscle weakness, primarily affecting the lower limbs. {ECO:0000269	PubMed:21870098}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 31, Otozomal Dominant, 610250, Otozomal Dominant; SPG31 (Otozomal Dominant Spastik Parapleji Tip 31) (REEP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 31, autosomal dominant, 610250, Autosomal dominant; SPG31 (Autosomal dominant spastic paraplegia type 31) (REEP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REEP1	REEP1, C2ORF23, SPG31, HMN8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuronopathy, distal hereditary motor, 5B (HMN5B) [MIM:614751]: A disorder characterized by distal muscular atrophy mainly affecting the upper extremities, in contrast to other distal motor neuronopathies. These constitute a heterogeneous group of neuromuscular diseases caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. HMN5B is characterized by onset in the first or second decade of distal muscle weakness and atrophy, primarily affecting the intrinsic hand muscles, but also affecting the lower legs, resulting in abnormal gait and pes cavus. {ECO:0000269	PubMed:22703882}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 33, Otozomal Dominant, 610244, Otozomal Dominant; SPG33 (ZFYVE27 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 33, autosomal dominant, 610244, Autosomal dominant; SPG33 (ZFYVE27 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFYVE27	ZFYVE27, SPG33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 33, autosomal dominant (SPG33) [MIM:610244]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269	PubMed:16826525}. Note=The disease is caused by mutations affecting the gene represented in this entry. According to PubMed:18606302, the properties of the variant Val- 191 and its frequency in some populations raise doubts on the implication of that gene in the disease.;	EDTA Blood Sample

Spastik Parapleji 33, Otozomal Dominant, 610244, Otozomal Dominant; SPG33 (Prenatal) (ZFYVE27 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spastic paraplegia 33, autosomal dominant, 610244, Autosomal dominant; SPG33 (ZFYVE27 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZFYVE27	ZFYVE27, SPG33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spastic paraplegia 33, autosomal dominant (SPG33) [MIM:610244]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. (ECO:0000269	PubMed:16826525). Note=The disease is caused by mutations affecting the gene represented in this entry. According to PubMed:18606302, the properties of the variant Val- 191 and its frequency in some populations raise doubts on the implication of that gene in the disease.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spastik Parapleji 35, Otozomal Resesif, 612319, Otozomal Resesif; SPG35 (Otozomal Resesif Spastik Parapleji Tip 35) (FA2H geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 35, autosomal recessive, 612319, Autosomal recessive; SPG35 (Autosomal recessive spastic paraplegia type 35) (FA2H gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FA2H	FA2H, FAAH, FAXDC1, FAH1, SCS7, SPG35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 35, autosomal recessive (SPG35) [MIM:612319]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG35 is a complicated form characterized by childhood onset of gait difficulties. It has a rapid progression and many patients become wheelchair-bound as young adults. Patients manifest cognitive decline associated with leukodystrophy. Other variable neurologic features, such as dystonia, optic atrophy, and seizures may also occur. (ECO:0000269|PubMed:19068277, ECO:0000269|PubMed:20104589, ECO:0000269|PubMed:20853438). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 39, Otozomal Resesif, 612020, Otozomal Resesif; SPG39 (Otozomal Resesif Spastik Parapleji Tip 39) (PNPLA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 39, autosomal recessive, 612020, Autosomal recessive; SPG39 (Autosomal recessive spastic paraplegia type 39) (PNPLA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PNPLA6	PNPLA6, NTE, SPG39, NTEMND, BNHS, LNMS, OMCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Boucher-Neuhauser syndrome (BNHS) [MIM:215470]: An autosomal recessive disorder characterized by spinocerebellar ataxia, hypogonadotropic hypogonadism, and visual impairment due to chorioretinal dystrophy. The age at onset is variable, but most patients develop 1 or more symptoms in the first decade of life. Chorioretinal dystrophy may not always be present. (ECO:0000269|PubMed:24357708, ECO:0000269|PubMed:25033069). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 3A, Otozomal Dominant, 182600, Otozomal Dominant (Otozomal Dominant Spastik Parapleji Tip 3) (ATL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 3A, autosomal dominant, 182600, Autosomal dominant (Autosomal dominant spastic paraplegia type 3) (ATL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATL1	ATL1, SPG3A, HSN1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary sensory, 1D (HSN1D) [MIM:613708]: A disease characterized by adult-onset distal axonal sensory neuropathy leading to mutilating ulcerations as well as hyporeflexia. Some patients may show features suggesting upper neuron involvement. (ECO:0000269	PubMed:21194679). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Spastik Parapleji 4, Otozomal Dominant, 182601, Otozomal Dominant; SPG4 (Otozomal Dominant Spastik Parapleji Tip 4) (SPAST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 4, autosomal dominant, 182601, Autosomal dominant; SPG4 (Autosomal dominant spastic paraplegia type 4) (SPAST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPAST	SPAST, SPG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 4, autosomal dominant (SPG4) [MIM:182601]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. (ECO:0000269|PubMed:10610178, ECO:0000269|PubMed:10699187, ECO:0000269|PubMed:11015453, ECO:0000269|PubMed:11039577, ECO:0000269|PubMed:11087788, ECO:0000269|PubMed:11309678, ECO:0000269|PubMed:11843700, ECO:0000269|PubMed:11985387, ECO:0000269|PubMed:12124993, ECO:0000269|PubMed:12161613, ECO:0000269|PubMed:12163196, ECO:0000269|PubMed:12202986, ECO:0000269|PubMed:12460147, ECO:0000269|PubMed:12552568, ECO:0000269|PubMed:12939659, ECO:0000269|PubMed:14732620, ECO:0000269|PubMed:15159500, ECO:0000269|PubMed:15210521, ECO:0000269|PubMed:15248095, ECO:0000269|PubMed:15326248, ECO:0000269|PubMed:15482961, ECO:0000269|PubMed:16682546, ECO:0000269|PubMed:16684598	EDTA Blood Sample
Spastik Parapleji 4, Otozomal Dominant, 182601, Otozomal Dominant; SPG4 (Otozomal Dominant Spastik Parapleji Tip 4) (MLPA)	Spastic paraplegia 4, autosomal dominant, 182601, Autosomal dominant; SPG4 (Autosomal dominant spastic paraplegia type 4) (MLPA)	SPAST	SPAST, SPG4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Spastic paraplegia 4, autosomal dominant (SPG4) [MIM:182601]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. (ECO:0000269|PubMed:10610178, ECO:0000269|PubMed:10699187, ECO:0000269|PubMed:11015453, ECO:0000269|PubMed:11039577, ECO:0000269|PubMed:11087788, ECO:0000269|PubMed:11309678, ECO:0000269|PubMed:11843700, ECO:0000269|PubMed:11985387, ECO:0000269|PubMed:12124993, ECO:0000269|PubMed:12161613, ECO:0000269|PubMed:12163196, ECO:0000269|PubMed:12202986, ECO:0000269|PubMed:12460147, ECO:0000269|PubMed:12552568, ECO:0000269|PubMed:12939659, ECO:0000269|PubMed:14732620, ECO:0000269|PubMed:15159500, ECO:0000269|PubMed:15210521, ECO:0000269|PubMed:15248095, ECO:0000269|PubMed:15326248, ECO:0000269|PubMed:15482961, ECO:0000269|PubMed:16682546, ECO:0000269|PubMed:16684598	EDTA Blood Sample
Spastik Parapleji 42, Otozomal Dominant, 612539, Otozomal Dominant; SPG42 (Otozomal Dominant Spastik Parapleji Tip 42) (SLC33A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 42, autosomal dominant, 612539, Autosomal dominant; SPG42 (Autosomal dominant spastic paraplegia type 42) (SLC33A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC33A1	SLC33A1, ACATN, AT1, SPG42, CCHLND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital cataracts, hearing loss, and neurodegeneration (CCHLND) [MIM:614482]: An autosomal recessive disorder characterized by congenital cataracts, severe psychomotor retardation, and hearing loss associated with decreased serum ceruloplasmin and copper. Brain MRI shows cerebral and cerebellar atrophy and hypomyelination. (ECO:0000269	PubMed:22243965). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Spastik Parapleji 43, Otozomal Resesif, 615043, Otozomal Resesif; SPG43 (Otozomal Resesif Spastik Parapleji Tip 43) (C19orf12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 43, autosomal recessive, 615043, Autosomal recessive; SPG43 (Autosomal recessive spastic paraplegia type 43) (C19orf12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C19orf12	C19orf12, NBIA4, SPG43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neurodegeneration with brain iron accumulation 4 (NBIA4) [MIM:614298]: A neurodegenerative disorder associated with iron accumulation in the brain, primarily in the basal ganglia. NBIA4 results in speech difficulty, extrapyramidal signs, oromandibular and generalized dystonia, and parkinsonism. Most patients have progressive involvement of the corticospinal tract, with spasticity, hyperreflexia, and extensor plantar responses. (ECO:0000269|PubMed:21981780, ECO:0000269|PubMed:22508347, ECO:0000269|PubMed:22584950, ECO:0000269|PubMed:23269600, ECO:0000269|PubMed:23521069, ECO:0000269|PubMed:23857908). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 44, Otozomal Resesif, 613206, Otozomal Resesif; SPG44 (Otozomal Resesif Spastik Parapleji Tip 44) (GJC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 44, autosomal recessive, 613206, Autosomal recessive; SPG44 (Autosomal recessive spastic paraplegia type 44) (GJC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJC2	GJC2, GJA12, CX47, PMLDAR, HLD2, SPG44, LMPH1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 44, autosomal recessive (SPG44) [MIM:613206]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. (ECO:0000269	PubMed:19058803). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 45, Otozomal Resesif, 613162, Otozomal Resesif; SPG45 (Otozomal Resesif Spastik Parapleji Tip 45) (NTSC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 45, autosomal recessive, 613162, Autosomal recessive; SPG45 (Autosomal recessive spastic paraplegia type 45) (NTSC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NTSC2	NTSC2, NTSB, PNTS, SPG45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 45, autosomal recessive (SPG45) [MIM:613162]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. Some SPG45 patients manifest mental retardation, contractures and learning disability. (ECO:0000269	PubMed:24482476). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 46, Otozomal Resesif, 614409, Otozomal Resesif; SPG46 (Otozomal Resesif Spastik Parapleji Tip 46) (GBA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 46, autosomal recessive, 614409, Autosomal recessive; SPG46 (Autosomal recessive spastic paraplegia type 46) (GBA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GBA2	GBA2, KIAA1605, SPG46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Spastik Parapleji 47, Otozomal Resesif, 614066, Otozomal Resesif; SPG47 (Ağır Mental Retardasyon ve Progresif Spastik Paraplejj) (AP4B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 47, autosomal recessive, 614066, Autosomal recessive; SPG47 (Severe intellectual disability and progressive spastic paraplegia) (AP4B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP4B1	AP4B1, SPG47, CPSQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample

Spastik Parapleji 48, Otozomal Resesif, 613647, Otozomal Resesif; SPG48 (Otozomal Resesif Spastik Parapleji Tip 48) (APSZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 48, autosomal recessive, 613647, Autosomal recessive; SPG48 (Autosomal recessive spastic paraplegia type 48) (APSZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	APSZ1	APSZ1, KIAA0415, SPG48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 48, autosomal recessive (SPG48) [MIM:613647]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:20613862}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 49, Otozomal Resesif, 615031, Otozomal Resesif; SPG49 (Otozomal Resesif Spastik Parapleji Tip 49) (TECPR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 49, autosomal recessive, 615031, Autosomal recessive; SPG49 (Autosomal recessive spastic paraplegia type 49) (TECPR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TECPR2	TECPR2, KIAA0329, SPG49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 50, Otozomal Resesif, 612936, Otozomal Resesif; SPG50 (Ağır Mental Retardasyon ve Progresif Spastik Parapleji) (AP4M1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 50, autosomal recessive, 612936, Autosomal recessive; SPG50 (Severe intellectual disability and progressive spastic paraplegia) (AP4M1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP4M1	AP4M1, SPG50, CPSQ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 51, Otozomal Resesif, 613744, Otozomal Resesif; SPG51 (Ağır Mental Retardasyon ve Progresif Spastik Parapleji) (AP4E1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 51, autosomal recessive, 613744, Autosomal recessive; SPG51 (Severe intellectual disability and progressive spastic paraplegia) (AP4E1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP4E1	AP4E1, SPG51, CPSQ4, STUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 52, Otozomal Resesif, 614067, Otozomal Resesif; SPG52 (Ağır Mental Retardasyon ve Progresif Spastik Parapleji) (AP4S1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 52, autosomal recessive, 614067, Autosomal recessive; SPG52 (Severe intellectual disability and progressive spastic paraplegia) (AP4S1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP4S1	AP4S1, CPSQ6, SPG52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 53, Otozomal Resesif, 614898, Otozomal Resesif; SPG53 (Otozomal Resesif Spastik Parapleji Tip 53) (VPS37A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 53, autosomal recessive, 614898, Autosomal recessive; SPG53 (Autosomal recessive spastic paraplegia type 53) (VPS37A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VPS37A	VPS37A, HCRP1, SPG53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 54, Otozomal Resesif, 615033, Otozomal Resesif; SPG54 (Otozomal Resesif Spastik Parapleji Tip 54) (DDHD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 54, autosomal recessive, 615033, Autosomal recessive; SPG54 (Autosomal recessive spastic paraplegia type 54) (DDHD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDHD2	DDHD2, KIAA0725, SPG54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 54, autosomal recessive (SPG54) [MIM:615033]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. Complicated forms are recognized by additional variable features including spastic quadriparesis, seizures, dementia, amyotrophy, extrapyramidal disturbance, cerebral or cerebellar atrophy, optic atrophy, and peripheral neuropathy, as well as by extra neurological manifestations. SPG54 patients have delayed psychomotor development, intellectual disability, and early-onset spasticity of the lower limbs. Brain MRI shows a thin corpus callosum and periventricular white matter lesions. {ECO:0000269|PubMed:23176823}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 55, Otozomal Resesif, 615035, Otozomal Resesif; SPG55 (Otozomal Resesif Spastik Parapleji Tip 55) (C12orf65 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 55, autosomal recessive, 615035, Autosomal recessive; SPG55 (Autosomal recessive spastic paraplegia type 55) (C12orf65 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C12orf65	C12orf65, COXPD7, SPG55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 7 (COXPD7) [MIM:613559]: A mitochondrial disease resulting in encephalomyopathy. Clinical manifestations include psychomotor delay and regression, ataxia, optic atrophy, nystagmus and muscle atrophy and weakness. {ECO:0000269|PubMed:20598281}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 56, Otozomal Resesif, 615030, Otozomal Resesif; SPG56 (Otozomal Resesif Spastik Parapleji Tip 56) (CYP2U1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 56, autosomal recessive, 615030, Autosomal recessive; SPG56 (Autosomal recessive spastic paraplegia type 56) (CYP2U1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2U1	CYP2U1, SPG56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 56, autosomal recessive (SPG56) [MIM:615030]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. Complicated forms are recognized by additional variable features including spastic quadriparesis, seizures, dementia, amyotrophy, extrapyramidal disturbance, cerebral or cerebellar atrophy, optic atrophy, and peripheral neuropathy, as well as by extra neurological manifestations. In SPG56, upper limbs are often also affected. Some SPG56 patients may have a subclinical axonal neuropathy. {ECO:0000269|PubMed:23176821}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 57, Otozomal Resesif, 615658, Otozomal Resesif; SPG57 (Otozomal Resesif Spastik Parapleji Tip 57) (TFG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 57, autosomal recessive, 615658, Autosomal recessive; SPG57 (Autosomal recessive spastic paraplegia type 57) (TFG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TFG	TFG, HMSNP, SPG57	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neuropathy, hereditary motor and sensory, Okinawa type (HMSNO) [MIM:604484]: A neurodegenerative disorder characterized by young adult onset of proximal muscle weakness and atrophy, muscle cramps, and fasciculations, with later onset of distal sensory impairment. The disorder is slowly progressive and clinically resembles amyotrophic lateral sclerosis. {ECO:0000269|PubMed:22883144}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 5A, Otozomal Resesif, 270800, Otozomal Resesif; SPG5A (Otozomal Resesif Spastik Parapleji Tip 5A) (CYP7B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 5A, autosomal recessive, 270800, Autosomal recessive; SPG5A (Autosomal recessive spastic paraplegia type 5A) (CYP7B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP7B1	CYP7B1, CBAS3, SPG5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital bile acid synthesis defect 3 (CBAS3) [MIM:613812]: A disorder resulting in severe cholestasis, cirrhosis and liver synthetic failure. Hepatic microsomal oxysterol 7-alpha-hydroxylase activity is undetectable. {ECO:0000269|PubMed:9802883}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 6, Otozomal Dominant, 600363, Otozomal Dominant; SPG6 (Otozomal Dominant Spastik Parapleji Tip 6) (NIPA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 6, autosomal dominant, 600363, Autosomal dominant; SPG6 (Autosomal dominant spastic paraplegia type 6) (NIPA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NIPA1	NIPA1, SPG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 61, Otozomal Resesif, 615685, Otozomal Resesif; SPG61 (Otozomal Resesif Spastik Parapleji Tip 61) (ARL6IP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 61, autosomal recessive, 615685, Autosomal recessive; SPG61 (Autosomal recessive spastic paraplegia type 61) (ARL6IP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARL6IP1	ARL6IP1, KIAA0069, SPG61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 61, autosomal recessive (SPG61) [MIM:615685]: A complicated form of spastic paraplegia with polysensory and motor neuropathy. Spastic paraplegia is a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:24482476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 62, 615681, Otozomal Resesif; SPG62 (Otozomal Resesif Spastik Parapleji Tip 62) (ERLIN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 62, 615681, Autosomal recessive; SPG62 (Autosomal recessive spastic paraplegia type 62) (ERLIN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERLIN1	ERLIN1, SPG62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 63, 615686, Otozomal Resesif; SPG63 (Otozomal Resesif Spastik Parapleji Tip 63) (AMPD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 63, 615686, Autosomal recessive; SPG63 (Autosomal recessive spastic paraplegia type 63) (AMPD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AMPD2	AMPD2, SPG63, PCH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 63, autosomal recessive (SPG63) [MIM:615686]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:24482476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 64, Otozomal Resesif, 615683, Otozomal Resesif; SPG64 (Otozomal Resesif Spastik Parapleji Tip 64) (ENTPD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 64, autosomal recessive, 615683, Autosomal recessive; SPG64 (Autosomal recessive spastic paraplegia type 64) (ENTPD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENTPD1	ENTPD1, CD39, SPG64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 64, autosomal recessive (SPG64) [MIM:615683]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. {ECO:0000269|PubMed:24482476}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 7, Otozomal Resesif, 607259, Otozomal Resesif, Otozomal Dominant; SPG7 (Spastik Parapleji Tip 7) (SPG7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 7, autosomal recessive, 607259, Autosomal recessive, Autosomal dominant; SPG7 (Spastic paraplegia type 7) (SPG7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG7	PGN, SPG7, CMAR, CAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in SPG7 may cause autosomal recessive osteogenesis imperfecta (OI). Osteogenesis imperfecta defines a group of connective tissue disorders characterized by bone fragility and low bone mass. Clinical features of SPG7-related osteogenesis imperfecta include recurrent fractures, mild bone deformities, delayed tooth eruption, normal hearing and white sclera.	EDTA Blood Sample
Spastik Parapleji 72, Otozomal Dominant, 615625, Otozomal Resesif, Otozomal Dominant (Otozomal Spastik Parapleji Tip 72) (REEP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 72, autosomal dominant, 615625, Autosomal recessive, Autosomal dominant (Autosomal spastic paraplegia type 72) (REEP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REEP2	REEP2, C5orf19, SPG72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 73, Otozomal Dominant, 616282, Otozomal Dominant; SPG73 (Otozomal Dominant Spastik Parapleji Tip 73) (CPT1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 73, autosomal dominant, 616282, Autosomal dominant; SPG73 (Autosomal dominant spastic paraplegia type 73) (CPT1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPT1C	CPT1C, SPG73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 74, Otozomal Resesif, 616451, Otozomal Resesif; SPG74 (Otozomal Resesif Spastik Parapleji Tip 74) (IBA57 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 74, autosomal recessive, 616451, Autosomal recessive; SPG74 (Autosomal recessive spastic paraplegia type 74) (IBA57 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IBA57	IBA57, C1orf69, MMDS3, SPG74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 74, autosomal recessive (SPG74) [MIM:616451]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG74 is characterized by a combination of spastic paraplegia, optic atrophy, and peripheral neuropathy with childhood-onset and slow progression into late adulthood. {ECO:0000269|PubMed:25609768}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Spastik Parapleji 75, Otozomal Resesif, 616680, Otozomal Resesif; SPG75 (Otozomal Resesif Spastik Parapleji Tip 75) (MAG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 75, autosomal recessive, 616680, Autosmal recessive; SPG75 (Autosomal recessive spastic paraplegia type 75) (MAG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAG	MAG, GMA, SPG75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 76, Otozomal Resesif, 616907, Otozomal Resesif; SPG76 (CAPN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 76, autosomal recessive, 616907, Autosmal recessive; SPG76 (CAPN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAPN1	CAPN1, SPG76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 77, Otozomal Resesif, 617046, Otozomal Resesif; SPG77 (Otozomal Resesif Spastik Parapleji Tip 77) (FARS2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 77, autosomal recessive, 617046, Autosmal recessive; SPG77 (Autosomal recessive spastic paraplegia type 77) (FARS2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FARS2	FARS2, FARS1, COXPD14, SPG77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Combined oxidative phosphorylation deficiency 14 (COXPD14) [MIM:614946]: A severe multisystemic autosomal recessive disorder characterized by neonatal onset of global developmental delay, refractory seizures, and lactic acidosis. Biochemical studies show deficiencies of multiple mitochondrial respiratory enzymes. {ECO:0000269|PubMed:22495541, ECO:0000269|PubMed:22833457}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 78, Otozomal Resesif, 617225, Otozomal Resesif; SPG78 (ATP13A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 78, autosomal recessive,, 617225, Autosmal recessive; SPG78 (ATP13A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP13A2	ATP13A2, PARK9, KRPPD, SPG78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 12 (CLN12) [MIM:606933]: A form of neuronal ceroid lipofuscinosis characterized by rapidly progressive visual loss due to retinal dystrophy, seizures, cerebellar ataxia, and cerebellar atrophy. Cognitive decline may also occur. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material. {ECO:0000269|PubMed:22388936}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 79, Otozomal Resesif, 615491, Otozomal Resesif; SPG79 (Erken Başlangıçlı Progresif Nörodejenerasyon-Körlük-Ataksi-Spastisite Sendromu) (UCHL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 79, autosomal recessive, 615491, Autosmal recessive; SPG79 (Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome) (UCHL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UCHL1	UCHL1, PARK5, SPG79, NDGOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Parkinson disease 5 (PARK5) [MIM:613643]: A complex neurodegenerative disorder with manifestations ranging from typical Parkinson disease to dementia with Lewy bodies. Clinical features include parkinsonian symptoms (resting tremor, rigidity, postural instability and bradykinesia), dementia, diffuse Lewy body pathology, autonomic dysfunction, hallucinations and paranoia. {ECO:0000269|PubMed:9774100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 79, Otozomal Resesif, 615491, Otozomal Resesif; SPG79 (Erken Başlangıçlı Progresif Nörodejenerasyon-Körlük-Ataksi-Spastisite Sendromu) (MLPA)	Spastic paraplegia 79, autosomal recessive, 615491, Autosmal recessive; SPG79 (Early-onset progressive neurodegeneration-blindness-ataxia-spasticity syndrome) (MLPA)	UCHL1	UCHL1, PARK5, SPG79, NDGOA	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Parkinson disease 5 (PARK5) [MIM:613643]: A complex neurodegenerative disorder with manifestations ranging from typical Parkinson disease to dementia with Lewy bodies. Clinical features include parkinsonian symptoms (resting tremor, rigidity, postural instability and bradykinesia), dementia, diffuse Lewy body pathology, autonomic dysfunction, hallucinations and paranoia. {ECO:0000269|PubMed:9774100}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spastik Parapleji 8, Otozomal Dominant, 603563, Otozomal Dominant; SPG8 (Otozomal Dominant Spastik Parapleji Tip 8) (WASHC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 8, autosomal dominant, 603563, Autosmal dominant; SPG8 (Autosomal dominant spastic paraplegia type 8) (WASHC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WASHC5	KIAA0196, SPG8, RTSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji 9A, Otozomal Dominant, 601162, Otozomal Dominant; SPG9A (Otozomal Dominant Komplekis Spastik Parapleji Tip 9A) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 9A, autosomal dominant, 601162, Autosmal dominant; SPG9A (Autosomal dominant complex spastic paraplegia type 9A) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SPG9A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, atheroid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. {ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18478038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji 9B, Otozomal Resesif, 616586, Otozomal Resesif; SPG9B (Otozomal Resesif Komplekis Spastik Parapleji Tip 9B) (ALDH18A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia 9B, autosomal recessive, 616586, Autosmal recessive; SPG9B (Autosomal recessive complex spastic paraplegia type 9B) (ALDH18A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH18A1	ALDH18A1, PYCS, GSAS, ARCL3A, SPG9A, SPG9B, ADCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal recessive, 3A (ARCL3A) [MIM:219150]: A syndrome characterized by facial dysmorphism with a progeroid appearance, large and late-closing fontanel, cutis laxa, joint hyperlaxity, atheroid movements and hyperreflexia, pre- and postnatal growth retardation, intellectual deficit, developmental delay, and ophthalmologic abnormalities. {ECO:0000269|PubMed:11092761, ECO:0000269|PubMed:18478038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spastik Parapleji, Psikomotor Retardasyon, ± Nöbet 616756, Otozomal Resesif; SPPRS (Spastik Parapleji-Ağır Gelişim Gecikmesi-Epilepsi Sendromu) (Prenatal) (HACE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia and psychomotor retardation with or without seizures, 616756, Autosomal recessive; SPPRS (Spastic paraplegia-severe developmental delay-epilepsy syndrome) (HACE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HACE1	HACE1, KIAA1320, SPPRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in HACE1 are a cause of Wilms tumor (WT). WT is a pediatric malignancy of kidney and one of the most common solid cancers in childhood. HACE1 is epigenetically down-regulated in sporadic Wilms tumor. Moreover, a t(5;6)(q21;q21) translocation that truncates HACE1 has been found in a child with bilateral, young-onset Wilms tumor (PubMed:19948536). {ECO:0000269|PubMed:17694067, ECO:0000269|PubMed:19948536}.;	EDTA Blood Sample
Spastik Parapleji, Psikomotor Retardasyon, ± Nöbet 616756, Otozomal Resesif; SPPRS (Spastik Parapleji-Ağır Gelişim Gecikmesi-Epilepsi Sendromu) (Prenatal) (HACE1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spastic paraplegia and psychomotor retardation with or without seizures, 616756, Autosmal recessive; SPPRS (Spastic paraplegia-severe developmental delay-epilepsy syndrome) (HACE1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HACE1	HACE1, KIAA1320, SPPRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Defects in HACE1 are a cause of Wilms tumor (WT). WT is a pediatric malignancy of kidney and one of the most common solid cancers in childhood. HACE1 is epigenetically down-regulated in sporadic Wilms tumor. Moreover, a t(5;6)(q21;q21) translocation that truncates HACE1 has been found in a child with bilateral, young-onset Wilms tumor (PubMed:19948536). {ECO:0000269|PubMed:17694067, ECO:0000269|PubMed:19948536}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spastik parapleji, kalıtsal (HSP) (SPG3A 14q21, SPAST 2p22) (MLPA)	Spastic paraplegia, hereditary (HSP) (SPG3A 14q21, SPAST 2p22) (MLPA)	SPG3A 14q21, SPAST 2p22	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Spastik Parapleji, Entellektüel Gerilik, Nistagmus ve Obezite, 617296, Otozomal Dominant; SINO (KIDINS220 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia, intellectual disability, nystagmus, and obesity, 617296, Autosmal dominant; SINO (KIDINS220 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIDINS220	KIDINS220, ARMS, SINO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Parapleji, Optik Atrofi, ve Nöropati, 609541, Otozomal Resesif; SPOAN (Spastik Parapleji-Optik Atrofi-Nöropati Sendromu) (KLC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic paraplegia, optic atrophy, and neuropathy, 609541, Autosmal recessive; SPOAN (Spastic paraplegia-optic atrophy-neuropathy syndrome) (KLC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLC2	KLC2, SPOAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Tetraplejİ, İnce Korpus Kallozum ve Progresif Mikrosefali, 616657, Otozomal Resesif; SPATCCM (Spastik Tetraplejİ-İnce Korpus Kallozum-Progresif Postnatal Mikrosefali Sendromu) (SLC1A4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657, Autosmal recessive; SPATCCM (Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome) (SLC1A4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC1A4	SLC1A4, SATT, ASCT1, SPATCCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spastik Tetraplejİ, İnce Korpus Kallozum ve Progresif Mikrosefali, 616657, Otozomal Resesif; SPATCCM (Spastik Tetraplejİ-İnce Korpus Kallozum-Progresif Postnatal Mikrosefali Sendromu) (Prenatal) (SLC1A4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spastic tetraplegia, thin corpus callosum, and progressive microcephaly, 616657, Autosmal recessive; SPATCCM (Spastic tetraplegia-thin corpus callosum-progressive postnatal microcephaly syndrome) (SLC1A4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC1A4	SLC1A4, SATT, ASCT1, SPATCCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spastisite, Çocuklukta Başlangıç, Hiperglisinemi, 616859, Otozomal Resesif; SPAHGC (Spastisite-Ataksi-Yürüme Anormallikleri Sendromu) (GLRX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spasticity, childhood-onset, with hyperglycinemia, 616859, Autosmal recessive; SPAHGC (Spasticity-ataxia-gait anomalies syndrome) (GLRX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLRX5	GLRX5, C14orf87, PRO1238, FLB4739, PRSA, SIDBA3, SPAHGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Anemia, sideroblastic, pyridoxine-refractory, autosomal recessive (PRARSA) [MIM:205950]: A form of sideroblastic anemia not responsive to pyridoxine. Sideroblastic anemia is characterized by anemia of varying severity, hypochromic peripheral erythrocytes, systemic iron overload secondary to chronic ineffective erythropoiesis, and the presence of bone marrow ringed sideroblasts. Sideroblasts are characterized by iron-loaded mitochondria clustered around the nucleus. {ECO:0000269|PubMed:17485548, ECO:0000269|PubMed:20364084, ECO:0000269|PubMed:25342667, ECO:0000269|PubMed:26100117}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spesifik Granül Eksikliği, 245480, Otozomal Resesif; SGD (Tekrarlayan Enfeksiyon, Spesifik Granül Eksikliğine Bağlı) (CEBPE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Specific granule deficiency, 245480, Autosmal recessive; SGD (Recurrent infection due to specific granule deficiency) (CEBPE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CEBPE	CEBPE, CRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spesifik Konuşma Bozukluğu 5, 615432, Otozomal Dominant; SLI5 (TM4SF20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Specific language impairment 5, 615432, Autosomal dominant; SLI5 (TM4SF20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TM4SF20	TM4SF20, SLI5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Konuşma-Dil Bozukluğu-1, 602081, Otozomal Dominant; SPCH1 (Çocukluk Çağ Konuşma Apraksisi) (FOXP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Speech-language disorder-1, 602081, Autosmal dominant; SPCH1 (Childhood apraxia of speech) (FOXP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXP2	FOXP2, SPCH1, TNRC10, CAGH44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Speech-language disorder 1 (SPCH1) [MIM:602081]: A disorder characterized by severe orofacial dyspraxia resulting in largely incomprehensible speech. Affected individuals have severe impairment in the selection and sequencing of fine orofacial movements which are necessary for articulation, and deficits in several facets of grammatical skills and language processing, such as the ability to break up words into their constituent phonemes. {ECO:0000269|PubMed:11586359}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sperm FISH	.	.	.	FISH	Sperm (Transport besi yeri içinde)		10	30	.	.	20	Postnatal	.	Sperm (Transport besi yeri içinde)
Spermatojenik bozukluk 10, 614822 (Sendromik Olmayan Erkek Infertilitesi, Sperm Motilite Bozukluğuna Bağlı) (SEPT12 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 10, 614822 (3) (Non-syndromic male infertility due to sperm motility disorder) (SEPT12 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SEPT12	SEPT12, SPGF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik 11, 615081, Otozomal Dominant; SPGF11 (Erkek Infertilitesi - Azoospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (KLHL10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 11, 615081, Autosomal dominant; SPGF11 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (KLHL10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KLHL10	KLHL10, SPGF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 11 (SPGF11) [MIM:615081]: An infertility disorder caused by spermatogenesis defects. It results in decreased sperm motility, concentration, and multiple sperm structural defects. Oligozoospermia is usually observed in SPGF11 patients. In addition to oligozoospermia, teratozoospermia and moderate asthenozoospermia is observed in some cases. {ECO:0000269|PubMed:17047026}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 12, 615413, Otozomal Dominant; SPGF12 (Erkek Infertilitesi-Teratozoospermi, Tek Gen Mutasyonuna Bağlı) (NANOS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 12, 615413, Autosomal dominant; SPGF12 (Male infertility with teratozoospermia due to single gene mutation) (NANOS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NANOS1	NANOS1, NOS1, SPGF12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 12 (SPGF12) [MIM:615413]: An infertility disorder caused by spermatogenesis defects. It results in decreased sperm motility, concentration, and multiple sperm structural defects. Non-obstructive azoospermia, oligozoospermia and oligo-astheno-teratozoospermia are features observed in SPGF12 patients. {ECO:0000269|PubMed:23315541}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Spermatojenik Yetmezlik 13, 615841, Otozomal Resesif; SPGF13 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (TAF4B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 13, 615841, Autosomal recessive; SPGF13 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (TAF4B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAF4B	TAF4B, TAF2C2, TAFII105, SPGF13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 13 (SPGF13) [MIM:615841]: A disorder resulting in the absence (azoospermia) or reduction (oligozoospermia) of sperm in the semen, leading to male infertility. {ECO:0000269	PubMed:24431330}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 14, 615842, Otozomal Resesif; SPGF14 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (ZMYND15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 14, 615842, Autosomal recessive; SPGF14 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (ZMYND15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZMYND15	ZMYND15, SPGF14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 14 (SPGF14) [MIM:615842]: A disorder resulting in the absence (azoospermia) or reduction (oligozoospermia) of sperm in the semen, leading to male infertility. {ECO:0000269	PubMed:24431330}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 15, 616950, Otozomal Resesif; SPGF15 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (SYCE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 15, 616950, Autosomal recessive; SPGF15 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (SYCE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYCE1	SYCE1, POF12, SPGF15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik 16, 617187, Otozomal Resesif; SPGF16 (SUN5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 16, 617187, Autosomal recessive; SPGF16 (SUN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUN5	SUN5, TSARG4, SPAG4L, SPGF16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik 17, 617214, Otozomal Resesif; SPGF17 (PLCZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 17, 617214, Autosomal recessive; SPGF17 (PLCZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLCZ1	PLCZ1, SPGF17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik 3, 606766, Otozomal Dominant; SPGF3 (Sendromik Olmayan Erkek İnfertilitesi, Sperm Motilite Bozukluğuna Bağlı) (SLC26A8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 3, 606766, Autosomal dominant; SPGF3 (Non-syndromic male infertility due to sperm motility disorder) (SLC26A8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC26A8	SLC26A8, TAT1, SPGF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 3 (SPGF3) [MIM:606766]: A disorder characterized by primary infertility, sperm morphologic abnormalities, and moderate to severe asthenozoospermia, condition in which the percentage of progressively motile sperm is abnormally low. {ECO:0000269	PubMed:23582645}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetersizlik 4, 270960, Otozomal Dominant; SPGF4 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (SYCP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 4, 270960, Autosomal dominant; SPGF4 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (SYCP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYCP3	SYCP3, SCP3, COR1, SPGF4, RPRGL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 4 (SPGF4) [MIM:270960]: An infertility disorder characterized by azoospermia, a condition of having no sperm present in the ejaculate. Testicular histology shows arrest of spermatogenesis at the pachytene stage of primary spermatocytes. {ECO:0000269	PubMed:14643120}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 5, 243060, Otozomal Resesif; SPGF5 (Erkek İnfertilitesi, Büyük Başlı Multiflagellar Poliploid Sperme Bağlı) (AURKC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 5, 243060, Autosomal recessive; SPGF5 (Male infertility due to large-headed multiflagellar polyploid spermatozoa) (AURKC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AURKC	AURKC, STK13, AIE2, SPGF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 5 (SPGF5) [MIM:243060]: An infertility disorder caused by spermatogenesis defects. Semen from affected men show close to 100% morphologically abnormal multiflagellar spermatozoa with low motility, oversized irregular heads, and abnormal midpiece and acrosome. {ECO:0000269|PubMed:17435757, ECO:0000269|PubMed:21733974}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 6, 102530, Otozomal Resesif (Erkek İnfertilitesi, Globozoospermiye Bağlı) (SPATA16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 6, 102530, Autosomal recessive (Male infertility due to globozoospermia) (SPATA16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPATA16	SPATA16, SPGF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik 7, 612997, Otozomal Resesif; SPGF7 (Sendromik Olmayan Erkek İnfertilitesi, Sperm Motilite Bozukluğuna Bağlı) (CATSPER1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 7, 612997, Autosomal recessive; SPGF7 (Non-syndromic male infertility due to sperm motility disorder) (CATSPER1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CATSPER1	CATSPER1, CATSPER, SPGF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spermatogenic failure 7 (SPGF7) [MIM:612997]: An infertility disorder characterized by non-motile sperm or sperm motility below the normal threshold, low sperm count, increased abnormally structured spermatozoa, and reduced semen volume. {ECO:0000269	PubMed:19344877}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spermatojenik Yetmezlik 8, 613957, Otozomal Resesif; SPGF8 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (NR5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 8, 613957, Autosomal recessive; SPGF8 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (NR5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, AD4BP, POF7, SRXY3, SPGF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. {ECO:0000269	PubMed:11038323}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spermatojenik Yetmezlik 8, 613957, Otozomal Resesif; SPGF8 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (MLPA)	Spermatogenic failure 8, 613957, Autosomal recessive; SPGF8 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (MLPA)	NR5A1	NR5A1, FTZF1, FTZ1, SF1, AD4BP, POF7, SRXY3, SPGF8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Adrenocortical insufficiency, without ovarian defect (ACIWOD) [MIM:184757]: A disorder that is characterized by severe 'slackness,' muscular hypotonia. There is decreased sodium, increased potassium and elevated ACTH. {ECO:0000269	PubMed:11038323}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spermatojenik Yetmezlik 9, 613958, Otozomal Resesif; SPGF9 (Erkek İnfertilitesi, Globozoospermiye Bağlı) (DPY19L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure 9, 613958, Autosomal recessive; SPGF9 (Male infertility due to globozoospermia) (DPY19L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPY19L2	DPY19L2, SPGF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik, Yatkınlık (DAZL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure, susceptibility to (DAZL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DAZL	DAZL, DAZH, SPGYLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik, 309120, X'e Bağlı, 2, X'e Bağlı resesif; SPGFX2 (Erkek İnfertilitesi - Azospermi Veya Oligozoospermi, Tek Gen Mutasyonuna Bağlı) (TEX11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spermatogenic failure, X-linked, 2, 309120, X-linked recessive; SPGFX2 (Male infertility with azoospermia or oligozoospermia due to single gene mutation) (TEX11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TEX11	TEX11, SPGFX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetersizlik, Y Kromozomal, 1, 400042 (Y Delesyonu-Parsiyel)	Spermatogenic failure, Y-linked, 1, 400042 (Partial chromosome Y deletion)	.	DELYq11, CYDEL q11, SPGFY1	PCR/ Jel elektroforezi	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Spermatojenik Yetmezlik, Y Kromozomal, 2, 415000, Y-linked; SPGFY2 (Y Delesyonu-Parsiyel) (Y kromozom mikrodelesyon testi) (SRY, AZFa, AZFb, AZFc)	Spermatogenic failure, Y-linked, 2, 415000, Y-linked; SPGFY2 (Partial chromosome Y deletion) (Y chromosome microdeletion test) (SRY, AZFa, AZFb, AZFc)	USP9Y	USP9Y, DFFRY, SPGFY2	PCR/ Jel elektroforezi	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spermatogenic failure Y-linked 2 (SPGFY2) [MIM:415000]: A disorder resulting in the absence (azoospermia) or reduction (oligozoospermia) of sperm in the semen, leading to male infertility. {ECO:0000269|PubMed:10581029}. Note=The disease may be caused by mutations affecting the gene represented in this entry. The role of USP9Y in spermatogenesis failure is uncertain {PubMed:19246359}. A 4-bp deletion in a splice-donor site, causing exon skipping and protein truncation has been observed in non- obstructive azoospermia {PubMed:10581029}. However, complete USP9Y deletion has been detected in individuals with no spermatogenic defects {PubMed:19246359}. {ECO:0000269|PubMed:10581029, ECO:0000269|PubMed:19246359};.	EDTA Blood Sample
Sferositozis, Tip 1, 182900, Otozomal Dominant; SPH1 (Kalıtsal Sferositoz) (ANK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spherocytosis, type 1, 182900, Autosomal dominant; SPH1 (Hereditary spherocytosis) (ANK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANK1	ANK1, SPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spherocytosis 1 (SPH1) [MIM:182900]: Spherocytosis is a hematologic disorder leading to chronic hemolytic anemia and characterized by numerous abnormally shaped erythrocytes which are generally spherocidal. SPH1 is characterized by severe hemolytic anemia. Inheritance is autosomal recessive. {ECO:0000269|PubMed:11029885, ECO:0000269|PubMed:8640229}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sferositozis, Tip 2, 616649, Otozomal Dominant; SPH2 (Kalıtsal Sferositoz) (SPTB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spherocytosis, type 2, 616649, Autosomal dominant; SPH2 (Hereditary spherocytosis) (SPTB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTB	SPTB, SPH2, EL3, HS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 3 (EL3) [MIM:182870]: A Rhesus-unlinked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. {ECO:0000269|PubMed:1975598, ECO:0000269|PubMed:7883966, ECO:0000269|PubMed:8018926, ECO:0000269|PubMed:8226774}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sferositozis, Tip 3, 270970, Otozomal Resesif; SPH3 (Kalıtsal Sferositoz) (SPTA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spherocytosis, type 3, 270970, Autosomal recessive; SPH3 (Hereditary spherocytosis) (SPTA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTA1	SPTA1, EL2, SPH3, HS3, HPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Elliptocytosis 2 (EL2) [MIM:130600]: A Rhesus-unlinked form of hereditary elliptocytosis, a genetically heterogeneous, autosomal dominant hematologic disorder. It is characterized by variable hemolytic anemia and elliptical or oval red cell shape. {ECO:0000269|PubMed:1541880, ECO:0000269|PubMed:1638030, ECO:0000269|PubMed:1679439, ECO:0000269|PubMed:1878597, ECO:0000269|PubMed:2384601, ECO:0000269|PubMed:2568861, ECO:0000269|PubMed:2568862, ECO:0000269|PubMed:2794061, ECO:0000269|PubMed:7772539, ECO:0000269|PubMed:8018926, ECO:0000269|PubMed:8193371, ECO:0000269|PubMed:8364215}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sferositozis, Tip 4, 612653, Otozomal Resesif; SPH4 (Kalıtsal Sferositoz) (SLC4A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spherocytosis, type 4, 612653, autosomal recessive; SPH4 (Hereditary spherocytosis) (SLC4A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC4A1	SLC4A1, AE1, EPB3, SPH4, SAO, CHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ovalocytosis, Southeast Asian (SAO) [MIM:166900]: A hereditary hematologic disorder characterized by ovalocytic erythrocytes that are rigid and exhibit reduced expression of many erythrocyte antigens. Clinical manifestations include mild hemolysis, intermittent jaundice and gallstones. However, the disorder is most often asymptomatic. {ECO:0000269|PubMed:1538405, ECO:0000269|PubMed:1722314}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sferositozis, Tip 5, 612690; SPH5 (Kalıtsal Sferositoz) (EPB42 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spherocytosis, type 5, 612690; SPH5 (Hereditary spherocytosis) (EPB42 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPB42	EPB42, SPH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spherocytosis 5 (SPH5) [MIM:612690]: Spherocytosis is a hematologic disorder leading to chronic hemolytic anemia and characterized by numerous abnormally shaped erythrocytes which are generally spherocidal. Absence of band 4.2 associated with spur or target erythrocytes has also been reported. {ECO:0000269|PubMed:10406914, ECO:0000269|PubMed:1558976, ECO:0000269|PubMed:7772513, ECO:0000269|PubMed:7810064, ECO:0000269|PubMed:8547071, ECO:0000269|PubMed:8547605}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spina Bifida, Folat Duyarlı, Yatkınlık, 601634, Otozomal Resesif (Nöral Tüp Kapanma Defekti) (MTHFD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spina bifida, folate-sensitive, susceptibility to, 601634, Autosomal recessive (Neural tube closure defect) (MTHFD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFD1	MTHFD, MTHFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Colorectal cancer (CRC) [MIM:114500]: A complex disease characterized by malignant lesions arising from the inner wall of the large intestine (the colon) and the rectum. Genetic alterations are often associated with progression from premalignant lesion (adenoma) to invasive adenocarcinoma. Risk factors for cancer of the colon and rectum include colon polyps, long-standing ulcerative colitis, and genetic family history. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Spina Bifida, Yatkınlık, 182940, Otozomal Dominant (Nöral Tüp Kapanma Defekti) (CCL2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spina bifida, susceptibility to, 182940, Autosomal dominant (Neural tube closure defect) (CCL2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCL2	CCL2, SCYA2, MCP1, MCAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Kennedy Tipi, Spinal ve Bulber Müsküler Atrofi, 313200, X'e Bağlı Resesif; SMAX1 (Kennedy Hastalığı) (Tekrar Sayısı)	Spinal and bulbar muscular atrophy of Kennedy, 313200, X-linked recessive; SMAX1 (Kennedy disease)(Repeat Analysis)	AR	AR, DHTR, TFM, SBMA, KD, SMAX1, HYSP1	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Androgen insensitivity syndrome (AIS) [MIM:300068]: An X- linked recessive form of pseudohermaphroditism due end-organ resistance to androgen. Affected males have female external genitalia, female breast development, blind vagina, absent uterus and female adnexa, and abdominal or inguinal testes, despite a normal 46,XY karyotype. {ECO:0000269|PubMed:10022458, ECO:0000269|PubMed:10221692, ECO:0000269|PubMed:10221770, ECO:0000269|PubMed:10404311, ECO:0000269|PubMed:10458483, ECO:0000269|PubMed:10571951, ECO:0000269|PubMed:10590024, ECO:0000269|PubMed:10690872, ECO:0000269|PubMed:11587068, ECO:0000269|PubMed:11744994, ECO:0000269|PubMed:1307250, ECO:0000269|PubMed:1316540, ECO:0000269|PubMed:1426313, ECO:0000269|PubMed:1430233, ECO:0000269|PubMed:1464650, ECO:0000269|PubMed:1480178, ECO:0000269|PubMed:1487249, ECO:0000269|PubMed:1569163, ECO:0000269|PubMed:1609793, ECO:0000269|PubMed:1775137, ECO:0000269|PubMed:1999491, ECO:0000269|PubMed:2082179, ECO:0000269|PubMed:2594783, ECO:0000269|PubMed:7537149, ECO:0000269|PubMed:7581399, ECO:0000269|PubMed:7633398,	EDTA Blood Sample
Spinal Müsküler Atrofi (SMA) (SMN1, SMN2, 5q13) (MLPA)	Spinal Muscular Atrophy (SMA) (SMN1, SMN2, 5q13) (MLPA)	SMN1, SMN2, 5q13	.	MLPA	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Spinal Müsküler Atrofi (SMA) (SMN1, SMN2, 5q13) (MLPA)	Spinal Muscular Atrophy (SMA) (SMN1, SMN2, 5q13) (MLPA)	SMN1, SMN2, 5q13	.	MLPA	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Spinal Müsküler Atrofi (SMA) (SMN1, SMN2, 5q13) (MLPA) (Prenatal)	Spinal Muscular Atrophy (SMA) (SMN1, SMN2, 5q13) (MLPA) (Prenatal)	SMN1, SMN2, 5q13	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi (SMA) (SMN1, SMN2, 5q13) (MLPA) (Prenatal)	Spinal Muscular Atrophy (SMA) (SMN1, SMN2, 5q13) (MLPA) (Prenatal)	SMN1, SMN2, 5q13	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-Konjenital Kemik Kırıkları 1, 616866, Otozomal Resesif; SMABF1 (TRIP4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy with congenital bone fractures 1, 616866, Autosomal recessive; SMABF1 (TRIP4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRIP4	TRIP4, ASC1, SMABF1, MDCDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinal Müsküler Atrofi-Konjenital Kemik Kırıkları 1, 616866, Otozomal Resesif; SMABF1 (Prenatal) (TRIP4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy with congenital bone fractures 1, 616866, Autosomal recessive; SMABF1 (TRIP4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRIP4	TRIP4, ASC1, SMABF1, MDCDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-Konjenital Kemik Kırıkları 2, 616867, Otozomal Resesif; SMABF2 (ASCC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy with congenital bone fractures 2, 616867, Autosomal recessive; SMABF2 (ASCC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASCC1	ASCC1, p50, SMABF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinal Müsküler Atrofi-Konjenital Kemik Kırıkları 2, 616867, Otozomal Resesif; SMABF2 (Prenatal) (ASCC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy with congenital bone fractures 2, 616867, Autosomal recessive; SMABF2 (ASCC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASCC1	ASCC1, p50, SMABF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-Progresif Miyoklonik Epilepsi, 159950, Otozomal Resesif; SMAPME (Spinal Müsküler Atrofi-Progresif Miyoklonik Epilepsi) (ASAH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950, Autosomal recessive; SMAPME (Spinal muscular atrophy-progressive myoclonic epilepsy) (ASAH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASAH1	ASAH1, AC, SMAPME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy with progressive myoclonic epilepsy (SMAPME) [MIM:159950]: An autosomal recessive neuromuscular disorder characterized by childhood onset of motor deficits and progressive myoclonic seizures, after normal developmental milestones. Proximal muscle weakness and generalized muscular atrophy are due to degeneration of spinal motor neurons. Myoclonic epilepsy is generally resistant to conventional therapy. The disease course is progressive and leads to respiratory muscle involvement and severe handicap or early death from respiratory insufficiency. {ECO:0000269|PubMed:22703880, ECO:0000269|PubMed:24164096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi-Progresif Miyoklonik Epilepsi, 159950, Otozomal Resesif; SMAPME (Spinal Müsküler Atrofi-Progresif Miyoklonik Epilepsi) (Prenatal) (ASAH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy with progressive myoclonic epilepsy, 159950, Autosomal recessive; SMAPME (Spinal muscular atrophy-progressive myoclonic epilepsy) (ASAH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ASAH1	ASAH1, AC, SMAPME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy with progressive myoclonic epilepsy (SMAPME) [MIM:159950]: An autosomal recessive neuromuscular disorder characterized by childhood onset of motor deficits and progressive myoclonic seizures, after normal developmental milestones. Proximal muscle weakness and generalized muscular atrophy are due to degeneration of spinal motor neurons. Myoclonic epilepsy is generally resistant to conventional therapy. The disease course is progressive and leads to respiratory muscle involvement and severe handicap or early death from respiratory insufficiency. {ECO:0000269|PubMed:22703880, ECO:0000269|PubMed:24164096}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-1, 253300, Otozomal Resesif; SMA1 (Proksimal Spinal Müsküler Atrofi) (SMN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy-1, 253300, Autosomal recessive; SMA1 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-1, 253300, Otozomal Resesif; SMA1 (Proksimal Spinal Müsküler Atrofi) (MLPA)	Spinal muscular atrophy-1, 253300, Autosomal recessive; SMA1 (Proximal spinal muscular atrophy) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-1, 253300, Otozomal Resesif; SMA1 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (SMN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy-1, 253300, Autosomal recessive; SMA1 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-1, 253300, Otozomal Resesif; SMA1 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (MLPA)	Spinal muscular atrophy-1, 253300, Autosomal recessive; SMA1 (Proximal spinal muscular atrophy) (Prenatal) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spinal Müsküler Atrofi-2, 253550, Otozomal Resesif; SMA2 (Proksimal Spinal Müsküler Atrofi) (SMN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy-2, 253550, Autosomal recessive; SMA2 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-2, 253550, Otozomal Resesif; SMA2 (Proksimal Spinal Müsküler Atrofi) (MLPA)	Spinal muscular atrophy-2, 253550, Autosomal recessive; SMA2 (Proximal spinal muscular atrophy) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-2, 253550, Otozomal Resesif; SMA2 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (SMN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy-2, 253550, Autosomal recessive; SMA2 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-2, 253550, Otozomal Resesif; SMA2 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (MLPA)	Spinal muscular atrophy-2, 253550, Autosomal recessive; SMA2 (Proximal spinal muscular atrophy) (Prenatal) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-3, 253400, Otozomal Resesif; SMA3 (Proksimal Spinal Müsküler Atrofi) (SMN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy-3, 253400, Autosomal recessive; SMA3 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-3, 253400, Otozomal Resesif; SMA3 (Proksimal Spinal Müsküler Atrofi) (MLPA)	Spinal muscular atrophy-3, 253400, Autosomal recessive; SMA3 (Proximal spinal muscular atrophy) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-3, 253400, Otozomal Resesif; SMA3 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (SMN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy-3, 253400, Autosomal recessive; SMA3 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-3, 253400, Otozomal Resesif; SMA3 (Proksimal Spinal Müsküler Atrofi) (Prenatal) (MLPA)	Spinal muscular atrophy-3, 253400, Autosomal recessive; SMA3 (Proximal spinal muscular atrophy) (Prenatal) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spinal Müsküler Atrofi-4, 271150, Otozomal Resesif; SMA4 (Proksimal Spinal Müskül Atrofi) (SMN1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy-4, 271150, Autosomal recessive; SMA4 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-4, 271150, Otozomal Resesif; SMA4 (Proksimal Spinal Müskül Atrofi) (MLPA)	Spinal muscular atrophy-4, 271150, Autosomal recessive; SMA4 (Proximal spinal muscular atrophy) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi-4, 271150, Otozomal Resesif; SMA4 (Proksimal Spinal Müskül Atrofi) (Prenatal) (SMN1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy-4, 271150, Autosomal recessive; SMA4 (Proximal spinal muscular atrophy) (SMN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi-4, 271150, Otozomal Resesif; SMA4 (Proksimal Spinal Müskül Atrofi) (Prenatal) (MLPA)	Spinal muscular atrophy-4, 271150, Autosomal recessive; SMA4 (Proximal spinal muscular atrophy) (Prenatal) (MLPA)	SMN1	SMN1, SMA1, SMA2, SMA3, SMA4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, Otozomal Resesif; 2, 605726, Otozomal Resesif; DSMA2 (Distal Kalıtsal Motor Nöropati, Jerash Tipi) (SIGMAR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, distal, autosomal recessive, 2, 605726, Autosomal recessive; DSMA2 (Distal hereditary motor neuropathy, Jerash type) (SIGMAR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIGMAR1	SIGMAR1, SRBP, ALS16, DSMA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Distal spinal muscular atrophy, autosomal recessive, 2 (DSMA2) [MIM:605726]: An autosomal recessive neuromuscular disorder characterized by onset of distal muscle weakness and wasting affecting the lower and upper limbs in the first decade. There is no sensory involvement. {ECO:0000269	PubMed:26078401}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, Otozomal Resesif; 2, 605726, Otozomal Resesif; DSMA2 (Distal Kalıtsal Motor Nöropati, Jerash Tipi) (Prenatal) (SIGMAR1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, distal, autosomal recessive, 2, 605726, Autosomal recessive; DSMA2 (Distal hereditary motor neuropathy, Jerash type) (SIGMAR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIGMAR1	SIGMAR1, SRBP, ALS16, DSMA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, autosomal recessive, 2 (DSMA2) [MIM:605726]: An autosomal recessive neuromuscular disorder characterized by onset of distal muscle weakness and wasting affecting the lower and upper limbs in the first decade. There is no sensory involvement. {ECO:0000269	PubMed:26078401}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi,Distal, Otozomal Resesif Otozomal Resesif, 4, 611067; DSMA4 (Otozomal Resesif Alt Motor Nöron Hastalığı-Çocukluk Çağı Başlangıçlı) (PLEKHG5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067, Autosomal recessive; DSMA4 (Autosomal recessive lower motor neuron disease with childhood onset) (PLEKHG5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLEKHG5	PLEKHG5, KIAA0720, DSMA4, CMTRIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease, recessive, intermediate type, C (CMTRIC) [MIM:613376]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:23777631, ECO:0000269|PubMed:23844677}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinal Müsküler Atrofi,Distal, Otozomal Resesif Otozomal Resesif, 4, 611067; DSMA4 (Otozomal Resesif Alt Motor Nöron Hastalığı-Çocukluk Çağı Başlangıçlı) (Prenatal) (PLEKHG5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, distal, autosomal recessive, 4, 611067, Autosomal recessive; DSMA4 (Autosomal recessive lower motor neuron disease with childhood onset) (PLEKHG5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PLEKHG5	PLEKHG5, KIAA0720, DSMA4, CMTRIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Charcot-Marie-Tooth disease, recessive, intermediate type, C (CMTRIC) [MIM:613376]: A form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Recessive intermediate forms of Charcot-Marie-Tooth disease are characterized by clinical and pathologic features intermediate between demyelinating and axonal peripheral neuropathies, and motor median nerve conduction velocities ranging from 25 to 45 m/sec. {ECO:0000269|PubMed:23777631, ECO:0000269|PubMed:23844677}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, Otozomal Resesif; 5, 614881, Otozomal Resesif; DSMA5 (Geniş Yetişkin Başlangıçlı Distal Kalıtsal Motor Nöropati) (DNAJB2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881, Autosomal recessive; DSMA5 (Young adult-onset distal hereditary motor neuropathy) (DNAJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNAJB2	DNAJB2, HSJ1, HSPF3, DSMA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2T (CMT2T) [MIM:616233]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. {ECO:0000269|PubMed:24627108, ECO:0000269|PubMed:25274842}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, Otozomal Resesif; 5, 614881, Otozomal Resesif; DSMA5 (Geniş Yetişkin Başlangıçlı Distal Kalıtsal Motor Nöropati) (Prenatal) (DNAJB2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, distal, autosomal recessive, 5, 614881, Autosomal recessive; DSMA5 (Young adult-onset distal hereditary motor neuropathy) (DNAJB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNAJB2	DNAJB2, HSJ1, HSPF3, DSMA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Charcot-Marie-Tooth disease 2T (CMT2T) [MIM:616233]: An axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. {ECO:0000269|PubMed:24627108, ECO:0000269|PubMed:25274842}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, X'e Bağlı 3, 300489, X'e Bağlı Resesif; SMAX3 (X'e Bağlı Distal Spinal Müsküler Atrofi Tip 3) (ATP7A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, distal, X-linked 3, 300489, X-linked recessive; SMAX3 (X-linked distal spinal muscular atrophy type 3) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample

Spinal Müsküler Atrofi, Distal, X'e Bağlı 3, 300489, X'e Bağlı Resesif; SMAX3 (X'e Bağlı Distal Spinal Müsküler Atrofi Tip 3) (MLPA)	Spinal muscular atrophy, distal, X-linked 3, 300489, X-linked recessive; SMAX3 (X-linked distal spinal muscular atrophy type 3) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, X'e Bağlı 3, 300489, X'e Bağlı Resesif; SMAX3 (X'e Bağlı Distal Spinal Müsküler Atrofi Tip 3) (Prenatal) (ATP7A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, distal, X-linked 3, 300489, X-linked recessive; SMAX3 (X-linked distal spinal muscular atrophy type 3) (ATP7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Distal, X'e Bağlı 3, 300489, X'e Bağlı Resesif; SMAX3 (X'e Bağlı Distal Spinal Müsküler Atrofi Tip 3) (Prenatal) (MLPA)	Spinal muscular atrophy, distal, X-linked 3, 300489, X-linked recessive; SMAX3 (X-linked distal spinal muscular atrophy type 3) (Prenatal) (MLPA)	ATP7A	ATP7A, MNK, MK, OHS, SMAX3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Distal spinal muscular atrophy, X-linked, 3 (DSMAX3) [MIM:300489]: A neuromuscular disorder. Distal spinal muscular atrophy, also known as distal hereditary motor neuronopathy, represents a heterogeneous group of neuromuscular disorders caused by selective degeneration of motor neurons in the anterior horn of the spinal cord, without sensory deficit in the posterior horn. The overall clinical picture consists of a classical distal muscular atrophy syndrome in the legs without clinical sensory loss. The disease starts with weakness and wasting of distal muscles of the anterior tibial and peroneal compartments of the legs. Later on, weakness and atrophy may expand to the proximal muscles of the lower limbs and/or to the distal upper limbs. {ECO:0000269	PubMed:20170900}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Jokela Tipi, 615048, Otozomal Dominant; SMAJ (Geç Yetişkin Başlangıçlı Alt Motor Nöron Sendromu) (CHCHD10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, Jokela type, 615048, Autosomal dominant; SMAJ (Lower motor neuron syndrome with late-adult onset) (CHCHD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHCHD10	CHCHD10, FTDALS2, SMAJ, IMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, Jokela type (SMAJ) [MIM:615048]: An autosomal dominant, slowly progressive, lower motor neuron disease. SMAJ is characterized by adult-onset of muscle cramps and fasciculations affecting the proximal and distal muscles of the upper and lower limbs. The disorder results in weakness and mild muscle atrophy later in life. {ECO:0000269	PubMed:25428574}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi, Jokela Tipi, 615048, Otozomal Dominant; SMAJ (Geç Yetişkin Başlangıçlı Alt Motor Nöron Sendromu) (Prenatal)) (CHCHD10 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, Jokela type, 615048, Autosomal dominant; SMAJ (Lower motor neuron syndrome with late-adult onset) (CHCHD10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHCHD10	CHCHD10, FTDALS2, SMAJ, IMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, Jokela type (SMAJ) [MIM:615048]: An autosomal dominant, slowly progressive, lower motor neuron disease. SMAJ is characterized by adult-onset of muscle cramps and fasciculations affecting the proximal and distal muscles of the upper and lower limbs. The disorder results in weakness and mild muscle atrophy later in life. {ECO:0000269	PubMed:25428574}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Geç Başlangıçlı, Finkel Tipi, 182980, Otozomal Dominant; SMAFK (Yetişkin Başlangıçlı Proksimal Spinal Müsküler Atrofi, Otozomal Dominant) (VAPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, late-onset, Finkel type, 182980, Autosomal dominant; SMAFK (Adult-onset proximal spinal muscular atrophy, autosomal dominant) (VAPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VAPB	VAPB, VAPC, ALS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, proximal, adult, autosomal dominant (SMAPAD) [MIM:182980]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMAPAD is characterized by proximal muscle weakness that begins in the lower limbs and then progresses to upper limbs, onset in late adulthood (after third decade) and a benign course. Most of the patients remain ambulatory 10 to 40 years after clinical onset. {ECO:0000269	PubMed:15372378}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi, Geç Başlangıçlı, Finkel Tipi, 182980, Otozomal Dominant; SMAFK (Yetişkin Başlangıçlı Proksimal Spinal Müsküler Atrofi, Otozomal Dominant) (Prenatal) (VAPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, late-onset, Finkel type, 182980, Autosomal dominant; SMAFK (Adult-onset proximal spinal muscular atrophy, autosomal dominant) (VAPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VAPB	VAPB, VAPC, ALS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, proximal, adult, autosomal dominant (SMAPAD) [MIM:182980]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMAPAD is characterized by proximal muscle weakness that begins in the lower limbs and then progresses to upper limbs, onset in late adulthood (after third decade) and a benign course. Most of the patients remain ambulatory 10 to 40 years after clinical onset. {ECO:0000269	PubMed:15372378}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Alt Ekstremite Dominant 1, AD, 158600, Otozomal Dominant; SMALED1 (Otozomal Dominant Çocukluk Çağı Başlangıçlı Proksimal Spinal Müsküler Atrofi-Kontraktür Olmadan) (DYNC1H1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600, Autosomal dominant; SMALED1 (Autosomal dominant childhood-onset proximal spinal muscular atrophy without contractures) (DYNC1H1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DYNC1H1	DYNC1H1, DNCL, DNECL, CMT20, MRD13, SMALED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 1, autosomal dominant (SMALED1) [MIM:158600]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMALED1 is characterized by muscle weakness predominantly affecting the proximal lower extremities. {ECO:0000269|PubMed:22459677, ECO:0000269|PubMed:22847149, ECO:0000269|PubMed:25512093}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi, Alt Ekstremite Dominant 1, AD, 158600, Otozomal Dominant; SMALED1 (Otozomal Dominant Çocukluk Çağı Başlangıçlı Proksimal Spinal Müsküler Atrofi-Kontraktür Olmadan) (Prenatal) (DYNC1H1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, lower extremity-predominant 1, AD, 158600, Autosomal dominant; SMALED1 (Autosomal dominant childhood-onset proximal spinal muscular atrophy without contractures) (DYNC1H1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DYNC1H1	DYNC1H1, DNCL, DNECL, CMT20, MRD13, SMALED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 1, autosomal dominant (SMALED1) [MIM:158600]: A form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. SMALED1 is characterized by muscle weakness predominantly affecting the proximal lower extremities. {ECO:0000269|PubMed:22459677, ECO:0000269|PubMed:22847149, ECO:0000269|PubMed:25512093}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Alt Ekstremite-Predominant, 2, OD, 615290, Otozomal Dominant; SMALED2 (Otozomal Dominant Çocukluk Çağı Başlangıçlı Proksimal Spinal Müsküler Atrofi-Kontraktürler) (BICD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290, Autosomal dominant; SMALED2 (Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures) (BICD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BICD2	BICD2, KIAA0699, SMALED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 2, autosomal dominant (SMALED2) [MIM:615290]: An autosomal dominant form of spinal muscular atrophy characterized by early-childhood onset of muscle weakness and atrophy predominantly affecting the proximal and distal muscles of the lower extremity, although some patients may show upper extremity involvement. The disorder results in delayed walking, waddling gait, difficulty walking, and loss of distal reflexes. Some patients may have foot deformities or hyperlordosis, and some show mild upper motor signs, such as spasticity. Sensation, bulbar function, and cognitive function are preserved. The disorder shows very slow progression throughout life. {ECO:0000269|PubMed:23664116, ECO:0000269|PubMed:23664119, ECO:0000269|PubMed:23664120}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi, Alt Ekstremite-Predominant, 2, OD, 615290, Otozomal Dominant; SMALED2 (Otozomal Dominant Çocukluk Çağı Başlangıçlı Proksimal Spinal Müsküler Atrofi-Kontraktürler) (Prenatal) (BICD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, lower extremity-predominant, 2, AD, 615290, Autosomal dominant; SMALED2 (Autosomal dominant childhood-onset proximal spinal muscular atrophy with contractures) (BICD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	BICD2	BICD2, KIAA0699, SMALED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy, lower extremity-predominant 2, autosomal dominant (SMALED2) [MIM:615290]: An autosomal dominant form of spinal muscular atrophy characterized by early-childhood onset of muscle weakness and atrophy predominantly affecting the proximal and distal muscles of the lower extremity, although some patients may show upper extremity involvement. The disorder results in delayed walking, waddling gait, difficulty walking, and loss of distal reflexes. Some patients may have foot deformities or hyperlordosis, and some show mild upper motor signs, such as spasticity. Sensation, bulbar function, and cognitive function are preserved. The disorder shows very slow progression throughout life. {ECO:0000269|PubMed:23664116, ECO:0000269|PubMed:23664119, ECO:0000269|PubMed:23664120}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Tip 3, Modifiye Edici, 253400, Otozomal Resesif (Proksimal Spinal Müsküler Atrofi Tip 3) (SMN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, type III, modifier of, 253400, Autosomal recessive (Proximal spinal muscular atrophy type 3) (SMN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SMN2	SMN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249925, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Spinal Müsküler Atrofi, Tip 3, Modifiye Edici, 253400, Otozomal Resesif (Proksimal Spinal Müsküler Atrofi Tip 3) (MLPA)	Spinal muscular atrophy, type III, modifier of, 253400, Autosomal recessive (Proximal spinal muscular atrophy type 3) (MLPA)	SMN2	SMN2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinal Müsküler Atrofi, Tip 3, Modifiye Edici, 253400, Otozomal Resesif (Proksimal Spinal Müsküler Atrofi Tip 3) (Prenatal) (SMN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, type III, modifier of, 253400, Autosomal recessive (Proximal spinal muscular atrophy type 3) (SMN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SMN2	SMN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, Tip 3, Modifiye Edici, 253400, Otozomal Resesif (Proksimal Spinal Müsküler Atrofi Tip 3) (Prenatal) (MLPA)	Spinal muscular atrophy, type III, modifier of, 253400, Autosomal recessive (Proximal spinal muscular atrophy type 3) (Prenatal) (MLPA)	SMN2	SMN2	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy 1 (SMA1) [MIM:253300]: A form of spinal muscular atrophy, a group of neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Autosomal recessive forms are classified according to the age of onset, the maximum muscular activity achieved, and survivorship. The severity of the disease is mainly determined by the copy number of SMN2, a copy gene which predominantly produces exon 7- skipped transcripts and only low amount of full-length transcripts that encode for a protein identical to SMN1. Only about 4% of SMA patients bear one SMN1 copy with an intragenic mutation. SMA1 is a severe form, with onset before 6 months of age. SMA1 patients never achieve the ability to sit. {ECO:0000269|PubMed:10732817, ECO:0000269|PubMed:15249625, ECO:0000269|PubMed:15580564, ECO:0000269|PubMed:7813012, ECO:0000269|PubMed:9147655}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinal Müsküler Atrofi, X'e Bağlı 2, İnfantil, 301830, X'e Bağlı Resesif; SMAX2 (X`e Bağlı Distal Artrogrifozis Multipleks Konjenita) (UBA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinal muscular atrophy, X-linked 2, infantile, 301830, X-linked recessive; SMAX2 (X-linked distal arthrogryposis multiplex congenita) (UBA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBA1	UBA1, UBE1, GXP1, A1ST, SMAX2, AMCX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinal muscular atrophy X-linked 2 (SMAX2) [MIM:301830]: A lethal infantile form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Clinical features include hypotonia, areflexia, and multiple congenital contractures. {ECO:0000269|PubMed:18179898, ECO:0000269|PubMed:23518311}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinal Müsküler Atrofi, X'e Bağlı 2, İnfantil, 301830, X'e Bağlı Resesif; SMAX2 (X`e Bağlı Distal Artrogrifozis Multipleks Konjenita) (Prenatal) (UBA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinal muscular atrophy, X-linked 2, infantile, 301830, X-linked recessive; SMAX2 (X-linked distal arthrogryposis multiplex congenita) (UBA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UBA1	UBA1, UBE1, GXP1, A1ST, SMAX2, AMCX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinal muscular atrophy X-linked 2 (SMAX2) [MIM:301830]: A lethal infantile form of spinal muscular atrophy, a neuromuscular disorder characterized by degeneration of the anterior horn cells of the spinal cord, leading to symmetrical muscle weakness and atrophy. Clinical features include hypotonia, areflexia, and multiple congenital contractures. {ECO:0000269|PubMed:18179898, ECO:0000269|PubMed:23518311}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi 1, 164400, Otozomal Dominant (Spinoserebellar Ataksi Tip 1) (ATXN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 1, 164400, Autosomal dominant (Spinocerebellar ataxia type 1) (ATXN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN1	ATXN1, ATX1, SCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 1 (SCA1) [MIM:164400]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA1 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA1 is caused by expansion of a CAG repeat in the coding region of ATXN1. Longer expansions result in earlier onset and more severe clinical manifestations of the disease. {ECO:0000269|PubMed:7647801, ECO:0000269|PubMed:7951322, ECO:0000269|PubMed:8634720}. Note=The disease is caused by mutations affecting the gene represented in this entry. The disease is caused by expansion of the polyglutamine tract to about 40-83 repeats, causing accumulation in neurons and exerting toxicity. {ECO:0000269|PubMed:7647801, ECO:0000269|PubMed:8634720}.;	EDTA Blood Sample
Spinoserebellar Ataksi 1; Sca1 (Spinoserebellar Ataksi Tip 1) (Tekrar Sayısı)	Spinocerebellar ataxia 1; SCA1 (Spinocerebellar ataxia type 1)(Repeat Analysis)	ATXN1		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 10, 603516, Otozomal Dominant (Spinoserebellar Ataksi Tip 10) (Tekrar Sayısı)	Spinocerebellar ataxia 10, 603516, Autosomal dominant (Spinocerebellar ataxia type 10)(Repeat Analysis)	ATXN10	ATXN10, SCA10	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Note=Defects in ATXN1 may be a cause of nephronophthisis a chronic tubulo-interstitial nephropathy that leads to anemia, polyuria, polydipsia, isosthenuria and death in uremia. {ECO:0000269|PubMed:21565611}.;	EDTA Blood Sample
Spinoserebellar Ataksi 10; Sca10 (Spinoserebellar Ataksi Tip 10) (Tekrar Sayısı)	Spinocerebellar ataxia 10; SCA10 (Spinocerebellar ataxia type 10)(Repeat Analysis)	ATXN10		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 11, 604432, Otozomal Dominant (Spinoserebellar Ataksi Tip 11) (TTBK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 11, 604432, Autosomal dominant (Spinocerebellar ataxia type 11) (TTBK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTBK2	TTBK2, SCA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 11 (SCA11) [MIM:604432]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA11 is an autosomal dominant cerebellar ataxia (ADCA). It is a relatively benign, late-onset, slowly progressive neurologic disorder. {ECO:0000269|PubMed:18037885}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 11; Sca11 (Spinoserebellar Ataksi Tip 11) (TTBK2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 11; SCA11 (Spinocerebellar ataxia type 11) (TTBK2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTBK2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 12, 604326, Otozomal Dominant (Spinoserebellar Ataksi Tip 12) (PPP2R2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 12, 604326, Autosomal dominant (Spinocerebellar ataxia type 12) (PPP2R2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPP2R2B	PPP2R2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 12 (SCA12) [MIM:604326]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA12 is an autosomal dominant cerebellar ataxia (ADCA). {ECO:0000269|PubMed:10581021}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 12; Sca12 (Spinoserebellar Ataksi Tip 12) (Tekrar Sayısı)	Spinocerebellar ataxia 12; SCA12 (Spinocerebellar ataxia type 12)(Repeat Analysis)	PPP2R2B		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 13, 605259, Otozomal Dominant (Spinoserebellar Ataksi Tip 13) (KCNC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 13, 605259, Autosomal dominant (Spinocerebellar ataxia type 13) (KCNC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNC3	KCNC3, SCA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 13 (SCA13) [MIM:605259]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA13 is an autosomal dominant cerebellar ataxia (ADCA) characterized by slow progression and variable age at onset, ranging from childhood to late adulthood. Mental retardation can be present in some patients. {ECO:0000269|PubMed:16501573, ECO:0000269|PubMed:19953606, ECO:0000269|PubMed:21479265, ECO:0000269|PubMed:23734863, ECO:0000269|PubMed:25152487, ECO:0000269|PubMed:25756792}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 13; Sca13 (Spinoserebellar Ataksi Tip 13) (KCNC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 13; SCA13 (Spinocerebellar ataxia type 13) (KCNC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNC3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 14, 605361, Otozomal Dominant (Spinoserebellar Ataksi Tip 14) (PRKCG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 14, 605361, Autosomal dominant (Spinocerebellar ataxia type 14) (PRKCG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCG	PRKCG, PKCC, PKCG, SCA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 14 (SCA14) [MIM:605361]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA14 is an autosomal dominant cerebellar ataxia (ADCA). {ECO:0000269|PubMed:12644968}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 14; Sca14 (Spinoserebellar Ataksi Tip 14) (PRKCG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 14; SCA14 (Spinocerebellar ataxia type 14) (PRKCG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKCG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample

Spinoserebellar Ataksi 15, 606658, Otozomal Dominant (Spinoserebellar Ataksi Tip 16) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 15, 606658, Autosomal dominant (Spinocerebellar ataxia type 16) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR1	ITPR1, SCA15, SCA16, SCA29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 29 (SCA29) [MIM:117360]: An autosomal dominant, congenital spinocerebellar ataxia characterized by early motor delay, hypotonia and mild cognitive delay. Affected individuals develop a very slowly progressive or non-progressive gait and limb ataxia associated with cerebellar atrophy on brain imaging. Additional variable features include nystagmus, dysarthria, and tremor. {ECO:0000269	PubMed:22986007}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 15; Sca15 (Spinoserebellar Ataksi Tip 16) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 15; SCA15 (Spinocerebellar ataxia type 16) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 17, 607136, Otozomal Dominant (Spinoserebellar Ataksi Tip 17) (Tekrar Sayısı)	Spinocerebellar ataxia 17, 607136, Autosomal dominant (Spinocerebellar ataxia type 17)(Repeat Analysis)	TBP	TBP, SCA17, HDL4	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 17 (SCA17) [MIM:607136]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA17 is an autosomal dominant cerebellar ataxia (ADCA) characterized by widespread cerebral and cerebellar atrophy, dementia and extrapyramidal signs. The molecular defect in SCA17 is the expansion of a CAG repeat in the coding region of TBP. Longer expansions result in earlier onset and more severe clinical manifestations of the disease. {ECO:0000269|PubMed:11313753, ECO:0000269|PubMed:11448935, ECO:0000269|PubMed:11939898}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 17; Sca17 (Spinoserebellar Ataksi Tip 17) (Tekrar Sayısı)	Spinocerebellar ataxia 17; SCA17 (Spinocerebellar ataxia type 17)(Repeat Analysis)	TBP		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 19, 607346, Otozomal Dominant (Spinoserebellar Ataksi Tip 19 / 22) (KCND3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 19, 607346, Autosomal dominant (Spinocerebellar ataxia type 19/22) (KCND3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCND3	KCND3, KCND3S, KCND3L, SCA19, SCA22, BRGDA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 19 (SCA19) [MIM:607346]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA19 is a relatively mild, cerebellar ataxic syndrome with cognitive impairment, pyramidal tract involvement, tremor and peripheral neuropathy, and mild atrophy of the cerebellar hemispheres and vermis. {ECO:0000269|PubMed:23280837, ECO:0000269|PubMed:23280838}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi 19; Sca19 (Spinoserebellar Ataksi Tip 19 / 22) (KCND3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 19; SCA19 (Spinocerebellar ataxia type 19/22) (KCND3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCND3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi 2, 183090, Otozomal Dominant (Spinoserebellar Ataksi Tip 2) (ATXN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 2, 183090, Autosomal dominant (Spinocerebellar ataxia type 2) (ATXN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN2	ATXN2, ATX2, SCA2, ASL13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 2 (SCA2) [MIM:183090]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to cerebellum degeneration with variable involvement of the brainstem and spinal cord. SCA2 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA2 is characterized by hyporeflexia, myoclonus and action tremor and dopamine-responsive parkinsonism. In some patients, SCA2 presents as pure familial parkinsonism without cerebellar signs. {ECO:0000269|PubMed:8896555, ECO:0000269|PubMed:8896556, ECO:0000269|PubMed:8896557}. Note=The disease is caused by mutations affecting the gene represented in this entry. SCA2 is caused by expansion of a CAG repeat resulting in about 36 to 52 repeats in some patients. Longer expansions result in earlier the expansion, onset of the disease.	EDTA Blood Sample
Spinoserebellar Ataksi 2; SCA2 (Spinoserebellar Ataksi Tip 2) (Tekrar Sayısı)	Spinocerebellar ataxia 2; SCA2 (Spinocerebellar ataxia type 2)(Repeat Analysis)	ATXN2		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi 20, 608687, Otozomal Dominant; SCA20 (Spinoserebellar Ataksi Tip 20) (2200)	Spinocerebellar ataxia 20, 608687, Autosomal dominant; SCA20 (Spinocerebellar ataxia type 20) (440)	.	SCA20, DUP11q12, C11DUPq12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Spinoserebellar Ataksi 21, 607454, Otozomal Dominant; SCA21 (Spinoserebellar Ataksi Tip 21) (TMEM240 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 21, 607454, Autosomal dominant; SCA21 (Spinocerebellar ataxia type 21) (TMEM240 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM240	TMEM240, C1orf70, SCA21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 21 (SCA21) [MIM:607454]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA21 is characterized by onset in the first decades of life of slowly progressive relatively mild cerebellar ataxia associated with slight extrapyramidal features predominant in older patients and cognitive impairment predominant in younger patients. Note=The disease is caused by mutations affecting the gene represented in this entry. {ECO:0000269	PubMed:25070513}.,	EDTA Blood Sample
Spinoserebellar Ataksi 21; Sca21 (Spinoserebellar Ataksi Tip 21) (TMEM240 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 21; SCA21 (Spinocerebellar ataxia type 21) (TMEM240 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM240		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi 23, 610245, Otozomal Dominant (Spinoserebellar Ataksi Tip 23) (PDYN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 23, 610245, Autosomal dominant (Spinocerebellar ataxia type 23) (PDYN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDYN	PDYN, SCA23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 23 (SCA23) [MIM:610245]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA23 is an adult-onset autosomal dominant form characterized by slowly progressive gait and limb ataxia, with variable additional features, including peripheral neuropathy and dysarthria. {ECO:0000269|PubMed:21035104, ECO:0000269|PubMed:21712028, ECO:0000269|PubMed:23108490, ECO:0000269|PubMed:23471613}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 23; Sca23 (Spinoserebellar Ataksi Tip 23) (PDYN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 23; SCA23 (Spinocerebellar ataxia type 23) (PDYN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDYN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 26, 609306, Otozomal Dominant (Spinoserebellar Ataksi Tip 26) (EEF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 26, 609306, Autosomal dominant (Spinocerebellar ataxia type 26) (EEF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EEF2	EEF2, EF2, SCA26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 26 (SCA26) [MIM:609306]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. {ECO:0000269	PubMed:23001565}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 27 609307, Otozomal Dominant (Spinoserebellar Ataksi Tip 27) (FGF14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 27, 609307, Autosomal dominant (Spinocerebellar ataxia type 27) (FGF14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF14	FGF14, FHF4, SCA27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 27; Sca27 (Spinoserebellar Ataksi Tip 27) (FGF14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 27; SCA27 (Spinocerebellar ataxia type 27) (FGF14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF14		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi 28 610246, Otozomal Dominant (Spinoserebellar Ataksi Tip 28) (AFG3L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 28, 610246, Autosomal dominant (Spinocerebellar ataxia type 28) (AFG3L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFG3L2	AFG3L2, SCA28, SPAX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic ataxia 5, autosomal recessive (SPAX5) [MIM:614487]: A neurodegenerative disorder characterized by early onset spasticity, peripheral neuropathy, ptosis, oculomotor apraxia, dystonia, cerebellar atrophy, and progressive myoclonic epilepsy. {ECO:0000269	PubMed:22022284}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 28; Sca28 (Spinoserebellar Ataksi Tip 28) (AFG3L2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 28; SCA28 (Spinocerebellar ataxia type 28) (AFG3L2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AFG3L2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 29, Konjenital Progresif Olmayan, 117360, Otozomal Dominant (Spinoserebellar Ataksi Tip 29) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 29, congenital nonprogressive, 117360, Autosomal dominant (Spinocerebellar ataxia type 29) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR1	ITPR1, SCA15, SCA16, SCA29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 29 (SCA29) [MIM:117360]: An autosomal dominant, congenital spinocerebellar ataxia characterized by early motor delay, hypotonia and mild cognitive delay. Affected individuals develop a very slowly progressive or non-progressive gait and limb ataxia associated with cerebellar atrophy on brain imaging. Additional variable features include nystagmus, dysarthria, and tremor. {ECO:0000269	PubMed:22986007}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 29; Sca29 (Spinoserebellar Ataksi Tip 29) (ITPR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 29; SCA29 (Spinocerebellar ataxia type 29) (ITPR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITPR1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 34, 133190, Otozomal Dominant (Spinoserebellar Ataksi Tip 34) (ELOVL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 34, 133190, Autosomal dominant (Spinocerebellar ataxia type 34) (ELOVL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL4	ELOVL4, ADMD, STGD2, STGD3, ISQMR, SCA34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 34 (SCA34) [MIM:133190]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA34 is an autosomal dominant form characterized by the association of progressive cerebellar ataxia with erythrokeratodermia variabilis. {ECO:0000269	PubMed:24566826}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Spinoserebellar Ataksi 34; Sca34 (Spinoserebellar Ataksi Tip 34) (ELOVL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 34; SCA34 (Spinocerebellar ataxia type 34) (ELOVL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample

Spinoserebellar Ataksi 35, 613908, Otozomal Dominant (Spinoserebellar Ataksi Tip 35) (TGM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 35, 613908, Autosomal dominant (Spinocerebellar ataxia type 35) (TGM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGM6	TGM6, TG6, TGY, SCA35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 35 (SCA35) [MIM:613908]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA35 patients commonly show upper limb involvement and torticollis. There is no cognitive impairment. {ECO:0000269|PubMed:21106500, ECO:0000269|PubMed:22554020, ECO:0000269|PubMed:23206699, ECO:0000269|PubMed:25253745}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 35; Sca35 (Spinoserebellar Ataksi Tip 35) (TGM6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 35; SCA35 (Spinocerebellar ataxia type 35) (TGM6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGM6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi 36, 614153, Otozomal Dominant (Spinoserebellar Ataksi Tip 36) (NOP56 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 36, 614153, Autosomal dominant (Spinocerebellar ataxia type 36) (NOP56 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOP56	NOP56, SCA36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 36 (SCA36) [MIM:614153]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA36 is characterized by complicated clinical features, with ataxia as the first symptom, followed by characteristic late-onset involvement of the motor neuron system. Ataxic symptoms, such as gait and truncal instability, ataxic dysarthria, and uncoordinated limbs, start in late forties to fifties. Characteristically, affected individuals exhibit tongue atrophy with fasciculation. Progression of motor neuron involvement is typically limited to the tongue and main proximal skeletal muscles in both upper and lower extremities. {ECO:0000269|PubMed:21683323}. Note=The disease is caused by mutations affecting the gene represented in this entry. Caused by large hexanucleotide CGCCTG repeat expansions within intron 1. These expansions induce RNA foci and sequester the RNA-binding protein SRSF2. In addition, the transcription of MIR1292, a microRNA gene located just 19 bp 3' of the GGCCTG repeat, is significantly decreased.;	EDTA Blood Sample
Spinoserebellar Ataksi 36; Sca36 (Spinoserebellar Ataksi Tip 36) (NOP56 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 36; SCA36 (Spinocerebellar ataxia type 36) (NOP56 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOP56		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 38, 615957, Otozomal Dominant (Spinoserebellar Ataksi Tip 38) (ELOVL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 38, 615957, Autosomal dominant (Spinocerebellar ataxia type 38) (ELOVL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL5	ELOVL5, HELO1, SCA38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 38; Sca38 (Spinoserebellar Ataksi Tip 38) (ELOVL5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 38; SCA38 (Spinocerebellar ataxia type 38) (ELOVL5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 40, 616053, Otozomal Dominant (Spinoserebellar Ataksi Tip 40) (CCDC88C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 40, 616053, Autosomal dominant (Spinocerebellar ataxia type 40) (CCDC88C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCDC88C	CCDC88C, HKRP2, DAPLE, KIAA1509, HYC, SCA40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hydrocephalus, non-syndromic, autosomal recessive 1 (HYC1) [MIM:236600]: A disease characterized by a disturbance of cerebrospinal fluid circulation causing accumulation of ventricular cerebrospinal fluid, which results in progressive ventricular dilatation with onset in utero. Affected individuals may have neurologic impairment. {ECO:0000269|PubMed:21031079, ECO:0000269|PubMed:23042809}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi 41, 616410, Otozomal Dominant (Spinoserebellar Ataksi Tip 41) (TRPC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 41, 616410, Autosomal dominant (Spinocerebellar ataxia type 41) (TRPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPC3	TRPC3, TRP3, SCA41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 42, 616795, Otozomal Dominant (Spinoserebellar Ataksi Tip 42) (CACNA1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 42, 616795, Autosomal dominant (Spinocerebellar ataxia type 42) (CACNA1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1G	CACNA1G, SCA42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 42; Sca42 (Spinoserebellar Ataksi Tip 42) (CACNA1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 42; SCA42 (Spinocerebellar ataxia type 42) (CACNA1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1G		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 43, 617018, Otozomal Dominant (MME geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 43, 617018, Autosomal dominant (MME gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MME	MME, CD10, CALLA, NEP, CMT27, SCA43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 5, 600224, Otozomal Dominant; SCA5 (Spinoserebellar Ataksi Tip 5) (SPTBN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 5, 600224, Autosomal dominant; SCA5 (Spinocerebellar ataxia type 5) (SPTBN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTBN2	SPTBN2, SCA5, SCAR14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 14 (SCAR14) [MIM:615386]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR14 is characterized by delayed psychomotor development, severe early onset gait ataxia, eye movement abnormalities, cerebellar atrophy on brain imaging, and intellectual disability. {ECO:0000269|PubMed:23236289, ECO:0000269|PubMed:23838597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 5; Sca5 (Spinoserebellar Ataksi Tip 5) (SPTBN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 5; SCA5 (Spinocerebellar ataxia type 5) (SPTBN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTBN2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 6, 183086, Otozomal Dominant (Spinoserebellar Ataksi Tip 6) (Tekrar Sayısı)	Spinocerebellar ataxia 6, 183086, Autosomal dominant (Spinocerebellar ataxia type 6)(Repeat Analysis)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 6, 183086, Otozomal Dominant (Spinoserebellar Ataksi Tip 6) (MLPA)	Spinocerebellar ataxia 6, 183086, Autosomal dominant (Spinocerebellar ataxia type 6) (MLPA)	CACNA1A	CACNA1A, CACNL1A4, SCA6, EIEE42	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Episodic ataxia 2 (EA2) [MIM:108500]: An autosomal dominant disorder characterized by acetozolamide-responsive attacks of ataxia, migraine-like symptoms, interictal nystagmus, and cerebellar atrophy. {ECO:0000269|PubMed:10987655, ECO:0000269|PubMed:11176968, ECO:0000269|PubMed:11723274, ECO:0000269|PubMed:12420090, ECO:0000269|PubMed:14718690, ECO:0000269|PubMed:15173248, ECO:0000269|PubMed:15293273, ECO:0000269|PubMed:18602318, ECO:0000269|PubMed:19232643, ECO:0000269|PubMed:20129625, ECO:0000269|PubMed:21696515, ECO:0000269|PubMed:8898206, ECO:0000269|PubMed:9302278}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 6; SCA6 (Spinoserebellar Ataksi Tip 6) (Tekrar Sayısı)	Spinocerebellar ataxia 6; SCA6 (Spinocerebellar ataxia type 6)(Repeat Analysis)	CACNA1A		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 7, 164500, Otozomal Dominant (Spinoserebellar Ataksi Tip 7) (ATXN7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia 7, 164500, Autosomal dominant (Spinocerebellar ataxia type 7) (ATXN7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATXN7	ATXN7, SCA7, OPCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 7 (SCA7) [MIM:164500] Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA7 belongs to the autosomal dominant cerebellar ataxias type II (ADCA II) which are characterized by cerebellar ataxia with retinal degeneration and pigmentary macular dystrophy. {ECO:0000269|PubMed:9288099}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 7; Sca7 (Spinoserebellar Ataksi Tip 7) (Tekrar Sayısı)	Spinocerebellar ataxia 7; SCA7 (Spinocerebellar ataxia type 7)(Repeat Analysis)	ATXN7		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi 8, 608768, Otozomal Dominant (Spinoserebellar Ataksi Tip 8) (Tekrar Sayısı)	Spinocerebellar ataxia 8, 608768, Autosomal dominant (Spinocerebellar ataxia type 8)(Repeat Analysis)	ATXN8	ATXN8	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 8 (SCA8) [MIM:608768] Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA8 is an autosomal dominant cerebellar ataxia (ADCA). It is caused by expansion of a CAG repeat in ATXN8, which is translated into a nearly pure polyglutamine protein which forms 1C2-positive inclusions in Purkinje cells and other neurons. {ECO:0000269|PubMed:16804541}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi 8; Sca8 (Spinoserebellar Ataksi Tip 8) (Tekrar Sayısı)	Spinocerebellar ataxia 8; SCA8 (Spinocerebellar ataxia type 8)(Repeat Analysis)	ATXN8		Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		20	90	.	.	60	Postnatal	.	EDTA Blood Sample

Spinoserebellar Ataksi, Otozomal Resesif 1, 606002, Otozomal Resesif (Spinoserebellar Ataksi-Aksonal Nöropati Tip 2) (SETX gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 1, 606002, Autosomal recessive (Spinocerebellar ataxia with axonal neuropathy type 2) (SETX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SETX	SETX, SCAR1, AOA2, ALS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 4 (ALS4) [MIM:602433]: A form of amyotrophic lateral sclerosis with childhood- or adolescent-onset, and characterized by slow disease progression and the sparing of bulbar and respiratory muscles. Amyotrophic lateral sclerosis is a neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:14770181, ECO:0000269|PubMed:15106121, ECO:0000269|PubMed:21190393, ECO:0000269|PubMed:24105744, ECO:0000269|PubMed:24244371}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 1, 606002, Otozomal Resesif (Spinoserebellar Ataksi-Aksonal Nöropati Tip 2) (MLPA)	Spinocerebellar ataxia, autosomal recessive 1, 606002, Autosomal recessive (Spinocerebellar ataxia with axonal neuropathy type 2) (MLPA)	SETX	SETX, SCAR1, AOA2, ALS4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Amyotrophic lateral sclerosis 4 (ALS4) [MIM:602433]: A form of amyotrophic lateral sclerosis with childhood- or adolescent-onset, and characterized by slow disease progression and the sparing of bulbar and respiratory muscles. Amyotrophic lateral sclerosis is a neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:14770181, ECO:0000269|PubMed:15106121, ECO:0000269|PubMed:21190393, ECO:0000269|PubMed:24105744, ECO:0000269|PubMed:24244371}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 10, 613728, Otozomal Resesif (Yetişkin Başlangıçlı Otozomal Resesif Serebellar Ataksi) (ANO10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 10, 613728, Autosomal recessive (Adult-onset autosomal recessive cerebellar ataxia) (ANO10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANO10	ANO10, TMEM16K, SCAR10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 11, 614229, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Psikomotor Retardasyon Sendromu) (SYT14 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 11, 614229, Autosomal recessive (Autosomal recessive cerebellar ataxia-psychomotor retardation syndrome) (SYT14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYT14	SYT14, SCAR11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 11, 614229, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Psikomotor Retardasyon Sendromu (Prenatal) (SYT14 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 11, 614229, Autosomal recessive (Autosomal recessive cerebellar ataxia-psychomotor retardation syndrome) (SYT14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SYT14	SYT14, SCAR11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 13, 614831, Otozomal Resesif (Otozomal Resesif Konjenital Serebellar Ataksi, MGLLR'I Eksikliğine Bağlı) (GRM1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 13, 614831, Autosomal recessive (Autosomal recessive congenital cerebellar ataxia due to MGLLR'I deficiency) (GRM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRM1	GRM1, MGLLR'I, GRM1A, SCAR13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 14, 615386, Otozomal Resesif, SCA14 (Spektrin Bağlantılı Otozomal Resesif Serebellar Ataksi) (SPTBN2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 14, 615386, Autosomal recessive; SCA14 (Spectrin-associated autosomal recessive cerebellar ataxia) (SPTBN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPTBN2	SPTBN2, SCA5, SCAR14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 14 (SCAR14) [MIM:615386]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR14 is characterized by delayed psychomotor development, severe early onset gait ataxia, eye movement abnormalities, cerebellar atrophy on brain imaging, and intellectual disability. {ECO:0000269|PubMed:23236289, ECO:0000269|PubMed:23838597}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 15, 615705, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu, KIAA0226 Eksikliğine Bağlı) (RUBCN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 15, 615705, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to KIAA0226 deficiency) (RUBCN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RUBCN	RUBCN, RUBICON, KIAA0226, SCAR15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 15 (SCAR15) [MIM:615705]: Spinocerebellar ataxia defines a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR15 patients manifest cerebellar ataxia in early childhood and delayed motor development with delayed walking. Additional features include dysarthria, upper limb involvement, abnormal eye movements, and hyporeflexia. {ECO:0000269	PubMed:20826435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 15, 615705, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu, KIAA0226 Eksikliğine Bağlı) (Prenatal) (RUBCN gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 15, 615705, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to KIAA0226 deficiency) (RUBCN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RUBCN	RUBCN, RUBICON, KIAA0226, SCAR15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 15 (SCAR15) [MIM:615705]: Spinocerebellar ataxia defines a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR15 patients manifest cerebellar ataxia in early childhood and delayed motor development with delayed walking. Additional features include dysarthria, upper limb involvement, abnormal eye movements, and hyporeflexia. {ECO:0000269	PubMed:20826435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 16, 615768, Otozomal Resesif (Otozomal Resesif Spinoserebellar Ataksi, STUB1 Eksikliğine Bağlı) (STUB1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 16, 615768, Autosomal recessive (Autosomal recessive cerebellar ataxia due to STUB1 deficiency) (STUB1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STUB1	STUB1, CHIP, SCAR16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 16 (SCAR16) [MIM:615768]: Spinocerebellar ataxia defines a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR16 is characterized by truncal and limb ataxia resulting in gait instability. Additionally, patients may show dysarthria, nystagmus, spasticity of the lower limbs, and mild peripheral sensory neuropathy. {ECO:0000269|PubMed:24113144, ECO:0000269|PubMed:24112599, ECO:0000269|PubMed:24719489, ECO:0000269|PubMed:24742043, ECO:0000269|PubMed:25250038}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 17, 616127, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi, CWF19L1 Eksikliğine Bağlı) (CWF19L1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 17, 616127, Autosomal recessive (Autosomal recessive cerebellar ataxia due to CWF19L1 deficiency) (CWF19L1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CWF19L1	CWF19L1, C19L1, SCAR17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 18, 616204, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Piramidal Bulgular-Nistagmus-Okulomotor Apraksi Sendromu) (GRID2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 18, 616204, Autosomal recessive (Autosomal recessive cerebellar ataxia-pyramidal signs-nystagmus-oculomotor apraxia syndrome) (GRID2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GRID2	GRID2, SCAR18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 18 (SCAR18) [MIM:616204]: Spinocerebellar ataxia defines a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR18 features include progressive cerebellar atrophy, delayed psychomotor development, severely impaired gait, ocular movement abnormalities, and intellectual disability. {ECO:0000269|PubMed:23611888, ECO:0000269|PubMed:24078737, ECO:0000269|PubMed:25841024}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 2, 213200, Otozomal Resesif (Otozomal Resesif Serebelloparankimal Bozukluk Tip 3) (PMPCA gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 2, 213200, Autosomal recessive (Autosomal recessive cerebelloparenchymal disorder type 3) (PMPCA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PMPCA	PMPCA, KIAA0123, SCAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 20, 616354, Otozomal Resesif (Mental Retardasyon-Kaba Yüz-Makroseftali-Serebellar Hipotrofi Sendromu) (SNX14 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 20, 616354, Autosomal recessive (Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome) (SNX14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNX14	SNX14, SCAR20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 20, 616354, Otozomal Resesif (Mental Retardasyon-Kaba Yüz-Makroseftali-Serebellar Hipotrofi Sendromu) (Prenatal) (SNX14 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 20, 616354, Autosomal recessive (Intellectual disability-coarse face-macrocephaly-cerebellar hypotrophy syndrome) (SNX14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNX14	SNX14, SCAR20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 21, 616719, Otozomal Resesif (Akut Karaciğer Yetmezliği İnfantil-Serebellar Ataksi-Periferal Duyusal Motor Nöropati Sendromu) (SCYL1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 21, 616719, Autosomal recessive (Acute infantile liver failure-cerebellar ataxia-peripheral sensory motor neuropathy syndrome) (SCYL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCYL1	SCYL1, NTKL, SCAR21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 22 616948, Otozomal Resesif (VWA3B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 22, 616948, Autosomal recessive (VWA3B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VWA3B	VWA3B, SCAR22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Defects in VWA3B may be a cause of cerebellar ataxia with intellectual disability. {ECO:0000269	PubMed:26157035}.;	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 23, 616949, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu, TUD Eksikliğine Bağlı) (TDP2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 23, 616949, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency) (TDP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TDP2	TDP2, TTRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Spinoserebellar Ataksi, Otozomal Resesif 23, 616949, Otozomal Resesif (Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu, TUD Eksik/Iğne Bağlı) (Prenatal) (TDP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 23, 616949, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to TUD deficiency) (TDP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TDP2	TDP2, TTRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 24, 617133, Otozomal Resesif (UBA5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 24, 617133, Autosomal recessive (UBA5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UBA5	UBA5, UBE1DC1, EIEE44, SCAR24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 3, 271250, Otozomal Resesif; SCAR3 (Otozomal Resesif Serebellar Ataksi-Körlük-Sağırlık Sendromu) (WASF3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 3, 271250, Autosomal recessive; SCAR3 (Autosomal recessive cerebellar ataxia-blindness-deafness syndrome) (WASF3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WASF3	SCAR3, SCABD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 7, 609270, Otozomal Resesif (Çocukluk Çağı Başlangıçlı Otozomal Resesif Yavaş Progresif Spinoserebellar Ataksi) (TPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 7, 609270, Autosomal recessive (Childhood-onset autosomal recessive slowly progressive spinocerebellar ataxia) (TPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPP1	TPP1, CLN2, SCAR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ceroid lipofuscinosis, neuronal, 2 (CLN2) [MIM:204500]: A form of neuronal ceroid lipofuscinosis. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipogigment pattern seen most often in CLN2 consists of curvilinear profiles. {ECO:0000269|PubMed:10330339, ECO:0000269|PubMed:10665500, ECO:0000269|PubMed:11241479, ECO:0000269|PubMed:11339651, ECO:0000269|PubMed:11589012, ECO:0000269|PubMed:12376936, ECO:0000269|PubMed:12414822, ECO:0000269|PubMed:12698559, ECO:0000269|PubMed:19201763, ECO:0000269|PubMed:20340139, ECO:0000269|PubMed:21990111, ECO:0000269|PubMed:22612257, ECO:0000269|PubMed:9295267}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 8, 610743, Otozomal Resesif (Otozomal Resesif Ataksi, Beauce Tipi) (SYNE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 8, 610743, Autosomal recessive (Autosomal recessive ataxia, Beauce type) (SYNE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SYNE1	SYNE1, KIAA0796, KIAA1756, KIAA1262, SCAR8, EDMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, 8 (SCAR8) [MIM:610743]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAR8 is an autosomal recessive form. {ECO:0000269|PubMed:17159980}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi, Aksonal Nöropati, Otozomal Resesif, 607250 (Spinoserebellar Ataksi Tip 1 - Aksonal Nöropati) (TDP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 (Spinocerebellar ataxia type 1 with axonal neuropathy) (TDP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TDP1	TDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia, autosomal recessive, with axonal neuropathy (SCAN1) [MIM:607250]: Spinocerebellar ataxia is a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCAN1 is an autosomal recessive cerebellar ataxia (ARCA) associated with peripheral axonal motor and sensory neuropathy, distal muscular atrophy, pes cavus and steppage gait as seen in Charcot-Marie-Tooth neuropathy. All affected individuals have normal intelligence. {ECO:0000269|PubMed:12244316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi, X'e Bağlı 1, 302500, X'e Bağlı (X'e Bağlı Progresif Serebellar Ataksi) (ATP2B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, X-linked 1, 302500, X-linked recessive (X-linked progressive cerebellar ataxia) (ATP2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP2B3	ATP2B3, PMCA3, SCAX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 12, 614322, Otozomal Resesif (WWOX Eksikliğine Bağlı Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu) (WWOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spinocerebellar ataxia, autosomal recessive 12, 614322, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency) (WWOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WWOX	WWOX, FOR, SCAR12, EIEE28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:11956080}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 12, 614322, Otozomal Resesif (WWOX Eksikliğine Bağlı Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu) (MLPA)	Spinocerebellar ataxia, autosomal recessive 12, 614322, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency) (MLPA)	WWOX	WWOX, FOR, SCAR12, EIEE28	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:11956080}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 12, 614322, Otozomal Resesif (WWOX Eksikliğine Bağlı Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu) (Prenatal) (WWOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 12, 614322, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency) (WWOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WWOX	WWOX, FOR, SCAR12, EIEE28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:11956080}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spinoserebellar Ataksi, Otozomal Resesif 12, 614322, Otozomal Resesif (WWOX Eksikliğine Bağlı Otozomal Resesif Serebellar Ataksi-Epilepsi-Mental Retardasyon Sendromu) (MLPA) (Prenatal)	Spinocerebellar ataxia, autosomal recessive 12, 614322, Autosomal recessive (Autosomal recessive cerebellar ataxia-epilepsy-intellectual disability syndrome due to WWOX deficiency) (Prenatal) (MLPA)	WWOX	WWOX, FOR, SCAR12, EIEE28	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Esophageal cancer (ESCR) [MIM:133239]: A malignancy of the esophagus. The most common types are esophageal squamous cell carcinoma and adenocarcinoma. Cancer of the esophagus remains a devastating disease because it is usually not detected until it has progressed to an advanced incurable stage. {ECO:0000269|PubMed:11956080}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spitz Nevus Veya Nevus Spilus Somatik, 137550 (Büyük Konjenital Melanositik Nevüs) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spitz nevus or nevus spilus, somatic, 137550 (Large congenital melanocytic nevus) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Yarık El / Ayak Malformasyonu, Mesoaxial Polidaktili, 616890, Otozomal Resesif (MAP3K20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Split-foot malformation with mesoaxial polydactyly, 616890, Autosomal recessive (MAP3K20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAP3K20	ZAK, MLTK, MRK, SFMMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yarık El / Ayak Malformasyonu, Mesoaxial Polidaktili, 616890, Otozomal Resesif (Prenatal) (MAP3K20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Split-foot malformation with mesoaxial polydactyly, 616890, Autosomal recessive (MAP3K20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAP3K20	ZAK, MLTK, MRK, SFMMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık El / Ayak Malformasyonu 1, Sensorinöral İşitme Kaybı, 220600, Otozomal Resesif; SHFM1D (Yarık El Ayak-Sağırlık Sendromu) (DLX5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Split-hand/foot malformation 1 with sensorineural hearing loss, 220600, Autosomal recessive; SHFM1D (Split hand-split foot-deafness syndrome) (DLX5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLX5	DLX5, SHFM1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Split-hand/foot malformation 1 with sensorineural hearing loss, autosomal recessive (SHFM1D) [MIM:220600]: A disease characterized by the association of split-hand/foot malformation with deafness. Split-hand/foot malformation is a limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of the hands and feet, and aplasia and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Some patients have been found to have mental retardation, ectodermal and craniofacial findings, and orofacial clefting. {ECO:0000269|PubMed:22121204}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Yarık El / Ayak Malformasyonu 1, Sensorinöral İşitme Kaybı, 220600, Otozomal Resesif; SHFM1D (Yarık El Ayak-Sağırlık Sendromu) (Prenatal) (DLX5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Split-hand/foot malformation 1 with sensorineural hearing loss, 220600, Autosomal recessive; SHFM1D (Split hand-split foot-deafness syndrome) (DLX5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLX5	DLX5, SHFM1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Split-hand/foot malformation 1 with sensorineural hearing loss, autosomal recessive (SHFM1D) [MIM:220600]: A disease characterized by the association of split-hand/foot malformation with deafness. Split-hand/foot malformation is a limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of the hands and feet, and aplasia and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Some patients have been found to have mental retardation, ectodermal and craniofacial findings, and orofacial clefting. {ECO:0000269|PubMed:22121204}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık El / Ayak Malformasyonu 3, Gen Duplikasyon Sendromu, 246560, Otozomal Dominant; SHFM3- Kromozom 10q24 duplikasyon sendromu (Distal Ekstremite Defektleri-Mikrognati Sendromu) (2200)	Split-hand/foot malformation 3, gene duplication syndrome, 246560, Autosomal dominant; SHFM3-CHROMOSOME 10q24 DUPLICATION SYNDROME (Distal limb deficiencies-micrognathia syndrome) (440)	.	SHFM3, SHSF3, DUP10q24, C10DUPq24	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Yarık El / Ayak Malformasyonu 3, Gen Duplikasyon Sendromu, 246560, Otozomal Dominant; SHFM3- Kromozom 10q24 duplikasyon sendromu (Distal Ekstremite Defektleri-Mikrognati Sendromu) (Prenatal)	Split-hand/foot malformation 3, gene duplication syndrome, 246560, Autosomal dominant; SHFM3-CHROMOSOME 10q24 DUPLICATION SYNDROME (Distal limb deficiencies-micrognathia syndrome) (Prenatal)	.	SHFM3, SHSF3, DUP10q24, C10DUPq24	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık El / Ayak Malformasyonu 4, 605289, Otozomal Dominant; SHFM4 (Yarık El / Ayak Maldormasyonu) (TP63 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Split-hand/foot malformation 4, 605289, Autosomal dominant; SHFM4 (Split hand-split foot malformation) (TP63 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269|PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Yarık El / Ayak Malformasyonu 4, 605289, Otozomal Dominant; SHFM4 (Yarık El / Ayak Maldormasyonu) (Prenatal) (TP63 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Split-hand/foot malformation 4, 605289, Autosomal dominant; SHFM4 (Split hand-split foot malformation) (TP63 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TP63	TP63, TP73L, KET, EEC3, SHFM4, LMS, RHS, OFC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acro-dermato-ungual-lacrimal-tooth syndrome (ADULT syndrome) [MIM:103285]: A form of ectodermal dysplasia. Ectodermal dysplasia defines a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. ADULT syndrome involves ectrodactyly, syndactyly, finger- and toenail dysplasia, hypoplastic breasts and nipples, intensive freckling, lacrimal duct atresia, frontal alopecia, primary hypodontia and loss of permanent teeth. ADULT syndrome differs significantly from EEC3 syndrome by the absence of facial clefting. {ECO:0000269|PubMed:11929852}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Yarık-El / Ayak Malformasyon 6, 225300, Otozomal Resesif; SHFM6 (Yarık El / Ayak Malformasyonu) (WNT10B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Split-hand/foot malformation 6, 225300, Autosomal recessive; SHFM6 (Split hand-split foot malformation) (WNT10B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT10B	WNT10B, SHFM6, STHAG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Split-hand/foot malformation 6 (SHFM6) [MIM:225300]: A limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of the hands and feet, and aplasia and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Some patients have been found to have mental retardation, ectodermal and craniofacial findings, and orofacial clefting. {ECO:0000269|PubMed:18515319}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Yarık-El / Ayak Malformasyon 6, 225300, Otozomal Resesif; SHFM6 (Yarık El / Ayak Malformasyonu) (Prenatal) (WNT10B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Split-hand/foot malformation 6, 225300, Autosomal recessive; SHFM6 (Split hand-split foot malformation) (WNT10B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT10B	WNT10B, SHFM6, STHAG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Split-hand/foot malformation 6 (SHFM6) [MIM:225300]: A limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of the hands and feet, and aplasia and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Some patients have been found to have mental retardation, ectodermal and craniofacial findings, and orofacial clefting. {ECO:0000269|PubMed:18515319}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yarık El / Ayak Malformasyonu-Uzun Kemik Eksikliği 3, 612576, Otozomal Dominant (Tibiyal Aplazi-Ektrodaktili Sendromu) (2200)	Split-hand/foot malformation with long bone deficiency 3, 612576, Autosomal dominant (Tibial aplasia-ectrodactyly syndrome) (440)	.	SHFLD3, DUP17p13.3, C17DUP13.3	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Yarık El / Ayak Malformasyonu-Uzun Kemik Eksikliği 3, 612576, Otozomal Dominant (Tibiyal Aplazi-Ektrodaktili Sendromu) (Prenatal)	Split-hand/foot malformation with long bone deficiency 3, 612576, Autosomal dominant (Tibial aplasia-ectrodactyly syndrome) (Prenatal)	.	SHFLD3, DUP17p13.3, C17DUP13.3	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilo-Megaepifizyal-Metafizyal Displazi, 613330, Otozomal Resesif; SMMD (Spondilo-Megaepifizyal-Metafizyal Displazi) (NKX3-2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330, Autosomal recessive; SMMD (Spondylo-megaepiphyseal-metaphyseal dysplasia) (NKX3-2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX3-2	NKX3-2, BAPX1, SMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondilo-Megaepifizyal-Metafizyal Displazi, 613330, Otozomal Resesif; SMMD (Spondilo-Megaepifizyal-Metafizyal Displazi) (Prenatal) (NKX3-2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylo-megaepiphyseal-metaphyseal dysplasia, 613330, Autosomal recessive; SMMD (Spondylo-megaepiphyseal-metaphyseal dysplasia) (NKX3-2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX3-2	NKX3-2, BAPX1, SMMD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloartropati, Yatkınlık 1, 106300, Multifaktöriyel; SPDA1 (HLA-B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloarthropathy, susceptibility to, 1, 106300, Multifactorial; SPDA1 (HLA-B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-B	HLA-B, SPDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondilotarsokarpal Sinostoz Sendromu, 272460, Otozomal Resesif; SCT (Spondilokarpotarsal Sinostoz) (FLNB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocarpotarsal synostosis syndrome, 272460, Autosomal recessive; SCT (Spondylocarpotarsal synostosis) (FLNB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269|PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilotarsokarpal Sinostoz Sendromu, 272460, Otozomal Resesif; SCT (Spondilokarpotarsal Sinostoz) (Prenatal) (FLNB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocarpotarsal synostosis syndrome, 272460, Autosomal recessive; SCT (Spondylocarpotarsal synostosis) (FLNB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FLNB	FLNB, SCT, AOI, LRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Atelosteogenesis 1 (AO1) [MIM:108720]: A lethal chondrodysplasia characterized by distal hypoplasia of the humeri and femurs, hypoplasia of the mid-thoracic spine, occasionally complete lack of ossification of single hand bones, and the finding in cartilage of multiple degenerated chondrocytes which are encapsulated in fibrous tissue. {ECO:0000269|PubMed:14991055}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilopeirodisplazi, Ehlers-Danlos Sendromu Benzeri, 612350, Otozomal Resesif (Ehlers-Danlos Sendromu, Spondilokarpotarsal Sinostoz) (SLC39A13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350, Autosomal recessive (Ehlers-Danlos syndrome, spondylocarpotarsal synostosis type) (SLC39A13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC39A13	SLC39A13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ehlers-Danlos syndrome-like spondylocheirodysplasia (SCD- EDS) [MIM:612350]: Spondylocheiro dysplastic form of Ehlers-Danlos syndrome. The syndrome consists of a generalized skeletal dysplasia involving mainly the spine (spondylo) and striking clinical abnormalities of the hands (cheiro) in addition to the EDS-like features. Clinical features included postnatal growth retardation, moderate short stature, protuberant eyes with bluish sclerae, hands with finely wrinkled palms, atrophy of the thenar muscles, and tapering fingers. Patients have thin, hyperelastic skin and hypermobile small joints consistent with an Ehlers- Danlos-like phenotype. Radiologic features included mild to moderate platyspondyly, mild to moderate osteoperia of the spine, small ileum, flat proximal femoral epiphyses, short, wide femoral necks, and broad metaphyses (elbows, knees, wrists, and interphalangeal joints). {ECO:0000269|PubMed:18513683}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilopeirodisplazi, Ehlers-Danlos Sendromu Benzeri, 612350, Otozomal Resesif (Ehlers-Danlos Sendromu, Spondilokarpotarsal Sinostoz) (SLC39A13 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocheirodysplasia, Ehlers-Danlos syndrome-like, 612350, Autosomal recessive (Ehlers-Danlos syndrome, spondylocarpotarsal synostosis type) (SLC39A13 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC39A13	SLC39A13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ehlers-Danlos syndrome-like spondylocheirodysplasia (SCD- EDS) [MIM:612350]: Spondylocheiro dysplastic form of Ehlers-Danlos syndrome. The syndrome consists of a generalized skeletal dysplasia involving mainly the spine (spondylo) and striking clinical abnormalities of the hands (cheiro) in addition to the EDS-like features. Clinical features included postnatal growth retardation, moderate short stature, protuberant eyes with bluish sclerae, hands with finely wrinkled palms, atrophy of the thenar muscles, and tapering fingers. Patients have thin, hyperelastic skin and hypermobile small joints consistent with an Ehlers- Danlos-like phenotype. Radiologic features included mild to moderate platyspondyly, mild to moderate osteoperia of the spine, small ileum, flat proximal femoral epiphyses, short, wide femoral necks, and broad metaphyses (elbows, knees, wrists, and interphalangeal joints). {ECO:0000269|PubMed:18513683}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilokostal Displazi 1, Otozomal Resesif, 277300, Otozomal Resesif; SCDO1 (Otozomal Resesif Spondilokostal Displazi) (DLL3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 1, autosomal recessive, 277300; Autosomal recessive; SCDO1 (Autosomal recessive spondylocostal dysostosis) (DLL3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLL3	DLL3, SCDO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylocostal dysostosis 1, autosomal recessive (SCDO1) [MIM:277300]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:10742114}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilokostal Displazi 1, Otozomal Resesif, 277300, Otozomal Resesif; SCDO1 (Otozomal Resesif Spondilokostal Displazi) (Prenatal) (DLL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 1, autosomal recessive, 277300, Autosomal recessive; SCDO1 (Autosomal recessive spondylocostal dysostosis) (DLL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLL3	DLL3, SCDO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylocostal dysostosis 1, autosomal recessive (SCDO1) [MIM:277300]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:10742114}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilokostal Displazi 2, Otozomal Resesif, 608681; SCDO2 (Otozomal Resesif Spondilokostal Displazi) (MESP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 2, autosomal recessive, 608681; SCDO2 (Autosomal recessive spondylocostal dysostosis) (MESP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MESP2	MESP2, SCDO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylocostal dysostosis 2, autosomal recessive (SCDO2) [MIM:608681]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:15122512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilokostal Displazi 2, Otozomal Resesif, 608681; SCDO2 (Otozomal Resesif Spondilokostal Displazi) (Prenatal) (MESP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 2, autosomal recessive, 608681; SCDO2 (Autosomal recessive spondylocostal dysostosis) (MESP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MESP2	MESP2, SCDO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylocostal dysostosis 2, autosomal recessive (SCDO2) [MIM:608681]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:15122512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilokostal Displazi 3, Otozomal Resesif, 609813; SCDO3 (Otozomal Resesif Spondilokostal Displazi) (LFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 3, autosomal recessive, 609813; SCDO3 (Autosomal recessive spondylocostal dysostosis) (LFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LFNG	LFNG, SCDO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylocostal dysostosis 3, autosomal recessive (SCDO3) [MIM:609813]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:16385447}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilokostal Displazi 3, Otozomal Resesif, 609813; SCDO3 (Otozomal Resesif Spondilokostal Displazi) (Prenatal) (LFNG geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 3, autosomal recessive, 609813; SCDO3 (Autosomal recessive spondylocostal dysostosis) (LFNG gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LFNG	LFNG, SCDO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylocostal dysostosis 3, autosomal recessive (SCDO3) [MIM:609813]: A condition of variable severity associated with vertebral and rib segmentation defects. The main skeletal malformations include fusion of vertebrae, hemivertebrae, fusion of certain ribs, and other rib malformations. Deformity of the chest and spine (severe scoliosis, kyphoscoliosis and lordosis) is a natural consequence of the malformation and leads to a dwarf- like appearance. As the thorax is small, infants frequently have respiratory insufficiency and repeated respiratory infections resulting in life-threatening complications in the first year of life. {ECO:0000269|PubMed:16385447}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spondilokostal Displazi 4, Otozomal Resesif, 613686, Otozomal Resesif; SCDO4 (Otozomal Resesif Spondilokostal Displazi) (HES7 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 4, autosomal recessive, 613686, Autosomal recessive; SCDO4 (Autosomal recessive spondylocostal dysostosis) (HES7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HES7	HES7, SCDO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylocostal dysostosis 4, autosomal recessive (SCDO4) [MIM:613686]: A rare condition of variable severity characterized by vertebral and costal anomalies. The main feature include dwarfism, vertebral fusion, hemivertebrae, posterior rib fusion, reduced rib number, and other rib malformations. (ECO:0000269|PubMed:18775957, ECO:0000269|PubMed:20087400). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilokostal Displazi 4, Otozomal Resesif, 613686, Otozomal Resesif; SCDO4 (Otozomal Resesif Spondilokostal Displazi) (Prenatal) (HES7 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 4, autosomal recessive, 613686, Autosomal recessive; SCDO4 (Autosomal recessive spondylocostal dysostosis) (HES7 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HES7	HES7, SCDO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylocostal dysostosis 4, autosomal recessive (SCDO4) [MIM:613686]: A rare condition of variable severity characterized by vertebral and costal anomalies. The main feature include dwarfism, vertebral fusion, hemivertebrae, posterior rib fusion, reduced rib number, and other rib malformations. (ECO:0000269|PubMed:18775957, ECO:0000269|PubMed:20087400). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilokostal Displazi 5, 122600, Otozomal Resesif, Otozomal Dominant; SCDO5 (Otozomal Dominant Spondilokostal Displazi) (TBX6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 5, 122600, Autosomal recessive, Autosomal dominant; SCDO5 (Autosomal dominant spondylocostal dysostosis) (TBX6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX6	TBX6, SCDO5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylocostal dysostosis 5, autosomal dominant (SCDO5) [MIM:122600]: A rare condition of variable severity characterized by vertebral and costal anomalies. The main feature include dwarfism, vertebral fusion, hemivertebrae, posterior rib fusion, reduced rib number, and other rib malformations. (ECO:0000269	PubMed:23335591). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilokostal Displazi 5, 122600, Otozomal Resesif, Otozomal Dominant; SCDO5 (Otozomal Dominant Spondilokostal Displazi) (Prenatal) (TBX6 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 5, 122600, Autosomal recessive, Autosomal dominant; SCDO5 (Autosomal dominant spondylocostal dysostosis) (TBX6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX6	TBX6, SCDO5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylocostal dysostosis 5, autosomal dominant (SCDO5) [MIM:122600]: A rare condition of variable severity characterized by vertebral and costal anomalies. The main feature include dwarfism, vertebral fusion, hemivertebrae, posterior rib fusion, reduced rib number, and other rib malformations. (ECO:0000269	PubMed:23335591). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilokostal Displazi 6, 616566, Otozomal Resesif, SCDO6 (Otozomal Resesif Spondilokostal Displazi) (RIPPLY2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylocostal dysostosis 6, 616566, Autosomal recessive; SCDO6 (Autosomal recessive spondylocostal dysostosis) (RIPPLY2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RIPPLY2	RIPPLY2, C6orf59, SCDO6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondilokostal Displazi 6, 616566, Otozomal Resesif, SCDO6 (Otozomal Resesif Spondilokostal Displazi) (Prenatal) (RIPPLY2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylocostal dysostosis 6, 616566, Autosomal recessive; SCDO6 (Autosomal recessive spondylocostal dysostosis) (RIPPLY2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RIPPLY2	RIPPLY2, C6orf59, SCDO6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloenkondrodisplazi, İmmün Disregülasyon, 607944, Otozomal Resesif; SPENCDI (Spondilometafizyal Displazi-Kombine İmmün Yetmezlik) (ACP5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloenchondrodysplasia with immune dysregulation, 607944, Autosomal recessive; SPENCDI (Spondylometaphyseal dysplasia with combined immunodeficiency) (ACP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACP5	ACP5, SPENCDI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondyloenchondrodysplasia with immune dysregulation (SPENCDI) [MIM:607944]: A disease characterized by vertebral and metaphyseal dysplasia, spasticity with cerebral calcifications, and strong predisposition to autoimmune diseases. The skeletal dysplasia is characterized by radiolucent and irregular spondylar and metaphyseal lesions that represent islands of chondroid tissue within bone. (ECO:0000269|PubMed:21217752, ECO:0000269|PubMed:21217755). Note=The disease is caused by mutations affecting the gene represented in this entry. ACP5 inactivating mutations result in a functional excess of phosphorylated osteopontin causing deregulation of osteopontin signaling and consequential autoimmune disease.;	EDTA Blood Sample
Spondiloenkondrodisplazi, İmmün Disregülasyon, 607944, Otozomal Resesif; SPENCDI (Spondilometafizyal Displazi-Kombine İmmün Yetmezlik) (Prenatal) (ACP5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloenchondrodysplasia with immune dysregulation, 607944, Autosomal recessive; SPENCDI (Spondylometaphyseal dysplasia with combined immunodeficiency) (ACP5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACP5	ACP5, SPENCDI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondyloenchondrodysplasia with immune dysregulation (SPENCDI) [MIM:607944]: A disease characterized by vertebral and metaphyseal dysplasia, spasticity with cerebral calcifications, and strong predisposition to autoimmune diseases. The skeletal dysplasia is characterized by radiolucent and irregular spondylar and metaphyseal lesions that represent islands of chondroid tissue within bone. (ECO:0000269|PubMed:21217752, ECO:0000269|PubMed:21217755). Note=The disease is caused by mutations affecting the gene represented in this entry. ACP5 inactivating mutations result in a functional excess of phosphorylated osteopontin causing deregulation of osteopontin signaling and consequential autoimmune disease.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepimetafizyel Displazi-Eklem Gevşekliği, Tip 1, ± Kırık, 271640, Otozomal Resesif; SEMDJL1 (Spondiloepimetafizyal Displazi-Eklem Gevşekliği) (B3GALT6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640, Autosomal recessive; SEMDJL1 (Spondyloepimetaphyseal dysplasia with joint laxity) (B3GALT6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	B3GALT6	B3GALT6, SEMDJL1, EDSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondyloepimetaphyseal dysplasia with joint laxity, 1, with or without fractures (SEMDJL1) [MIM:271640]: A bone disease characterized by vertebral abnormalities and ligamentous laxity that result in spinal misalignment and progressive severe kyphoscoliosis, thoracic asymmetry, and respiratory compromise resulting in early death. Additional skeletal features include elbow deformities with radial head dislocation, dislocated hips, clubfeet, and tapered fingers with spatulate distal phalanges. Many affected children have an oval face, flat midface, prominent eyes with blue sclerae, and a long philtrum. Palatal abnormalities and congenital heart disease are also observed. (ECO:0000269|PubMed:23664117, ECO:0000269|PubMed:23664118). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondiloepimetafizyel Displazi-Eklem Gevşekliği, Tip 1, ± Kırık, 271640, Otozomal Resesif; SEMDJL1 (Spondiloepimetafizyal Displazi-Eklem Gevşekliği) (Prenatal) (B3GALT6 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640, Autosomal recessive; SEMDJL1 (Spondyloepimetaphyseal dysplasia with joint laxity) (B3GALT6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	B3GALT6	B3GALT6, SEMDJL1, EDSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondyloepimetaphyseal dysplasia with joint laxity, 1, with or without fractures (SEMDJL1) [MIM:271640]: A bone disease characterized by vertebral abnormalities and ligamentous laxity that result in spinal misalignment and progressive severe kyphoscoliosis, thoracic asymmetry, and respiratory compromise resulting in early death. Additional skeletal features include elbow deformities with radial head dislocation, dislocated hips, clubfeet, and tapered fingers with spatulate distal phalanges. Many affected children have an oval face, flat midface, prominent eyes with blue sclerae, and a long philtrum. Palatal abnormalities and congenital heart disease are also observed. (ECO:0000269|PubMed:23664117, ECO:0000269|PubMed:23664118). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepimetafizyel Displazi- Eklem Gevşekliği, Tip 2, 603546, Otozomal Dominant; SEMDJL2 (Spondiloepimetafizyal Displazi-Multipl Dislokasyon) (KIF22 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546, Autosomal dominant; SEMDJL2 (Spondyloepimetaphyseal dysplasia with multiple dislocations) (KIF22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KIF22	KIF22, KNSL4, KID, OBP, SEMDJL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondyloepimetaphyseal dysplasia with joint laxity, 2 (SEMDJL2) [MIM:603546]: A bone disease characterized by short stature, distinctive midface retrusion, progressive knee malalignment (genu valgum and/or varum), generalized ligamentous laxity, and mild spinal deformity. Intellectual development is not impaired. Radiographic characteristics include significantly retarded epiphyseal ossification that evolves into epiphyseal dysplasia and precocious osteoarthritis, metaphyseal irregularities and vertical striations, constricted femoral neck, slender metacarpals and metatarsals, and mild thoracolumbar kyphosis or scoliosis with normal or mild platyspondyly. The most distinctive features for differential diagnosis of SEMDJL2 are the slender metacarpals and phalanges and the progressive degeneration of carpal bones; however, these 2 features are evident only in older children and young adults. The soft consistency of cartilage in the airways leads to laryngotracheomalacia with proneness to respiratory obstruction and inspiratory stridor in infancy and childhood. (ECO:0000269|PubMed:22152677, ECO:0000269|PubMed:22152678). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondiloepimetafizyel Displazi- Eklem Gevşekliği, Tip 2, 603546, Otozomal Dominant; SEMDJL2 (Spondiloepimetafizyal Displazi-Multipl Dislokasyon) (Prenatal) (KIF22 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepimetaphyseal dysplasia with joint laxity, type 2, 603546, Autosomal dominant; SEMDJL2 (Spondyloepimetaphyseal dysplasia with multiple dislocations) (KIF22 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KIF22	KIF22, KNSL4, KID, OBP, SEMDJL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondyloepimetaphyseal dysplasia with joint laxity, 2 (SEMDJL2) [MIM:603546]: A bone disease characterized by short stature, distinctive midface retrusion, progressive knee malalignment (genu valgum and/or varum), generalized ligamentous laxity, and mild spinal deformity. Intellectual development is not impaired. Radiographic characteristics include significantly retarded epiphyseal ossification that evolves into epiphyseal dysplasia and precocious osteoarthritis, metaphyseal irregularities and vertical striations, constricted femoral neck, slender metacarpals and metatarsals, and mild thoracolumbar kyphosis or scoliosis with normal or mild platyspondyly. The most distinctive features for differential diagnosis of SEMDJL2 are the slender metacarpals and phalanges and the progressive degeneration of carpal bones; however, these 2 features are evident only in older children and young adults. The soft consistency of cartilage in the airways leads to laryngotracheomalacia with proneness to respiratory obstruction and inspiratory stridor in infancy and childhood. (ECO:0000269|PubMed:22152677, ECO:0000269|PubMed:22152678). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepimetafizyel Displazi, 608728, Otozomal Resesif (Spondiloepimetafizyal Displazi, Matrilin-3 Tip) (MATN3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepimetaphyseal dysplasia, 608728, Autosomal recessive (Spondyloepimetaphyseal dysplasia, matrilin-3 type) (MATN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MATN3	MATN3, EDM5, HOA, OS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteoarthritis 2 (OS2) [MIM:140600]: A degenerative disease of the joints characterized by degradation of the hyaline articular cartilage and remodeling of the subchondral bone with sclerosis. Clinical symptoms include pain and joint stiffness often leading to significant disability and joint replacement. In the hand, osteoarthritis can develop in the distal interphalangeal and the first carpometacarpal (base of thumb) and proximal interphalangeal joints. Patients with osteoarthritis may have one, a few, or all of these sites affected. [ECO:0000269	PubMed:12736871). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondiloepimetafizyel Displazi, 608728, Otozomal Resesif (Spondiloepimetafizyal Displazi, Matrilin-3 Tip) (Prenatal) (MATN3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepimetaphyseal dysplasia, 608728, Autosomal recessive (Spondyloepimetaphyseal dysplasia, matrilin-3 type) (MATN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MATN3	MATN3, EDM5, HOA, OS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteoarthritis 2 (OS2) [MIM:140600]: A degenerative disease of the joints characterized by degradation of the hyaline articular cartilage and remodeling of the subchondral bone with sclerosis. Clinical symptoms include pain and joint stiffness often leading to significant disability and joint replacement. In the hand, osteoarthritis can develop in the distal interphalangeal and the first carpometacarpal (base of thumb) and proximal interphalangeal joints. Patients with osteoarthritis may have one, a few, or all of these sites affected. [ECO:0000269	PubMed:12736871). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteochondritis dissecans short stature and early-onset osteoarthritis (OD) [MIM:165800]: A type of osteochondritis defined as a separation of cartilage and subchondral bone from the surrounding tissue, primarily affecting the knee, ankle and elbow joints. It is clinically characterized by multiple osteochondritic lesions in knees and/or hips and/or elbows, disproportionate short stature and early-onset osteoarthritis. {ECO:0000269|PubMed:20137779}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (MLPA)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (Prenatal) (MLPA)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (Prenatal) (MLPA)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (Prenatal) (MLPA)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital; Sedc (Spondiloepifizyal Displazi Konjenita) (Prenatal) (MLPA)	SPONDYLOEPIPHYSEAL DYSPLASIA CONGENITA; SEDC (Spondyloepiphyseal dysplasia congenita) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi Tarda-Progresif Artropati, 208230, Otozomal Resesif (Çocukluk Çağı Progresif Psödoromatoid Artropatisi (Progresif Artropati İle Spondiloepifizyal Displazi Tarda)) (WISP3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230, Autosomal recessive (Progressive pseudorheumatoid arthropathy of childhood) (WISP3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WISP3	WISP3, PPAC, PPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondiloepifizyal Displazi Tarda-Progresif Artropati, 208230, Otozomal Resesif (Çocukluk Çağı Progresif Psödoromatoid Artropatisi (Progresif Artropati İle Spondiloepifizyal Displazi Tarda)) (Prenatal) (WISP3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230, Autosomal recessive (Progressive pseudorheumatoid arthropathy of childhood) (WISP3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WISP3	WISP3, PPAC, PPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi Tarda, 313400, X'e Bağlı Resesif; SEDT (Spondiloepifizyal Displazia Tarda) (TRAPPC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia tarda, 313400, X-linked recessive; SEDT (Spondyloepiphyseal dysplasia tarda) (TRAPPC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRAPPC2	TRAPPC2, SEDL, SEDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondiloepifizyal Displazia Tarda, 313400, X'e Bağlı Resesif; SEDT (Spondiloepifizyal Displazia Tarda) (Prenatal) (TRAPPC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia tarda, 313400, X-linked recessive; SEDT (Spondyloepiphyseal dysplasia tarda) (TRAPPC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRAPPC2	TRAPPC2, SEDL, SEDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spondiloepifizyal Displazi, Konjenital Eklem Dislokasyonlari, 143095, Otozomal Resesif; SEDCJD (CHST3 ilgili Iskelet Displazisi) (CHST3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095, Autosomal recessive; SEDCJD (CHST3-related skeletal dysplasia) (CHST3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHST3	CHST3, CGST, CGST1, HSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Spondiloepifizyal Displazi, Konjenital Eklem Dislokasyonlari, 143095, Otozomal Resesif; SEDCJD (CHST3 ilgili Iskelet Displazisi) (Prenatal) (CHST3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia with congenital joint dislocations, 143095, Autosomal recessive; SEDCJD (CHST3-related skeletal dysplasia) (CHST3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CHST3	CHST3, CGST, CGST1, HSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Kimberley Tipi, 608361, Otozomal Dominant; SEDK (Spondiloepifizyal Displazi, Kimberley Tipi) (ACAN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia, Kimberley type, 608361, Autosomal dominant; SEDK (Spondyloepiphyseal dysplasia, Kimberley type) (ACAN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACAN	ACAN, AGC1, CSPG1, MSK16, SEDK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Osteochondritis dissecans short stature and early-onset osteoarthritis (OD) [MIM:165800]: A type of osteochondritis defined as a separation of cartilage and subchondral bone from the surrounding tissue, primarily affecting the knee, ankle and elbow joints. It is clinically characterized by multiple osteochondritic lesions in knees and/or hips and/or elbows, disproportionate short stature and early-onset osteoarthritis. {ECO:0000269|PubMed:20137779}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondiloepifizyal Displazi, Kimberley Tipi, 608361, Otozomal Dominant; SEDK (Spondiloepifizyal Displazi, Kimberley Tipi) (Prenatal) (ACAN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia, Kimberley type, 608361, Autosomal dominant; SEDK (Spondyloepiphyseal dysplasia, Kimberley type) (ACAN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ACAN	ACAN, AGC1, CSPG1, MSK16, SEDK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Osteochondritis dissecans short stature and early-onset osteoarthritis (OD) [MIM:165800]: A type of osteochondritis defined as a separation of cartilage and subchondral bone from the surrounding tissue, primarily affecting the knee, ankle and elbow joints. It is clinically characterized by multiple osteochondritic lesions in knees and/or hips and/or elbows, disproportionate short stature and early-onset osteoarthritis. {ECO:0000269|PubMed:20137779}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Stanescu Tipi, 616583, Otozomal Dominant; SEDSTN (Spondiloepifizyal Displazi, Stanescu Tipi) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia, Stanescu type, 616583, autosomal dominant; SEDSTN (Spondyloepiphyseal dysplasia, Stanescu type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondiloepifizyal Displazi, Stanescu Tipi, 616583, Otozomal Dominant; SEDSTN (Spondiloepifizyal Displazi, Stanescu Tipi) (MLPA)	Spondyloepiphyseal dysplasia, Stanescu type, 616583, autosomal dominant; SEDSTN (Spondyloepiphyseal dysplasia, Stanescu type) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondiloepifizyal Displazi, Stanescu Tipi, 616583, Otozomal Dominant; SEDSTN (Spondiloepifizyal Displazi, Stanescu Tipi) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia, Stanescu type, 616583, autosomal dominant; SEDSTN (Spondyloepiphyseal dysplasia, Stanescu type) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepifizyal Displazi, Stanescu Tipi, 616583, Otozomal Dominant; SEDSTN (Spondiloepifizyal Displazi, Stanescu Tipi) (MLPA)	Spondyloepiphyseal dysplasia, Stanescu type, 616583, autosomal dominant; SEDSTN (Spondyloepiphyseal dysplasia, Stanescu type) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometaepifizyal Displazi, Ksa Ekstremite-El Tipi, 271665, Otozomal Resesif (Spondiloepimetafizyal Displazi-Ksa Ekstremite-Anormal Kalsifikasyon Sendromu) (DDR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665, Autosomal recessive (Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome) (DDR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDR2	DDR2, NTRKR3, TKT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondyloepimetaphyseal dysplasia short limb-hand type (SEMD-SL) [MIM:271665]: A bone disease characterized by short- limbed dwarfism, a narrow chest with pectus excavatum, brachydactyly in the hands and feet, a characteristic craniofacial appearance and premature calcifications. The radiological findings are distinctive and comprise short long bones throughout the skeleton with striking epiphyses that are stippled, flattened and fragmented and flared, irregular metaphyses. Platyspondyly in the spine with wide intervertebral spaces is observed and some vertebral bodies are pear-shaped with central humps, anterior protrusions and posterior scalloping. {ECO:0000269|PubMed:19110212, ECO:0000269|PubMed:20223752}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilometaepifizyal Displazi, Ksa Ekstremite-El Tipi, 271665, Otozomal Resesif (Spondiloepimetafizyal Displazi-Ksa Ekstremite-Anormal Kalsifikasyon Sendromu) (Prenatal) (DDR2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaepiphyseal dysplasia, short limb-hand type, 271665, Autosomal recessive (Spondyloepimetaphyseal dysplasia-short limb-abnormal calcification syndrome) (DDR2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDR2	DDR2, NTRKR3, TKT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondyloepimetaphyseal dysplasia short limb-hand type (SEMD-SL) [MIM:271665]: A bone disease characterized by short- limbed dwarfism, a narrow chest with pectus excavatum, brachydactyly in the hands and feet, a characteristic craniofacial appearance and premature calcifications. The radiological findings are distinctive and comprise short long bones throughout the skeleton with striking epiphyses that are stippled, flattened and fragmented and flared, irregular metaphyses. Platyspondyly in the spine with wide intervertebral spaces is observed and some vertebral bodies are pear-shaped with central humps, anterior protrusions and posterior scalloping. {ECO:0000269|PubMed:19110212, ECO:0000269|PubMed:20223752}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, Kon-Rod Distrofisi İle 608940, Otozomal Resesif; SMDCRD (Spondilometafizyal Displazi-Kon Rod Distrofi Sendromu) (PCYT1A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940, Autosomal recessive; SMDCRD (Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome) (PCYT1A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCYT1A	PCYT1A, CTPCT, PCYT1, SMDCRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylometaphyseal dysplasia with cone-rod dystrophy (SMDCRD) [MIM:608940]: A disorder characterized by postnatal growth deficiency resulting in profound short stature, rhizomelia with bowing of the lower extremities, platyspondyly with anterior vertebral protrusions, progressive metaphyseal irregularity and cupping with shortened tubular bones, and early-onset progressive visual impairment associated with a pigmentary maculopathy and electroretinographic evidence of cone-rod dysfunction. {ECO:0000269|PubMed:24387990, ECO:0000269|PubMed:24387991}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Spondilometafizyal Displazi, Kon-Rod Distrofisi İle 608940, Otozomal Resesif; SMDCRD (Spondilometafizyal Displazi-Kon Rod Distrofi Sendromu) (Prenatal) (PCYT1A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaphyseal dysplasia with cone-rod dystrophy, 608940, Autosomal recessive; SMDCRD (Spondylometaphyseal dysplasia-cone-rod dystrophy syndrome) (PCYT1A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PCYT1A	PCYT1A, CTPCT, PCYT1, SMDCRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylometaphyseal dysplasia with cone-rod dystrophy (SMDCRD) [MIM:608940]: A disorder characterized by postnatal growth deficiency resulting in profound short stature, rhizomelia with bowing of the lower extremities, platyspondyly with anterior vertebral protrusions, progressive metaphyseal irregularity and cupping with shortened tubular bones, and early-onset progressive visual impairment associated with a pigmentary maculopathy and electroretinographic evidence of cone-rod dysfunction. {ECO:0000269|PubMed:24387990, ECO:0000269|PubMed:24387991}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Spondilometafizyal Displazi, 'Köşe Kırığı' Tipi (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaphyseal dysplasia, 'corner fracture' type (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilometafizyal Displazi, 'Köşe Kırığı' Tipi (MLPA)	Spondylometaphyseal dysplasia, 'corner fracture' type (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilometafizyal Displazi, 'Köşe Kırığı' Tipi (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaphyseal dysplasia, 'corner fracture' type (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, 'Köşe Kırığı' Tipi (Prenatal) (MLPA)	Spondylometaphyseal dysplasia, 'corner fracture' type (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, Megarbane-Dagher-Melike Tipi, 613320, Otozomal Resesif; SMDMDM (Otozomal Resesif Spondilometafizyal Displazi, Megarbane Tipi) (PAM16 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320, Autosomal recessive; SMDMDM (Autosomal recessive spondylometaphyseal dysplasia, Megarbané type) (PAM16 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAM16	PAM16, MAGMAS, SMDMDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type (SMDMDM) [MIM:613320]: An autosomal recessive disease characterized by pre- and postnatal short stature, developmental delay, dysmorphic facial appearance, narrow chest, prominent abdomen, platyspondyly, and short limbs. {ECO:0000269|PubMed:24786642}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilometafizyal Displazi, Megarbane-Dagher-Melike Tipi, 613320, Otozomal Resesif; SMDMDM (Otozomal Resesif Spondilometafizyal Displazi, Megarbane Tipi) (Prenatal) (PAM16 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type, 613320, Autosomal recessive; SMDMDM (Autosomal recessive spondylometaphyseal dysplasia, Megarbané type) (PAM16 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAM16	PAM16, MAGMAS, SMDMDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type (SMDMDM) [MIM:613320]: An autosomal recessive disease characterized by pre- and postnatal short stature, developmental delay, dysmorphic facial appearance, narrow chest, prominent abdomen, platyspondyly, and short limbs. {ECO:0000269|PubMed:24786642}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, Schmidt Tipi (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaphyseal dysplasia, Schmidt type (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilometafizyal Displazi, Schmidt Tipi (MLPA)	Spondylometaphyseal dysplasia, Schmidt type (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondilometafizyal Displazi, Schmidt Tipi (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaphyseal dysplasia, Schmidt type (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, Schmidt Tipi (Prenatal) (MLPA)	Spondylometaphyseal dysplasia, Schmidt type (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilometafizyal Displazi, Sedaghatian Tipi, 250220, Otozomal Resesif; SMDS (Spondilometafizyal Displazi, Sedaghatian Tipi) (GPX4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondylometaphyseal dysplasia, Sedaghatian type, 250220, Autosomal recessive; SMDS (Spondylometaphyseal dysplasia, Sedaghatian type) (GPX4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPX4	GPX4, SMDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Spondylometafizyal Displazi, Sedaghatian Tip, 250220, Otozomal Resesif; SMDS (Spondilometafizyal Displazi, Sedaghatian Tip) (Prenatal) (GPX4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondylometaphyseal dysplasia, Sedaghatian type, 250220, Autosomal recessive; SMDS (Spondylometaphyseal dysplasia, Sedaghatian type) (GPX4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GPX4	GPX4, SMDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondilooküler Sendrom, 605822, Otozomal Resesif; SOS (Spondilo-Oküler Sendrom) (XYLT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloocular syndrome, 605822, Autosomal recessive; SOS (Spondylo-ocular syndrome) (XYLT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XYLT2	XYLT2, XT2, SOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:16571645}. Note=The gene represented in this entry acts as a disease modifier. PXE patients carrying causative ABCC6 mutations, manifest a more severe disease course characterized by earlier onset, frequent skin lesions and higher organ involvement, in the presence of XYLT2 variants. {ECO:0000269|PubMed:16571645};.	EDTA Blood Sample
Spondilooküler Sendrom, 605822, Otozomal Resesif; SOS (Spondilo-Oküler Sendrom) (Prenatal) (XYLT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloocular syndrome, 605822, Autosomal recessive; SOS (Spondylo-ocular syndrome) (XYLT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XYLT2	XYLT2, XT2, SOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pseudoxanthoma elasticum (PXE) [MIM:264800]: A multisystem disorder characterized by accumulation of mineralized and fragmented elastic fibers in the skin, vascular walls, and Burch membrane in the eye. Clinically, patients exhibit characteristic lesions of the posterior segment of the eye including peau d'orange, angioid streaks, and choroidal neovascularizations, of the skin including soft, ivory colored papules in a reticular pattern that predominantly affect the neck and large flexor surfaces, and of the cardiovascular system with peripheral and coronary arterial occlusive disease as well as gastrointestinal bleedings. {ECO:0000269|PubMed:16571645}. Note=The gene represented in this entry acts as a disease modifier. PXE patients carrying causative ABCC6 mutations, manifest a more severe disease course characterized by earlier onset, frequent skin lesions and higher organ involvement, in the presence of XYLT2 variants. {ECO:0000269|PubMed:16571645};.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepiferal Displazi, 271700, Otozomal Dominant (Spondiloepiferal Displazi-Kısa Ulna Sendromu) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Spondyloepiphyseal dysplasia, 271700, autosomal dominant (Spondyloepiphyseal dysplasia-short ulna syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondiloepiferal Displazi, 271700, Otozomal Dominant (Spondiloepiferal Displazi-Kısa Ulna Sendromu) (MLPA)	Spondyloepiphyseal dysplasia, 271700, autosomal dominant (Spondyloepiphyseal dysplasia-short ulna syndrome) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Spondiloepiferal Displazi, 271700, Otozomal Dominant (Spondiloepiferal Displazi-Kısa Ulna Sendromu) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Spondyloepiphyseal dysplasia, 271700, autosomal dominant (Spondyloepiphyseal dysplasia-short ulna syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Spondiloepiferal Displazi, 271700, Otozomal Dominant (Spondiloepiferal Displazi-Kısa Ulna Sendromu) (Prenatal) (MLPA)	Spondyloepiphyseal dysplasia, 271700, autosomal dominant (Spondyloepiphyseal dysplasia-short ulna syndrome) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Skuamöz Hücre Karsinomu, Yanık Skarı İle İlgili, Somatik (FAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Squamous cell carcinoma, burn scar-related, somatic (FAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAS	FAS, TNFRSF6, APT1, CD95, ALPS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Autoimmune lymphoproliferative syndrome 1A (ALPS1A) [MIM:601859]: A disorder of apoptosis that manifests in early childhood and results in the accumulation of autoreactive lymphocytes. It is characterized by non-malignant lymphadenopathy with hepatosplenomegaly, and autoimmune hemolytic anemia, thrombocytopenia and neutropenia. {ECO:0000269|PubMed:10090885, ECO:0000269|PubMed:10340403, ECO:0000269|PubMed:10515860, ECO:0000269|PubMed:11418480, ECO:0000269|PubMed:17336828, ECO:0000269|PubMed:7540117, ECO:0000269|PubMed:8929361, ECO:0000269|PubMed:9028321, ECO:0000269|PubMed:9028957, ECO:0000269|PubMed:9322534, ECO:0000269|PubMed:9821419, ECO:0000269|PubMed:9927496}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Skuamöz Hücre Karsinomu, Baş ve Boyun, 275355, Otozomal Resesif; HNSCC (Baş ve Boyun Skuamöz Hücre Karsinomu) (TNFRSF10B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Squamous cell carcinoma, head and neck, 275355, Autosomal recessive; HNSCC (Squamous cell carcinoma of head and neck) (TNFRSF10B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNFRSF10B	TNFRSF10B, DR5, TRAILR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Squamous cell carcinoma of the head and neck (HNSCC) [MIM:275355]: A non-melanoma skin cancer affecting the head and neck. The hallmark of cutaneous SCC is malignant transformation of normal epidermal keratinocytes. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Skuamöz Hücreli Karsinoma, Baş ve Boyun, Somatik, 275355 (Baş ve Boyun Skuamöz Hücre Karsinomu) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Squamous cell carcinoma, head and neck, somatic, 275355 (Squamous cell carcinoma of head and neck) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Skuamöz Hücre Karsinomu, Baş ve Boyun, Somatik, 275355 (Baş ve Boyun Skuamöz Hücre Karsinomu) (ING1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Squamous cell carcinoma, head and neck, somatic, 275355 (Squamous cell carcinoma of head and neck) (ING1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ING1	ING1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Squamous cell carcinoma of the head and neck (HNSCC) [MIM:275355]: A non-melanoma skin cancer affecting the head and neck. The hallmark of cutaneous SCC is malignant transformation of normal epidermal keratinocytes. {ECO:0000269	PubMed:10866301}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
SRY (Yp11.3) (FISH)	SRY (Yp11.3) (FISH)	Yp11.3	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample

SRY Geni Delesyon ve varlığı	SRY gene deletion and presence	SRY	.	PCR/ Jel elektroforezi	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	DISEASE: 46,XY sex reversal 1 (SRXY1) [MIM:400044]: A condition characterized by male-to-female sex reversal in the presence of a normal 46,XY karyotype. Patients manifest rapid and early degeneration of their gonads, which are present in the adult as 'streak gonads', consisting mainly of fibrous tissue and variable amounts of ovarian stroma. As a result these patients do not develop secondary sexual characteristics at puberty. The external genitalia in these subjects are completely female, and Muellerian structures are normal. {ECO:0000269|PubMed:10670762, ECO:0000269|PubMed:10721678, ECO:0000269|PubMed:10843173, ECO:0000269|PubMed:10852465, ECO:0000269|PubMed:12107262, ECO:0000269|PubMed:12793612, ECO:0000269|PubMed:1339396, ECO:0000269|PubMed:1415266, ECO:0000269|PubMed:1483689, ECO:0000269|PubMed:1570829, ECO:0000269|PubMed:17063144, ECO:0000269|PubMed:2247149, ECO:0000269|PubMed:7717397, ECO:0000269|PubMed:7776083, ECO:0000269|PubMed:7985018, ECO:0000269|PubMed:8019555, ECO:0000269|PubMed:8105086, ECO:0000269|PubMed:8353496, ECO:0000269|PubMed:8447323, ECO:0000269|PubMed:9450909, ECO:0000269|PubMed:9521592, ECO:0000269|PubMed:9678356, ECO:0000269|Ref.41, ECO:0000269|Ref.47}. Note=The disease is caused by	EDTA Blood Sample
Stapes Ankilozu-Geniş Parmak ve Ayak Parmakları, 184460, Otozomal Dominant (Stapes Ankilozu-Geniş El ve Ayak Başparmak) (NOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stapes ankylosis with broad thumb and toes, 184460, Autosomal dominant (Stapes ankylosis with broad thumbs and toes) (NOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOG	NOG, SYM1, SYNS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stapes ankylosis with broad thumb and toes (SABTS) [MIM:184460]: Congenital autosomal dominant disorder that includes hyperopia, a hemicylindrical nose, broad thumbs, great toes, and other minor skeletal anomalies but lacked carpal and tarsal fusion and symphalangism. {ECO:0000269|PubMed:12095654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
STAR Sendromu, 300707, X'e Bağlı Dominant (Sindaktili-Telekantus-Anogenital-Renal Malformasyonlar Sendromu) (FAM58A gen) (Tüm Gen Dizi Analizi) (Postnatal)	STAR syndrome, 300707, X-linked dominant (Syndactyly-telecanthus-anogenital and renal malformations syndrome) (FAM58A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAM58A	FAM58A, STAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Toe syndactyly, telecanthus, and anogenital and renal malformations (STAR) [MIM:300707]: A syndrome characterized by anal, genital and renal tract anomalies, facial dysmorphism and syndactyly. Features include anal stenosis, a rectovaginal fistula, clitoral hypertrophy, a pelvic right kidney, toe syndactyly, and telecanthus. {ECO:0000269|PubMed:18297069}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Stargardt Hastalığı 1, 248200, Otozomal Resesif; STGD1 (Stargardt Hastalığı) (ABCA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stargardt disease 1, 248200, Autosomal recessive; STGD1 (Stargardt disease) (ABCA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA4	ABCA4, ABCR, STGD1, FFM, RP19, CORD3, ARMD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Macular degeneration, age-related, 2 (ARMD2) [MIM:153800]: A form of age-related macular degeneration, a multifactorial eye disease and the most common cause of irreversible vision loss in the developed world. In most patients, the disease is manifest as ophthalmoscopically visible yellowish accumulations of protein and lipid that lie beneath the retinal pigment epithelium and within an elastin-containing structure known as Bruch membrane. {ECO:0000269|PubMed:19028736, ECO:0000269|PubMed:9295268}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Stargardt Hastalığı 3, 600110, Otozomal Dominant; STGD3 (Stargardt Hastalığı) (ELOVL4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stargardt disease 3, 600110, Autosomal dominant; STGD3 (Stargardt disease) (ELOVL4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELOVL4	ELOVL4, ADMD, STGD2, STGD3, ISQMR, SCA34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spinocerebellar ataxia 34 (SCA34) [MIM:133190]: A form of spinocerebellar ataxia, a clinically and genetically heterogeneous group of cerebellar disorders. Patients show progressive incoordination of gait and often poor coordination of hands, speech and eye movements, due to degeneration of the cerebellum with variable involvement of the brainstem and spinal cord. SCA34 is an autosomal dominant form characterized by the association of progressive cerebellar ataxia with erythrokeratodermia variabilis. {ECO:0000269|PubMed:24566826}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Stargardt Hastalığı, 603786; STGD4 (Stargardt Hastalığı) (PROM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stargardt disease 4, 603786; STGD4 (Stargardt disease) (PROM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROM1	PROM1, PROML1, AC133, RP41, CORD12, CD133, MCDR2, STGD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stargardt disease 4 (STGD4) [MIM:603786]: A common hereditary macular degeneration. It is characterized by decreased central vision, atrophy of the macula and underlying retinal pigment epithelium, and frequent presence of prominent flecks in the posterior pole of the retina. {ECO:0000269|PubMed:18654668}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Kalıtsal hiperekpleksia (Startle hastalığı) paneli (GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1) (MLPA)	Startle disease, Hyperekplexia (GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1) (MLPA)	GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Kalıtsal hiperekpleksia (Startle hastalığı) paneli (GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1) (MLPA) (Prenatal)	Startle disease, Hyperekplexia (GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1) (MLPA) (Prenatal)	GLRB 4q32.1, GLRA1 5q33.1, SLC6A5 11p15.1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Steel Sendromu, 615155, Otozomal Resesif; STLS (Steel Sendromu) (COL27A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Steel syndrome, 615155, Autosomal recessive; STLS (Steel syndrome) (COL27A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL27A1	COL27A1, KIAA1870, STLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Steel syndrome (STLS) [MIM:615155]: A syndrome characterized by dislocated hips and radial heads, fusion of carpal bones, short stature, scoliosis, and cervical spine anomalies. Facial features include prominent forehead, long oval-shaped face, hypertelorism and broad nasal bridge. {ECO:0000269|PubMed:24986830}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Steel Sendromu, 615155, Otozomal Resesif; STLS (Steel Sendromu) (Prenatal) (COL27A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Steel syndrome, 615155, Autosomal recessive; STLS (Steel syndrome) (COL27A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL27A1	COL27A1, KIAA1870, STLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Steel syndrome (STLS) [MIM:615155]: A syndrome characterized by dislocated hips and radial heads, fusion of carpal bones, short stature, scoliosis, and cervical spine anomalies. Facial features include prominent forehead, long oval-shaped face, hypertelorism and broad nasal bridge. {ECO:0000269|PubMed:24986830}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Steroid Sülfataz Eksikliği (Xp22.31 Mikrodelesyonu) (FISH)	Steroid sulphatase deficiency (Xp22.31 microdeletion) (FISH)	Xp22.31	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal		Heparinized blood sample
Steroid Sülfataz Eksikliği (Xp22 31 Mikrodelesyonu) (FISH) (Prenatal)	Steroid sulphatase deficiency (Xp22 31 microdeletion) (Prenatal) (FISH)	Xp22.31	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stevens-Johnson Sendromu, Yatkınlık, 608579 (Stevens-Johnson Sendromu (HLA-B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stevens-Johnson syndrome, susceptibility to, 608579 (Stevens-Johnson syndrome) (HLA-B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-B	HLA-B, SPDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Stickler Sendromu, Tip 1, 108300, Otozomal Dominant; STL1 (Stickler Sendromu) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type I, 108300, autosomal dominant; STL1 (Stickler syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 1, 108300, Otozomal Dominant; STL1 (Stickler Sendromu) (MLPA)	Stickler syndrome, type I, 108300, autosomal dominant; STL1 (Stickler syndrome) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 1, 108300, Otozomal Dominant; STL1 (Stickler Sendromu) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type I, 108300, autosomal dominant; STL1 (Stickler syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİŞİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Stickler Sendromu, Tip 1, 108300, Otozomal Dominant; STL1 (Stickler Sendromu) (Prenatal) (MLPA)	Stickler syndrome, type I, 108300, autosomal dominant; STL1 (Stickler syndrome) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, Tip 1, Sendromik Olmayan Oküler, 609508, Otozomal Dominant (Stickler Sendromu) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type I, nonsyndromic ocular, 609508, autosomal dominant (Stickler syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 1, Sendromik Olmayan Oküler, 609508, Otozomal Dominant (Stickler Sendromu) (MLPA)	Stickler syndrome, type I, nonsyndromic ocular, 609508, autosomal dominant (Stickler syndrome) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 1, Sendromik Olmayan Oküler, 609508, Otozomal Dominant (Stickler Sendromu) (Prenatal) (COL2A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type I, nonsyndromic ocular, 609508, autosomal dominant (Stickler syndrome) (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, Tip 1, Sendromik Olmayan Oküler, 609508, Otozomal Dominant (Stickler Sendromu) (Prenatal) (MLPA)	Stickler syndrome, type I, nonsyndromic ocular, 609508, autosomal dominant (Stickler syndrome) (Prenatal) (MLPA)	COL2A1	COL2A1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 1 (STL1) [MIM:108300]: An autosomal dominant form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:11007540, ECO:0000269|PubMed:20513134, ECO:0000269|PubMed:7977371}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, Tip 2, 604841, Otozomal Dominant; STL2 (Stickler Sendromu) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type II, 604841, Autosomal dominant; STL2 (Stickler syndrome) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Marshall syndrome (MRSHS) [MIM:154780]: An autosomal dominant disorder characterized by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia. Clinical features include short stature; flat or retruded midface with short, depressed nose, flat nasal bridge and anteverted nares; cleft palate with or without the Pierre Robin sequence; appearance of large eyes with ocular hypertelorism; cataracts, either congenital or juvenile; esotropia; high myopia; sensorineural hearing loss; spondyloepiphyseal abnormalities; calcification of the falx cerebri; ectodermal abnormalities, including defects in sweating and dental structures. {ECO:0000269|PubMed:10486316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 2, 604841, Otozomal Dominant; STL2 (Stickler Sendromu) (Prenatal) (COL11A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type II, 604841, Autosomal dominant; STL2 (Stickler syndrome) (COL11A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A1	COL11A1, STL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Marshall syndrome (MRSHS) [MIM:154780]: An autosomal dominant disorder characterized by ocular abnormalities, deafness, craniofacial anomalies, and anhidrotic ectodermal dysplasia. Clinical features include short stature; flat or retruded midface with short, depressed nose, flat nasal bridge and anteverted nares; cleft palate with or without the Pierre Robin sequence; appearance of large eyes with ocular hypertelorism; cataracts, either congenital or juvenile; esotropia; high myopia; sensorineural hearing loss; spondyloepiphyseal abnormalities; calcification of the falx cerebri; ectodermal abnormalities, including defects in sweating and dental structures. {ECO:0000269|PubMed:10486316}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, Tip 3, 184840, Otozomal Dominant (Stickler Sendromu Tip 3) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type III, 184840, Autosomal dominant (Stickler syndrome type 3) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 3 (STL3) [MIM:184840]: An autosomal dominant non-ocular form of Stickler syndrome. Classical Stickler syndrome associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular symptoms are absent. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:9506662}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, Tip 3, 184840, Otozomal Dominant (Stickler Sendromu Tip 3) (Prenatal) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type III, 184840, Autosomal dominant (Stickler syndrome type 3) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 3 (STL3) [MIM:184840]: An autosomal dominant non-ocular form of Stickler syndrome. Classical Stickler syndrome associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular symptoms are absent. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:9506662}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, Tip 4, 614134; STL4 (Stickler Sendromu) (COL9A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type IV, 614134; STL4 (Stickler syndrome) (COL9A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A1	COL9A1, EDM6, STL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stickler syndrome 4 (STL4) [MIM:614134]: An autosomal recessive form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylisis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:16909383}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Stickler Sendromu, Tip 4, 614134; STL4 (Stickler Sendromu) (Prenatal) (COL9A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type IV, 614134; STL4 (Stickler syndrome) (COL9A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL9A1	COL9A1, EDM6, STL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stickler syndrome 4 (STL4) [MIM:614134]: An autosomal recessive form of Stickler syndrome, an inherited disorder that associates ocular signs with more or less complete forms of Pierre Robin sequence, bone disorders and sensorineural deafness. Ocular disorders may include juvenile cataract, myopia, strabismus, vitreoretinal or chorioretinal degeneration, retinal detachment, and chronic uveitis. Pierre Robin sequence includes an opening in the roof of the mouth (a cleft palate), a large tongue (macroglossia), and a small lower jaw (micrognathia). Bones are affected by slight platyspondylosis and large, often defective epiphyses. Juvenile joint laxity is followed by early signs of arthrosis. The degree of hearing loss varies among affected individuals and may become more severe over time. Syndrome expressivity is variable. {ECO:0000269|PubMed:16909383}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stickler Sendromu, 5 Tipi, 614284, Otozomal Resesif; STL5 (Stickler Sendromu) (COL9A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stickler syndrome, type V, 614284, Autosomal recessive; STL5 (Stickler syndrome) (COL9A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL9A2	COL9A2, EDM2, STL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Multiple epiphyseal dysplasia 2 (EDM2) [MIM:600204]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10364514}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Stickler Sendromu, 5 Tipi, 614284, Otozomal Resesif; STL5 (Stickler Sendromu) (Prenatal) (COL9A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stickler syndrome, type V, 614284, Autosomal recessive; STL5 (Stickler syndrome) (COL9A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL9A2	COL9A2, EDM2, STL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Multiple epiphyseal dysplasia 2 (EDM2) [MIM:600204]: A generalized skeletal dysplasia associated with significant morbidity. Joint pain, joint deformity, waddling gait, and short stature are the main clinical signs and symptoms. Radiological examination of the skeleton shows delayed, irregular mineralization of the epiphyseal ossification centers and of the centers of the carpal and tarsal bones. Multiple epiphyseal dysplasia is broadly categorized into the more severe Fairbank and the milder Ribbing types. The Fairbank type is characterized by shortness of stature, short and stubby fingers, small epiphyses in several joints, including the knee, ankle, hand, and hip. The Ribbing type is confined predominantly to the hip joints and is characterized by hands that are normal and stature that is normal or near-normal. {ECO:0000269|PubMed:10364514}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sert Deri Sendromu, 184900, Otozomal Dominant; SSKS (Sert Deri Sendromu) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stiff skin syndrome, 184900, Autosomal dominant; SSKS (Stiff skin syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683323}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sert Deri Sendromu, 184900, Otozomal Dominant; SSKS (Sert Deri Sendromu) (MLPA)	Stiff skin syndrome, 184900, Autosomal dominant; SSKS (Stiff skin syndrome) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683323}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Sert Deri Sendromu, 184900, Otozomal Dominant; SSKS (Sert Deri Sendromu) (Prenatal) (FBN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stiff skin syndrome, 184900, Autosomal dominant; SSKS (Stiff skin syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683323}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sert Deri Sendromu, 184900, Otozomal Dominant; SSKS (Sert Deri Sendromu) (Prenatal) (MLPA)	Stiff skin syndrome, 184900, Autosomal dominant; SSKS (Stiff skin syndrome) (Prenatal) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Acromicric dysplasia (ACMICD) [MIM:102370]: An autosomal dominant disorder characterized by severe short stature, short hands and feet, joint limitations, and skin thickening. Radiologic features include delayed bone age, cone-shaped epiphyses, shortened long tubular bones, and ovoid vertebral bodies. Affected individuals have distinct facial features, including round face, well-defined eyebrows, long eyelashes, bulbous nose with anteverted nostrils, long and prominent philtrum, and thick lips with a small mouth. Other characteristic features include hoarse voice and pseudomuscular build, and there are distinct skeletal features as well, including an internal notch of the femoral head, internal notch of the second metacarpal, and external notch of the fifth metacarpal. {ECO:0000269|PubMed:21683323}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
STING İlişkili Vaskülopati, İnfantil Başlangıçlı, 615934, Otozomal Dominant; SAVI (STING İlişkili Vaskülopati, İnfantil Başlangıçlı) (TMEM173 geni) (Tüm Gen Dizi Analizi) (Postnatal)	STING-associated vasculopathy, infantile-onset, 615934, Autosomal dominant; SAVI (STING-associated vasculopathy with onset in infancy) (TMEM173 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TMEM173	TMEM173, STING, MPYS, SAVI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: STING-associated vasculopathy, infantile-onset (SAVI) [MIM:615934]: An autoinflammatory disease characterized by early- onset systemic inflammation and cutaneous vasculopathy, resulting in severe skin lesions. Violaceous, scaling lesions of fingers, toes, nose, cheeks and ears progress to acral necrosis in most of the patients. Some patients have severe interstitial lung disease. {ECO:0000269|PubMed:25029355}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Stocco Dos Santos Sendromu, Mental Retardasyon, X'e Bağlı 300434 (X'e Bağlı Mental Retardasyon, Stocco Dos Santos Tipi) (SHROOM4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stocco dos Santos X-linked mental retardation syndrome, 300434 (X-linked intellectual disability, Stocco Dos Santos type) (SHROOM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SHROOM4	SHROOM4, KIAA1202, SDSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving SHROOM4 is a cause of X-linked mental retardation (XLMR). Translocation t(X;8)(p11.22;p23.3) with FBXO25.	EDTA Blood Sample
Stocco Dos Santos Sendromu, Mental Retardasyon, X'e Bağlı 300434 (X'e Bağlı Mental Retardasyon, Stocco Dos Santos Tipi) (Prenatal) (SHROOM4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stocco dos Santos X-linked mental retardation syndrome, 300434 (X-linked intellectual disability, Stocco Dos Santos type) (SHROOM4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SHROOM4	SHROOM4, KIAA1202, SDSX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving SHROOM4 is a cause of X-linked mental retardation (XLMR). Translocation t(X;8)(p11.22;p23.3) with FBXO25.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Stomatin Eksikliği Olan Kryohidrositozis, Nörolojik Defektler, 608885, Otozomal Dominant; SDCHCN (Kalıtsal Kriyohidrositozis-Azalmış Stomatin) (SLC2A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stomatin-deficient cryohydrocytosis with neurologic defects, 608885, Autosomal dominant; SDCHCN (Hereditary cryohydrocytosis with reduced stomatin) (SLC2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269|PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Stomatin Eksikliği Olan Kryohidrositozis, Nörolojik Defektler, 608885, Otozomal Dominant; SDCHCN (Kalıtsal Kriyohidrositozis-Azalmış Stomatin) (MLPA)	Stomatin-deficient cryohydrocytosis with neurologic defects, 608885, Autosomal dominant; SDCHCN (Hereditary cryohydrocytosis with reduced stomatin) (MLPA)	SLC2A1	SLC2A1, GLUT1, DYT18, PED, GLUT1DS, EIG12, DYT9, SDCHCN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269|PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.,	EDTA Blood Sample
Stormorken Sendromu, 185070, Otozomal Dominant; STRMK (Stormorken-Sjaastad-Langslet Sendromu) (STIM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stormorken syndrome, 185070, Autosomal dominant; STRMK (Stormorken-Sjaastad-Langslet syndrome) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. {ECO:0000269|PubMed:23332920}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Stormorken Sendromu, 185070, Otozomal Dominant; STRMK (Stormorken-Sjaastad-Langslet Sendromu) (Prenatal) (STIM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stormorken syndrome, 185070, Autosomal dominant; STRMK (Stormorken-Sjaastad-Langslet syndrome) (STIM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STIM1	STIM1, TAM1, IMD10, STRMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Myopathy, tubular aggregate, 1 (TAM1) [MIM:160565]: A rare congenital myopathy characterized by regular arrays of membrane tubules on muscle biopsies without additional histopathological hallmarks. Tubular aggregates in muscle are structures of variable appearance consisting of an outer tubule containing either one or more microtubule-like structures or amorphous material. They may occur in a variety of circumstances, including inherited myopathies, alcohol- and drug-induced myopathies, exercise-induced cramps or muscle weakness. {ECO:0000269	PubMed:23332920}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Striatal Dejenerasyon, Otozomal Dominant, 609161, Otozomal Dominant (Otozomal Dominant Striatal Nörodejenerasyon) (PDE8B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Striatal degeneration, autosomal dominant, 609161, Autosomal dominant (Autosomal dominant striatal neurodegeneration) (PDE8B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE8B	PDE8B, PPNAD3, ADSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Striatal degeneration autosomal dominant (ADSD) [MIM:609161]: A movement disorder affecting the striatal part of the basal ganglia and characterized by bradykinesia, dysarthria and muscle rigidity. These symptoms resemble idiopathic Parkinson disease, but tremor is not present. {ECO:0000269	PubMed:20085714}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Striatal Dejenerasyon, Otozomal Dominant, 609161, Otozomal Dominant (Otozomal Dominant Striatal Nörodejenerasyon) (Prenatal) (PDE8B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Striatal degeneration, autosomal dominant, 609161, Autosomal dominant (Autosomal dominant striatal neurodegeneration) (PDE8B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDE8B	PDE8B, PPNAD3, ADSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Striatal degeneration autosomal dominant (ADSD) [MIM:609161]: A movement disorder affecting the striatal part of the basal ganglia and characterized by bradykinesia, dysarthria and muscle rigidity. These symptoms resemble idiopathic Parkinson disease, but tremor is not present. {ECO:0000269	PubMed:20085714}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Striatal Dejenerasyon, Otozomal Dominant, 616922, Otozomal Dominant (PDE10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Striatal degeneration, autosomal dominant, 616922, Autosomal dominant (PDE10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE10A	PDE10A, IOLOD, ADSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Striatal Dejenerasyon, Otozomal Dominant, 616922, Otozomal Dominant (Prenatal) (PDE10A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Striatal degeneration, autosomal dominant, 616922, Autosomal dominant (PDE10A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PDE10A	PDE10A, IOLOD, ADSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Striatonigral Dejenerasyon, Çocuklukta Başlayan, 617054, Otozomal Resesif; SNDC (VAC14 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Striatonigral degeneration, childhood-onset, 617054, Autosomal recessive; SNDC (VAC14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VAC14	VAC14, TAX1BP2, TRX, SNDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Striatonigral Dejenerasyon, Çocuklukta Başlayan, 617054, Otozomal Resesif; SNDC (Prenatal) (VAC14 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Striatonigral degeneration, childhood-onset, 617054, Autosomal recessive; SNDC (VAC14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VAC14	VAC14, TAX1BP2, TRX, SNDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Striatonigral Dejenerasyon, İnfantil, 271930, Otozomal Resesif (Ailesel İnfantil Bilateral Striatal Nekroz) (NUP62 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Striatonigral degeneration, infantile, 271930, Autosomal recessive (Familial infantile bilateral striatal necrosis) (NUP62 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUP62	NUP62, SNDI, IBSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Infantile striatonigral degeneration (SNDI) [MIM:271930]: Neurological disorder characterized by symmetrical degeneration of the caudate nucleus, putamen, and occasionally the globus pallidus, with little involvement of the rest of the brain. The clinical features include developmental regression, choreoathetosis, dystonia, spasticity, dysphagia, failure to thrive, nystagmus, optic atrophy, and mental retardation. {ECO:0000269	PubMed:16786527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Striatonigral Dejenerasyon, İnfantil, 271930, Otozomal Resesif (Ailesel İnfantil Bilateral Striatal Nekroz) (Prenatal) (NUP62 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Striatonigral degeneration, infantile, 271930, Autosomal recessive (Familial infantile bilateral striatal necrosis) (NUP62 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NUP62	NUP62, SNDI, IBSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Infantile striatonigral degeneration (SNDI) [MIM:271930]: Neurological disorder characterized by symmetrical degeneration of the caudate nucleus, putamen, and occasionally the globus pallidus, with little involvement of the rest of the brain. The clinical features include developmental regression, choreoathetosis, dystonia, spasticity, dysphagia, failure to thrive, nystagmus, optic atrophy, and mental retardation. {ECO:0000269	PubMed:16786527}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Strok, Hemorajik, 614519 (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, hemorrhagic, 614519 (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Renal tubular dysgenesis (RTD) [MIM:267430]: Autosomal recessive severe disorder of renal tubular development characterized by persistent fetal anuria and perinatal death, probably due to pulmonary hypoplasia from early-onset oligohydramnios (the Potter phenotype). {ECO:0000269	PubMed:16116425}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Strok, İskemik, 614519 (ACE geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, ischemic, 614519 (ACE gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACE	ACE, DCP1, ACE1, MVCD3, ICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to activated protein C resistance (THPHC) [MIM:188055]: A hemostatic disorder due to defective degradation of factor V by activated protein C. It is characterized by a poor anticoagulant response to activated protein C resulting in tendency to thrombosis. {ECO:0000269|PubMed:11435304, ECO:0000269|PubMed:11858490, ECO:0000269|PubMed:14617013, ECO:0000269|PubMed:9454742}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Strok, İskemik, Yatkınlık, 601367, Multifaktöryel (FS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, ischemic, susceptibility to, 601367, Multifactorial (FS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FS	FS, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to thrombin defect (THPH1) [MIM:188050]: A multifactorial disorder of hemostasis characterized by abnormal platelet aggregation in response to various agents and recurrent thrombi formation. {ECO:0000269	PubMed:2825773}. Note=The disease is caused by mutations affecting the gene represented in this entry. A common genetic variation in the 3-prime untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increased risk of venous thrombosis.	EDTA Blood Sample
Strok, İskemik, Yatkınlık, 601367, Multifaktöryel (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, ischemic, susceptibility to, 601367, Multifactorial (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Acrodysostosis 2, with or without hormone resistance (ACRDYS2) [MIM:614613]: A pleiotropic disorder characterized by skeletal, endocrine, and neurological abnormalities. Skeletal features include brachycephaly, midface hypoplasia with a small upturned nose, brachydactyly, and lumbar spinal stenosis. Endocrine abnormalities include hypothyroidism and hypogonadism in males and irregular menses in females. Developmental disability is a common finding but is variable in severity and can be associated with significant behavioral problems. {ECO:0000269|PubMed:22464250, ECO:0000269|PubMed:22464252, ECO:0000269|PubMed:23033274, ECO:0000269|PubMed:23043190}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
İnme, Yatkınlık 1, 606799 (Akrodisostozis) (PDE4D geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, susceptibility to, 1, 606799 (Acrodysostosis) (PDE4D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDE4D	PDE4D, DPDE3, STRK1, ACRDYS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ischemic stroke (ISCHSTR) [MIM:601367]: A stroke is an acute neurologic event leading to death of neural tissue of the brain and resulting in loss of motor, sensory and/or cognitive function. Ischemic strokes, resulting from vascular occlusion, is considered to be a highly complex disease consisting of a group of heterogeneous disorders with multiple genetic and environmental risk factors. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
İnme, Yatkınlık, 601367, Multifaktöryel (ALOXSAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stroke, susceptibility to, 601367, Multifactorial (ALOXSAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALOXSAP	ALOXSAP, FLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 31 (CILD31) [MIM:616369]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269	PubMed:25564561}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Stromme Sendromu, 243605, Otozomal Resesif; STROMS (Letal Fetal Beyin Malformasyonu-Duodenal Atrezi-Bilateral Renal Hipoplazi Sendromu (Stromme Sendromu)) (CENPF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stromme syndrome, 243605, Autosomal recessive; STROMS (Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome) (CENPF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CENPF	CENPF, CILD31, STROMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ciliary dyskinesia, primary, 31 (CILD31) [MIM:616369]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269	PubMed:25564561}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Stromme Sendromu, 243605, Otozomal Resesif; STROMS (Letal Fetal Beyin Malformasyonu-Duodenal Atrezi-Bilateral Renal Hipoplazi Sendromu (Stromme Sendromu)) (Prenatal) (CENPF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stromme syndrome, 243605, Autosomal recessive; STROMS (Lethal fetal brain malformation-duodenal atresia-bilateral renal hypoplasia syndrome) (CENPF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CENPF	CENPF, CILD31, STROMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ciliary dyskinesia, primary, 31 (CILD31) [MIM:616369]: A disorder characterized by abnormalities of motile cilia. Respiratory infections leading to chronic inflammation and bronchiectasis are recurrent, due to defects in the respiratory cilia. Patients may exhibit randomization of left-right body asymmetry and situs inversus, due to dysfunction of monocilia at the embryonic node. Primary ciliary dyskinesia associated with situs inversus is referred to as Kartagener syndrome. {ECO:0000269	PubMed:25564561}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sturge Weber Sendromu, Somatik, Mozaik, 185300; SWS (Sturge-Weber Sendromu) (GNAQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sturge-Weber syndrome, somatic, mosaic, 185300; SWS (Sturge-Weber syndrome) (GNAQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAQ	GNAQ, SWS, CMC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Capillary malformations, congenital (CMC) [MIM:163000]: A form of vascular malformations that are present from birth, tend to grow with the individual, do not regress spontaneously, and show normal rates of endothelial cell turnover. Capillary malformations are distinct from capillary hemangiomas, which are highly proliferative lesions that appear shortly after birth and show rapid growth, slow involution, and endothelial hypercellularity. {ECO:0000269	PubMed:23656586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Biopsy of affected site-transport media
Kekemelik, Ailesel Dirençli, 1, 184450, Otozomal Dominant; STUT1 (Ağır Mental Retardasyon ve Progresif Spastik Paraplejiyle) (AP4E1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stuttering, familial persistent, 1, 184450, Autosomal dominant; STUT1 (Severe intellectual disability and progressive spastic paraplegia) (AP4E1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AP4E1	AP4E1, SPG51, CPSQ4, STUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Stuve-Wiedemann Sendromu / Schwartz-Jampel Tip 2 Sendromu, 601559, Otozomal Resesif (Stüve-Wiedemann Sendromu) (LIFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559, Autosomal recessive (Stüve-Wiedemann syndrome) (LIFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIFR	LIFR, STWS, SWS, SJS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stueve-Wiedemann syndrome (STWS) [MIM:601559]: Severe autosomal recessive condition and belongs to the group of the bent-bone dysplasias. SWS is characterized by bowing of the lower limbs, with internal cortical thickening, wide metaphyses with abnormal trabecular pattern, and camptodactyly. Additional features include feeding and swallowing difficulties, as well as respiratory distress and hyperthermic episodes, which cause death in the first months of life. The rare survivors develop progressive scoliosis, spontaneous fractures, bowing of the lower limbs, with prominent joints and dysautonomia symptoms, including temperature instability, absent corneal and patellar reflexes, and smooth tongue. {ECO:0000269	PubMed:14740318}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Stuve-Wiedemann Sendromu / Schwartz-Jampel Tip 2 Sendromu, 601559, Otozomal Resesif (Stüve-Wiedemann Sendromu) (Prenatal) (LIFR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559, Autosomal recessive (Stüve-Wiedemann syndrome) (LIFR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIFR	LIFR, STWS, SWS, SJS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Stueve-Wiedemann syndrome (STWS) [MIM:601559]: Severe autosomal recessive condition and belongs to the group of the bent-bone dysplasias. SWS is characterized by bowing of the lower limbs, with internal cortical thickening, wide metaphyses with abnormal trabecular pattern, and camptodactyly. Additional features include feeding and swallowing difficulties, as well as respiratory distress and hyperthermic episodes, which cause death in the first months of life. The rare survivors develop progressive scoliosis, spontaneous fractures, bowing of the lower limbs, with prominent joints and dysautonomia symptoms, including temperature instability, absent corneal and patellar reflexes, and smooth tongue. {ECO:0000269	PubMed:14740318}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Subkortikal Laminal Heteropia, X'e Bağlı, 300067, X'e Bağlı (Subkortikal Bant Heterotopisi) (DCX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Subcortical laminal heteropia, X-linked, 300067, X-linked (Subcortical band heterotopia) (DCX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCX	DCX, DBCN, LISX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Subkortikal Laminal Heteropia, X'e Bağlı, 300067, X'e Bağlı (Subkortikal Bant Heterotopisi) (MLPA)	Subcortical laminal heteropia, X-linked, 300067, X-linked (Subcortical band heterotopia) (MLPA)	DCX	DCX, DBCN, LISX	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Subkortikal Laminal Heteropia, X'e Bağlı, 300067, X'e Bağlı (Subkortikal Bant Heterotopisi) (Prenatal) (DCX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Subcortical laminal heteropia, X-linked, 300067, X-linked (Subcortical band heterotopia) (DCX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCX	DCX, DBCN, LISX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Subkortikal Laminal Heteropia, X'e Bağlı, 300067, X'e Bağlı (Subkortikal Bant Heterotopisi) (Prenatal) (MLPA)	Subcortical laminal heteropia, X-linked, 300067, X-linked (Subcortical band heterotopia) (Prenatal) (MLPA)	DCX	DCX, DBCN, LISX	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lissencephaly, X-linked 1 (LISX1) [MIM:300067]: A classic lissencephaly characterized by mental retardation and seizures that are more severe in male patients. Affected boys show an abnormally thick cortex with absent or severely reduced gyri. Clinical manifestations include feeding problems, abnormal muscular tone, seizures and severe to profound psychomotor retardation. Female patients display a less severe phenotype referred to as 'doublecortex'. {ECO:0000269|PubMed:11468322, ECO:0000269|PubMed:12552055, ECO:0000269|PubMed:9489699, ECO:0000269|PubMed:9489700, ECO:0000269|PubMed:9668176, ECO:0000269|PubMed:9817918}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Subkortikal Laminer Heterotopi, 607432, Sporadik Vakalar (Subkortikal Bant Heterotopisi) (PAFAH1B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Subcortical laminar heterotopia, 607432, Isolated cases (Subcortical band heterotopia) (PAFAH1B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAFAH1B1	PAFAH1B1, LIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Miller-Dieker lissencephaly syndrome (MDLS) [MIM:247200]: A contiguous gene deletion syndrome of chromosome 17p13.3, characterized by classical lissencephaly and distinct facial features. Additional congenital malformations can be part of the condition. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Subkortikal Laminer Heterotopi, 607432, Sporadik Vakalar (Subkortikal Bant Heterotopisi) (Prenatal) (PAFAH1B1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Subcortical laminar heterotopia, 607432, Isolated cases (Subcortical band heterotopia) (PAFAH1B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAFAH1B1	PAFAH1B1, LIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Miller-Dieker lissencephaly syndrome (MDLS) [MIM:247200]: A contiguous gene deletion syndrome of chromosome 17p13.3, characterized by classical lissencephaly and distinct facial features. Additional congenital malformations can be part of the condition. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sublingual Nitrogliserin, Kötü Yanıt Yatkınlık (ALDH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sublingual nitroglycerin, susceptibility to poor response to (ALDH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH2	ALDH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Subtelomerik Delesyon Analizi (FISH)	Subtelomeric deletion analysis (FISH)			FISH	Heparinli Kan (2-4 ml)		10	30			20	Postnatal		Heparinized blood sample
Süksinik Semialdehid Dehidrogenaz Eksikliği, 271980, Otozomal Resesif, SSADHD (Süksinik Semialdehid Dehidrogenaz Eksikliği) (ALDH5A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Succinic semialdehyde dehydrogenase deficiency, 271980, Autosomal recessive; SSADHD (Succinic semialdehyde dehydrogenase deficiency) (ALDH5A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ALDH5A1	ALDH5A1, SSADH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Süksinik Semialdehid Dehidrogenaz Eksikliği, 271980, Otozomal Resesif, SSADHD (Süksinik Semialdehid Dehidrogenaz Eksikliği) (Prenatal) (ALDH5A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Succinic semialdehyde dehydrogenase deficiency, 271980, Autosomal recessive; SSADHD (Succinic semialdehyde dehydrogenase deficiency) (ALDH5A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ALDH5A1	ALDH5A1, SSADH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Süksinil-CoA 3-Oksoasit-CoA Transferaz Eksikliği, 245050; SCOTD (SCOT Eksikliği) (Süksinil-CoA 3-Ketoasit-CoA Transferaz Eksikliği) (OXCT1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050; SCOTD (SCOT deficiency) (Succinyl-CoA:3-ketoacid CoA transferase deficiency) (OXCT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OXCT1	OXCT1, OXCT, SCOT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Süksinil-CoA 3-Oksoasit-CoA Transferaz Eksikliği, 245050; SCOTD (SCOT Eksikliği) (Süksinil-CoA 3-Ketoasit-CoA Transferaz Eksikliği) (Prenatal) (OXCT1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Succinyl CoA:3-oxoacid CoA transferase deficiency, 245050; SCOTD (SCOT deficiency) (Succinyl-CoA:3-ketoacid CoA transferase deficiency) (OXCT1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	OXCT1	OXCT1, OXCT, SCOT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sükroz-İzomaltaz Eksikliği, Konjenital, 222900, Otozomal Resesif; CSID (Konjenital Sukroz-İzomaltaz Eksikliği) (SI geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sucrase-isomaltase deficiency, congenital, 222900, Autosomal recessive; CSID (Congenital sucrase-isomaltase deficiency) (SI gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SI	SI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Congenital sucrase-isomaltase deficiency (CSID) [MIM:222900]: Autosomal recessive intestinal disorder that is clinically characterized by fermentative diarrhea, abdominal pain, and cramps upon ingestion of sugar. The symptoms are the consequence of absent or drastically reduced enzymatic activities of sucrase and isomaltase. The prevalence of CSID is 0.02 % in individuals of European descent and appears to be much higher in Greenland, Alaskan, and Canadian native people. CSID arises due to post-translational perturbations in the intracellular transport, polarized sorting, aberrant processing, and defective function of SI. {ECO:0000269|PubMed:10903344, ECO:0000269|PubMed:11340066, ECO:0000269|PubMed:14724820, ECO:0000269|PubMed:16329100, ECO:0000269|PubMed:8609217}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sükroz-İzomaltaz Eksikliği, Konjenital, 222900, Otozomal Resesif; CSID (Konjenital Sukroz-İzomaltaz Eksikliği) (Prenatal) (SI geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sucrase-isomaltase deficiency, congenital, 222900, Autosomal recessive; CSID (Congenital sucrase-isomaltase deficiency) (SI gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SI	SI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Congenital sucrase-isomaltase deficiency (CSID) [MIM:222900]: Autosomal recessive intestinal disorder that is clinically characterized by fermentative diarrhea, abdominal pain, and cramps upon ingestion of sugar. The symptoms are the consequence of absent or drastically reduced enzymatic activities of sucrase and isomaltase. The prevalence of CSID is 0.02 % in individuals of European descent and appears to be much higher in Greenland, Alaskan, and Canadian native people. CSID arises due to post-translational perturbations in the intracellular transport, polarized sorting, aberrant processing, and defective function of SI. {ECO:0000269|PubMed:10903344, ECO:0000269|PubMed:11340066, ECO:0000269|PubMed:14724820, ECO:0000269|PubMed:16329100, ECO:0000269|PubMed:8609217}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ani Kalp Yetmezliği, Alkole Bağlı, 617223, Otozomal Resesif; SCFAI (PPA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sudden cardiac failure, alcohol-induced, 617223, Autosomal recessive; SCFAI (PPA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPA2	PPA2, SCFI, SCFAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ani Kalp Yetmezliği, İnfantil, 617222, Otozomal Resesif; SCFI (PPA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sudden cardiac failure, infantile, 617222, Autosomal recessive; SCFI (PPA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PPA2	PPA2, SCFI, SCFAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ani Kalp Yetmezliği, İnfantil, 617222, Otozomal Resesif; SCFI (Prenatal) (PPA2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sudden cardiac failure, infantile, 617222, Autosomal recessive; SCFI (PPA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PPA2	PPA2, SCFI, SCFAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ani Bebek Ölümü, Yatkınlık, 272120, Otozomal Resesif (Brugada Sendromu) (SCN5A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sudden infant death syndrome, susceptibility to, 272120, Autosomal recessive (Brugada syndrome) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Atrial standstill 1 (ATRST1) [MIM:108770]: A rare arrhythmia characterized by the absence of electrical and mechanical activity in the atria. Electrocardiographically, it is characterized by bradycardia, the absence of P waves, and a junctional narrow complex escape rhythm. {ECO:0000269	PubMed:12522116}. Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry. A mutation in SCN5A has been detected in combination with a rare GJA5 genotype in a large family with atrial standstill.	EDTA Blood Sample

Ani Bebek Ölümü, Yatkınlık, 272120, Otozomal Resesif (Brugada Sendromu) (Prenatal) (SCN5A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sudden infant death syndrome, susceptibility to, 272120, Autosomal recessive (Brugada syndrome) (SCN5A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCN5A	SCN5A, LQT3, VF1, HB1, SSS1, CMD1E, CDCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Atrial standstill 1 (ATRST1) [MIM:108770]: A rare arrhythmia characterized by the absence of electrical and mechanical activity in the atria. Electrocardiographically, it is characterized by bradycardia, the absence of P waves, and a junctional narrow complex escape rhythm. (ECO:0000269	PubMed:12522116). Note=The disease may be caused by mutations affecting distinct genetic loci, including the gene represented in this entry. A mutation in SCN5A has been detected in combination with a rare GJA5 genotype in a large family with atrial standstill.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ani Çocuk Ölümü, Testis Disgenezisi Sendromu, 608800, Otozomal Resesif; SIDDT (Ani Bebek Ölümü-Testis Disgenezisi Sendromu) (TSPYL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sudden infant death with dysgenesis of the testes syndrome, 608800, Autosomal recessive; SIDDT (Sudden infant death-dysgenesis of the testes syndrome) (TSPYL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSPYL1	TSPYL1, TSPYL, SIDDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ani Çocuk Ölümü, Testis Disgenezisi Sendromu, 608800, Otozomal Resesif; SIDDT (Ani Bebek Ölümü-Testis Disgenezisi Sendromu) (Prenatal) (TSPYL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sudden infant death with dysgenesis of the testes syndrome, 608800, Autosomal recessive; SIDDT (Sudden infant death-dysgenesis of the testes syndrome) (TSPYL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSPYL1	TSPYL1, TSPYL, SIDDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sülfit Oksidaz Eksikliği, 272300, Otozomal Resesif (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (SUOX geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sulfite oxidase deficiency, 272300, Autosomal recessive (Encephalopathy due to sulfite oxidase deficiency) (SUOX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SUOX	SUOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Isolated sulfite oxidase deficiency (ISOD) [MIM:272300]: Characterized by neurological abnormalities including multicystic leukoencephalopathy with brain atrophy. Patients often suffer from seizures. Often leads to death at an early age. (ECO:0000269|PubMed:10519592, ECO:0000269|PubMed:12112661, ECO:0000269|PubMed:12368985, ECO:0000269|PubMed:9428520, ECO:0000269|PubMed:9600976). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sülfit Oksidaz Eksikliği, 272300, Otozomal Resesif (Ensefalopati, Sülfit Oksidaz Eksikliğine Bağlı) (Prenatal) (SUOX geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sulfite oxidase deficiency, 272300, Autosomal recessive (Encephalopathy due to sulfite oxidase deficiency) (SUOX gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SUOX	SUOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Isolated sulfite oxidase deficiency (ISOD) [MIM:272300]: Characterized by neurological abnormalities including multicystic leukoencephalopathy with brain atrophy. Patients often suffer from seizures. Often leads to death at an early age. (ECO:0000269|PubMed:10519592, ECO:0000269|PubMed:12112661, ECO:0000269|PubMed:12368985, ECO:0000269|PubMed:9428520, ECO:0000269|PubMed:9600976). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Supranükleer Palsi, Progresif Atipik, 260540, Otozomal Resesif (Progresif Supranükleer Palsi) (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Supranuclear palsy, progressive atypical, 260540, Autosomal recessive (Progressive supranuclear palsy) (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. (ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Supranükleer Palsi, Progresif Atipik, 260540, Otozomal Resesif (Progresif Supranükleer Palsi) (MLPA)	Supranuclear palsy, progressive atypical, 260540, Autosomal recessive (Progressive supranuclear palsy) (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. (ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Supranükleer Palsi, Progresif, 601104, Otozomal Dominant; PSNP1 (Progresif Supranükleer Palsi) (MAPT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Supranuclear palsy, progressive, 601104, Autosomal dominant; PSNP1 (Progressive supranuclear palsy) (MAPT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MAPT	MAPT, MTBT1, DDPAC, MSTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. (ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Supranükleer Palsi, Progresif, 601104, Otozomal Dominant; PSNP1 (Progresif Supranükleer Palsi) (MLPA)	Supranuclear palsy, progressive, 601104, Autosomal dominant; PSNP1 (Progressive supranuclear palsy) (MLPA)	MAPT	MAPT, MTBT1, DDPAC, MSTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pick disease of the brain (PIDB) [MIM:172700]: A rare form of dementia pathologically defined by severe atrophy, neuronal loss and gliosis. It is characterized by the occurrence of tau-positive inclusions, swollen neurons (Pick cells) and argentophilic neuronal inclusions known as Pick bodies that disproportionally affect the frontal and temporal cortical regions. Clinical features include aphasia, apraxia, confusion, anomia, memory loss and personality deterioration. (ECO:0000269|PubMed:10604746, ECO:0000269|PubMed:11089577, ECO:0000269|PubMed:11117542, ECO:0000269|PubMed:11601501, ECO:0000269|PubMed:11891833). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Supravalvar Aort Stenozu, 185500, Otozomal Dominant; SVAS (Supravalvüler Aort Darlığı) (ELN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Supravalvar aortic stenosis, 185500, Autosomal dominant; SVAS (Supravalvular aortic stenosis) (ELN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELN	ELN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cutis laxa, autosomal dominant, 1 (ADCL1) [MIM:123700]: A connective tissue disorder characterized by loose, hyperextensible skin with decreased resilience and elasticity leading to a premature aged appearance. Face, hands, feet, joints, and torso may be differentially affected. Additional variable clinical features are gastrointestinal diverticula, hernia, and genital prolapse. Rare manifestations are pulmonary artery stenosis, aortic aneurysm, bronchiectasis, and emphysema. (ECO:0000269	PubMed:9873040). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 1, 265120, Otozomal Resesif; SMDP1 (Neonatal Akut Solunum Sıkıntısı, SP-B Eksikliğine Bağlı) (SFTPB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Surfactant metabolism dysfunction, pulmonary, 1, 265120, Autosomal recessive; SMDP1 (Neonatal acute respiratory distress due to SP-B deficiency) (SFTPB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFTPB	SFTPB, SFTB3, SMDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary surfactant metabolism dysfunction 1 (SMDP1) [MIM:265120]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocculear material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. (ECO:0000269	PubMed:7491219). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 1, 265120, Otozomal Resesif; SMDP1 (Neonatal Akut Solunum Sıkıntısı, SP-B Eksikliğine Bağlı) (Prenatal) (SFTPB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Surfactant metabolism dysfunction, pulmonary, 1, 265120, Autosomal recessive; SMDP1 (Neonatal acute respiratory distress due to SP-B deficiency) (SFTPB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SFTPB	SFTPB, SFTB3, SMDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary surfactant metabolism dysfunction 1 (SMDP1) [MIM:265120]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocculear material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. (ECO:0000269	PubMed:7491219). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 2, 610913, Otozomal Dominant; SMDP2 (Kronik Solunum Güçlüğü-Surfaktan Metabolizması Eksikliği) (SFTPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Surfactant metabolism dysfunction, pulmonary, 2, 610913, Autosomal dominant; SMDP2 (Chronic respiratory distress with surfactant metabolism deficiency) (SFTPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SFTPC	SFTPC, SFTP2, SMDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Respiratory distress syndrome in premature infants (RDS) [MIM:267450]: A lung disease affecting usually premature newborn infants. It is characterized by deficient gas exchange, diffuse atelectasis, high-permeability lung edema and fibrin-rich alveolar deposits called 'hyaline membranes'. (ECO:0000269	PubMed:14735158). Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 2, 610913, Otozomal Dominant; SMDP2 (Kronik Solunum Güçlüğü-Surfaktan Metabolizması Eksikliği) (Prenatal) (SFTPC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Surfactant metabolism dysfunction, pulmonary, 2, 610913, Autosomal dominant; SMDP2 (Chronic respiratory distress with surfactant metabolism deficiency) (SFTPC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SFTPC	SFTPC, SFTP2, SMDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Respiratory distress syndrome in premature infants (RDS) [MIM:267450]: A lung disease affecting usually premature newborn infants. It is characterized by deficient gas exchange, diffuse atelectasis, high-permeability lung edema and fibrin-rich alveolar deposits called 'hyaline membranes'. (ECO:0000269	PubMed:14735158). Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 3, 610921, Otozomal Resesif; SMDP3 (Interstisyel Akciğer Hastalığı , ABCA3 Eksikliğine Bağlı) (ABCA3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Surfactant metabolism dysfunction, pulmonary, 3, 610921, Autosomal recessive; SMDP3 (Interstitial lung disease due to ABCA3 deficiency) (ABCA3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA3	ABCA3, ABC3, SMDP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary surfactant metabolism dysfunction 3 (SMDP3) [MIM:610921]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocculear material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. (ECO:0000269	PubMed:15044640). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 3, 610921, Otozomal Resesif; SMDP3 (Interstisyel Akciğer Hastalığı , ABCA3 Eksikliğine Bağlı) (Prenatal) (ABCA3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Surfactant metabolism dysfunction, pulmonary, 3, 610921, Autosomal recessive; SMDP3 (Interstitial lung disease due to ABCA3 deficiency) (ABCA3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCA3	ABCA3, ABC3, SMDP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary surfactant metabolism dysfunction 3 (SMDP3) [MIM:610921]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocculear material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. (ECO:0000269	PubMed:15044640). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 4, 300770; SMDP4 (Kalıtsal Pulmoner Alveolar Proteinoz) (CSF2RA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Surfactant metabolism dysfunction, pulmonary, 4, 300770; SMDP4 (Hereditary pulmonary alveolar proteinosis) (CSF2RA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSF2RA	CSF2RA, SMDP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary surfactant metabolism dysfunction 4 (SMDP4) [MIM:300770]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocculear material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. (ECO:0000269|PubMed:18955567, ECO:0000269|PubMed:18955570). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 4, 300770; SMDP4 (Kalıtsal Pulmoner Alveolar Proteinoz) (Prenatal) (CSF2RA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Surfactant metabolism dysfunction, pulmonary, 4, 300770; SMDP4 (Hereditary pulmonary alveolar proteinosis) (CSF2RA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSF2RA	CSF2RA, SMDP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary surfactant metabolism dysfunction 4 (SMDP4) [MIM:300770]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocular material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. {ECO:0000269|PubMed:18955567, ECO:0000269|PubMed:18955570}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 5, 614370, Otozomal Resesif; SMDP5 (Kalıtsal Pulmoner Alveolar Proteinoz) (CSF2RB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Surfactant metabolism dysfunction, pulmonary, 5, 614370, Autosomal recessive; SMDP5 (Hereditary pulmonary alveolar proteinosis) (CSF2RB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CSF2RB	CSF2RB, SMDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary surfactant metabolism dysfunction 5 (SMDP5) [MIM:614370]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocular material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. {ECO:0000269|PubMed:21075760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sürfaktan Metabolizma Disfonksiyonu, Pulmoner, 5, 614370, Otozomal Resesif; SMDP5 (Kalıtsal Pulmoner Alveolar Proteinoz) (Prenatal) (CSF2RB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Surfactant metabolism dysfunction, pulmonary, 5, 614370, Autosomal recessive; SMDP5 (Hereditary pulmonary alveolar proteinosis) (CSF2RB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CSF2RB	CSF2RB, SMDP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pulmonary surfactant metabolism dysfunction 5 (SMDP5) [MIM:614370]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocular material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. {ECO:0000269|PubMed:21075760}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sveinsson Korioretinal Atrofi, 108985, Otozomal Dominant; SCRA (Helikoid Peripapiller Koryoretinal Dejenerasyon (Sveinsson Koryoretinal Atrofi)) (TEAD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sveinsson chorioretinal atrophy, 108985, Autosomal dominant; SCRA (Helicoid peripapillary chorioretinal degeneration) (TEAD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TEAD1	TEAD1, TCF13, REF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kistik Fibrozis Olmayan Ter Klor Yüksekliği (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Sweat chloride elevation without CF (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	EDTA Blood Sample
Kistik Fibrozis Olmayan Ter Klor Yüksekliği (MLPA)	Sweat chloride elevation without CF (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	EDTA Blood Sample
Kistik Fibrozis Olmayan Ter Klor Yüksekliği (Prenatal) (CFTR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Sweat chloride elevation without CF (CFTR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kistik Fibrozis Olmayan Ter Klor Yüksekliği (Prenatal) (MLPA)	Sweat chloride elevation without CF (Prenatal) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. {ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Konjenital Simetrik Dairesel Deri Kıvrıklıkları, 1, 156610, Otozomal Dominant; CSCSC1 (Multipl Benign Çembersel Eksitremite Deri Kattanları 2) (TUBB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Symmetric circumferential skin creases, congenital, 1, 156610, Autosomal dominant; CSCSC1 (Multiple benign circumferential skin creases on limbs) (TUBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TUBB	TUBB, TUBB5, M40, CDCBM6, CSCSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 6 (CDCBM6) [MIM:615771]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Affected individuals have microcephaly, ataxia, and severe delayed psychomotor development. Brain imaging shows variable malformations of cortical development, including white matter streaks, dysmorphic basal ganglia, corpus callosum abnormalities, brainstem and cerebellar hypoplasia, cortical dysplasia, polymicrogyria. {ECO:0000269|PubMed:23246003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Konjenital Simetrik Dairesel Deri Kırışıklıkları, 1, 156610, Otozomal Dominant; CSCSC1 (Multipl Benign Çemberseel Ekstremite Deri Kattantları 2) (Prenatal) (TUBB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Symmetric circumferential skin creases, congenital, 1, 156610, Autosomal dominant; CSCSC1 (Multiple benign circumferential skin creases on limbs) (TUBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TUBB	TUBB, TUBB5, M40, CDCBM6, CSCSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Cortical dysplasia, complex, with other brain malformations 6 (CDCBM6) [MIM:615771]: A disorder of aberrant neuronal migration and disturbed axonal guidance. Affected individuals have microcephaly, ataxia, and severe delayed psychomotor development. Brain imaging shows variable malformations of cortical development, including white matter streaks, dysmorphic basal ganglia, corpus callosum abnormalities, brainstem and cerebellar hypoplasia, cortical dysplasia, polymicrogyria. {ECO:0000269	PubMed:23346003}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Simetrik Çevresel Deri Kırışıklıkları, Konjenital, 2, 616734, Otozomal Dominant (Multipl Benign Çemberseel Ekstremite Deri Kattantları 2) (MAPRE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Symmetric circumferential skin creases, congenital, 2, 616734, Autosomal dominant (Multiple benign circumferential skin creases on limbs) (MAPRE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAPRE2	MAPRE2, EB2, RP1, CSCSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Simetrik Çevresel Deri Kırışıklıkları, Konjenital, 2, 616734, Otozomal Dominant (Multipl Benign Çemberseel Ekstremite Deri Kattantları 2) (Prenatal) (MAPRE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Symmetric circumferential skin creases, congenital, 2, 616734, Autosomal dominant (Multiple benign circumferential skin creases on limbs) (MAPRE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MAPRE2	MAPRE2, EB2, RP1, CSCSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Sinfalanjizm, Proksimal, 1A, 185800, Otozomal Dominant; SYM1A (Proksimal Sinfalanjizmi) (NOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Symphalangism, proximal, 1A, 185800, Autosomal dominant; SYM1A (Proximal symphalangism) (NOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOG	NOG, SYM1, SYNS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stapes ankylosis with broad thumb and toes (SABTS) [MIM:184460]: Congenital autosomal dominant disorder that includes hypermobile, a hemicylindrical nose, broad thumbs, great toes, and other minor skeletal anomalies but lacked carpal and tarsal fusion and symphalangism. {ECO:0000269	PubMed:12089654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sinfalanjizm, Proksimal, 1B, 615298; SYM1B (Proksimal Sinfalanjizmi) (GDF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Symphalangism, proximal, 1B, 615298; SYM1B (Proximal symphalangism) (GDF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF5	GDF5, CDMP1, SYNS2, OS5, BDA1C, SYM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly A2 (BDA2) [MIM:112600]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. In brachydactyly type A2 shortening of the middle phalanges is confined to the index finger and the second toe, all other digits being more or less normal. Because of a rhomboid or triangular shape of the affected middle phalanx, the end of the second finger usually deviates radially. {ECO:0000269|PubMed:16127465, ECO:0000269|PubMed:18203755, ECO:0000269|PubMed:21976273}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sindaktili, Mesoaksiyel Sinostotik, Falangeal Redüksiyon, 609432, Otozomal Resesif; MSSD (Mezoaksiyel Sinostotik Sindaktili-Falangeal Redüksiyon) (BHLHA9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Syndactyly, mesoaxial synostotic, with phalangeal reduction, 609432, Autosomal recessive; MSSD (Mesoaxial synostotic syndactyly with phalangeal reduction) (BHLHA9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BHLHA9	BHLHA9, BHLHF42, MSSD, CCSPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Syndactyly, mesoaxial synostotic, with phalangeal reduction (MSSD) [MIM:609432]: An autosomal recessive, non-syndromic digit anomaly characterized by mesoaxial osseous synostosis at a metacarpal level, reduction of one or more phalanges, hypoplasia of distal phalanges of preaxial and postaxial digits, clinodactyly of fifth fingers, and preaxial fusion of toes. {ECO:0000269	PubMed:25466284}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sindaktili, Tip 1, ± Kraniosinostozis 185900, Otozomal Dominant (Kranyosinostozis, Philadelphia Tipi) (2200)	Syndactyly, type 1, with or without craniosynostosis, 185900, Autosomal dominant (Craniosynostosis, Philadelphia type) (440)	.	DUP2q35, C2DUPq35, SDTY1, SD1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Sindaktili, Tip 3 (Sindaktili Tip 3) (GJA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	SYNDACTYLY, TYPE III (Syndactyly type 3) (GJA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJA1	GJA1, CX43, ODDD, SDTY3, ODOD, HSS, AVSD3, HLHS1, CMDR, EKVP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Oculodentodigital dysplasia (ODDD) [MIM:164200]: A disease characterized by a typical facial appearance and variable involvement of the eyes, dentition, and fingers. Characteristic facial features include a narrow, pinched nose with hypoplastic alae nasi, prominent columella and thin anteverted nares together with a narrow nasal bridge, and prominent epicanthic folds giving the impression of hypertelorism. The teeth are usually small and carious. Typical eye findings include microphthalmia and microcornea. The characteristic digital malformation is complete syndactyly of the fourth and fifth fingers (syndactyly type III) but the third finger may be involved and associated camptodactyly is a common finding. Cardiac abnormalities are observed in rare instances. {ECO:0000269|PubMed:12457340, ECO:0000269|PubMed:14729836, ECO:0000269|PubMed:15108203, ECO:0000269|PubMed:15637728, ECO:0000269|PubMed:16219735, ECO:0000269|PubMed:16222672, ECO:0000269|PubMed:16378922, ECO:0000269|PubMed:16709485, ECO:0000269|PubMed:16813608, ECO:0000269|PubMed:16816024, ECO:0000269|PubMed:17509830, ECO:0000269|PubMed:18161618, ECO:0000269|PubMed:19338053, ECO:0000269|PubMed:21670345, ECO:0000269|PubMed:23550541, ECO:0000269|PubMed:24508941}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sindaktili, Tip 4, 186200, Otozomal Dominant; SDTY4 (Sindaktili Tip 4) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Syndactyly, type IV, 186200, Autosomal dominant; SDTY4 (Syndactyly type 4) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159}.;	EDTA Blood Sample
Sindaktili, Tip 5, 186300, Otozomal Dominant; SDTY5 (Sindaktili Tip 5) (HOXD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Syndactyly, type V, 186300, Autosomal dominant; SDTY5 (Syndactyly type 5) (HOXD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sindaktili, Tip 5, 186300, Otozomal Dominant; SDTY5 (Sindaktili Tip 5) (MLPA)	Syndactyly, type V, 186300, Autosomal dominant; SDTY5 (Syndactyly type 5) (MLPA)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sinovit, Kronik, Yatkınlık (HLA-B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Synovitis, chronic, susceptibility to (HLA-B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-B	HLA-B, SPDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sinpolidaktili 1, 186000, Otozomal Dominant; SPD1 (Sinpolidaktili Tip 1) (HOXD13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Synpolydactyly 1, 186000, Autosomal dominant; SPD1 (Synpolydactyly type 1) (HOXD13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sinpolidaktili 1, 186000, Otozomal Dominant; SPD1 (Sinpolidaktili Tip 1) (MLPA)	Synpolydactyly 1, 186000, Autosomal dominant; SPD1 (Synpolydactyly type 1) (MLPA)	HOXD13	HOXD13, HOX4I, SPD1, BDSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Brachydactyly D (BDD) [MIM:113200]: A form of brachydactyly. Brachydactyly defines a group of inherited malformations characterized by shortening of the digits due to abnormal development of the phalanges and/or the metacarpals. Brachydactyly type D is characterized by short and broad terminal phalanges of the thumbs and big toes. {ECO:0000269	PubMed:12649808}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Sinpolidaktili, 3 / 3'4, Metakarpal ve Metatarsal Sinostozis İle İlişkili, 608180, Otozomal Dominant; SPD2 (Sinpolidaktili Tip 2) (FBLN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180, Autosomal dominant; SPD2 (Synpolydactyly type 2) (FBLN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBLN1	FBLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving FBLN1 is found in a complex type of synpolydactyly referred to as 3/3-prime/4 synpolydactyly associated with metacarpal and metatarsal synostoses. Reciprocal translocation t(12:22)(p11.2;q13.3) with RASSF8. Fibroblasts derived from a patient with synpolydactyly displayed alterations in the level of isoform D splice variant incorporated into the ECM and secreted into the conditioned culture medium. By contrast, the expression of isoform C was not perturbed in the patients fibroblasts. Furthermore, no aberrant polypeptides were detected in extracts of cultured patients fibroblasts. The translocation t(12;22) may result in haploinsufficiency of the isoform D splice variant, which could lead to the observed limb malformation. {ECO:0000269	PubMed:11836357}.	EDTA Blood Sample
Sistemik Lupus Eritematosus 16, 614420, Otozomal Resesif; SLEB16 (Otozomal Resesif Sistemik Lupus Eritematozus) (DNASE1L3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus 16, 614420, Autosomal recessive; SLEB16 (Autosomal recessive systemic lupus erythematosus) (DNASE1L3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNASE1L3	DNASE1L3, SLEB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Sistemik Lupus Eritematosus Yatkınlık, 152700, Otozomal Dominant (PTPN22 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus susceptibility to, 152700, Autosomal dominant (PTPN22 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTPN22	PTPN22, PEP, PTPN8, LYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269	PubMed:15273934}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

Sistemik Lupus Eritematosus, Direnç, 601744 (TLR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, resistance to, 601744 (TLR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR5	TLR5, TIL3, SLEB1, MELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 1 (SLEB1) [MIM:601744]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık 1, 601744; SLEB1 (TLR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 1, 601744; SLEB1 (TLR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TLR5	TLR5, TIL3, SLEB1, MELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 1 (SLEB1) [MIM:601744]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 10, 612251; SLEB10 (IRF5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 10, 612251; SLEB10 (IRF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF5	IRF5, IBD14, SLEB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis (RA) [MIM:180300]: An inflammatory disease with autoimmune features and a complex genetic component. It primarily affects the joints and is characterized by inflammatory changes in the synovial membranes and articular structures, widespread fibrinoid degeneration of the collagen fibers in mesenchymal tissues, and by atrophy and rarefaction of bony structures. {ECO:0000269	PubMed:17599733}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 11, 612253; SLEB11 (STAT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 11, 612253; SLEB11 (STAT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STAT4	STAT4, SLEB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Rheumatoid arthritis (RA) [MIM:180300]: An inflammatory disease with autoimmune features and a complex genetic component. It primarily affects the joints and is characterized by inflammatory changes in the synovial membranes and articular structures, widespread fibrinoid degeneration of the collagen fibers in mesenchymal tissues, and by atrophy and rarefaction of bony structures. {ECO:0000269	PubMed:17604942}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 152700, Otozomal Dominant (FCGR2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 152700, Autosomal dominant (FCGR2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2B	FCGR2B, CD32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:12115230, ECO:0000269|PubMed:20395827}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 152700, Otozomal Dominant (DNASE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 152700, Autosomal dominant (DNASE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNASE1	DNASE1, DNL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269	PubMed:11479590}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 152700, Otozomal Dominant (CTLA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 152700, Autosomal dominant (CTLA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CTLA4	CTLA4, IDDM12, CELIAC3, ALPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:15138458, ECO:0000269|PubMed:15688186}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 152700, Otozomal Dominant (Chilblain Lupus) (TREX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 152700, Autosomal dominant (Chilblain lupus) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:17660818, ECO:0000269|PubMed:20131292}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Enhanced immune sensing of oxidized DNA may be involved in the phototoxicity experienced by SLE patients. Exposure to UV- light produces DNA oxidative damage. Oxidized DNA being a poor TREX1 substrate, it accumulates in skin, leading to enhanced auto- immune reactivity and eventually skin lesions (PubMed:23993650). {ECO:0000269|PubMed:23993650}.	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık 2, 605218; SLEB2 (PDCD1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 2, 605218; SLEB2 (PDCD1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDCD1	PDCD1, SLEB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 2 (SLEB2) [MIM:605218]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269	PubMed:12420038}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritematozus, Yatkınlık, 9, 610927; SLEB9 (CR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, susceptibility to, 9, 610927; SLEB9 (CR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CR2	CR2, C3DR, SLEB9, CVID7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency, common variable, 7 (CVID7) [MIM:614699]: A primary immunodeficiency characterized by antibody deficiency, hypogammaglobulinemia, recurrent bacterial infections and an inability to mount an antibody response to antigen. The defect results from a failure of B-cell differentiation and impaired secretion of immunoglobulins; the numbers of circulating B-cells is usually in the normal range, but can be low. {ECO:0000269	PubMed:22035880}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Sistemik Lupus Eritemtl, Yatkınlık, 609939 (ITGAM geni) (Tüm Gen Dizi Analizi) (Postnatal)	Systemic lupus erythematosus, association with susceptibility to, 6, 609939 (ITGAM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGAM	ITGAM, CR3A, CD11B, MAC1A, SLEB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus 6 (SLEB6) [MIM:609939]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:18204446, ECO:0000269|PubMed:18204448}. Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry.;	EDTA Blood Sample
MLL (KMT2A) (11q23) Breakapart / Tüm MLL Translokasyonları IFISH)	MLL (KMT2A) (11q23) Breakapart / MLL Translocations IFISH)	11q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(11;14) (q13;q32) (IgH/ CCND1) (FISH)	t (11;14) (q13;q32) (IgH / CCND1) (FISH)	11q13-14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(12;21) (p13;q22) (TEL/ AML1) (FISH)	t (12;21) (p13;q22) (TEL / AML1) (FISH)	12p13-21q22	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(12;21) (p13;q22) (TEL/ AML1) (REAL - TIME PCR)	t (12;21) (p13;q22) (TEL / AML1) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
t(14;18) (q32;q21) (IgH/ BCL2) (FISH)	t (14;18) (q32;q21) (IgH / BCL2) (FISH)	14q32-18q21	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(15;17) (q22;q21) (PML/ RARA) (FISH)	t (15;17) (q22;q21) (PML / RARA) (FISH)	15q22-17q21	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(15;17) (q22;q21) (PML/ RARA) (REAL - TIME PCR)	t (15;17) (q22;q21) (PML / RARA) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
t(8;14) (q14-q32) (? - IGH) (FISH)	t (8;14) (q24;q32) (?-IGH) (FISH)	8q14-14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(8;21) (q22;q22) (ETO/ AML1) (FISH)	t (8;21) (q22;q22) (ETO / AML1) (FISH)	8q22-21q22	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(8;21) (q22;q22) (ETO/ AML1) (REAL - TIME PCR)	t (8;21) (q22;q22) (ETO / AML1) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)		5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
t(9;22) (q34;q11.2) (BCR/ ABL) (Philadelphia Kromozomu) (FISH)	t (9;22) (q34;q11.2) (BCR/ABL) (Philadelphia chromosome) (FISH)	9q34-22q11.2	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)	*	10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(9;22) (q34;q11.2) (BCR/ ABL) (Philadelphia Kromozomu) (p190 ve p210) (REAL - TIME PCR)	t (9;22) (q34;q11.2) (BCR/ABL) (Philadelphia chromosome) (p190 ve p210) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)	*	5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
T-Hücreli Akut Lenfoblastik Lösemi (MYB geni) (Tüm Gen Dizi Analizi) (Postnatal)	T-cell acute lymphoblastic leukemia (MYB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYB	MYB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
T-Hücreli Akut Lenfoblastik Lösemi, Somatik, 613065 (Akut Lenfoblastik Lösemi) (BAX geni) (Tüm Gen Dizi Analizi) (Postnatal)	T-cell acute lymphoblastic leukemia, somatic, 613065 (Acute lymphoblastic leukemia) (BAX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAX	BAX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
T-Hücreli İmmün Yetmezliği, Konjenital Alopesi ve Tırnak Distrofisi, 601705, Otozomal Resesif, TIIAC (Alenfoid Kistik Timik Displazisi) (FOXN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705, Autosomal recessive, TIIAC (Alymphoid cystic thymic dysgenesis) (FOXN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXN1	FOXN1, WHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

T-Hücreli İmmün Yetmeliği, Konjenital Alopesi ve Tırnak Distrofisi, 601705, Otozomal Resesif; TIIAC (Alenfoid Kistik Timik Disgenezis) (Prenatal) (FOXN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705, Autosomal recessive; TIIAC (Alymphoid cystic thymic dysgenesis) (FOXN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FOXN1	FOXN1, WHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
T-Hücre İmmün Yetmezliği, Nükseden Enfeksiyonlar, Otoimmünite ve Kardiyak Malformasyonlar, 614868 (Kombine İmmün Yetmezlik, STK4 Eksikliğine Bağlı) (STK4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (Combined immunodeficiency due to STK4 deficiency) (STK4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STK4	STK4, MST1, KRS2, TIIAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: T-cell immunodeficiency, recurrent infections, and autoimmunity with or without cardiac malformations (TIIAC) [MIM:614868]: A primary T-cell immunodeficiency syndrome characterized by progressive loss of naive T-cells, recurrent bacterial, viral, and fungal infections, warts, and abscesses, autoimmune manifestations, and cardiac malformations, including atrial septal defect. {ECO:0000269|PubMed:22294732}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
T-Hücre İmmün Yetmezliği, Nükseden Enfeksiyonlar, Otoimmünite ve Kardiyak Malformasyonlar, 614868 (Kombine İmmün Yetmezlik, STK4 Eksikliğine Bağlı) (Prenatal) (STK4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 (Combined immunodeficiency due to STK4 deficiency) (STK4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STK4	STK4, MST1, KRS2, TIIAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: T-cell immunodeficiency, recurrent infections, and autoimmunity with or without cardiac malformations (TIIAC) [MIM:614868]: A primary T-cell immunodeficiency syndrome characterized by progressive loss of naive T-cells, recurrent bacterial, viral, and fungal infections, warts, and abscesses, autoimmune manifestations, and cardiac malformations, including atrial septal defect. {ECO:0000269|PubMed:22294732}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
T-Hücresi Prolenfositik Lösemi, Somatik (ATM geni) (Tüm Gen Dizi Analizi) (Postnatal)	T-cell prolymphocytic leukemia, somatic (ATM gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATM	ATM, ATA, AT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	DISEASE: Ataxia telangiectasia (AT) [MIM:208900]: A rare recessive disorder characterized by progressive cerebellar ataxia, dilation of the blood vessels in the conjunctiva and eyeballs, immunodeficiency, growth retardation and sexual immaturity. Patients have a strong predisposition to cancer; about 30% of patients develop tumors, particularly lymphomas and leukemias. Cells from affected individuals are highly sensitive to damage by ionizing radiation and resistant to inhibition of DNA synthesis following irradiation. {ECO:0000269|PubMed:10234507, ECO:0000269|PubMed:10425038, ECO:0000269|PubMed:10817650, ECO:0000269|PubMed:10873394, ECO:0000269|PubMed:7792600, ECO:0000269|PubMed:8698354, ECO:0000269|PubMed:8755918, ECO:0000269|PubMed:8789452, ECO:0000269|PubMed:8797579, ECO:0000269|PubMed:8808599, ECO:0000269|PubMed:8845835, ECO:0000269|PubMed:9043869, ECO:0000269|PubMed:9150358, ECO:0000269|PubMed:9443866, ECO:0000269|PubMed:9450874, ECO:0000269|PubMed:9463314, ECO:0000269|PubMed:9497252, ECO:0000269|PubMed:9521587, ECO:0000269|PubMed:9711876, ECO:0000269|PubMed:9792409, ECO:0000269|PubMed:9792410, ECO:0000269|PubMed:9872980, ECO:0000269|PubMed:9887333}. Note=The disease is caused	EDTA blood sample / EDTA bone marrow
t(11;14)(q13;q32) (IGH/ MYEOV) Multipl Myeloma (FISH)	t(11;14)(q13;q32) (IGH/MYEOV) Multiple Myeloma (FISH)	11q13-14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(14;16)(q32.3;q23) (IGH/ MAF) (FISH)	t(14;16)(q32.3;q23) (IGH/ MAF) (FISH)	14q32.3-16q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
t(4;14)(p16;q32) (IgH/ FGRF3) (FISH)	t(4;14)(p16;q32) (IgH/ FGRF3) (FISH)	4p16-14q32	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Takenouchi-Kosaki Sendromu, 616737; TKS (CDC42 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Takenouchi-Kosaki syndrome, 616737; TKS (CDC42 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDC42	CDC42, TKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Takenouchi-Kosaki Sendromu, 616737; TKS (Prenatal) (CDC42 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Takenouchi-Kosaki syndrome, 616737; TKS (CDC42 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CDC42	CDC42, TKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tangier Hastalığı, 205400, Otozomal Resesif; TGD (Tangier Hastalığı) (ABCA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tangier disease, 205400, Autosomal recessive; TGD (Tangier disease) (ABCA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ABCA1	ABCA1, ABC1, HDLDT1, TGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: High density lipoprotein deficiency 1 (HDLD1) [MIM:205400]: Recessive disorder characterized by absence of high density lipoprotein (HDL) cholesterol from plasma, accumulation of cholesteryl esters, premature coronary artery disease (CAD), hepatosplenomegaly, recurrent peripheral neuropathy and progressive muscle wasting and weakness. {ECO:0000269|PubMed:10431236, ECO:0000269|PubMed:10431237, ECO:0000269|PubMed:10706591, ECO:0000269|PubMed:10938021, ECO:0000269|PubMed:11086027, ECO:0000269|PubMed:11257260, ECO:0000269|PubMed:11476961, ECO:0000269|PubMed:11476965, ECO:0000269|PubMed:11785958, ECO:0000269|PubMed:12111371, ECO:0000269|PubMed:12111381, ECO:0000269|PubMed:12407001, ECO:0000269|PubMed:14576201, ECO:0000269|PubMed:15019541, ECO:0000269|PubMed:15158913, ECO:0000269|PubMed:15262183, ECO:0000269|PubMed:15297675, ECO:0000269|PubMed:15520867}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tangier Hastalığı, 205400, Otozomal Resesif; TGD (Tangier Hastalığı) (Prenatal) (ABCA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tangier disease, 205400, Autosomal recessive; TGD (Tangier disease) (ABCA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ABCA1	ABCA1, ABC1, HDLDT1, TGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: High density lipoprotein deficiency 1 (HDLD1) [MIM:205400]: Recessive disorder characterized by absence of high density lipoprotein (HDL) cholesterol from plasma, accumulation of cholesteryl esters, premature coronary artery disease (CAD), hepatosplenomegaly, recurrent peripheral neuropathy and progressive muscle wasting and weakness. {ECO:0000269|PubMed:10431236, ECO:0000269|PubMed:10431237, ECO:0000269|PubMed:10706591, ECO:0000269|PubMed:10938021, ECO:0000269|PubMed:11086027, ECO:0000269|PubMed:11257260, ECO:0000269|PubMed:11476961, ECO:0000269|PubMed:11476965, ECO:0000269|PubMed:11785958, ECO:0000269|PubMed:12111371, ECO:0000269|PubMed:12111381, ECO:0000269|PubMed:12407001, ECO:0000269|PubMed:14576201, ECO:0000269|PubMed:15019541, ECO:0000269|PubMed:15158913, ECO:0000269|PubMed:15262183, ECO:0000269|PubMed:15297675, ECO:0000269|PubMed:15520867}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
TARP Sendromu, 311900, X'e Bağlı; TARPS (TARP Sendromu) (RBM10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	TARP syndrome, 311900, X-linked recessive; TARPS (TARP syndrome) (RBM10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RBM10	RBM10, DXS8237E, KIAA0122, TARPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: TARP syndrome (TARPS) [MIM:311900]: A disorder characterized by the Robin sequence (micrognathia, glossoptosis and cleft palate), talipes equinovarus and cardiac defects. {ECO:0000269|PubMed:20451169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
TARP Sendromu, 311900, X'e Bağlı; TARPS (TARP Sendromu) (Prenatal) (RBM10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	TARP syndrome, 311900, X-linked recessive; TARPS (TARP syndrome) (RBM10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RBM10	RBM10, DXS8237E, KIAA0122, TARPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: TARP syndrome (TARPS) [MIM:311900]: A disorder characterized by the Robin sequence (micrognathia, glossoptosis and cleft palate), talipes equinovarus and cardiac defects. {ECO:0000269|PubMed:20451169}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tarsal-Karpal Koalisyon Sendromu, 186570, Otozomal Dominant; TCC (Tarsal-Karpal Koalisyon Sendromu) (NOG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tarsal-carpal coalition syndrome, 186570, Autosomal dominant; TCC (Tarsal-carpal coalition syndrome) (NOG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOG	NOG, SYM1, SYNS1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Stapes ankylosis with broad thumb and toes (SABTS) [MIM:184460]: Congenital autosomal dominant disorder that includes hyperopia, a hemicylindrical nose, broad thumbs, great toes, and other minor skeletal anomalies but lacked carpal and tarsal fusion and symphalangism. {ECO:0000269|PubMed:12089654}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tatton-Brown-Rahman Sendromu, 615879, Otozomal Dominant; TBRS (Uzun Boy-Mental Retardasyon-Fasiyal Dismorfizm Sendromu) (DNMT3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tatton-Brown-Rahman syndrome, 615879, Autosomal dominant; TBRS (Tall stature-intellectual disability-facial dysmorphism syndrome) (DNMT3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DNMT3A	DNMT3A, TBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tatton-Brown-Rahman syndrome (TBRS) [MIM:615879]: An overgrowth syndrome characterized by a distinctive facial appearance, tall stature and intellectual disability. Facial gestalt is characterized by a round face, heavy horizontal eyebrows and narrow palpebral fissures. Less common features include atrial septal defects, seizures, umbilical hernia, and scoliosis. {ECO:0000269|PubMed:24614070}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tatton-Brown-Rahman Sendromu, 615879, Otozomal Dominant; TBRS (Uzun Boy-Mental Retardasyon-Fasiyal Dismorfizm Sendromu) (Prenatal) (DNMT3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tatton-Brown-Rahman syndrome, 615879, Autosomal dominant; TBRS (Tall stature-intellectual disability-facial dysmorphism syndrome) (DNMT3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DNMT3A	DNMT3A, TBRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tatton-Brown-Rahman syndrome (TBRS) [MIM:615879]: An overgrowth syndrome characterized by a distinctive facial appearance, tall stature and intellectual disability. Facial gestalt is characterized by a round face, heavy horizontal eyebrows and narrow palpebral fissures. Less common features include atrial septal defects, seizures, umbilical hernia, and scoliosis. {ECO:0000269|PubMed:24614070}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Tay-Sachs Hastalığı, 272800, Otozomal Resesif; TSD (Tay-Sachs Hastalığı) (HEXA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tay-Sachs disease, 272800, Autosomal recessive; TSD (Tay-Sachs disease) (HEXA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HEXA	HEXA, TSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tay-Sachs Hastalığı, 272800, Otozomal Resesif; TSD (Tay-Sachs Hastalığı) (MLPA)	Tay-Sachs disease, 272800, Autosomal recessive; TSD (Tay-Sachs disease) (MLPA)	HEXA	HEXA, TSD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tay-Sachs Hastalığı, 272800, Otozomal Resesif; TSD (Tay-Sachs Hastalığı) (Prenatal) (HEXA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tay-Sachs disease, 272800, Autosomal recessive; TSD (Tay-Sachs disease) (HEXA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HEXA	HEXA, TSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tay-Sachs Hastalığı, 272800, Otozomal Resesif; TSD (Tay-Sachs Hastalığı) (Prenatal) (MLPA)	Tay-Sachs disease, 272800, Autosomal recessive; TSD (Tay-Sachs disease) (Prenatal) (MLPA)	HEXA	HEXA, TSD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: GM2-gangliosidosis 1 (GM2G1) [MIM:272800]: An autosomal recessive lysosomal storage disease marked by the accumulation of GM2 ganglosides in the neuronal cells. It is characterized by GM2 ganglosides accumulation in the absence of HEXA activity, leading to neurodegeneration and, in the infantile form, death in early childhood. It exists in several forms: infantile (most common and most severe), juvenile and adult (late-onset). {ECO:0000269|PubMed:1301189, ECO:0000269|PubMed:1301190, ECO:0000269|PubMed:1302612, ECO:0000269|PubMed:14566483, ECO:0000269|PubMed:1532289, ECO:0000269|PubMed:1837283, ECO:0000269|PubMed:2144098, ECO:0000269|PubMed:2522679, ECO:0000269|PubMed:2970528, ECO:0000269|PubMed:7717398, ECO:0000269|PubMed:7837766, ECO:0000269|PubMed:7898712, ECO:0000269|PubMed:7951261, ECO:0000269|PubMed:8445615, ECO:0000269|PubMed:8490625, ECO:0000269|PubMed:8581357, ECO:0000269|PubMed:8757036, ECO:0000269|PubMed:9150157, ECO:0000269|PubMed:9338583, ECO:0000269|PubMed:9375850, ECO:0000269|PubMed:9401008, ECO:0000269|PubMed:9603435}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Telenjiektazi, Herediter Hemorajik, Tip 1, 187300, Otozomal Dominant; HHT (Rendu - Osler - Weber Sendromu) (ENG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Telangiectasia, hereditary hemorrhagic, type 1, 187300, Autosomal dominant; HHT (Rendu - Osler - Weber Syndrome) (ENG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ENG	ENG, END, HHT1, ORW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Telenjiektazi, Herediter Hemorajik, Tip 1, 187300, Otozomal Dominant; HHT (Rendu - Osler - Weber Sendromu) (MLPA)	Telangiectasia, hereditary hemorrhagic, type 1, 187300, Autosomal dominant; HHT (Rendu - Osler - Weber Syndrome) (MLPA)	ENG	ENG, END, HHT1, ORW	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal		EDTA Blood Sample
Telenjiektazi, Herediter Hemorajik, Tip 2, 600376, Otozomal Dominant; HHT2 (Kalıtsal Hemorajik Telenjiektazi) (ACVRL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Telangiectasia, hereditary hemorrhagic, type 2, 600376, Autosomal dominant; HHT2 (Hereditary hemorrhagic telangiectasia) (ACVRL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACVRL1	ACVRL1, ACVRLK1, ALK1, HHT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Telangiectasia, hereditary hemorrhagic, 2 (HHT2) [MIM:600376]: A multisystemic vascular dysplasia leading to dilation of permanent blood vessels and arteriovenous malformations of skin, mucosa, and viscera. The disease is characterized by recurrent epistaxis and gastro-intestinal hemorrhage. Visceral involvement includes arteriovenous malformations of the lung, liver, and brain. {ECO:0000269|PubMed:10694922, ECO:0000269|PubMed:10767348, ECO:0000269|PubMed:11170071, ECO:0000269|PubMed:11484689, ECO:0000269|PubMed:14684682, ECO:0000269|PubMed:15024723, ECO:0000269|PubMed:15712270, ECO:0000269|PubMed:16525724, ECO:0000269|PubMed:16752392, ECO:0000269|PubMed:20414677, ECO:0000269|PubMed:8640225, ECO:0000269|PubMed:9245985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Telenjiektazi, Herediter Hemorajik, Tip 2, 600376, Otozomal Dominant; HHT2 (Kalıtsal Hemorajik Telenjiektazi) (MLPA)	Telangiectasia, hereditary hemorrhagic, type 2, 600376, Autosomal dominant; HHT2 (Hereditary hemorrhagic telangiectasia) (MLPA)	ACVRL1	ACVRL1, ACVRLK1, ALK1, HHT2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Telangiectasia, hereditary hemorrhagic, 2 (HHT2) [MIM:600376]: A multisystemic vascular dysplasia leading to dilation of permanent blood vessels and arteriovenous malformations of skin, mucosa, and viscera. The disease is characterized by recurrent epistaxis and gastro-intestinal hemorrhage. Visceral involvement includes arteriovenous malformations of the lung, liver, and brain. {ECO:0000269|PubMed:10694922, ECO:0000269|PubMed:10767348, ECO:0000269|PubMed:11170071, ECO:0000269|PubMed:11484689, ECO:0000269|PubMed:14684682, ECO:0000269|PubMed:15024723, ECO:0000269|PubMed:15712270, ECO:0000269|PubMed:16525724, ECO:0000269|PubMed:16752392, ECO:0000269|PubMed:20414677, ECO:0000269|PubMed:8640225, ECO:0000269|PubMed:9245985}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Telenjiektazi, Herediter Hemoraji, Tip 5, 615506, Otozomal Dominant; HHT5 (Kalıtsal Hemoraji Telenjiektazi) (GDF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Telangiectasia, hereditary hemorrhagic, type 5, 615506, Autosomal dominant; HHT5 (Hereditary hemorrhagic telangiectasia) (GDF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF2	GDF2, BMP9, HHT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Telangiectasia, hereditary hemorrhagic, 5 (HHT5) [MIM:615506]: A multisystemic vascular dysplasia leading to dilation of permanent blood vessels and arteriovenous malformations of skin, mucosa, and viscera. The disease is characterized by recurrent epistaxis and gastro-intestinal hemorrhage. Visceral involvement includes arteriovenous malformations of the lung, liver, and brain. {ECO:0000269|PubMed:23972370}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Temple-Baraitser Sendromu, 611816, Otozomal Dominant; TMBTS (Temple-Baraitser Sendromu) (KCNH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Temple-Baraitser syndrome, 611816, Autosomal dominant; TMBTS (Temple-Baraitser syndrome) (KCNH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNH1	KCNH1, EAG, TMBTS, ZLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Zimmermann-Laband syndrome 1 (ZLS1) [MIM:135500]: A disorder characterized by gingival fibromatosis, dysplastic or absent nails, finger abnormalities, hepatosplenomegaly, and abnormalities of the cartilage of the nose and/or ears. {ECO:0000269|PubMed:25915598}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Temple-Baraitser Sendromu, 611816, Otozomal Dominant; TMBTS (Temple-Baraitser Sendromu) (Prenatal) (KCNH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Temple-Baraitser syndrome, 611816, Autosomal dominant; TMBTS (Temple-Baraitser syndrome) (KCNH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNH1	KCNH1, EAG, TMBTS, ZLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Zimmermann-Laband syndrome 1 (ZLS1) [MIM:135500]: A disorder characterized by gingival fibromatosis, dysplastic or absent nails, finger abnormalities, hepatosplenomegaly, and abnormalities of the cartilage of the nose and/or ears. {ECO:0000269|PubMed:25915598}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Temtamy Preaksiyel Brakidaktili Sendromu, 605282, Otozomal Resesif; TPBS (Temtamy Preaksiyel Brakidaktili Sendromu) (CHSY1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Temtamy preaxial brachydactyly syndrome, 605282, Autosomal recessive; TPBS (Temtamy preaxial brachydactyly syndrome) (CHSY1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CHSY1	CHSY1, KIAA0990, TPBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Temtamy Sendromu, 218340, Otozomal Resesif; TEMTYS (Temtamy Sendromu) (C12orf57 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Temtamy syndrome, 218340, Autosomal recessive; TEMTYS (Temtamy syndrome) (C12orf57 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C12orf57	C12orf57, C10, TEMTYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Temtamy Sendromu, 218340, Otozomal Resesif; TEMTYS (Temtamy Sendromu) (Prenatal) (C12orf57 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Temtamy syndrome, 218340, Autosomal recessive; TEMTYS (Temtamy syndrome) (C12orf57 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	C12orf57	C12orf57, C10, TEMTYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tenorio Sendromu, 616260, Otozomal Dominant; TNORS (RNF125 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tenorio syndrome, 616260, Autosomal dominant; TNORS (RNF125 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF125	RNF125, TRAC1, TNORS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tenorio Sendromu, 616260, Otozomal Dominant; TNORS (Prenatal) (RNF125 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tenorio syndrome, 616260, Autosomal dominant; TNORS (RNF125 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RNF125	RNF125, TRAC1, TNORS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Terminal Osseöz Displazi, 300244; TOD (Terminal Kemik Displazisi-Pigment Defekt Sendromu) (FLNA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Terminal osseous dysplasia, 300244; TOD (Terminal osseous dysplasia-pigmentary defects syndrome) (FLNA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Terminal Osseöz Displazi, 300244; TOD (Terminal Kemik Displazisi-Pigment Defekt Sendromu) (MLPA)	Terminal osseous dysplasia, 300244; TOD (Terminal osseous dysplasia-pigmentary defects syndrome) (MLPA)	FLNA	FLNA, FLN1, NHBP, OPD1, OPD2, FMD, MNS, CVD1, CSBS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Intestinal pseudoobstruction, neuronal, chronic idiopathic, X-linked (IPOX) [MIM:300048]: A disease characterized by a severe abnormality of gastrointestinal motility due to primary qualitative defects of enteric ganglia and nerve fibers. Affected individuals manifest recurrent signs of intestinal obstruction in the absence of any mechanical lesion. {ECO:0000269|PubMed:17357080}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Testis Anomalileri, ± Konjenital Kalp Hastalığı, 615542, Otozomal Dominant; TACHD (46,XY Parsiyel Gonadal Disgenesis) (GATA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Testicular anomalies with or without congenital heart disease, 615542, Autosomal dominant; TACHD (46,XY partial gonadal dysgenesis) (GATA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Testis Anomalileri, ± Konjenital Kalp Hastalığı, 615542, Otozomal Dominant; TACHD (46,XY Parsiyel Gonadal Disgenesis) (MLPA)	Testicular anomalies with or without congenital heart disease, 615542, Autosomal dominant; TACHD (46,XY partial gonadal dysgenesis) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Testis Tümörü, Somatik, 273300 (STK11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Testicular tumor, somatic, 273300 (STK11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	STK11	STK11, PJS, LKB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinde doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Peutz-Jeghers syndrome (PJS) [MIM:175200]: An autosomal dominant disorder characterized by melanocytic macules of the lips, multiple gastrointestinal hamartomatous polyps and an increased risk for various neoplasms, including gastrointestinal cancer. {ECO:0000269|PubMed:10408777, ECO:0000269|PubMed:12372054, ECO:0000269|PubMed:21411391, ECO:0000269|PubMed:9425897, ECO:0000269|PubMed:9428765, ECO:0000269|PubMed:9760200, ECO:0000269|PubMed:9837816}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Tetra-Amelia Sendromu, 273395, Otozomal Resesif; TETAMS (Tetraameli-Multipl Malformasyon Sendromu) (WNT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetra-amelia syndrome, 273395, Autosomal recessive; TETAMS (Tetraamelia-multiple malformations syndrome) (WNT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT3	WNT3, INT4, TETAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetraamelia syndrome, autosomal recessive (TETAMS) [MIM:273395]: A rare human genetic disorder characterized by complete absence of all four limbs and other anomalies such as craniofacial, nervous system, pulmonary, skeletal and urogenital defects. {ECO:0000269|PubMed:14872406}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tetra-Amelia Sendromu, 273395, Otozomal Resesif; TETAMS (Tetraameli-Multipl Malformasyon Sendromu) (Prenatal) (WNT3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetra-amelia syndrome, 273395, Autosomal recessive; TETAMS (Tetraamelia-multiple malformations syndrome) (WNT3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT3	WNT3, INT4, TETAMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetraamelia syndrome, autosomal recessive (TETAMS) [MIM:273395]: A rare human genetic disorder characterized by complete absence of all four limbs and other anomalies such as craniofacial, nervous system, pulmonary, skeletal and urogenital defects. {ECO:0000269|PubMed:14872406}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:14517948, ECO:0000269|PubMed:20807224}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (TBX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (TBX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: DiGeorge syndrome (DGS) [MIM:188400]: A congenital syndrome characterized by a wide spectrum of characteristics including parathyroid hypoplasia resulting in hypocalcemia, thymic hypoplasia resulting in T-cell immunodeficiency, defects in the outflow tract of the heart, and craniofacial anomalies. Disturbance of cervical neural crest migration into the derivatives of the pharyngeal arches and pouches can account for the phenotype. {ECO:0000269|PubMed:14585638}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (JAG1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (JAG1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	JAG1	JAG1, AGS1, AHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. {ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (GDF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (GATA6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (GATA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GATA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (MLPA)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (MLPA)	JAG1	JAG1, AGS1, AHD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. {ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (MLPA)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (ZFPM2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (ZFPM2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZFPM2	ZFPM2, FOG2, DIH3, SRXY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:14517949, ECO:0000269|PubMed:20807224}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (TBX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (TBX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: DiGeorge syndrome (DGS) [MIM:188400]: A congenital syndrome characterized by a wide spectrum of characteristics including parathyroid hypoplasia resulting in hypocalcemia, thymic hypoplasia resulting in T-cell immunodeficiency, defects in the outflow tract of the heart, and craniofacial anomalies. Disturbance of cervical neural crest migration into the derivatives of the pharyngeal arches and pouches can account for the phenotype. {ECO:0000269	PubMed:14585638}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (JAG1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (JAG1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	JAG1	JAG1, AGS1, AHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. {ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (GDF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269	PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (GATA6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GATA6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA6	GATA6, AVSD5, ASD9, AVSD5, PACHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:20581743, ECO:0000269|PubMed:20631719}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (GATA4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (GATA4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (MLPA)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (Prenatal) (MLPA)	JAG1	JAG1, AGS1, AHD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Alagille syndrome 1 (ALGS1) [MIM:118450]: A form of Alagille syndrome, an autosomal dominant multisystem disorder. It is clinically defined by hepatic bile duct paucity and cholestasis in association with cardiac, skeletal, and ophthalmologic manifestations. There are characteristic facial features and less frequent clinical involvement of the renal and vascular systems. {ECO:0000269|PubMed:10220506, ECO:0000269|PubMed:10533065, ECO:0000269|PubMed:11058898, ECO:0000269|PubMed:11139247, ECO:0000269|PubMed:11157803, ECO:0000269|PubMed:11180599, ECO:0000269|PubMed:12442286, ECO:0000269|PubMed:12497640, ECO:0000269|PubMed:15712272, ECO:0000269|PubMed:16575836, ECO:0000269|PubMed:23801938, ECO:0000269|PubMed:9207788, ECO:0000269|PubMed:9585603}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant (Fallot Tetralojisi) (Prenatal) (MLPA)	Tetralogy of Fallot, 187500, Autosomal dominant (Tetralogy of Fallot) (Prenatal) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:18672102, ECO:0000269|PubMed:21110066, ECO:0000269|PubMed:24000169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant; TOF (Fallot Tetralojisi) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tetralogy of Fallot, 187500, Autosomal dominant; TOF (Tetralogy of Fallot) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Fallot Tetralojisi, 187500, Otozomal Dominant; TOF (Fallot Tetralojisi) (Prenatal) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tetralogy of Fallot, 187500, Autosomal dominant; TOF (Tetralogy of Fallot) (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:10587520, ECO:0000269|PubMed:11714651, ECO:0000269|PubMed:14607454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemi Hb Lepore Nedeniyle (HBD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thalassemia due to Hb Lepore (HBD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBD	HBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample

Talasemi-Beta, Dominant İnhibizyon Cisimciği, 603902 (Dominant Beta Talasemi) (HBB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thalassemia-beta, dominant inclusion-body, 603902 (Dominant beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemi-Beta, Dominant İnhibizyon Cisimciği, 603902 (Dominant Beta Talasemi) (MLPA)	Thalassemia-beta, dominant inclusion-body, 603902 (Dominant beta-thalassemia) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)	.	20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemi-Beta, Dominant İnhibizyon Cisimciği, 603902 (Dominant Beta Talasemi) (Prenatal) (HBB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Thalassemia-beta, dominant inclusion-body, 603902 (Dominant beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemi-Beta, Dominant İnhibizyon Cisimciği, 603902 (Dominant Beta Talasemi) (Prenatal) (MLPA)	Thalassemia-beta, dominant inclusion-body, 603902 (Dominant beta-thalassemia) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemi, Alfa-, 604131 (Heinz Cisimciği Anemisi) (MLPA)	Thalassemia, alpha-, 604131 (Heinz body anemia) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemi, Alfa-, 604131 (Heinz Cisimciği Anemisi) (Prenatal) (MLPA)	Thalassemia, alpha-, 604131 (Heinz body anemia) (Prenatal) (MLPA)	HBA1-HBA2	HBA2, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemi, Delta (HBD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thalassemia, delta- (HBD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBD	HBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Talasemi, Delta (Prenatal) (HBD gen) (Tüm Gen Dizi Analizi) (Prenatal)	Thalassemia, delta- (HBD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBD	HBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemiler, Alfa-, 604131 (Alfa-Talasemi) (MLPA)	Thalassemias, alpha-, 604131 (Alpha-thalassemia) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemiler, Alfa-, 604131 (Alfa-Talasemi) (Prenatal) (MLPA)	Thalassemias, alpha-, 604131 (Alpha-thalassemia) (Prenatal) (MLPA)	HBA1-HBA2	HBA1, HBH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269	PubMed:2833478). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Talasemiler, Beta-, 613985 (Beta-Talasemi) (HBB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thalassemias, beta-, 613985 (Beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemiler, Beta-, 613985 (Beta-Talasemi) (MLPA)	Thalassemias, beta-, 613985 (Beta-thalassemia) (MLPA)	HBB	HBB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemiler, Beta-, 613985 (Beta-Talasemi) (Prenatal) (HBB gen) (Tüm Gen Dizi Analizi) (Prenatal)	Thalassemias, beta-, 613985 (Beta-thalassemia) (HBB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBB	HBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. (ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193). Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Talasemiler, Beta-, 613985 (Beta-Talasemi) (Prenatal) (MLPA)	Thalassemias, beta-, 613985 (Beta-thalassemia) (Prenatal) (MLPA)	HBB	HBB	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269|PubMed:186485, ECO:0000269|PubMed:2599881, ECO:0000269|PubMed:6259091, ECO:0000269|PubMed:8704193}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tanatoforik Displazi, Tip 1; Td1 (Tanatoforik Displazi) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	THANATOPHORIC DYSPLASIA, TYPE I; TD1 (Thanatophoric dysplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tanatoforik Displazi, Tip 1; Td1 (Tanatoforik Displazi) (MLPA)	THANATOPHORIC DYSPLASIA, TYPE I; TD1 (Thanatophoric dysplasia) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tanatoforik Displazi, Tip 1; Td1 (Tanatoforik Displazi) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	THANATOPHORIC DYSPLASIA, TYPE I; TD1 (Thanatophoric dysplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tanatoforik Displazi, Tip 1; Td1 (Tanatoforik Displazi) (Prenatal) (MLPA)	THANATOPHORIC DYSPLASIA, TYPE I; TD1 (Thanatophoric dysplasia) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tanatoforik Displazi, Tip 2; Td2 (Tanatoforik Displazi) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	THANATOPHORIC DYSPLASIA, TYPE II; TD2 (Thanatophoric dysplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tanatoforik Displazi, Tip 2; Td2 (Tanatoforik Displazi) (MLPA)	THANATOPHORIC DYSPLASIA, TYPE II; TD2 (Thanatophoric dysplasia) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tanatoforik Displazi, Tip 2; Td2 (Tanatoforik Displazi) (Prenatal) (FGFR3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	THANATOPHORIC DYSPLASIA, TYPE II; TD2 (Thanatophoric dysplasia) (FGFR3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR3	FGFR3, ACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tanatoforik Displazi, Tip 2; Td2 (Tanatoforik Displazi) (Prenatal) (MLPA)	THANATOPHORIC DYSPLASIA, TYPE II; TD2 (Thanatophoric dysplasia) (Prenatal) (MLPA)	FGFR3	FGFR3, ACH	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Achondroplasia (ACH) [MIM:100800]: A frequent form of short-limb dwarfism. It is characterized by a long, narrow trunk, short extremities, particularly in the proximal (rhizomelic) segments, a large head with frontal bossing, hypoplasia of the midface and a trident configuration of the hands. {ECO:0000269|PubMed:7758520, ECO:0000269|PubMed:7847369, ECO:0000269|PubMed:8078586}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Thauvin-Robinet-Faivre Sendromu, 617107, Otozomal Resesif; TROFAS (FIBP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thauvin-Robinet-Faivre syndrome, 617107, Autosomal recessive; TROFAS (FIBP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIBP	FIBP, TROFAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Thauvin-Robinet-Faivre Sendromu, 617107, Otozomal Resesif; TROFAS (Prenatal) (FIBP geni) (Tüm Gen Dizi Analizi) (Prenatal)	Thauvin-Robinet-Faivre syndrome, 617107, Autosomal recessive; TROFAS (FIBP gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FIBP	FIBP, TROFAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İnsülin benzeri büyüme faktörü 1 reseptörü (IGF1R) paneli (IGFR1, IGFBP3.) (MLPA)	The insulin-like growth factor 1 receptor (IGF1R) panel (IGFR1, IGFBP3.) (MLPA)	IGFR1, IGFBP3.	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Sarkom-kanser paneli (Kromozom 12, MDM2, CDK4, HMGA2 genleri) (MLPA)	The sarcoma-cancer panel (chromosome 12, MDM2, CDK4, HMGA2 genes) (MLPA)	Kromozom 12, MDM2, CDK4, HMGA2 genleri	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 2 (Biyotin Veya Tiyamin Yanıtlı Ensefalopati Tip 2), 607483, Otozomal Resesif; THMD2 (Tiamin Duyarlı Ensefalopati) (SLC19A3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483, Autosomal recessive; THMD2 (Thiamine-responsive encephalopathy) (SLC19A3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC19A3	SLC19A3, THMD2, BBGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 2 (Biyotin Veya Tiyamin Yanıtlı Ensefalopati Tip 2), 607483, Otozomal Resesif; THMD2 (Tiamin Duyarlı Ensefalopati) (Prenatal) (SLC19A3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Thiamine metabolism dysfunction syndrome 2 (biotin- or thiamine-responsive encephalopathy type 2), 607483, Autosomal recessive; THMD2 (Thiamine-responsive encephalopathy) (SLC19A3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC19A3	SLC19A3, THMD2, BBGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 4 (Progresif Polinöropati Tipi), 613710, Otozomal Resesif (Progresif Demiyelinizan Nöropati-Bilateral Striatal Nekroz) (SLC25A19 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710, Autosomal recessive (Progressive demyelinating neuropathy with bilateral striatal necrosis) (SLC25A19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC25A19	SLC25A19, DNC, MUP1, MCPHA, THMD3, THMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thiamine metabolism dysfunction syndrome 4, bilateral striatal degeneration and progressive polyneuropathy type (THMD4) [MIM:613710]: A disease characterized by recurrent episodes of flaccid paralysis and encephalopathy associated with bilateral striatal necrosis and chronic progressive polyneuropathy. {ECO:0000269|PubMed:19798730}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 4 (Progresif Polinöropati Tipi), 613710, Otozomal Resesif (Progresif Demiyelinizan Nöropati-Bilateral Striatal Nekroz) (Prenatal) (SLC25A19 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Thiamine metabolism dysfunction syndrome 4 (progressive polyneuropathy type), 613710, Autosomal recessive (Progressive demyelinating neuropathy with bilateral striatal necrosis) (SLC25A19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC25A19	SLC25A19, DNC, MUP1, MCPHA, THMD3, THMD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Thiamine metabolism dysfunction syndrome 4, bilateral striatal degeneration and progressive polyneuropathy type (THMD4) [MIM:613710]: A disease characterized by recurrent episodes of flaccid paralysis and encephalopathy associated with bilateral striatal necrosis and chronic progressive polyneuropathy. {ECO:0000269|PubMed:19798730}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 5 (Episodik Ensefalopati Tipi), 614458, Otozomal Resesif (Çocukluk Çağı Ensefalopatisi, Tiamin Profosfokinaz Eksikliğine Bağlı) (TPK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458, Autosomal recessive (Childhood encephalopathy due to thiamine pyrophosphokinase deficiency) (TPK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPK1	TPK1, THMD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiamin Metabolizma Disforiksiyon Sendromu 5 (Episodik Ensefalopati Tipi), 614458, Otozomal Resesif (Çocukluk Çağı Ensefalopatisi, Tiamin Profosfokinaz Eksikliğine Bağlı) (Prenatal) (TPK1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Thiamine metabolism dysfunction syndrome 5 (episodic encephalopathy type), 614458, Autosomal recessive (Childhood encephalopathy due to thiamine pyrophosphokinase deficiency) (TPK1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TPK1	TPK1, THMD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tiamin-Yanıtlı Megaloblastik Anemi Sendromu, 249270, Otozomal Resesif; TRMA (Tiamin Duyarlı Megaloblastik Anemi Sendromu) (SLC19A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiamine-responsive megaloblastic anemia syndrome, 249270, Autosomal recessive; TRMA (Thiamine-responsive megaloblastic anemia syndrome) (SLC19A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC19A2	SLC19A2, THTR1, TRMA, THMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiamin-Yanıtlı Megaloblastik Anemi Sendromu, 249270, Otozomal Resesif; TRMA (Tiamin Duyarlı Megaloblastik Anemi Sendromu) (Prenatal) (SLC19A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Thiamine-responsive megaloblastic anemia syndrome, 249270, Autosomal recessive; TRMA (Thiamine-responsive megaloblastic anemia syndrome) (SLC19A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SLC19A2	SLC19A2, THTR1, TRMA, THMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tiyopurin, S-metiltransferaz (TPMT, S-adenosil-L-metionin: tiopurin S-transferaz) (TPMT 6p22) (MLPA)	Thiopurine, S-methyltransferase (TPMT, S-adenosyl-L-methionine: thiopurine S-transferase) (TPMT 6p22) (MLPA)	TPMT 6p22	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Tiopurinler, Zayıf Metabolizma, 1, 610460, Otozomal Resesif; THPM1 (Azatiyoprin ve 6-Mercaptopurine Toksisitesi veya Doz Seçimi) (TPMT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiopurines, poor metabolism of, 1, 610460, Autosomal recessive; THPM1 (Azathioprine or 6-mercaptopurine toxicity or dose selection) (TPMT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPMT	TPMT, TPMTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thiopurine S-methyltransferase deficiency (TPMT deficiency) [MIM:610460]: Enzyme involved in the normal metabolic inactivation of thiopurine drugs. These drugs are generally used as immunosuppressants or cytotoxic drugs and are prescribed for a variety of clinical conditions including leukemia, autoimmune disease and organ transplantation. Patients with intermediate or no TPMT activity are at risk of toxicity after receiving standard doses of thiopurine drugs and it is shown that inter-individual differences in response to these drugs are largely determined by genetic variation at the TPMT locus. (ECO:0000269|PubMed:10208641, ECO:0000269|PubMed:10751626, ECO:0000269|PubMed:15819814, ECO:0000269|PubMed:16220112, ECO:0000269|PubMed:16476125, ECO:0000269|PubMed:16789994, ECO:0000269|PubMed:7862671, ECO:0000269|PubMed:8561894, ECO:0000269|PubMed:8644731, ECO:0000269|PubMed:9246020, ECO:0000269|PubMed:9336428, ECO:0000269|PubMed:9711875, ECO:0000269|PubMed:9931345, ECO:0000269|PubMed:9931346). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiopurinler, Zayıf Metabolizma, 1, 610460, Otozomal Resesif; THPM1 (Azatiyoprin ve 6-Mercaptopurine Toksisitesi veya Doz Seçimi) (MLPA)	Thiopurines, poor metabolism of, 1, 610460, Autosomal recessive; THPM1 (Azathioprine or 6-mercaptopurine toxicity or dose selection) (MLPA)	TPMT	TPMT, TPMTD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Thiopurine S-methyltransferase deficiency (TPMT deficiency) [MIM:610460]: Enzyme involved in the normal metabolic inactivation of thiopurine drugs. These drugs are generally used as immunosuppressants or cytotoxic drugs and are prescribed for a variety of clinical conditions including leukemia, autoimmune disease and organ transplantation. Patients with intermediate or no TPMT activity are at risk of toxicity after receiving standard doses of thiopurine drugs and it is shown that inter-individual differences in response to these drugs are largely determined by genetic variation at the TPMT locus. (ECO:0000269|PubMed:10208641, ECO:0000269|PubMed:10751626, ECO:0000269|PubMed:15819814, ECO:0000269|PubMed:16220112, ECO:0000269|PubMed:16476125, ECO:0000269|PubMed:16789994, ECO:0000269|PubMed:7862671, ECO:0000269|PubMed:8561894, ECO:0000269|PubMed:8644731, ECO:0000269|PubMed:9246020, ECO:0000269|PubMed:9336428, ECO:0000269|PubMed:9711875, ECO:0000269|PubMed:9931345, ECO:0000269|PubMed:9931346). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiopurinler, Bir Metabolizma, 2, 616903, Otozomal Dominant; THPM2 (Azatiyoprin ve 6-Mercaptopurine Toksisitesi veya Doz Seçimi) (NUDT15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thiopurines, poor metabolism of, 2, 616903, Autosomal dominant; THPM2 (Azathioprine or 6-mercaptopurine toxicity or dose selection) (NUDT15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NUDT15	NUDT15, MTH2, NUDT15D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trombositemi 1, 187950, Otozomal Dominant, THCYT1 (Esansiyel Trombositemi) (THPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocythemia 1, 187950, Autosomal dominant; THCYT1 (Essential thrombocythemia) (THPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THPO	THPO, MGDF, MPLLG, TPO, THCYT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombocythemia 1 (THCYT1) [MIM:187950]: A myeloproliferative disorder characterized by excessive platelet production, resulting in increased numbers of circulating platelets. It can be associated with spontaneous hemorrhages and thrombotic episodes. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositemi 2, 601977, Otozomal Dominant, Somatik Mutasyon; THCYT2 (Esansiyel Trombositemi) (MPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocythemia 2, 601977, Autosomal dominant, Somatic mutation; THCYT2 (Essential thrombocythemia) (MPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPL	MPL, TPOR, MPLV, THCYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Trombositemi 3, 614521, Otozomal Dominant, Somatik Mutasyon; THCYT3 (Esansiyel Trombositemi) V617F Mutasyonu ve (Ekzon 12-14) (JAK2 geni) (Dizi Analizi) (Postnatal)	Thrombocythemia 3, 614521, Autosomal dominant, Somatic mutation; THCYT3 (Essential thrombocythemia) V617F Mutation and (Exon 12-14) (JAK2 gene) (Dizi Analizi) (Postnatal)	JAK2	JAK2, THCYT3	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Trombositemi, Somatik, 187950 (Esansiyel Trombositemi) (SH2B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocythemia, somatic, 187950 (Essential thrombocythemia) (SH2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SH2B3	SH2B3, LNK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Trombositemi, Somatik, 187950 (Esansiyel Trombositemi) (CALR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocythemia, somatic, 187950 (Essential thrombocythemia) (CALR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALR	CALR, SSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Trombositopeni 2, 188000, Otozomal Dominant; THC2 (Otozomal Trombositopeni-Normal Trombositler) (ANKRD26 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia 2, 188000, Autosomal dominant; THC2 (Autosomal thrombocytopenia with normal platelets) (ANKRD26 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ANKRD26	ANKRD26, KIAA1074, THC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trombositopeni 4, 612004, Otozomal Dominant; THC4 (Otozomal Trombositopeni-Normal Trombositler9 (CYCS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia 4, 612004, Autosomal dominant; THC4 (Autosomal thrombocytopenia with normal platelets) (CYCS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYCS	CYCS, CYC, THC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombocytopenia 4 (THC4) [MIM:612004]: Thrombocytopenia is defined by a decrease in the number of platelets in circulating blood, resulting in the potential for increased bleeding and decreased ability for clotting. (ECO:0000269	PubMed:18345000). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni 5, 616216, Otozomal Dominant; THC5 (Allesel Trombosit Sendromu-Akut Myelositler Lösemi Eğilimi) (ETV6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia 5, 616216, Autosomal dominant; THC5 (Familial platelet syndrome with predisposition to acute myelogenous leukemia) (ETV6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ETV6	ETV6, TEL, THC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. (ECO:0000269	PubMed:15806161). Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Trombositopeni 6, 616937, Otozomal Dominant; THC6 (Kalıtsal Trombositopeni-Erken Başlangıçlı Miyelofibrozi (SRC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia 6, 616937, Autosomal dominant; THC6 (Hereditary thrombocytopenia with early-onset myelofibrosis) (SRC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRC	SRC, ASV, SRC1, THC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=SRC kinase activity has been shown to be increased in several tumor tissues and tumor cell lines such as colon carcinoma cells.;	EDTA Blood Sample
Trombositopeni-Beta Talasemi, X'e Bağlı, 314050, X'e Bağlı Resesif; XLTT (Beta-Talasemi-X'e Bağlı Trombositopeni Sendromu) (GATA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia with beta-thalassemia, X-linked, 314050, X-linked recessive; XLTT (Beta-thalassemia-X-linked thrombocytopenia syndrome) (GATA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA1	GATA1, GF1, ERYF1, NFE1, XLTDA, XLTT, XLANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked dyserythropoietic anemia and thrombocytopenia (XDAT) [MIM:300367]: Disorder characterized by erythrocytes with abnormal size and shape, and paucity of platelets in peripheral blood. The bone marrow contains abundant and abnormally small megakaryocytes. (ECO:0000269|PubMed:10700180, ECO:0000269|PubMed:11418466, ECO:0000269|PubMed:11675338, ECO:0000269|PubMed:11809723). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombositopeni-Radius Yokluğu (TAR) Sendromu, 274000, Otozomal Resesif; TAR (Trombositopeni-Radius Yokluğu Sendromu) (2200)	Thrombocytopenia-absent radius syndrome, 274000, Autosomal recessive; TAR (Thrombocytopenia-absent radius syndrome) (440)	RBM8A	RBM8A, RBM8B, TAR, C1DEL q21.1, DEL1q21.1	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal		EDTA Blood Sample and Heparinized blood sample
Trombositopeni, Konjenital Amegakaryositik, 604498, Otozomal Resesif (Konjenital Amegakaryositik Trombositopeni) (MPL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, congenital amegakaryocytic, 604498, Autosomal recessive (Congenital amegakaryocytic thrombocytopenia) (MPL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPL	MPL, TPOR, MPLV, THCYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Myelofibrosis with myeloid metaplasia (MMM) [MIM:254450]: A chronic myeloproliferative disorder characterized by replacement of the bone marrow by fibrous tissue, extramedullary hematopoiesis, anemia, leukoerythroblastosis and hepatosplenomegaly. (ECO:0000269|PubMed:16834459, ECO:0000269|PubMed:16868251). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni Neonatal Alloimmün (Glanzmann Trombastenisi) (ITGB3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, neonatal alloimmune (Glanzmann thrombasthenia) (ITGB3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGB3	ITGB3, GP3A, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (BDPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. (ECO:0000269	PubMed:18065693). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni, Neonatal Alloimmün, BAK Antijeni İle Bağlantılı (ITGA2B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, neonatal alloimmune, BAK antigen related (ITGA2B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ITGA2B	ITGA2B, GP2B, CD41B, GT, BDPLT2, BDPLT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bleeding disorder, platelet-type 16 (BDPLT16) [MIM:187800]: An autosomal dominant form of congenital macrothrombocytopenia associated with platelet anisocytosis. It is a disorder of platelet production. Affected individuals may have no or only mildly increased bleeding tendency. In vitro studies show mild platelet functional abnormalities. (ECO:0000269|PubMed:21454453, ECO:0000269|PubMed:9834222). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni, X'e Bağlı, 313900, X'e Bağlı Resesif; THC1 (Kalıtsal Trombositopeni-Normal Trombositler) (WAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, X-linked, 313900, X-linked recessive; THC1 (Hereditary thrombocytopenia with normal platelets) (WAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital, X-linked (XLN) [MIM:300299]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. (ECO:0000269	PubMed:11242115). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni, X'e Bağlı, İntermittant, 313900, X'e Bağlı Resesif (X' e Bağlı Trombositopeni-Normal Trombositler) (WAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, X-linked, intermittent, 313900, X-linked recessive (X-linked thrombocytopenia with normal platelets) (WAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Neutropenia, severe congenital, X-linked (XLN) [MIM:300299]: A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below 0.5 x 10(9)/l and early onset of severe bacterial infections. (ECO:0000269	PubMed:11242115). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombositopeni, X'e Bağlı, ± Diseritropoietik Anemi, 300367, X'e Bağlı Resesif; XLTDA (Trombositopeni-Konjenital Diseritropoietik Anemi) (GATA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenia, X-linked, with or without dyserythropoietic anemia, 300367, X-linked recessive; XLTDA (Thrombocytopenia with congenital dyserythropoietic anemia) (GATA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA1	GATA1, GF1, ERYF1, NFE1, XLTDA, XLTT, XLANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: X-linked dyserythropoietic anemia and thrombocytopenia (XDAT) [MIM:300367]: Disorder characterized by erythrocytes with abnormal size and shape, and paucity of platelets in peripheral blood. The bone marrow contains abundant and abnormally small megakaryocytes. (ECO:0000269|PubMed:10700180, ECO:0000269|PubMed:11418466, ECO:0000269|PubMed:11675338, ECO:0000269|PubMed:11809723). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Trombositopenik Purpura, Otoimmün, 188030, Otozomal Dominant, AITP (İmmün Trombositopenik Purpura) (FCGR2C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombocytopenic purpura, autoimmune, 188030, Autosomal dominant; AITP (immune thrombocytopenic purpura) (FCGR2C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FCGR2C	FCGR2C, CD32C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tromboembolizm, Yatkınlık, 188050, Otozomal Dominant (MTHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thromboembolism, susceptibility to, 188050, Autosomal dominant (MTHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679844, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tromboembolizm, Yatkınlık, 188050, Otozomal Dominant (MLPA)	Thromboembolism, susceptibility to, 188050, Autosomal dominant (MLPA)	MTHFR	MTHFR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679844, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombofili, Aktive Protein C Direncine Bağlı, 188055, Otozomal Dominant; THPH2 (F5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to activated protein C resistance, 188055, Autosomal dominant; THPH2 (F5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to activated protein C resistance (THPH2) [MIM:188055]: A homostatic disorder due to defective degradation of factor V by activated protein C. It is characterized by a poor anticoagulant response to activated protein C resulting in tendency to thrombosis. {ECO:0000269|PubMed:11435304, ECO:0000269|PubMed:11858490, ECO:0000269|PubMed:14617013, ECO:0000269|PubMed:9454742}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombofili, Antitrombin 3 Eksikliğine Bağlı 613118, Otozomal Resesif, Otozomal Dominant; AT3D (Kalıtsal Trombofili, Konjenital Antitrombin Eksikliğine Bağlı) (SERPINC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to antithrombin III deficiency, 613118, Autosomal recessive, Autosomal dominant; AT3D (Hereditary thrombophilia due to congenital antithrombin deficiency) (SERPINC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINC1	SERPINC1, AT3, AT3D, THPH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Antithrombin III deficiency (AT3D) [MIM:613118]: An important risk factor for hereditary thrombophilia, a hemostatic disorder characterized by a tendency to recurrent thrombosis. Antithrombin-III deficiency is classified into 4 types. Type I: characterized by a 50% decrease in antigenic and functional levels. Type II: has defects affecting the thrombin-binding domain. Type III: alteration of the heparin-binding domain. Plasma AT-III antigen levels are normal in type II and III. Type IV: consists of miscellaneous group of unclassifiable mutations. {ECO:0000269|PubMed:10997998, ECO:0000269|PubMed:11713457, ECO:0000269|PubMed:11794707, ECO:0000269|PubMed:12253073, ECO:0000269|PubMed:12595305, ECO:0000269|PubMed:12894857, ECO:0000269|PubMed:15164384, ECO:0000269|PubMed:1547341, ECO:0000269|PubMed:1555650, ECO:0000269|PubMed:16908819, ECO:0000269|PubMed:1906811, ECO:0000269|PubMed:2229057, ECO:0000269|PubMed:2365065, ECO:0000269|PubMed:23910795, ECO:0000269|PubMed:2781509, ECO:0000269|PubMed:3080419, ECO:0000269|PubMed:3162733, ECO:0000269|PubMed:3179438, ECO:0000269|PubMed:3191114, ECO:0000269|PubMed:3805013, ECO:0000269|PubMed:6582486, ECO:0000269|PubMed:7734359, ECO:0000269|PubMed:7932182	EDTA Blood Sample
Trombofili, Artmış HRG Nedeniyle, 613116, Otozomal Dominant (HRG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to elevated HRG, 613116, Autosomal dominant (HRG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRG	HRG, THPH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to histidine-rich glycoprotein deficiency (THPH11) [MIM:613116]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269|PubMed:11057869, ECO:0000269|PubMed:9414276}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombofili Heparin Kofaktörü 2 Eksikliği, 612356, Otozomal Dominant (SERPIND1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to heparin cofactor II deficiency, 612356, Autosomal dominant (SERPIND1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPIND1	HCF2, HC2, SERPIND1, THPH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to heparin cofactor 2 deficiency (THPH10) [MIM:612356]: A hemostatic disorder characterized by a tendency to recurrent thrombosis. {ECO:0000269|PubMed:10391209, ECO:0000269|PubMed:11204559, ECO:0000269|PubMed:15337701, ECO:0000269|PubMed:2647747}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombofili HRG Eksikliğine Bağlı, 613116, Otozomal Dominant; THPH11 (Kalıtsal Trombofili, Konjenital Histidin Açısından Zengin (Poli-L) Glikoprotein Eksikliğine Bağlı) (HRG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to HRG deficiency, 613116, Autosomal dominant; THPH11 (Hereditary thrombophilia due to congenital histidine-rich (poly-L) glycoprotein deficiency) (HRG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRG	HRG, THPH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to histidine-rich glycoprotein deficiency (THPH11) [MIM:613116]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269|PubMed:11057869, ECO:0000269|PubMed:9414276}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombofili, Protein C Eksikliğine Bağlı, Otozomal Dominant 176860, Otozomal Dominant (Kalıtsal Trombofili, Konjenital Protein C Yetersizliğine Bağlı) (PROC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to protein C deficiency, autosomal dominant, 176860, Autosomal dominant; THPH3 (Hereditary thrombophilia due to congenital protein C deficiency) (PROC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROC	PROC, PC, THPH3, THPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to protein C deficiency, autosomal dominant (THPH3) [MIM:176860]: A hemostatic disorder characterized by impaired regulation of blood coagulation and a tendency to recurrent venous thrombosis. Individuals with decreased amounts of protein C are classically referred to as having type I protein C deficiency and those with normal amounts of a functionally defective protein as having type II deficiency. {ECO:0000269|PubMed:1301959, ECO:0000269|PubMed:1347706, ECO:0000269|PubMed:1511989, ECO:0000269|PubMed:1868249, ECO:0000269|PubMed:2437584, ECO:0000269|PubMed:25618265, ECO:0000269|PubMed:25748729, ECO:0000269|PubMed:2602169, ECO:0000269|PubMed:7792728, ECO:0000269|PubMed:7865674, ECO:0000269|PubMed:8292730, ECO:0000269|PubMed:8398832, ECO:0000269|PubMed:8499568, ECO:0000269|PubMed:8560401, ECO:0000269|PubMed:8829639, ECO:0000269|PubMed:9798967}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombofili, Protein C Eksikliğine Bağlı, Otozomal Resesif, 612304, Otozomal Resesif; THPH4 (Kalıtsal Trombofili, Konjenital Protein C Yetersizliğine Bağlı) (PROC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to protein C deficiency, autosomal recessive, 612304, Autosomal recessive; THPH4 (Hereditary thrombophilia due to congenital protein C deficiency) (PROC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROC	PROC, PC, THPH3, THPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to protein C deficiency, autosomal dominant (THPH3) [MIM:176860]: A hemostatic disorder characterized by impaired regulation of blood coagulation and a tendency to recurrent venous thrombosis. Individuals with decreased amounts of protein C are classically referred to as having type I protein C deficiency and those with normal amounts of a functionally defective protein as having type II deficiency. {ECO:0000269|PubMed:1301959, ECO:0000269|PubMed:1347706, ECO:0000269|PubMed:1511989, ECO:0000269|PubMed:1868249, ECO:0000269|PubMed:2437584, ECO:0000269|PubMed:25618265, ECO:0000269|PubMed:25748729, ECO:0000269|PubMed:2602169, ECO:0000269|PubMed:7792728, ECO:0000269|PubMed:7865674, ECO:0000269|PubMed:8292730, ECO:0000269|PubMed:8398832, ECO:0000269|PubMed:8499568, ECO:0000269|PubMed:8560401, ECO:0000269|PubMed:8829639, ECO:0000269|PubMed:9798967}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombofili, Protein S Eksikliği, Otozomal Dominant 612336, Otozomal Dominant; THPH5 (Kalıtsal Trombofili, Konjenital Protein S Eksikliğine Bağlı) (PROS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to protein S deficiency, autosomal dominant, 612336, Autosomal dominant; THPH5 (Hereditary thrombophilia due to congenital protein S deficiency) (PROS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROS1	PROS1, THPH5, THPH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to protein S deficiency, autosomal dominant (THPH5) [MIM:612336]: A hemostatic disorder characterized by impaired regulation of blood coagulation and a tendency to recurrent venous thrombosis. Based on the plasma levels of total and free PROS1 as well as the serine protease-activated protein C cofactor activity, three types of THPH5 have been described: type I, characterized by reduced total and free PROS1 levels together with reduced anticoagulant activity; type II, in which only free PROS1 antigen and PROS1 activity levels are reduced; and the rare type II which is characterized by normal concentrations of both total and free PROS1 antigen, but low cofactor activity. {ECO:0000269|PubMed:10447256, ECO:0000269|PubMed:10613647, ECO:0000269|PubMed:10706858, ECO:0000269|PubMed:10790208, ECO:0000269|PubMed:11372770, ECO:0000269|PubMed:11776305, ECO:0000269|PubMed:11858485, ECO:0000269|PubMed:11927129, ECO:0000269|PubMed:12351389, ECO:0000269|PubMed:12632031, ECO:0000269|PubMed:15238143, ECO:0000269|PubMed:15712227, ECO:0000269|PubMed:7482398, ECO:0000269|PubMed:7545463, ECO:0000269|PubMed:7579449, ECO:0000269|PubMed:7803790, ECO:0000269|PubMed:8298131, ECO:0000269|PubMed:8639833, ECO:0000269|PubMed:8701404, ECO:0000269|PubMed:8765219, ECO:0000269|PubMed:8781426	EDTA Blood Sample

Trombofili, Protein S Eksikliğine Bağlı, Otozomal Resesif, 614514, Otozomal Resesif; THPH6 (Kalıtsal Trombofili, Konjenital Protein S Eksikliğine Bağlı) (PROS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to protein S deficiency, autosomal recessive, 614514, Autosomal recessive; THPH6 (Hereditary thrombophilia due to congenital protein S deficiency) (PROS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PROS1	PROS1, THPH5, THPH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to protein S deficiency, autosomal dominant (THPH5) [MIM:612336]: A hemostatic disorder characterized by impaired regulation of blood coagulation and a tendency to recurrent venous thrombosis. Based on the plasma levels of total and free PROS1 as well as the serine protease-activated protein C cofactor activity, three types of THPH5 have been described: type I, characterized by reduced total and free PROS1 levels together with reduced anticoagulant activity; type II, in which only free PROS1 antigen and PROS1 activity levels are reduced; and the rare type III which is characterized by normal concentrations of both total and free PROS1 antigen, but low cofactor activity. {ECO:0000269|PubMed:10447256, ECO:0000269|PubMed:10613647, ECO:0000269|PubMed:10706858, ECO:0000269|PubMed:10792008, ECO:0000269|PubMed:11372770, ECO:0000269|PubMed:11776305, ECO:0000269|PubMed:11858485, ECO:0000269|PubMed:11927129, ECO:0000269|PubMed:12351389, ECO:0000269|PubMed:12632031, ECO:0000269|PubMed:15238143, ECO:0000269|PubMed:15712227, ECO:0000269|PubMed:7482398, ECO:0000269|PubMed:7545463, ECO:0000269|PubMed:7579449, ECO:0000269|PubMed:7803790, ECO:0000269|PubMed:8298131, ECO:0000269|PubMed:8639833, ECO:0000269|PubMed:8701404, ECO:0000269|PubMed:8765219, ECO:0000269|PubMed:8781426.	EDTA Blood Sample
Trombofili, Trombin Defektine Bağlı, 188050, Otozomal Dominant; THPH1 (F2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to thrombin defect, 188050, Autosomal dominant; THPH1 (F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F2	F2, THPH1, RPRGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to thrombin defect (THPH1) [MIM:188050]: A multifactorial disorder of hemostasis characterized by abnormal platelet aggregation in response to various agents and recurrent thrombi formation. {ECO:0000269	PubMed:2825773}. Note=The disease is caused by mutations affecting the gene represented in this entry. A common genetic variation in the 3-prime untranslated region of the prothrombin gene is associated with elevated plasma prothrombin levels and an increased risk of venous thrombosis.	EDTA Blood Sample
Trombofili, Trombomodulin Defektine Bağlı, 614488, THPH12 (Trombomodulin İlişkili Kanama Bozukluğu) (THBD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia due to thrombomodulin defect, 614488, THPH12 (Thrombomodulin-related bleeding disorder) (THBD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THBD	THBD, THRM, AHUS6, THPH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hemolytic uremic syndrome atypical 6 (AHUS6) [MIM:612926]: An atypical form of hemolytic uremic syndrome. It is a complex genetic disease characterized by microangiopathic hemolytic anemia, thrombocytopenia, renal failure and absence of episodes of enterocolitis and diarrhea. In contrast to typical hemolytic uremic syndrome, atypical forms have a poorer prognosis, with higher death rates and frequent progression to end-stage renal disease. {ECO:0000269|PubMed:19625716, ECO:0000269|PubMed:20513133}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Other genes may play a role in modifying the phenotype.;	EDTA Blood Sample
Trombofili, Allesel, Azalmış PLAT Salınımına Bağlı, 612348 (PLAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia, familial, due to decreased release of PLAT, 612348 (PLAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PLAT	PLAT, TPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Increased activity of TPA results in increased fibrinolysis of fibrin blood clots that is associated with excessive bleeding. Defective release of TPA results in hypofibrinolysis that can lead to thrombosis or embolism. {ECO:0000269|PubMed:17621441}.	EDTA Blood Sample
Trombofili, Yatkınlık, Faktör 5 Leiden'e Bağlı, 188055, Otozomal Dominant (F5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia, susceptibility to, due to factor V Leiden, 188055, Autosomal dominant (F5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F5	F5, THPH2, RPRGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia due to activated protein C resistance (THPH2) [MIM:188055]: A hemostatic disorder due to defective degradation of factor V by activated protein C. It is characterized by a poor anticoagulant response to activated protein C resulting in tendency to thrombosis. {ECO:0000269|PubMed:11435304, ECO:0000269|PubMed:11858490, ECO:0000269|PubMed:14617013, ECO:0000269|PubMed:9454742}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trombofili, X'e Bağlı, Faktör 9 Defekti Nedeniyle, 300807; THPH8 (F9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombophilia, X-linked, due to factor IX defect, 300807; THPH8 (F9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F9	F9, HEMB, THPH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombophilia, X-linked, due to factor IX defect (THPH8) [MIM:300807]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269|PubMed:19846852}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trombofili, X'e Bağlı, Faktör 9 Defekti Nedeniyle, 300807, THPH8 (MLPA)	Thrombophilia, X-linked, due to factor IX defect, 300807, THPH8 (MLPA)	F9	F9, HEMB, THPH8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Thrombophilia, X-linked, due to factor IX defect (THPH8) [MIM:300807]: A hemostatic disorder characterized by a tendency to thrombosis. {ECO:0000269|PubMed:19846852}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tromboz, Hiperhomosisteinemik, 236200, Otozomal Resesif (Klasik Homosistinüri) (CBS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombosis, hyperhomocysteinemic, 236200, Autosomal recessive (Classic homocystinuria) (CBS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CBS	CBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trombotik Trombositopenik Purpura, Allesel, 274150, Otozomal Resesif; TTP (Trombotik Trombositopenik Purpura) (ADAMTS13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thrombotic thrombocytopenic purpura, familial, 274150, Autosomal recessive; TTP (Thrombotic thrombocytopenic purpura) (ADAMTS13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTS13	ADAMTS13, VWFCP, TTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thrombotic thrombocytopenic purpura congenital (TTP) [MIM:274150]: A hematologic disease characterized by hemolytic anemia with fragmentation of erythrocytes, thrombocytopenia, diffuse and non-focal neurologic findings, decreased renal function and fever. recessive. {ECO:0000269|PubMed:11586351, ECO:0000269|PubMed:12181489, ECO:0000269|PubMed:12393505, ECO:0000269|PubMed:12614216, ECO:0000269|PubMed:12753286, ECO:0000269|PubMed:14512317, ECO:0000269|PubMed:14563640, ECO:0000269|PubMed:15009458, ECO:0000269|PubMed:15126318, ECO:0000269|PubMed:15327386, ECO:0000269|PubMed:16449289, ECO:0000269|PubMed:16453338, ECO:0000269|PubMed:16796708, ECO:0000269|PubMed:16807643, ECO:0000269|PubMed:17003922, ECO:0000269|PubMed:18443791, ECO:0000269|PubMed:19055667, ECO:0000269|PubMed:19116307, ECO:0000269|PubMed:22075512}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tromboksan Sentaz Eksikliği, 614158, Otozomal Dominant (TBXAS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thromboxane synthase deficiency, 614158, Autosomal dominant (TBXAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBXAS1	TBXAS1, GHOSAL, CYP5, BDPLT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Thromboxane synthetase deficiency has been detected in some patients with a bleeding disorder due to platelet dysfunction. {ECO:0000269|PubMed:6101498}.;	EDTA Blood Sample
Tiroid Adenomu, Hiperfonksiyone, Somatik (TSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid adenoma, hyperfunctioning, somatic (TSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHR	TSHR, CHNG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. {ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15331543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Biopsy of affected site-transport media
Tiroid Kanseri, Medüller Olmayan, 1, 188550, Otozomal Dominant; NMTC1 (Diferansiye Tiroid Kanseri) (NKX2-1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid cancer, monmedullary, 1, 188550, Autosomal dominant; NMTC1 (Differentiated thyroid carcinoma) (NKX2-1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Chorea, hereditary benign (BHC) [MIM:118700]: A rare autosomal dominant movement disorder, defined by early onset in childhood, a stable or non-progressive course of chorea, and no mental deterioration. Chorea is characterized by involuntary, forcible, rapid, jerky movements that may be subtle or become confluent, markedly altering normal patterns of movement. {ECO:0000269|PubMed:11971878, ECO:0000269|PubMed:15955952}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiroid Kanseri, Medüller Olmayan, 1, 188550, Otozomal Dominant; NMTC1 (Diferansiye Tiroid Kanseri) (MLPA)	Thyroid cancer, monmedullary, 1, 188550, Autosomal dominant; NMTC1 (Differentiated thyroid carcinoma) (MLPA)	NKX2-1	NKX2-1, TITF1, NKX2A, TTF1, NMTC1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Chorea, hereditary benign (BHC) [MIM:118700]: A rare autosomal dominant movement disorder, defined by early onset in childhood, a stable or non-progressive course of chorea, and no mental deterioration. Chorea is characterized by involuntary, forcible, rapid, jerky movements that may be subtle or become confluent, markedly altering normal patterns of movement. {ECO:0000269|PubMed:11971878, ECO:0000269|PubMed:15955952}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiroid Kanseri, Medüller Olmayan, 2, 188470, Otozomal Dominant; NMTC2 (Allesel Papiller veya Foliküler Tiroid Karsinomu) (SRGAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid cancer, nonmedullary, 2, 188470, Autosomal dominant; NMTC2 (Familial papillary or follicular thyroid carcinoma) (SRGAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SRGAP1	SRGAP1, KIAA1304, NMTC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Kanseri, Medüller Olmayan, 4, 616534, Otozomal Dominant; NMTC4 (Allesel Papiller veya Foliküler Tiroid Karsinomu) (FOXE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid cancer, nonmedullary, 4, 616534, Autosomal dominant; NMTC4 (Familial papillary or follicular thyroid carcinoma) (FOXE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. {ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Kanseri, Medüller Olmayan, 4, 616534, Otozomal Dominant; NMTC4 (Allesel Papiller veya Foliküler Tiroid Karsinomu) (MLPA)	Thyroid cancer, nonmedullary, 4, 616534, Autosomal dominant; NMTC4 (Familial papillary or follicular thyroid carcinoma) (MLPA)	FOXE1	FOXE1, FKHL15, TITF2, TTF2, NMTC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bamforth-Lazarus syndrome (BLS) [MIM:241850]: A disease characterized by thyroid agenesis, cleft palate and choanal atresia. {ECO:0000269|PubMed:12165566, ECO:0000269|PubMed:9697705}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Tiroid Kanseri, Medüller Olmayan, 5, 616535, Otozomal Dominant; NMTC5 (Ailesel Papiller veya Foliküler Tiroid Karsinomu) (HABP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid cancer, nonmedullary, 5, 616535, Autosomal dominant; NMTC5 (Familial papillary or follicular thyroid carcinoma) (HABP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HABP2	HABP2, PHBP, HGFAL, FSAP, NMTC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Karsinoma-Tirotoksikoz (TSHR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid carcinoma with thyrotoxicosis (TSHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSHR	TSHR, CHNG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. (ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiroid Karsinoma-Tirotoksikoz (MLPA)	Thyroid carcinoma with thyrotoxicosis (MLPA)	TSHR	TSHR, CHNG1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypothyroidism, congenital, non-goitrous, 1 (CHNG1) [MIM:275200]: A non-autoimmune condition characterized by resistance to thyroid-stimulating hormone (TSH) leading to increased levels of plasma TSH and low levels of thyroid hormone. It presents variable severity depending on the completeness of the defect. Most patients are euthyroid and asymptomatic, with a normal sized thyroid gland. Only a subset of patients develop hypothyroidism and present a hypoplastic thyroid gland. (ECO:0000269|PubMed:10720030, ECO:0000269|PubMed:11095460, ECO:0000269|PubMed:11442002, ECO:0000269|PubMed:12050212, ECO:0000269|PubMed:14725684, ECO:0000269|PubMed:15531543, ECO:0000269|PubMed:7528344, ECO:0000269|PubMed:8954020, ECO:0000269|PubMed:9100579, ECO:0000269|PubMed:9185526, ECO:0000269|PubMed:9329388). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiroid Karsinomu, Foliküler, 188470, Otozomal Dominant (Ailesel Papiller veya Foliküler Tiroid Karsinomu) (MINPP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid carcinoma, follicular, 188470, Autosomal dominant (Familial papillary or follicular thyroid carcinoma) (MINPP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MINPP1	MINPP1, HIPER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Karsinomu, Foliküler, Somatik, 188470 (Ailesel Papiller veya Folliküler Tiroid Karsinomu) (NRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid carcinoma, follicular, somatic, 188470 (Familial papillary or follicular thyroid carcinoma) (NRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NRAS	NRAS, ALPS4, NS6, CMNS, NCMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Tiroid Karsinomu, Foliküler, Somatik, 188470 (Ailesel Papiller veya Folliküler Tiroid Karsinomu) (HRAS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid carcinoma, follicular, somatic, 188470 (Familial papillary or follicular thyroid carcinoma) (HRAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HRAS	HRAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Etiklenmiş bölgeden alınan biyopsi-Transport besi yeri içinde		30	120	.	.	60	Postnatal	.	Biopsy of affected site-transport media
Tiroid Karsinomu, Hurthle Hücreli, 607464 (Diferansiye Tiroid Kanseri) (NDUFA13 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid carcinoma, Hurthle cell, 607464 (Differentiated thyroid carcinoma) (NDUFA13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NDUFA13	NDUFA13, GRIM19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hurthle cell thyroid carcinoma (HCTC) [MIM:607464]: A rare type of thyroid cancer accounting for only about 3-10% of all differentiated thyroid cancers. These neoplasms are considered a variant of follicular carcinoma of the thyroid and are referred to as follicular carcinoma, oxyphilic type. (ECO:0000269	PubMed:15841082). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry..	EDTA Blood Sample
Tiroid disgenezisi paneli (TPO, PAX8, FOXE1, NKX2-1, TSHR) (MLPA)	Thyroid dysgenesis panel (TPO, PAX8, FOXE1, NKX2-1, TSHR) (MLPA)	TPO, PAX8, FOXE1, NKX2-1, TSHR	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Tiroid Dishormonogenezis 1, 274400, Otozomal Resesif; TDH1 (Ailesel Tiroid Dishormonogenezisi) (SLC5A5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 1, 274400, Autosomal recessive; TDH1 (Familial thyroid dyshormonogenesis) (SLC5A5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC5A5	SLC5A5, NIS, TDH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Dishormonogenezis 2A, 274500, Otozomal Resesif; TDH2A (Ailesel Tiroid Dishormonogenezisi) (TPO geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 2A, 274500, Autosomal recessive; TDH2A (Familial thyroid dyshormonogenesis) (TPO gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPO	TPO, TPX, TDH2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroid dyshormonogenesis 2A (TDH2A) [MIM:274500]: A disorder due to defective conversion of accumulated iodide to organically bound iodine. The iodide organification defect can be partial or complete. (ECO:0000269|PubMed:10084596, ECO:0000269|PubMed:10468986, ECO:0000269|PubMed:11061528, ECO:0000269|PubMed:11415848, ECO:0000269|PubMed:11874711, ECO:0000269|PubMed:11916616, ECO:0000269|PubMed:12213873, ECO:0000269|PubMed:12490071, ECO:0000269|PubMed:12843174, ECO:0000269|PubMed:12864797, ECO:0000269|PubMed:12938097, ECO:0000269|PubMed:16284446, ECO:0000269|PubMed:16684826, ECO:0000269|PubMed:7550241, ECO:0000269|PubMed:9024270, ECO:0000269|PubMed:9924196). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Dishormonogenezis 2A, 274500, Otozomal Resesif; TDH2A (Ailesel Tiroid Dishormonogenezisi) (MLPA)	Thyroid dyshormonogenesis 2A, 274500, Autosomal recessive; TDH2A (Familial thyroid dyshormonogenesis) (MLPA)	TPO	TPO, TPX, TDH2A	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Thyroid dyshormonogenesis 2A (TDH2A) [MIM:274500]: A disorder due to defective conversion of accumulated iodide to organically bound iodine. The iodide organification defect can be partial or complete. (ECO:0000269|PubMed:10084596, ECO:0000269|PubMed:10468986, ECO:0000269|PubMed:11061528, ECO:0000269|PubMed:11415848, ECO:0000269|PubMed:11874711, ECO:0000269|PubMed:11916616, ECO:0000269|PubMed:12213873, ECO:0000269|PubMed:12490071, ECO:0000269|PubMed:12843174, ECO:0000269|PubMed:12864797, ECO:0000269|PubMed:12938097, ECO:0000269|PubMed:16284446, ECO:0000269|PubMed:16684826, ECO:0000269|PubMed:7550241, ECO:0000269|PubMed:9024270, ECO:0000269|PubMed:9924196). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Dishormonogenezis 3, 274700, Otozomal Resesif; TDH3 (Ailesel Tiroid Dishormonogenezisi) (TG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 3, 274700, Autosomal recessive; TDH3 (Familial thyroid dyshormonogenesis) (TG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TG	TG, AITD3, TDH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroid dyshormonogenesis 3 (TDH3) [MIM:274700]: A disorder due to thyroid dyshormonogenesis, causing large goiters of elastic and soft consistency in the majority of patients. Although the degree of thyroid dysfunction varies considerably among patients with defective thyroglobulin synthesis, patients usually have a relatively high serum free triiodothyronine (T3) concentration with disproportionately low free tetraiodothyronine (T4) level. The maintenance of relatively high free T3 levels prevents profound tissue hypothyroidism except in brain and pituitary, which are dependent on T4 supply, resulting in neurologic and intellectual defects in some cases. (ECO:0000269|PubMed:10199792, ECO:0000269|PubMed:16477365, ECO:0000269|PubMed:17244789, ECO:0000269|PubMed:17532758, ECO:0000269|PubMed:19509106). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tiroid Dishormonogenezis 4, 274800, Otozomal Resesif; TDH4 (Ailesel Tiroid Dishormonogenezisi) (IYD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 4, 274800, Autosomal recessive; TDH4 (Familial thyroid dyshormonogenesis) (IYD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IYD	IYD, DEHAL1, TDH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Dishormonogenezis 5, 274900, Otozomal Resesif; TDH5 (Ailesel Tiroid Dishormonogenezisi) (DUOXA2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 5, 274900, Autosomal recessive; TDH5 (Familial thyroid dyshormonogenesis) (DUOXA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DUOXA2	DUOXA2, TDH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroid dyshormonogenesis 5 (TDH5) [MIM:274900]: A disorder due to thyroid dyshormonogenesis, causing hypothyroidism, goiter, and variable mental deficits derived from unrecognized and untreated hypothyroidism. (ECO:0000269|PubMed:18042646, ECO:0000269|PubMed:25675383). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Dishormonogenezis 6, 607200, Otozomal Resesif; TDH6 (Ailesel Tiroid Dishormonogenezisi) (DUOX2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid dyshormonogenesis 6, 607200, Autosomal recessive; TDH6 (Familial thyroid dyshormonogenesis) (DUOX2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DUOX2	DUOX2, THOX2, TDH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyroid dyshormonogenesis 6 (TDH6) [MIM:607200]: A disorder due to a defective conversion of accumulated iodide to organically bound iodine. The iodide organification defect can be partial or complete. (ECO:0000269|PubMed:12110737, ECO:0000269|PubMed:16134168, ECO:0000269|PubMed:16322276, ECO:0000269|PubMed:20187165). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Hormon Metabolizması, Anormal, 609698 (Kısa Boy-Gecikmiş Kemik Yaşı, Tiroid Hormon Metabolizması Eksikliğine Bağlı) (SECISBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid hormone metabolism, abnormal, 609698 (Short stature-delayed bone age due to thyroid hormone metabolism deficiency) (SECISBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SECISBP2	SECISBP2, SBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tiroid Hormonu Direnci, 188570, Otozomal Dominant; GRTH (Jeneralize Tiroid Hormon Direnci) (THRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid hormone resistance, 188570, Autosomal dominant; GRTH (Generalized resistance to thyroid hormone) (THRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THRB	THRB, ERBA2, THR1, PRTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Selective pituitary thyroid hormone resistance (PRTH) [MIM:145650]: Variant form of thyroid hormone resistance and is characterized by clinical hyperthyroidism, with elevated free thyroid hormones, but inappropriately normal serum TSH. Unlike GRTH, where the syndrome usually segregates with a dominant allele, the mode of inheritance in PRTH has not been established. (ECO:0000269|PubMed:7528740, ECO:0000269|PubMed:8381821). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Tiroid Hormonu Direnci, Otozomal Resesif, 274300, Otozomal Resesif; GRTH (Jeneralize Tiroid Hormon Direnci) (THRB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid hormone resistance, autosomal recessive, 274300, Autosomal recessive; GRTH (Generalized resistance to thyroid hormone) (THRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THRB	THRB, ERBA2, THR1, PRTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Selective pituitary thyroid hormone resistance (PRTH) [MIM:145650]: Variant form of thyroid hormone resistance and is characterized by clinical hyperthyroidism, with elevated free thyroid hormones, but inappropriately normal serum TSH. Unlike GRTH, where the syndrome usually segregates with a dominant allele, the mode of inheritance in PRTH has not been established. {ECO:0000269|PubMed:7528740, ECO:0000269|PubMed:8381821}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tiroid Hormonu Direnci, Selektif Hipofiz, 145650, Otozomal Dominant; PRTH (Tiroid Hormonuna Karşı Seçici Hipofizer Direnç) (THRB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thyroid hormone resistance, selective pituitary, 145650, Autosomal dominant; PRTH (Selective pituitary resistance to thyroid hormone) (THRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	THRB	THRB, ERBA2, THR1, PRTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Selective pituitary thyroid hormone resistance (PRTH) [MIM:145650]: Variant form of thyroid hormone resistance and is characterized by clinical hyperthyroidism, with elevated free thyroid hormones, but inappropriately normal serum TSH. Unlike GRTH, where the syndrome usually segregates with a dominant allele, the mode of inheritance in PRTH has not been established. {ECO:0000269|PubMed:7528740, ECO:0000269|PubMed:8381821}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tirotoksik Periodik Paraliz, Yatkınlık, 1, 1, Yatkınlık, 188580, İzole vakalar; TTPP1 (Tirotoksik Periodik Paraliz) (CACNA1S gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thyrotoxic periodic paralysis, susceptibility to, 1, 188580, Isolated cases; TTPP1 (Thyrotoxic periodic paralysis) (CACNA1S gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tirotoksik Periodik Paraliz, Yatkınlık, 1, 1, Yatkınlık, 188580, İzole vakalar; TTPP1 (Tirotoksik Periodik Paraliz) (MLPA)	Thyrotoxic periodic paralysis, susceptibility to, 1, 188580, Isolated cases; TTPP1 (Thyrotoxic periodic paralysis) (MLPA)	CACNA1S	CACNA1S, CACNL1A3, CCHL1A3, TTPP1, HOKPP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Periodic paralysis hypokalemic 1 (HOKPP1) [MIM:170400]: An autosomal dominant disorder manifested by episodic flaccid generalized muscle weakness associated with falls of serum potassium levels. {ECO:0000269|PubMed:17418573, ECO:0000269|PubMed:18162704, ECO:0000269|PubMed:19118277, ECO:0000269|PubMed:7987325, ECO:0000269|PubMed:8004673}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tirotoksik Periyodik Paralizi, Yatkınlık, 2, 613239, Sporadik Vakalar; TTPP2 (Tirotoksik Periodik Paralizi) (KCNJ18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Thyrotoxic periodic paralysis, susceptibility to, 2, 613239, Isolated cases; TTPP2 (Thyrotoxic periodic paralysis) (KCNJ18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ18	KCNJ18, KIR2.6, TTPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Thyrotoxic periodic paralysis 2 (TTPP2) [MIM:613239]: A sporadic muscular disorder characterized by episodic weakness and hypokalemia during a thyrotoxic state. It is clinically similar to hereditary hypokalemic periodic paralysis, except for the fact that hyperthyroidism is an absolute requirement for disease manifestation. The disease presents with recurrent episodes of acute muscular weakness of the four extremities that vary in severity from paresis to complete paralysis. Attacks are triggered by ingestion of a high carbohydrate load or strenuous physical activity followed by a period of rest. Thyrotoxic periodic paralysis can occur in association with any cause of hyperthyroidism, but is most commonly associated with Graves disease. {ECO:0000269	PubMed:20074522}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Tirotropin Salgılatan Hormon Eksikliği, 275120, Otozomal Resesif Izole Tirotropin Salgılayan Hormon Eksikliği (TRH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thyrotropin-releasing hormone deficiency, 275120, Autosomal recessive (Isolated thyrotropin-releasing hormone deficiency) (TRH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRH	TRH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tirotropin-Salgılatıcı Hormon Reseptörü; Trhr (TRHR gen) (Tüm Gen Dizi Analizi) (Postnatal)	THYROTROPIN-RELEASING HORMONE RECEPTOR; TRHR (TRHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRHR	TRHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tirotropin Salgılatan Hormon Direnci, Jeneralize (TRHR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thyrotropin-releasing hormone resistance, generalized (TRHR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRHR	TRHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tibia Yokluğu Veya Hipoplazisi-Poldaktili, Thyp (Tibia Yokluğu-Polidaktili Sendromu) (LMBR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	TIBIA, HYPOPLASIA OR APLASIA OF, WITH POLYDACTYLY; THYP (Absent tibia-polydactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159}.;	EDTA Blood Sample
Tibia Yokluğu Veya Hipoplazisi-Poldaktili, Thyp (Tibia Yokluğu-Polidaktili Sendromu) (Prenatal) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	TIBIA, HYPOPLASIA OR APLASIA OF, WITH POLYDACTYLY; THYP (Absent tibia-polydactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). {ECO:0000269|PubMed:24456159}. Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. {ECO:0000269|PubMed:24456159}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tibiyal Müsküler Distrofi, Yavaş Gelişen (Tardive), 600334, Otozomal Dominant; TMD (Tibial Müsküler Distrofi) (TTN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tibial muscular dystrophy, tardive, 600334, Autosomal dominant; TMD (Tibial muscular dystrophy) (TTN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. {ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tibiyal Müsküler Distrofi, Yavaş Gelişen (Tardive), 600334, Otozomal Dominant; TMD (Tibial Müsküler Distrofi) (Prenatal) (TTN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tibial muscular dystrophy, tardive, 600334, Autosomal dominant; TMD (Tibial muscular dystrophy) (TTN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TTN	TTN, CMD1G, TMD, LGMD2J, MPRM, HMERF, EOMFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tardive tibial muscular dystrophy (TMD) [MIM:600334]: Autosomal dominant, late-onset distal myopathy. Muscle weakness and atrophy are usually confined to the anterior compartment of the lower leg, in particular the tibialis anterior muscle. Clinical symptoms usually occur at age 35-45 years or much later. {ECO:0000269|PubMed:12145747, ECO:0000269|PubMed:12891679}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tietz Albinizm-Sağırlık Sendromu, 103500, Otozomal Dominant; TADS (Tietz Sendromu) (MITF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tietz albinism-deafness syndrome, 103500, Autosomal dominant; TADS (Tietz syndrome) (MITF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Waardenburg syndrome 2, with ocular albinism, autosomal recessive (WS2-OA) [MIM:103470]: A disorder characterized by the association of features typical of Waardenburg syndrome type 2 with ocular albinism. Patients manifest reduced visual acuity, albinotic fundus, deafness, hypomelanosis. {ECO:0000269	PubMed:9647758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tietz Albinizm-Sağırlık Sendromu, 103500, Otozomal Dominant; TADS (Tietz Sendromu) (MLPA)	Tietz albinism-deafness syndrome, 103500, Autosomal dominant; TADS (Tietz syndrome) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Waardenburg syndrome 2, with ocular albinism, autosomal recessive (WS2-OA) [MIM:103470]: A disorder characterized by the association of features typical of Waardenburg syndrome type 2 with ocular albinism. Patients manifest reduced visual acuity, albinotic fundus, deafness, hypomelanosis. {ECO:0000269	PubMed:9647758}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Timidilat Sentaz (TS) (28bp Delesyon/ insersiyon Polimorfizmi) (Dizi Analizi) (TYMS geni) (Dizi Analizi) (Postnatal)	Timidilate synthetase (28bp deletion/insertion polymorphism) (Sequence analysis) (TYMS gene) (Dizi Analizi) (Postnatal)	TYMS	TS	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Timothy Sendromu, 601005, Otozomal Dominant; TS (Timothy Sendromu) (CACNA1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Timothy syndrome, 601005, Autosomal dominant; TS (Timothy syndrome) (CACNA1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CACNA1C	CACNA1C, CACNL1A1, CCHL1A1, TS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Timothy syndrome (TS) [MIM:601005]: Disorder characterized by multiorgan dysfunction including lethal arrhythmias, webbing of fingers and toes, congenital heart disease, immune deficiency, intermittent hypoglycemia, cognitive abnormalities and autism. {ECO:0000269|PubMed:15454078, ECO:0000269|PubMed:15863812, ECO:0000269|PubMed:25260352}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tn Poliaglütinasyon Sendromu, Somatik, 300622 (C1GALT1C1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tn polyagglutination syndrome, somatic, 300622 (C1GALT1C1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	C1GALT1C1	C1GALT1C1, COSMC, C1GALT2, TNPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)/ EDTA'lı Kemik İliği (2-3 ml)		30	120	.	.	60	Postnatal	.	EDTA blood sample / EDTA bone marrow
Ayak Parmağı Sindaktilisi, Telekantus, Anogenital ve Renal Malformasyonlar; Star (Sindaktili-Telekantus-Anogenital-Renal Malformasyonlar Sendromu) (CCNQ geni) (Tüm Gen Dizi Analizi) (Postnatal)	TOE SYNDACTYLY, TELECANTHUS, AND ANOGENITAL AND RENAL MALFORMATIONS; STAR (Syndactyly-telecanthus-anogenital and renal malformations syndrome) (CCNQ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCNQ	FAM58A, STAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ayak Parmağı Sindaktilisi, Telekantus, Anogenital ve Renal Malformasyonlar; Star (Sindaktili-Telekantus-Anogenital-Renal Malformasyonlar Sendromu) (Prenatal) (CCNQ geni) (Tüm Gen Dizi Analizi) (Prenatal)	TOE SYNDACTYLY, TELECANTHUS, AND ANOGENITAL AND RENAL MALFORMATIONS; STAR (Syndactyly-telecanthus-anogenital and renal malformations syndrome) (CCNQ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CCNQ	FAM58A, STAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ayak Tırnağı Distrofisi, İzole, 607523, Otozomal Dominant (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Toenail dystrophy, isolated, 607523, Autosomal dominant (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Transient bullous dermolysis of the newborn (TBDN) [MIM:131705]: TBDN is a neonatal form of dystrophic epidermolysis bullosa characterized by sub-epidermal blisters, reduced or abnormal anchoring fibrils at the derme-epidermal junction, and electron-dense inclusions in keratinocytes. TBDN heals spontaneously or strongly improves within the first months and years of life. {ECO:0000269	PubMed:9856844}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Ayak Tırnağı Distrofisi, İzole, 607523, Otozomal Dominant (MLPA)	Toenail dystrophy, isolated, 607523, Autosomal dominant (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Transient bullous dermolysis of the newborn (TBDN) [MIM:131705]: TBDN is a neonatal form of dystrophic epidermolysis bullosa characterized by sub-epidermal blisters, reduced or abnormal anchoring fibrils at the derme-epidermal junction, and electron-dense inclusions in keratinocytes. TBDN heals spontaneously or strongly improves within the first months and years of life. {ECO:0000269	PubMed:9858644}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tolbutamid Yavaş Metabolizması (CYP2C9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tolbutamide poor metabolizer (CYP2C9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C9	CYP2C9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tolbutamid Yavaş Metabolizması (MLPA)	Tolbutamide poor metabolizer (MLPA)	CYP2C9	CYP2C9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 1, ± Veya Orofasiyal Yarık, 106600, Otozomal Dominant; STHAG1 (Oligodonti) (MSX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600, Autosomal dominant; STHAG1 (Oligodontia) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 1 (STHAG1) [MIM:106600]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). Tooth agenesis selective type 1 can be associated with orofacial cleft in some patients. {ECO:0000269|PubMed:12097313, ECO:0000269|PubMed:8696335}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 3, 604625, Otozomal Dominant; STHAG3 (Oligodonti) (PAX9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 3, 604625, Autosomal dominant; STHAG3 (Oligodontia) (PAX9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX9	PAX9, STHAG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 3 (STHAG3) [MIM:604625]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). {ECO:0000269	PubMed:12786960}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 4, 150400, Otozomal Resesif, Otozomal Dominant; STHAG4 (Oligodonti) (WNT10A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 4, 150400, Autosomal recessive, Autosomal dominant; STHAG4 (Oligodontia) (WNT10A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT10A	WNT10A, SSPS, STHAG4, OODD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tooth agenesis selective 4 (STHAG4) [MIM:150400]: A form of selective tooth agenesis, a common anomaly characterized by the congenital absence of one or more teeth. Selective tooth agenesis without associated systemic disorders has sometimes been divided into 2 types: oligodontia, defined as agenesis of 6 or more permanent teeth, and hypodontia, defined as agenesis of less than 6 teeth. The number in both cases does not include absence of third molars (wisdom teeth). In STHAG4, the upper lateral incisors are absent or peg-shaped. {ECO:0000269|PubMed:21484994, ECO:0000269|PubMed:22581971}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 7, 616724, Otozomal Dominant; STHAG7 (Oligodonti) (LRP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 7, 616724, Autosomal dominant; STHAG7 (Oligodontia) (LRP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP6	LRP6, ADCAD2, STHAG7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Coronary artery disease, autosomal dominant, 2 (ADCAD2) [MIM:610947]: A common heart disease characterized by reduced or absent blood flow in one or more of the arteries that encircle and supply the heart. Its most important complication is acute myocardial infarction. {ECO:0000269	PubMed:17332414}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 8, 617073, Otozomal Dominant; STHAG8 (Oligodonti) (WNT10B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 8, 617073, Autosomal dominant; STHAG8 (Oligodontia) (WNT10B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT10B	WNT10B, SHFM6, STHAG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Split-hand/foot malformation 6 (SHFM6) [MIM:225300]: A limb malformation involving the central rays of the autopod and presenting with syndactyly, median clefts of the hands and feet, and aplasia and/or hypoplasia of the phalanges, metacarpals, and metatarsals. Some patients have been found to have mental retardation, ectodermal and craniofacial findings, and orofacial clefting. {ECO:0000269	PubMed:18515319}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Diş Agenezisi, Seçkitif, 9, 617275, Otozomal Dominant; STHAG9 (GREM2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, 9, 617275, Autosomal dominant; STHAG9 (GREM2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GREM2	GREM2, PRDC, STHAG9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Diş Agenezisi, Seçkitif, X'e Bağlı 1, 313500, X'e Bağlı Dominant; STHAGX1 (Oligodonti) (EDA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tooth agenesis, selective, X-linked 1, 313500, X-linked dominant; STHAGX1 (Oligodontia) (EDA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDA	EDA, ED1, ECTD1, EDA, HED1, STHAGX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ectodermal dysplasia 1, hypohidrotic, X-linked (XHED) [MIM:305100]: A form of ectodermal dysplasia, a heterogeneous group of disorders due to abnormal development of two or more ectodermal structures. Characterized by sparse hair (atrichosis or hypotrichosis), abnormal or missing teeth and the inability to sweat due to the absence of sweat glands. It is the most common form of over 150 clinically distinct ectodermal dysplasias. {ECO:0000269|PubMed:10469321, ECO:0000269|PubMed:10951256, ECO:0000269|PubMed:11279189, ECO:0000269|PubMed:11295832, ECO:0000269|PubMed:11343303, ECO:0000269|PubMed:11378824, ECO:0000269|PubMed:12225002, ECO:0000269|PubMed:12932274, ECO:0000269|PubMed:17256800, ECO:0000269|PubMed:18231121, ECO:0000269|PubMed:19127222, ECO:0000269|PubMed:19438931, ECO:0000269|PubMed:20486090, ECO:0000269|PubMed:20979233, ECO:0000269|PubMed:22008666, ECO:0000269|PubMed:22350046, ECO:0000269|PubMed:8696334, ECO:0000269|PubMed:9507389, ECO:0000269|PubMed:9630076, ECO:0000269|PubMed:9683615, ECO:0000269|PubMed:9736768}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tourette Sendromu, 137580, Otozomal Dominant (SLITRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tourette syndrome, 137580, Autosomal dominant (SLITRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLITRK1	SLITRK1, KIAA1910, TTM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gilles de la Tourette syndrome (GTS) [MIM:137580]: Neurologic disorder manifested particularly by motor and vocal tics and associated with behavioral abnormalities. {ECO:0000269	PubMed:16234024}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Townes-Brocks Brankiootorenal Benzeri Sendrom, 107480, Otozomal Dominant (Townes-Brocks Sendromu) (SALL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Townes-Brocks branchiootorenal-like syndrome, 107480, Autosomal dominant (Townes-Brocks syndrome) (SALL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SALL1	SALL1, HSAL1, TBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Townes-Brocks syndrome (TBS) [MIM:107480]: Rare, autosomal dominant malformation syndrome with a combination of imperforate anus, triphalangeal and supernumerary thumbs, malformed ears and sensorineural hearing loss. {ECO:0000269|PubMed:10533063, ECO:0000269|PubMed:9425907, ECO:0000269|PubMed:9973281}. Note=The disease is caused by mutations affecting the gene represented in this entry. Some individuals with SALL1 mutations manifest a phenotype overlapping with TBS and broncho-oto-renal syndrome. Clinical features include dysplastic ears, hypoplastic kidneys with impaired renal function, gastroesophageal reflux, hypermetropia, hypospadias, and mild developmental delay. Affected individuals lack the characteristic anal or hand malformations of TBS. {ECO:0000269|PubMed:10928856}.;	EDTA Blood Sample
Townes-Brocks Brankiootorenal Benzeri Sendrom, 107480, Otozomal Dominant (Townes-Brocks Sendromu) (Prenatal) (SALL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Townes-Brocks branchiootorenal-like syndrome, 107480, Autosomal dominant (Townes-Brocks syndrome) (SALL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SALL1	SALL1, HSAL1, TBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Townes-Brocks syndrome (TBS) [MIM:107480]: Rare, autosomal dominant malformation syndrome with a combination of imperforate anus, triphalangeal and supernumerary thumbs, malformed ears and sensorineural hearing loss. {ECO:0000269|PubMed:10533063, ECO:0000269|PubMed:9425907, ECO:0000269|PubMed:9973281}. Note=The disease is caused by mutations affecting the gene represented in this entry. Some individuals with SALL1 mutations manifest a phenotype overlapping with TBS and broncho-oto-renal syndrome. Clinical features include dysplastic ears, hypoplastic kidneys with impaired renal function, gastroesophageal reflux, hypermetropia, hypospadias, and mild developmental delay. Affected individuals lack the characteristic anal or hand malformations of TBS. {ECO:0000269|PubMed:10928856}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Townes-Brocks Sendromu, 107480, Otozomal Dominant; TBS (Townes-Brocks Sendromu) (SALL1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Townes-Brocks syndrome, 107480, Autosomal dominant; TBS (Townes-Brocks syndrome) (SALL1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SALL1	SALL1, HSAL1, TBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Townes-Brocks syndrome (TBS) [MIM:107480]: Rare, autosomal dominant malformation syndrome with a combination of imperforate anus, triphalangeal and supernumerary thumbs, malformed ears and sensorineural hearing loss. {ECO:0000269|PubMed:10533063, ECO:0000269|PubMed:9425907, ECO:0000269|PubMed:9973281}. Note=The disease is caused by mutations affecting the gene represented in this entry. Some individuals with SALL1 mutations manifest a phenotype overlapping with TBS and broncho-oto-renal syndrome. Clinical features include dysplastic ears, hypoplastic kidneys with impaired renal function, gastroesophageal reflux, hypermetropia, hypospadias, and mild developmental delay. Affected individuals lack the characteristic anal or hand malformations of TBS. {ECO:0000269|PubMed:10928856}.;	EDTA Blood Sample
Townes-Brocks Sendromu, 107480, Otozomal Dominant; TBS (Townes-Brocks Sendromu) (Prenatal) (SALL1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Townes-Brocks syndrome, 107480, Autosomal dominant; TBS (Townes-Brocks syndrome) (SALL1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SALL1	SALL1, HSAL1, TBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Townes-Brocks syndrome (TBS) [MIM:107480]: Rare, autosomal dominant malformation syndrome with a combination of imperforate anus, triphalangeal and supernumerary thumbs, malformed ears and sensorineural hearing loss. {ECO:0000269|PubMed:10533063, ECO:0000269|PubMed:9425907, ECO:0000269|PubMed:9973281}. Note=The disease is caused by mutations affecting the gene represented in this entry. Some individuals with SALL1 mutations manifest a phenotype overlapping with TBS and broncho-oto-renal syndrome. Clinical features include dysplastic ears, hypoplastic kidneys with impaired renal function, gastroesophageal reflux, hypermetropia, hypospadias, and mild developmental delay. Affected individuals lack the characteristic anal or hand malformations of TBS. {ECO:0000269|PubMed:10928856}.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Toksik Epidermal Nekroliz, Yatkınlık, 608579 (Stevens-Johnson Sendromu) (HLA-B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Toxic epidermal necrolysis, susceptibility to, 608579 (Stevens-Johnson syndrome) (HLA-B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HLA-B	HLA-B, SPDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample

TPMT Polimorfizmleri (238 G>C, 460 G>A, 719 A>G) (Tiopurin Grubu İlaç Metabolizması) (TPMT geni) (Dizi Analizi) (Postnatal)	TPMT polymorphisms (238 G>C, 460 G>A, 719 A>G) (Thiopurine Drug Metabolism) (TPMT gene) (Sequence Analysis) (Postnatal)	TPMT	.	Dizi Analizi/ Sequence Analysis	Lösemilerde kemik iliği/ Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	DISEASE: Thiopurine S-methyltransferase deficiency (TPMT deficiency) [MIM:610460]: Enzyme involved in the normal metabolic inactivation of thiopurine drugs. These drugs are generally used as immunosuppressants or cytotoxic drugs and are prescribed for a variety of clinical conditions including leukemia, autoimmune disease and organ transplantation. Patients with intermediate or no TPMT activity are at risk of toxicity after receiving standard doses of thiopurine drugs and it is shown that inter-individual differences in response to these drugs are largely determined by genetic variation at the TPMT locus. {ECO:0000269|PubMed:10208641, ECO:0000269|PubMed:10751626, ECO:0000269|PubMed:15819814, ECO:0000269|PubMed:16220112, ECO:0000269|PubMed:16476125, ECO:0000269|PubMed:16789994, ECO:0000269|PubMed:7862671, ECO:0000269|PubMed:8561894, ECO:0000269|PubMed:8644731, ECO:0000269|PubMed:9246020, ECO:0000269|PubMed:9336428, ECO:0000269|PubMed:9711875, ECO:0000269|PubMed:9931345, ECO:0000269|PubMed:9931346}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Lösemilerde kemik iliği/ Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Traboutsi Sendromu, 601552, Otozomal Resesif; FDLAB (Fasiyal Dismorfizm-Lens Dislokasyonu-Ön Segment Anormallikleri-Spontan Filtrelenen Bleb Sendromu (Traboutsi Sendromu)) (ASPH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Traboutsi syndrome, 601552, Autosomal recessive; FDLAB (Facial dysmorphism-lens dislocation-anterior segment abnormalities-spontaneous filtering bleb syndrome) (ASPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ASPH	ASPH, HAAH, FDLAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Transaldolaz Eksikliği, 606003, Otozomal Resesif (Transaldolaz Eksikliği) (TALDO1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transaldolase deficiency, 606003, Autosomal recessive (Transaldolase deficiency) (TALDO1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TALDO1	TALDO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Transaldolase deficiency (TALDOD) [MIM:606003]: An inborn error of the pentose phosphate pathway resulting in early-onset multisystem disease. Clinical features include growth retardation, dysmorphic features, cutis laxa, congenital heart disease, hepatosplenomegaly, telangiectases of the skin, pancytopenia, and bleeding tendency. {ECO:0000269|PubMed:11283793}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Transaldolaz Eksikliği, 606003, Otozomal Resesif (Transaldolaz Eksikliği) (Prenatal) (TALDO1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Transaldolase deficiency, 606003, Autosomal recessive (Transaldolase deficiency) (TALDO1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TALDO1	TALDO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Transaldolase deficiency (TALDOD) [MIM:606003]: An inborn error of the pentose phosphate pathway resulting in early-onset multisystem disease. Clinical features include growth retardation, dysmorphic features, cutis laxa, congenital heart disease, hepatosplenomegaly, telangiectases of the skin, pancytopenia, and bleeding tendency. {ECO:0000269|PubMed:11283793}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Transkobalamin 2 Eksikliği, 275350, Otozomal Resesif (Transkobalamin 2 Eksikliği) (TCN2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transcobalamin II deficiency, 275350, Autosomal recessive (Transcobalamin deficiency) (TCN2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCN2	TCN2, TC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Transcobalamin II deficiency (TCN2 deficiency) [MIM:275350]: Results in various forms of anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Transkobalamin 2 Eksikliği, 275350, Otozomal Resesif (Transkobalamin 2 Eksikliği) (Prenatal) (TCN2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Transcobalamin II deficiency, 275350, Autosomal recessive (Transcobalamin deficiency) (TCN2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCN2	TCN2, TC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Transcobalamin II deficiency (TCN2 deficiency) [MIM:275350]: Results in various forms of anemia. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Plazminojen Aktivatör İnhibitörü Transkripsiyonu, Modülatör (Konjenital Plazminojen Aktivatör İnhibitörü Tip 1 Eksikliği) (SERPINE1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transcription of plasminogen activator inhibitor, modulator of (Congenital plasminogen activator inhibitor type 1 deficiency) (SERPINE1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SERPINE1	PAI1, PLANH1, SERPINE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Plasminogen activator inhibitor-1 deficiency (PAI-1D) [MIM:613329]: A hematologic disorder characterized by increased bleeding after trauma, injury, or surgery. Affected females have menorrhagia. The bleeding defect is due to increased fibrinolysis of fibrin blood clots due to deficiency of plasminogen activator inhibitor-1, which inhibits tissue and urinary activators of plasminogen. {ECO:0000269|PubMed:9207454}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Neonatalin Geçici Büllöz Dermolizisi; Tbdn (Neonatalin Geçici Büllöz Dermolizi) (COL7A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN; TBDN (Transient bullous dermolysis of the newborn) (COL7A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL7A1	COL7A1, NDNC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Transient bullous dermolysis of the newborn (TBDN) [MIM:131705]: TBDN is a neonatal form of dystrophic epidermolysis bullosa characterized by sub-epidermal blisters, reduced or abnormal anchoring fibrils at the dermo-epidermal junction, and electron-dense inclusions in keratinocytes. TBDN heals spontaneously or strongly improves within the first months and years of life. {ECO:0000269|PubMed:9856844}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Neonatalin Geçici Büllöz Dermolizisi; Tbdn (Neonatalin Geçici Büllöz Dermoliz) (MLPA)	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN; TBDN (Transient bullous dermolysis of the newborn) (MLPA)	COL7A1	COL7A1, NDNC8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Transient bullous dermolysis of the newborn (TBDN) [MIM:131705]: TBDN is a neonatal form of dystrophic epidermolysis bullosa characterized by sub-epidermal blisters, reduced or abnormal anchoring fibrils at the dermo-epidermal junction, and electron-dense inclusions in keratinocytes. TBDN heals spontaneously or strongly improves within the first months and years of life. {ECO:0000269|PubMed:9856844}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Çocukluk Çağı Geçici Eritroblastopenisi, 227050, Otozomal Resesif; TEC (Çocukluk Çağı Geçici Eritroblastopeni) (TEC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transient erythroblastopenia of childhood, 227050, Autosomal recessive; TEC (Transient erythroblastopenia of childhood) (TEC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TEC	TEC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Büyük Arterlerin Transpozisyonu, Dekstro-Looplu 3, 613854, Otozomal Dominant; DTGA3 (Büyük Arterlerin Konjenital Düzeltilmemiş Transpozisyonu) (GDF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transposition of great arteries, dextro-looped 3, 613854, Autosomal dominant; DTGA3 (Congenitally uncorrected transposition of the great arteries) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Büyük Arterlerin Transpozisyonu, Dekstro-Looplu 3, 613854, Otozomal Dominant; DTGA3 (Büyük Arterlerin Konjenital Düzeltilmemiş Transpozisyonu) (Prenatal) (GDF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Transposition of great arteries, dextro-looped 3, 613854, Autosomal dominant; DTGA3 (Congenitally uncorrected transposition of the great arteries) (GDF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GDF1	GDF1, DTGA3, DORV, RAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tetralogy of Fallot (TOF) [MIM:187500]: A congenital heart anomaly which consists of pulmonary stenosis, ventricular septal defect, dextroposition of the aorta (aorta is on the right side instead of the left) and hypertrophy of the right ventricle. In this condition, blood from both ventricles (oxygen-rich and oxygen-poor) is pumped into the body often causing cyanosis. {ECO:0000269|PubMed:17924340}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Büyük Arterlerin Transpozisyonu, Dekstro-Looplu, 608808, 1, Otozomal Dominant; DTGA1 (Büyük Arterlerin Konjenital Düzeltilmemiş Transpozisyonu) (MED13L geni) (Tüm Gen Dizi Analizi) (Postnatal)	Transposition of the great arteries, dextro-looped 1, 608808, Autosomal dominant; DTGA1 (Congenitally uncorrected transposition of the great arteries) (MED13L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MED13L	MED13L, THRAP2, PROSIT240, TRAP240L, KIAA1025, MRFACD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving MED13L is found in a patient with transposition of the great arteries, dextro-looped and mental retardation. Translocation t(12;17)(q24.1;q21). {ECO:0000269|PubMed:14638541};.	EDTA Blood Sample
Büyük Arterlerin Transpozisyonu, Dekstro-Looplu, 608808, 1, Otozomal Dominant; DTGA1 (Büyük Arterlerin Konjenital Düzeltilmemiş Transpozisyonu) (Prenatal) (MED13L geni) (Tüm Gen Dizi Analizi) (Prenatal)	Transposition of the great arteries, dextro-looped 1, 608808, Autosomal dominant; DTGA1 (Congenitally uncorrected transposition of the great arteries) (MED13L gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MED13L	MED13L, THRAP2, PROSIT240, TRAP240L, KIAA1025, MRFACD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=A chromosomal aberration involving MED13L is found in a patient with transposition of the great arteries, dextro-looped and mental retardation. Translocation t(12;17)(q24.1;q21). {ECO:0000269|PubMed:14638541};.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Treacher Collins Sendromu 1, 154500, Otozomal Dominant; TCS1 (Treacher-Collins Sendromu) (TCOF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Treacher Collins syndrome 1, 154500, Autosomal dominant; TCS1 (Treacher-Collins syndrome) (TCOF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCOF1	TCOF1, MFD1, TCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Treacher Collins syndrome 1 (TCS1) [MIM:154500]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269|PubMed:9042910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Treacher Collins Sendromu 1, 154500, Otozomal Dominant; TCS1 (Treacher-Collins Sendromu) (MLPA)	Treacher Collins syndrome 1, 154500, Autosomal dominant; TCS1 (Treacher-Collins syndrome) (MLPA)	TCOF1	TCOF1, MFD1, TCS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Treacher Collins syndrome 1 (TCS1) [MIM:154500]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269|PubMed:9042910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Treacher Collins Sendromu 1, 154500, Otozomal Dominant; TCS1 (Treacher-Collins Sendromu) (Prenatal) (TCOF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Treacher Collins syndrome 1, 154500, Autosomal dominant; TCS1 (Treacher-Collins syndrome) (TCOF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TCOF1	TCOF1, MFD1, TCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Treacher Collins syndrome 1 (TCS1) [MIM:154500]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269|PubMed:9042910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Treacher Collins Sendromu 1, 154500, Otozomal Dominant; TCS1 (Treacher-Collins Sendromu) (Prenatal) (MLPA)	Treacher Collins syndrome 1, 154500, Autosomal dominant; TCS1 (Treacher-Collins syndrome) (Prenatal) (MLPA)	TCOF1	TCOF1, MFD1, TCS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Treacher Collins syndrome 1 (TCS1) [MIM:154500]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269|PubMed:9042910}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Treacher Collins Sendromu 2, 613717, Otozomal Dominant; TCS2 (Treacher-Collins Sendromu) (POLR1D gen) (Tüm Gen Dizi Analizi) (Postnatal)	Treacher Collins syndrome 2, 613717, Autosomal dominant; TCS2 (Treacher-Collins syndrome) (POLR1D gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR1D	POLR1D, RPA16, RPAC2, TCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Treacher Collins syndrome 2 (TCS2) [MIM:613717]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269	PubMed:21131976}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Treacher Collins Sendromu 2, 613717, Otozomal Dominant; TCS2 (Treacher-Collins Sendromu) (Prenatal) (POLR1D gen) (Tüm Gen Dizi Analizi) (Prenatal)	Treacher Collins syndrome 2, 613717, Autosomal dominant; TCS2 (Treacher-Collins syndrome) (POLR1D gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR1D	POLR1D, RPA16, RPAC2, TCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Treacher Collins syndrome 2 (TCS2) [MIM:613717]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269	PubMed:21131976}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Treacher Collins Sendromu 3, 248390, Otozomal Resesif; TCS3 (Treacher-Collins Sendromu) (POLR1C gen) (Tüm Gen Dizi Analizi) (Postnatal)	Treacher Collins syndrome 3, 248390, Autosomal recessive; TCS3 (Treacher-Collins syndrome) (POLR1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLR1C	POLR1C, RPA39, RPA40, RPAC1, RPA5, TCS3, HLD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Treacher Collins syndrome 3 (TCS3) [MIM:248390]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269	PubMed:21131976}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Treacher Collins Sendromu 3, 248390, Otozomal Resesif; TCS3 (Treacher-Collins Sendromu) (Prenatal) (POLR1C gen) (Tüm Gen Dizi Analizi) (Prenatal)	Treacher Collins syndrome 3, 248390, Autosomal recessive; TCS3 (Treacher-Collins syndrome) (POLR1C gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLR1C	POLR1C, RPA39, RPA40, RPAC1, RPA5, TCS3, HLD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Treacher Collins syndrome 3 (TCS3) [MIM:248390]: A form of Treacher Collins syndrome, a disorder of craniofacial development. Treacher Collins syndrome is characterized by a combination of bilateral downward slanting of the palpebral fissures, colobomas of the lower eyelids with a paucity of eyelashes medial to the defect, hypoplasia of the facial bones, cleft palate, malformation of the external ears, atresia of the external auditory canals, and bilateral conductive hearing loss. {ECO:0000269	PubMed:21131976}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trehaloz Eksikliği, 612119 (Trehaloz Eksikliğine Bağlı Diyare-Kusma) (TREH gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trehalase deficiency, 612119 (Diarrhea-vomiting due to trehalase deficiency) (TREH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREH	TREH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=Deficiency of TREH results in isolated trehalose intolerance that causes gastrointestinal symptoms after ingestion of edible mushrooms.;	EDTA Blood Sample
Trehaloz Eksikliği, 612119 (Trehaloz Eksikliğine Bağlı Diyare-Kusma) (Prenatal) (TREH gen) (Tüm Gen Dizi Analizi) (Prenatal)	Trehalase deficiency, 612119 (Diarrhea-vomiting due to trehalase deficiency) (TREH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TREH	TREH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Note=Deficiency of TREH results in isolated trehalose intolerance that causes gastrointestinal symptoms after ingestion of edible mushrooms.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Titreme (Tremor), Kalıtsal Esansiyel, 4, 614782, Otozomal Dominant; ETM4 (FUS gen) (Tüm Gen Dizi Analizi) (Postnatal)	Tremor, hereditary essential, 4, 614782, Autosomal dominant; ETM4 (FUS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUS	FUS, TLS, ALS6, ETM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amyotrophic lateral sclerosis 6, with or without frontotemporal dementia (ALS6) [MIM:608030]: A neurodegenerative disorder affecting upper motor neurons in the brain and lower motor neurons in the brain stem and spinal cord, resulting in fatal paralysis. Sensory abnormalities are absent. The pathologic hallmarks of the disease include pallor of the corticospinal tract due to loss of motor neurons, presence of ubiquitin-positive inclusions within surviving motor neurons, and deposition of pathologic aggregates. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of the cases. {ECO:0000269|PubMed:19251627, ECO:0000269|PubMed:19251628, ECO:0000269|PubMed:19861302, ECO:0000269|PubMed:20124201}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Titreme, Kalıtsal Esansiyel, 5, 616736, Otozomal Dominant; ETM5 (TENM4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Tremor, hereditary essential, 5, 616736, Autosomal dominant; ETM5 (TENM4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TENM4	TENM4, ODZ4, TNM4, DOC4, KIAA1302, ETM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trikodontoosseoz Sendrom, 190320, Otozomal Dominant; TDO (Trikodontoosseoz Sendromu) (DLX3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichodontoosseous syndrome, 190320, Autosomal dominant; TDO (Tricho-dento-osseous syndrome) (DLX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DLX3	DLX3, TDO, AI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Amelogenesis imperfecta 4 (AI4) [MIM:104510]: An autosomal dominant defect of enamel formation associated with enlarged pulp chambers. Enamel is thin, teeth are small and widely spaced. {ECO:0000269	PubMed:15666299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trikodontoosseoz Sendrom, 190320, Otozomal Dominant; TDO (Trikodontoosseoz Sendromu) (Prenatal) (DLX3 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Trichodontoosseous syndrome, 190320, Autosomal dominant; TDO (Tricho-dento-osseous syndrome) (DLX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DLX3	DLX3, TDO, AI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Amelogenesis imperfecta 4 (AI4) [MIM:104510]: An autosomal dominant defect of enamel formation associated with enlarged pulp chambers. Enamel is thin, teeth are small and widely spaced. {ECO:0000269	PubMed:15666299}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trikoepitelioma, Multipl Ailesel, 1, 601606, Otozomal Dominant (Ailesel Multipl Trikoepitelyoma) (CYLD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichoepithelioma, multiple familial, 1, 601606, Autosomal dominant (Familial multiple trichoepithelioma) (CYLD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYLD	CYLD, CDMT, EAC, MFT1, KIAA0849, BRSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cylindromatosis, familial (FCYL) [MIM:132700]: A disorder characterized by multiple skin tumors that develop from skin appendages, such as hair follicles and sweat glands. Affected individuals typically develop large numbers of tumors called cylindromas that arise predominantly in hairy parts of the body with approximately 90% on the head and neck. In severely affected individuals, cylindromas may combine into a confluent mass which may ulcerate or become infected (turban tumor syndrome). Individuals with familial cylindromatosis occasionally develop other types of tumors including spiradenomas that begin in sweat glands, and trichoepithelomas arising from hair follicles. {ECO:0000269|PubMed:12190880, ECO:0000269|PubMed:16922728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trikoepitelyoma, Multipl Ailesel, 2, 612099 (Ailesel Multipl Trikoepitelyoma) (CYLD gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichoepithelioma, multiple familial, 2, 612099 (Familial multiple trichoepithelioma) (CYLD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYLD	MFT2, TEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cylindromatosis, familial (FCYL) [MIM:132700]: A disorder characterized by multiple skin tumors that develop from skin appendages, such as hair follicles and sweat glands. Affected individuals typically develop large numbers of tumors called cylindromas that arise predominantly in hairy parts of the body with approximately 90% on the head and neck. In severely affected individuals, cylindromas may combine into a confluent mass which may ulcerate or become infected (turban tumor syndrome). Individuals with familial cylindromatosis occasionally develop other types of tumors including spiradenomas that begin in sweat glands, and trichoepithelomas arising from hair follicles. {ECO:0000269|PubMed:12190880, ECO:0000269|PubMed:16922728}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trikohepatoenterik Sendrom 1, 222470, Otozomal Resesif; THES1 (Sendromik Diyare) (TTC37 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichohepatoenteric syndrome 1, 222470, Autosomal recessive; THES1 (Syndromic diarrhea) (TTC37 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TTC37	TTC37, KIAA0372	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trikohepatoenterik Sendrom 2, 614602, Otozomal Resesif; THES2 (Sendromik Diyare) (SKIV2L gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichohepatoenteric syndrome 2, 614602, Autosomal recessive; THES2 (Syndromic diarrhea) (SKIV2L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SKIV2L	SKIV2L, SKI2, SKI2W, THES2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Trichohepatoenteric syndrome 2 (THES2) [MIM:614602]: A syndrome characterized by intrauterine growth retardation, severe diarrhea in infancy requiring total parenteral nutrition, facial dysmorphism, immunodeficiency, and hair abnormalities, mostly trichorrhexis nodosa. Hepatic involvement contributes to the poor prognosis of affected patients. {ECO:0000269	PubMed:23444670}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trikomegali, 190330, Otozomal Resesif; TCMGLY (Ailesel İzole Trikomegali) (FGF5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichomegaly, 190330, Autosomal recessive; TCMGLY (Familial isolated trichomegaly) (FGF5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF5	FGF5, TCMGLY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Trichomegaly (TCMGLY) [MIM:190330]: A morphologic trait characterized by unusually long eyelashes and mild hypertrichosis of eyebrows. It can be observed in association with corneal irritation, cataracts, and hereditary spherocytosis. {ECO:0000269	PubMed:24989505}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trikorinofalangeal Sendrom, Tip 1, 190350, Otozomal Dominant; TRPS1 (Trikorinofalangeal Sendrom Tip 1 ve Tip 3) (TRPS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Trichorhinophalangeal syndrome, type I, 190350, Autosomal dominant; TRPS1 (Trichorhinophalangeal syndrome type 1 and 3) (TRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPS1	TRPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tricho-rhino-phalangeal syndrome 2 (TRPS2) [MIM:150230]: A syndrome that combines the clinical features of tricho-rhino- phalangeal syndrome type 1 and multiple exostoses type 1. Affected individuals manifest multiple dysmorphic facial features including large, laterally protruding ears, a bulbous nose, an elongated upper lip, as well as sparse scalp hair, winged scapulae, multiple cartilaginous exostoses, redundant skin, and mental retardation. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration resulting in the loss of functional copies of TRPS1 and EXT1 has been found in TRPS2 patients.	EDTA Blood Sample
Trikorinofalangeal Sendrom, Tip 1, 190350, Otozomal Dominant; TRPS1 (Trikorinofalangeal Sendrom Tip 1 ve Tip 3) (Prenatal) (TRPS1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Trichorhinophalangeal syndrome, type I, 190350, Autosomal dominant; TRPS1 (Trichorhinophalangeal syndrome type 1 and 3) (TRPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPS1	TRPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tricho-rhino-phalangeal syndrome 2 (TRPS2) [MIM:150230]: A syndrome that combines the clinical features of tricho-rhino- phalangeal syndrome type 1 and multiple exostoses type 1. Affected individuals manifest multiple dysmorphic facial features including large, laterally protruding ears, a bulbous nose, an elongated upper lip, as well as sparse scalp hair, winged scapulae, multiple cartilaginous exostoses, redundant skin, and mental retardation. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration resulting in the loss of functional copies of TRPS1 and EXT1 has been found in TRPS2 patients.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trikorinofalangeal Sendrom, Tip 2, 150230, Otozomal Dominant; TRPS2 (Langer-Giedion Sendromu) (Kromozom 8q24.1 delesyon sendromu) (2200)	Trichorhinophalangeal syndrome, type II, 150230, Autosomal dominant; TRPS2 (Langer-Giedion syndrome) (CHROMOSOME 8q24.1 DELETION SYNDROME) (440)	Array-CGH	TRPS2, LGCR, LGS	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Trikorinofalangeal Sendrom, Tip 2, 150230, Otozomal Dominant; TRPS2 (Langer-Giedion Sendromu) (Kromozom 8q24.1 delesyon sendromu) (Prenatal)	Trichorhinophalangeal syndrome, type II, 150230, Autosomal dominant; TRPS2 (Langer-Giedion syndrome) (CHROMOSOME 8q24.1 DELETION SYNDROME) (Prenatal)	Array-CGH	TRPS2, LGCR, LGS	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Trikorinofalangeal Sendrom, Otozomal Dominant Tip 3, 190351; TRPS3 (Trikorinofalangeal Sendrom Tip 1 ve Tip 3) (TRPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichorhinophalangeal syndrome, type III, 190351, Autosomal dominant; TRPS3 (Trichorhinophalangeal syndrome type 1 and 3) (TRPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRPS1	TRPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tricho-rhino-phalangeal syndrome 2 (TRPS2) [MIM:150230]: A syndrome that combines the clinical features of tricho-rhino- phalangeal syndrome type 1 and multiple exostoses type 1. Affected individuals manifest multiple dysmorphic facial features including large, laterally protruding ears, a bulbous nose, an elongated upper lip, as well as sparse scalp hair, winged scapulae, multiple cartilaginous exostoses, redundant skin, and mental retardation. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration resulting in the loss of functional copies of TRPS1 and EXT1 has been found in TRPS2 patients.	EDTA Blood Sample
Trikorinofalangeal Sendrom, Otozomal Dominant Tip 3, 190351; TRPS3 (Trikorinofalangeal Sendrom Tip 1 ve Tip 3) (Prenatal) (TRPS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Trichorhinophalangeal syndrome, type III, 190351, Autosomal dominant; TRPS3 (Trichorhinophalangeal syndrome type 1 and 3) (TRPS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TRPS1	TRPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tricho-rhino-phalangeal syndrome 2 (TRPS2) [MIM:150230]: A syndrome that combines the clinical features of tricho-rhino- phalangeal syndrome type 1 and multiple exostoses type 1. Affected individuals manifest multiple dysmorphic facial features including large, laterally protruding ears, a bulbous nose, an elongated upper lip, as well as sparse scalp hair, winged scapulae, multiple cartilaginous exostoses, redundant skin, and mental retardation. Note=The gene represented in this entry is involved in disease pathogenesis. A chromosomal aberration resulting in the loss of functional copies of TRPS1 and EXT1 has been found in TRPS2 patients.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trikotiyodistrofi 1, Fotosensitivite, 601675, Otozomal Resesif; TTD1 (Trikotiyodistrofi) (ERCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 1, photosensitive, 601675, Autosomal recessive; TTD1 (Trichothiodystrophy) (ERCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC2	ERCC2, EM9, XPD, COFS2, TTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Xeroderma pigmentosum complementation group D (XP-D) [MIM:278730]: An autosomal recessive pigmentary skin disorder characterized by solar hypersensitivity of the skin, high predisposition for developing cancers on areas exposed to sunlight and, in some cases, neurological abnormalities. The skin develops marked freckling and other pigmentation abnormalities. Some XP-D patients present features of Cockayne syndrome, including cachectic dwarfism, pigmentary retinopathy, ataxia, decreased nerve conduction velocities. The phenotype combining xeroderma pigmentosum and Cockayne syndrome traits is referred to as XP-CS complex. {ECO:0000269|PubMed:11706541, ECO:0000269|PubMed:7585650, ECO:0000269|PubMed:7825573, ECO:0000269|PubMed:7849702, ECO:0000269|PubMed:9101292}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trikotiyodistrofi 2, Işığa Duyarlı, 616390, Otozomal Resesif; TTD2 (Trikotiyodistrofi) (ERCC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 2, photosensitive, 616390, Autosomal recessive; TTD2 (Trichothiodystrophy) (ERCC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC3	ERCC3, XPB, TTD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Xeroderma pigmentosum complementation group B (XP-B) [MIM:610651]: An autosomal recessive pigmentary skin disorder characterized by solar hypersensitivity of the skin, high predisposition for developing cancers on areas exposed to sunlight and, in some cases, neurological abnormalities. The skin develops marked freckling and other pigmentation abnormalities. Some XP-B patients present features of Cockayne syndrome, including cachectic dwarfism, pigmentary retinopathy, ataxia, decreased nerve conduction velocities. The phenotype combining xeroderma pigmentosum and Cockayne syndrome traits is referred to as XP-CS complex. {ECO:0000269|PubMed:16947863, ECO:0000269|PubMed:8304337}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trikotiyodistrofi 3, Işığa Duyarlı, 616395; TTD3 (Trikotiyodistrofi) (GTF2H5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 3, photosensitive, 616395; TTD3 (Trichothiodystrophy) (GTF2H5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GTF2H5	GTF2H5, TTD3, TF85, C6orf175, TTDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Trichothiodystrophy 3, photosensitive (TTD3) [MIM:616395]: A form of trichothiodystrophy, an autosomal recessive disease characterized by sulfur-deficient brittle hair and multisystem variable abnormalities. The spectrum of clinical features varies from mild disease with only hair involvement to severe disease with cutaneous, neurologic and profound developmental defects. Ichthyosis, intellectual and developmental disabilities, decreased fertility, abnormal characteristics at birth, ocular abnormalities, short stature, and infections are common manifestations. There are both photosensitive and non-photosensitive forms of the disorder. {ECO:0000269|PubMed:15220921}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trikotiyodistrofi 4, Işığa Duyarlı Olmayan, 234050, Otozomal Resesif; TTD4 (BIDS Sendromu) (MPLKIP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 4, nonphotosensitive, 234050, Autosomal recessive; TTD4 (BIDS syndrome) (MPLKIP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MPLKIP	MPLKIP, C7orf11, ABHS, TTDN1, TTD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Trichothiodystrophy 4, non-photosensitive (TTD4) [MIM:234050]: A form of trichothiodystrophy, an autosomal recessive disease characterized by sulfur-deficient brittle hair and multisystem variable abnormalities. The spectrum of clinical features varies from mild disease with only hair involvement to severe disease with cutaneous, neurologic and profound developmental defects. Ichthyosis, intellectual and developmental disabilities, decreased fertility, abnormal characteristics at birth, ocular abnormalities, short stature, and infections are common manifestations. There are both photosensitive and non-photosensitive forms of the disorder. TTD4 patients do not manifest cutaneous photosensitivity. {ECO:0000269|PubMed:15645369}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Trikotiyodistrofi 5, Fotosensitif Olmayan, 300953, X'e Bağlı Dominant; TTD5 (Trikotiyodistrofi) (RNF113A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 5, nonphotosensitive, 300953, X-linked dominant; TTD5 (Trichothiodystrophy) (RNF113A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RNF113A	RNF113A, ZNF183, TTD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trikotiyodistrofi 6, Işığa Duyarlı Olmayan, 616943, Otozomal Resesif; TTD6 (Trikotiyodistrofi) (GTF2E2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichothiodystrophy 6, nonphotosensitive, 616943, Autosomal recessive; TTD6 (Trichothiodystrophy) (GTF2E2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GTF2E2	GTF2E2, TTD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trikotillomani 613229, Otozomal Dominant, Multifaktöriyel; TTM (SLITRK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trichotillomania, 613229, Autosomal dominant, Multifactorial; TTM (SLITRK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLITRK1	SLITRK1, KIAA1910, TTM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Gilles de la Tourette syndrome (GTS) [MIM:137580]: Neurologic disorder manifested particularly by motor and vocal tics and associated with behavioral abnormalities. {ECO:0000269|PubMed:16224024}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Üç İşlevli Protein Eksikliği, 609015, Otozomal Resesif (Mitokondriyal Üç Fonksiyonlu Protein Eksikliği) (HADHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trifunctional protein deficiency, 609015, Autosomal recessive (Mitochondrial trifunctional protein deficiency) (HADHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHB	HADHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269|PubMed:12754706, ECO:0000269|PubMed:8651282, ECO:0000269|PubMed:9259266}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Üç İşlevli Protein Eksikliği, 609015, Otozomal Resesif (Mitokondriyal Üç Fonksiyonlu Protein Eksikliği) (Prenatal) (HADHB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Trifunctional protein deficiency, 609015, Autosomal recessive (Mitochondrial trifunctional protein deficiency) (HADHB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADHB	HADHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269|PubMed:12754706, ECO:0000269|PubMed:8651282, ECO:0000269|PubMed:9259266}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Üç İşlevli Protein Eksikliği, 609015, Otozomal Resesif; MTPD (Mitokondriyal Üç Fonksiyonlu Protein Eksikliği) (HADHA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trifunctional protein deficiency, 609015, Autosomal recessive; MTPD (Mitochondrial trifunctional protein deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269|PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Üç İşlevli Protein Eksikliği, 609015, Otozomal Resesif; MTPD (Mitokondriyal Üç Fonksiyonlu Protein Eksikliği) (Prenatal) (HADHA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Trifunctional protein deficiency, 609015, Autosomal recessive; MTPD (Mitochondrial trifunctional protein deficiency) (HADHA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HADHA	HADHA, MTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitochondrial trifunctional protein deficiency (MTPD) [MIM:609015]: A disease biochemically characterized by loss of all enzyme activities of the mitochondrial trifunctional protein complex. Variable clinical manifestations include hypoglycemia, cardiomyopathy, delayed psychomotor development, sensorimotor axonopathy, generalized weakness, hepatic dysfunction, respiratory failure. Sudden infant death may occur. Most patients die from heart failure. {ECO:0000269|PubMed:9739053}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trigonosefali 1, 190440, Otozomal Dominant; TRIGNO1 (İzole Trigonosefali) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trigonocephaly 1, 190440, Autosomal dominant; TRIGNO1 (Isolated trigonocephaly) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269|PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trigonosefali 1, 190440, Otozomal Dominant; TRIGNO1 (İzole Trigonosefali) (MLPA)	Trigonocephaly 1, 190440, Autosomal dominant; TRIGNO1 (Isolated trigonocephaly) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269|PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trigonosefali 1, 190440, Otozomal Dominant; TRIGNO1 (İzole Trigonosefali) (Prenatal) (FGFR1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Trigonocephaly 1, 190440, Autosomal dominant; TRIGNO1 (Isolated trigonocephaly) (FGFR1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). {ECO:0000269|PubMed:7874169}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Trigonosefali 1, 190440, Otozomal Dominant; TRIGNO1 (özle Trigonosefali) (Prenatal) (MLPA)	Trigonocephaly 1, 190440, Autosomal dominant; TRIGNO1 (isolated trigonocephaly) (Prenatal) (MLPA)	FGFR1	FGFR1, FLT2, OGD, KAL2, HH2, HRTFDS, ECCL	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Pfeiffer syndrome (PS) [MIM:101600]: A syndrome characterized by the association of craniosynostosis, broad and deviated thumbs and big toes, and partial syndactyly of the fingers and toes. Three subtypes are known: mild autosomal dominant form (type 1); cloverleaf skull, elbow ankylosis, early death, sporadic (type 2); craniosynostosis, early demise, sporadic (type 3). ECO:0000269	PubMed:7874169, Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trigonosefali 2, 614485, Otozomal Dominant; TRIGNO2 (özle Trigonosefali) (FREM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trigonocephaly 2, 614485, Autosomal dominant; TRIGNO2 (isolated trigonocephaly) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Manitoba oculotrichoanal syndrome (MOTA) [MIM:248450]: A rare condition defined by eyelid colobomas, cryptophthalmos, and anophthalmia/microphthalmia, an aberrant hairline, a bifid or broad nasal tip, and gastrointestinal anomalies such as omphalocele and anal stenosis. (ECO:0000269	PubMed:21507892). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Trigonosefali 2, 614485, Otozomal Dominant; TRIGNO2 (özle Trigonosefali) (Prenatal) (FREM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Trigonocephaly 2, 614485, Autosomal dominant; TRIGNO2 (isolated trigonocephaly) (FREM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FREM1	FREM1, C9orf154, BNAR, MOTA, TRIGNO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Manitoba oculotrichoanal syndrome (MOTA) [MIM:248450]: A rare condition defined by eyelid colobomas, cryptophthalmos, and anophthalmia/microphthalmia, an aberrant hairline, a bifid or broad nasal tip, and gastrointestinal anomalies such as omphalocele and anal stenosis. (ECO:0000269	PubMed:21507892). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trimetilaminüri, 602079, Otozomal Resesif (Alır Primer Trimetilaminürü) (FMO3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trimethylaminuria, 602079, Autosomal recessive (Severe primary trimethylaminuria) (FMO3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FMO3	FMO3, TMAU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Trifalangeal Başparmak-Polisindaktili Sendromu, 174500, Otozomal Dominant (Triphalangeal thumb-polysyndactyly syndrome) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Triphalangeal thumb-polysyndactyly syndrome, 174500, Autosomal dominant (Triphalangeal thumb-polysyndactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269|PubMed:24456159). Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. (ECO:0000269|PubMed:24456159)..	EDTA Blood Sample
Triphalangeal Başparmak, Tip 1, 174500, Otozomal Dominant (Triphalangeal thumb-polysyndactyly syndrome) (LMBR1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Triphalangeal thumb, type I, 174500, Autosomal dominant (Triphalangeal thumb-polysyndactyly syndrome) (LMBR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LMBR1	LMBR1, ACHP, C7orf2, PPD2, THYP, LSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Laurin-Sandrow syndrome (LSS) [MIM:135750]: A rare autosomal dominant disorder characterized by polysyndactyly of hands and/or feet, mirror image duplication of the feet, nasal defects, and loss of identity between fibula and tibia. Some patients do not have nasal abnormalities (segmental Laurin-Sandrow syndrome). (ECO:0000269|PubMed:24456159). Note=The disease is caused by mutations affecting the gene represented in this entry. Disease-causing mutations consists of duplications (16-75 kb) involving the ZPA regulatory sequence (ZRS), a SHH long-range cis- regulatory element, located in LMBR1 intron 5. The mutations do not alter normal LMBR1 expression and function, but affect SHH limb expression. (ECO:0000269|PubMed:24456159)..	EDTA Blood Sample
Trismus-Psödokamptodaktili Sendromu, 158300, Otozomal Dominant (Trismus-Psödokamptodaktili Sendromu) (MYHB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trismus-pseudocamptodactyly syndrome, 158300, Autosomal dominant (Trismus-pseudocamptodactyly syndrome) (MYHB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYHB	MYHB, DA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Arthrogryposis, distal, 7 (DA7) [MIM:158300]: A form of distal arthrogryposis, a disease characterized by congenital joint contractures that mainly involve two or more distal parts of the limbs, in the absence of a primary neurological or muscle disease. DA7 is characterized by an inability to open the mouth fully (trismus) and pseudocamptodactyly in which wrist dorsiflexion, but not volarflexion, produces involuntary flexion contracture of distal and proximal interphalangeal joints. Additional features include shortened hamstring muscles and short stature. (ECO:0000269|PubMed:15282353, ECO:0000269|PubMed:20949528). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Trizomi 10 (FISH)	Trisomy 10 (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Trizomi 12 (FISH)	Trisomy 12 (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Tropik Kalsifiye Pankreatit, 608189, Otozomal Resesif, Otozomal Dominant (Tropikal Pankreatit) (SPINK1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tropical calcific pancreatitis, 608189, Autosomal recessive, Autosomal dominant (Tropical pancreatitis) (SPINK1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. (ECO:0000269|PubMed:10691414, ECO:0000269|PubMed:10835640, ECO:0000269|PubMed:12974284). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Tropik Kalsifiye Pankreatit, 608189, Otozomal Resesif, Otozomal Dominant (Tropikal Pankreatit) (MLPA)	Tropical calcific pancreatitis, 608189, Autosomal recessive, Autosomal dominant (Tropical pancreatitis) (MLPA)	SPINK1	SPINK1, PSTI, PCTT, TATI, TCP	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. (ECO:0000269|PubMed:10691414, ECO:0000269|PubMed:10835640, ECO:0000269|PubMed:12974284). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Troyer Sendromu, 275900, Otozomal Resesif; SPG20 (Otozomal Resesif Spastik Paraplej Tip 20) (SPG20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Troyer syndrome, 275900, Autosomal recessive; SPG20 (Autosomal recessive spastic paraplegia type 20) (SPG20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SPG20	SPG20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Spastic paraplegia 20, autosomal recessive (SPG20) [MIM:275900]: A form of spastic paraplegia, a neurodegenerative disorder characterized by a slow, gradual, progressive weakness and spasticity of the lower limbs. Rate of progression and the severity of symptoms are quite variable. Initial symptoms may include difficulty with balance, weakness and stiffness in the legs, muscle spasms, and dragging the toes when walking. In some forms of the disorder, bladder symptoms (such as incontinence) may appear, or the weakness and stiffness may spread to other parts of the body. SPG20 is characterized by dysarthria, distal amyotrophy, mild developmental delay and short stature. (ECO:0000269	PubMed:12134148). Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
Tripsinojen Eksikliği, 614044, Otozomal Resesif (PRSS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Trypsinogen deficiency, 614044, Autosomal recessive (PRSS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRSS1	PRSS1, TRY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. (ECO:0000269|PubMed:10204851, ECO:0000269|PubMed:10381903, ECO:0000269|PubMed:10930381, ECO:0000269|PubMed:11073545, ECO:0000269|PubMed:11788572, ECO:0000269|PubMed:11866271, ECO:0000269|PubMed:14695529, ECO:0000269|PubMed:15776435, ECO:0000269|PubMed:8841182, ECO:0000269|PubMed:9322498, ECO:0000269|PubMed:9633818). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Tripsinojen Eksikliği, 614044, Otozomal Resesif (MLPA)	Trypsinogen deficiency, 614044, Autosomal recessive (MLPA)	PRSS1	PRSS1, TRY1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Pancreatitis, hereditary (PCTT) [MIM:167800]: A disease characterized by pancreas inflammation, permanent destruction of the pancreatic parenchyma, maldigestion, and severe abdominal pain attacks. (ECO:0000269|PubMed:10204851, ECO:0000269|PubMed:10381903, ECO:0000269|PubMed:10930381, ECO:0000269|PubMed:11073545, ECO:0000269|PubMed:11788572, ECO:0000269|PubMed:11866271, ECO:0000269|PubMed:14695529, ECO:0000269|PubMed:15776435, ECO:0000269|PubMed:8841182, ECO:0000269|PubMed:9322498, ECO:0000269|PubMed:9633818). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
TSC2 Anjiyomyolipomalar, Renal, Modifiye, 613254, Otozomal Dominant (Tüberoz Skleroz Kompleksi) (IFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	TSC2 angiomyolipomas, renal, modifier of, 613254, Autosomal dominant (Tuberous sclerosis complex) (IFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aplastic anemia (AA) [MIM:609135]: A form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. It is characterized by peripheral pancytopenia and marrow hypoplasia. (ECO:0000269	PubMed:15327519). Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry..	EDTA Blood Sample
Tüberküloz Enfeksiyonu, Karşı Koruma, 607948 (Tüberküloz) (TIRAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberculosis infection, protection against, 607948 (Tuberculosis) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209950]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. (ECO:0000269|PubMed:10811650, ECO:0000269|PubMed:9389728). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tüberküloz, Karşı Koruma, 607948 (Tüberküloz) (TIRAP geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberculosis, protection against, 607948 (Tuberculosis) (TIRAP gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIRAP	TIRAP, BACTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal		EDTA Blood Sample
Tüberküloz, Karşı Koruma, 607948 (Tüberküloz) (IFNG geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberculosis, protection against, 607948 (Tuberculosis) (IFNG gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNG	IFNG, IFG, IFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Aplastic anemia (AA) [MIM:609135]: A form of anemia in which the bone marrow fails to produce adequate numbers of peripheral blood elements. It is characterized by peripheral pancytopenia and marrow hypoplasia. (ECO:0000269	PubMed:15327519). Note=Disease susceptibility may be associated with variations affecting the gene represented in this entry..	EDTA Blood Sample

Tüberküloz, Yatkınlık, 607948 (Otozomal Resesif Mikobakteriyel Hastalıklara Mendelyen Yatkınlık, Parsiyel İfngamar1 Eksikliğine Bağlı) (IFNGR1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberculosis, susceptibility to, 607948 (Autosomal recessive mendelian susceptibility to mycobacterial diseases due to partial IFNgammaR1 deficiency) (IFNGR1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IFNGR1	IFNGR1, IMD27A, IMD27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Immunodeficiency 27A (IMD27A) [MIM:209550]: A form of Mendelian susceptibility to mycobacterial disease, a rare condition caused by impairment of interferon-gamma mediated immunity. It is characterized by predisposition to illness caused by moderately virulent mycobacterial species, such as Bacillus Calmette-Guerin (BCG) vaccine, environmental non-tuberculous mycobacteria, and by the more virulent Mycobacterium tuberculosis. Other microorganisms rarely cause severe clinical disease in individuals with susceptibility to mycobacterial infections, with the exception of Salmonella which infects less than 50% of these individuals. Clinical outcome severity depends on the degree of impairment of interferon-gamma mediated immunity. Some patients die of overwhelming mycobacterial disease with lepromatous-like lesions in early childhood, whereas others develop, later in life, disseminated but curable infections with tuberculoid granulomas. (ECO:0000269|PubMed:10811850, ECO:0000269|PubMed:9389726). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tüberküloz, Yatkınlık, 607948 (Tüberküloz (CISH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberculosis, susceptibility to, 607948 (Tuberculosis (CISH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CISH	CISH, BACTS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tüberoz Skleroz-1, 191100, Otozomal Dominant, TSC1 (Tuberoz Skleroz Kompleksi) (TSC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberous sclerosis-1, 191100, Autosomal dominant; TSC1 (Tuberous sclerosis complex) (TSC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSC1	TSC1, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. (ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tüberoz Skleroz-1, 191100, Otozomal Dominant, TSC1 (Tuberoz Skleroz Kompleksi) (MLPA)	Tuberous sclerosis-1, 191100, Autosomal dominant; TSC1 (Tuberous sclerosis complex) (MLPA)	TSC1	TSC1, LAM	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. (ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tüberoz Skleroz-1, 191100, Otozomal Dominant, TSC1 (Tuberoz Skleroz Kompleksi) (Prenatal) (TSC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tuberous sclerosis-1, 191100, Autosomal dominant; TSC1 (Tuberous sclerosis complex) (TSC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSC1	TSC1, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. (ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tüberoz Skleroz-1, 191100, Otozomal Dominant, TSC1 (Tuberoz Skleroz Kompleksi) (Prenatal) (MLPA)	Tuberous sclerosis-1, 191100, Autosomal dominant; TSC1 (Tuberous sclerosis complex) (Prenatal) (MLPA)	TSC1	TSC1, LAM	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tuberous sclerosis 1 (TSC1) [MIM:191100]: An autosomal dominant multi-system disorder that affects especially the brain, kidneys, heart, and skin. It is characterized by hamartomas (benign overgrowths predominantly of a cell or tissue type that occurs normally in the organ) and hamartias (developmental abnormalities of tissue combination). Clinical manifestations include epilepsy, learning difficulties, behavioral problems, and skin lesions. Seizures can be intractable and premature death can occur from a variety of disease-associated causes. (ECO:0000269|PubMed:10227394, ECO:0000269|PubMed:10533069, ECO:0000269|PubMed:10570911, ECO:0000269|PubMed:10874311, ECO:0000269|PubMed:18830229, ECO:0000269|PubMed:22161988, ECO:0000269|PubMed:9328481). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tüberoz Skleroz-2, 613254, Otozomal Dominant (Tuberoz Skleroz Kompleksi) (TSC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tuberous sclerosis-2, 613254, Autosomal dominant (Tuberous sclerosis complex) (TSC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TSC2	TSC2, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Lymphangioleiomyomatosis (LAM) [MIM:606690]: Progressive and often fatal lung disease characterized by a diffuse proliferation of abnormal smooth muscle cells in the lungs. It affects almost exclusively young women and can occur as an isolated disorder or in association with tuberous sclerosis complex. (ECO:0000269	PubMed:10823953). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tüberoz Skleroz-2, 613254, Otozomal Dominant (Tuberoz Skleroz Kompleksi) (MLPA)	Tuberous sclerosis-2, 613254, Autosomal dominant (Tuberous sclerosis complex) (MLPA)	TSC2	TSC2, LAM	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Lymphangioleiomyomatosis (LAM) [MIM:606690]: Progressive and often fatal lung disease characterized by a diffuse proliferation of abnormal smooth muscle cells in the lungs. It affects almost exclusively young women and can occur as an isolated disorder or in association with tuberous sclerosis complex. (ECO:0000269	PubMed:10823953). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tüberoz Skleroz-2, 613254, Otozomal Dominant (Tuberoz Skleroz Kompleksi) (Prenatal) (TSC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tuberous sclerosis-2, 613254, Autosomal dominant (Tuberous sclerosis complex) (TSC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TSC2	TSC2, LAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphangioleiomyomatosis (LAM) [MIM:606690]: Progressive and often fatal lung disease characterized by a diffuse proliferation of abnormal smooth muscle cells in the lungs. It affects almost exclusively young women and can occur as an isolated disorder or in association with tuberous sclerosis complex. (ECO:0000269	PubMed:10823953). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tüberoz Skleroz-2, 613254, Otozomal Dominant (Tuberoz Skleroz Kompleksi) (Prenatal) (MLPA)	Tuberous sclerosis-2, 613254, Autosomal dominant (Tuberous sclerosis complex) (Prenatal) (MLPA)	TSC2	TSC2, LAM	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Lymphangioleiomyomatosis (LAM) [MIM:606690]: Progressive and often fatal lung disease characterized by a diffuse proliferation of abnormal smooth muscle cells in the lungs. It affects almost exclusively young women and can occur as an isolated disorder or in association with tuberous sclerosis complex. (ECO:0000269	PubMed:10823953). Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tümör paneli (BRAF, HRAS, KRAS, NRAS) (MLPA)	Tumor panel (BRAF, HRAS, KRAS, NRAS) (MLPA)	BRAF, HRAS, KRAS, NRAS	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Tümör Yatkınlık Sendromu, 614327, Otozomal Dominant; TPDS (BAP1 İlişkili Tümör Yatkınlık Sendromu) (BAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tumor predisposition syndrome, 614327, Autosomal dominant; TPDS (BAP1-related tumor predisposition syndrome) (BAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BAP1	BAP1, TPDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mesothelioma, malignant (MESOM) [MIM:156240]: An aggressive neoplasm of the serosal lining of the chest. It appears as broad sheets of cells, with some regions containing spindle-shaped, sarcoma-like cells and other regions showing adenomatous patterns. Pleural mesotheliomas have been linked to exposure to asbestos. (ECO:0000269|PubMed:21642991, ECO:0000269|PubMed:21874000). Note=The gene represented in this entry is involved in disease pathogenesis.	EDTA Blood Sample
Tümöral Kalsinoz, Ailesel, Normofosfatemik, 610455, Otozomal Resesif; NFTC (Ailesel Normofosfatemik Tümöral Kalsinozis) (SAMD9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tumoral calcinosis, familial, normophosphatemic, 610455, Autosomal recessive; NFTC (Familial normophosphatemic tumoral calcinosis) (SAMD9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SAMD9	SAMD9, NFTC, MIRAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tumoral calcinosis, normophosphatemic, familial (NFTC) [MIM:610455]: An uncommon, life-threatening disorder characterized by progressive deposition of calcified masses in cutaneous and subcutaneous tissues. Serum phosphate levels are normal. Clinical features include painful calcified ulcerative lesions and massive calcium deposition in the mid- and lower dermis, severe skin and bone infections, erythematous papular skin eruption in infancy, conjunctivitis, and gingivitis. NFTC shows a striking resemblance to acquired dystrophic calcinosis, in which tissue calcification occurs as a consequence of tissue injury/inflammation. (ECO:0000269|PubMed:16968814, ECO:0000269|PubMed:18094730). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tümöral Kalsinoz, Hiperfosfatemik, 211900, Otozomal Resesif; HFTC (Ailesel Tümöral Kalsinozis) (KL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tumoral calcinosis, hyperphosphatemic, 211900, Autosomal recessive; HFTC (Familial tumoral calcinosis) (KL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KL	KL, KLOTHO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tumoral calcinosis, hyperphosphatemic, familial (HFTC) [MIM:211900]: A severe metabolic disorder that manifests with hyperphosphatemia and massive calcium deposits in the skin and subcutaneous tissues. Some patients manifest recurrent, transient, painful swellings of the long bones associated with the radiographic findings of periosteal reaction and cortical hyperostosis and absence of skin involvement. (ECO:0000269	PubMed:17710231). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tümöral Kalsinoz, Hiperfosfatemik, 211900, Otozomal Resesif (Ailesel Normofosfatemik Tümöral Kalsinozis / Hiperfosfatemik Hiperostoz Sendromu) (GALNT3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tumoral calcinosis, hyperphosphatemic, familial, 211900, Autosomal recessive (Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome) (GALNT3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GALNT3	GALNT3, HHS, HFTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tumoral calcinosis, hyperphosphatemic, familial (HFTC) [MIM:211900]: A severe metabolic disorder that manifests with hyperphosphatemia and massive calcium deposits in the skin and subcutaneous tissues. Some patients manifest recurrent, transient, painful swellings of the long bones associated with the radiographic findings of periosteal reaction and cortical hyperostosis and absence of skin involvement. (ECO:0000269|PubMed:15133511, ECO:0000269|PubMed:15599692). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample

Tümöral Kalsinoz, Hiperfosfatemik, 211900, Otozomal Resesif (Ailesel Normofosfatemik Tümöral Kalsinozlu / Hiperfosfatemik Hiperostoz Sendromu) (FGF23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tumoral calcinosis, hyperphosphatemic, familial, 211900, Autosomal recessive (Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome) (FGF23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FGF23	FGF23, ADHR, HPDR2, PHPTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. [ECO:0000269	PubMed:11062477]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Tümöral Kalsinoz, Hiperfosfatemik, 211900, Otozomal Resesif (Ailesel Normofosfatemik Tümöral Kalsinozlu / Hiperfosfatemik Hiperostoz Sendromu) (MLPA)	Tumoral calcinosis, hyperphosphatemic, familial, 211900, Autosomal recessive (Familial hyperphosphatemic tumoral calcinosis/Hyperphosphatemic hyperostosis syndrome) (MLPA)	FGF23	FGF23, ADHR, HPDR2, PHPTC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Hypophosphatemic rickets, autosomal dominant (ADHR) [MIM:193100]: A disease characterized by isolated renal phosphate wasting, hypophosphatemia, and inappropriately normal 1,25- dihydroxyvitamin D3 (calcitriol) levels. Patients frequently present with bone pain, rickets, and tooth abscesses. [ECO:0000269	PubMed:11062477]. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Özofagus Kanseri, Tilozis, 148500, Otozomal Dominant; TOC (Palmoplantar Keratoderma-Özofageal Karsinoma Sendromu) (RHBDF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tylosis with esophageal cancer, 148500, Autosomal dominant; TOC (Palmoplantar keratoderma-esophageal carcinoma syndrome) (RHBDF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RHBDF2	RHBDF2, IRHOM2, TOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tirozinemi, Tip 1, 276700, Otozomal Resesif; TYRSN1 (Tirozinemi Tip 1) (FAH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tyrosinemia, type I, 276700, Autosomal recessive; TYRSN1 (Tyrosinemia type 1) (FAH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAH	FAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tirozinemi, Tip 1, 276700, Otozomal Resesif; TYRSN1 (Tirozinemi Tip 1) (Prenatal) (FAH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tyrosinemia, type I, 276700, Autosomal recessive; TYRSN1 (Tyrosinemia type 1) (FAH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAH	FAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tirozinemi, Tip 1b (GSTZ1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tyrosinemia, type Ib (GSTZ1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GSTZ1	GSTZ1, MAAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tirozinemi, Tip 1b (Prenatal) (GSTZ1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tyrosinemia, type Ib (GSTZ1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GSTZ1	GSTZ1, MAAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tirozinemi, Tip 2, 276600, Otozomal Resesif; TYRSN2 (Tirozinemi Tip 2) (TAT geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tyrosinemia, type II, 276600, Autosomal recessive; TYRSN2 (Tyrosinemia type 2) (TAT gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAT	TAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Tyrosinemia 2 (TYRSN2) [MIM:276600]: An inborn error of metabolism characterized by elevations of tyrosine in the blood and urine, and oculocutaneous manifestations. Typical features include palmoplantar keratosis, painful corneal ulcers, and mental retardation. [ECO:0000269	PubMed:1357662]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tirozinemi, Tip 2, 276600, Otozomal Resesif; TYRSN2 (Tirozinemi Tip 2) (Prenatal) (TAT geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tyrosinemia, type II, 276600, Autosomal recessive; TYRSN2 (Tyrosinemia type 2) (TAT gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TAT	TAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Tyrosinemia 2 (TYRSN2) [MIM:276600]: An inborn error of metabolism characterized by elevations of tyrosine in the blood and urine, and oculocutaneous manifestations. Typical features include palmoplantar keratosis, painful corneal ulcers, and mental retardation. [ECO:0000269	PubMed:1357662]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tirozinemi, Tip 3, 276710, Otozomal Resesif; TYRSN3 (Tirozinemi Tip 3) (HPD geni) (Tüm Gen Dizi Analizi) (Postnatal)	Tyrosinemia, type III, 276710, Autosomal recessive; TYRSN3 (Tyrosinemia type 3) (HPD gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPD	HPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hawkinsinuria (HAWK) [MIM:140350]: An inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin, in the urine. [ECO:0000269	PubMed:11073718]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Tirozinemi, Tip 3, 276710, Otozomal Resesif; TYRSN3 (Tirozinemi Tip 3) (Prenatal) (HPD geni) (Tüm Gen Dizi Analizi) (Prenatal)	Tyrosinemia, type III, 276710, Autosomal recessive; TYRSN3 (Tyrosinemia type 3) (HPD gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPD	HPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Hawkinsinuria (HAWK) [MIM:140350]: An inborn error of tyrosine metabolism characterized by failure to thrive, persistent metabolic acidosis, fine and sparse hair, and excretion of the unusual cyclic amino acid metabolite, hawkinsin, in the urine. [ECO:0000269	PubMed:11073718]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 1, 254090, Otozomal Resesif, Otozomal Dominant (Konjenital Müsküler Distrofi, Ullrich Tipi) (COL6A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ullrich congenital muscular dystrophy 1, 254090, Autosomal recessive, Autosomal dominant (Congenital muscular dystrophy, Ullrich type) (COL6A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A1	COL6A1, BTHLM1, UCHMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:15955946, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 1, 254090, Otozomal Resesif, Otozomal Dominant (Konjenital Müsküler Distrofi, Ullrich Tipi) (Prenatal) (COL6A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ullrich congenital muscular dystrophy 1, 254090, Autosomal recessive, Autosomal dominant (Congenital muscular dystrophy, Ullrich type) (COL6A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL6A1	COL6A1, BTHLM1, UCHMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:15955946, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 1, 254090, Otozomal Resesif, Otozomal Dominant; UCMD1 (Konjenital Müsküler Distrofi, Ullrich Tipi) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ullrich congenital muscular dystrophy 1, 254090, Autosomal recessive, Autosomal dominant; UCMD1 (Congenital muscular dystrophy, Ullrich type) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 1, 254090, Otozomal Resesif, Otozomal Dominant; UCMD1 (Konjenital Müsküler Distrofi, Ullrich Tipi) (Prenatal) (COL6A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ullrich congenital muscular dystrophy 1, 254090, Autosomal recessive, Autosomal dominant; UCMD1 (Congenital muscular dystrophy, Ullrich type) (COL6A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL6A2	COL6A2, BTHLM1, UCMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bethlem myopathy 1 (BTHLM1) [MIM:158810]: A benign proximal myopathy characterized by early childhood onset and joint contractures most frequently affecting the elbows and ankles. (ECO:0000269|PubMed:11865138, ECO:0000269|PubMed:15689448, ECO:0000269|PubMed:17886299, ECO:0000269|PubMed:8782832). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 2, 616470 (Konjenital Müsküler Distrofi, Ullrich Tipi) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ullrich congenital muscular dystrophy 2, 616470 (Congenital muscular dystrophy, Ullrich type) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ullrich congenital muscular dystrophy 2 (UCMD2) [MIM:616470]: A form of Ullrich muscular dystrophy, a congenital myopathy characterized by muscle weakness and multiple joint contractures, generally noted at birth or early infancy. The clinical course is more severe than in Bethlem myopathy. (ECO:0000269	PubMed:24334604). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 2, 616470 (Konjenital Müsküler Distrofi, Ullrich Tipi) (Prenatal) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ullrich congenital muscular dystrophy 2, 616470 (Congenital muscular dystrophy, Ullrich type) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ullrich congenital muscular dystrophy 2 (UCMD2) [MIM:616470]: A form of Ullrich muscular dystrophy, a congenital myopathy characterized by muscle weakness and multiple joint contractures, generally noted at birth or early infancy. The clinical course is more severe than in Bethlem myopathy. (ECO:0000269	PubMed:24334604). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 2; Ucmd2 (Konjenital Müsküler Distrofi, Ullrich Tipi) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	ULLRICH CONGENITAL MUSCULAR DYSTROPHY 2; UCMD2 (Congenital muscular dystrophy, Ullrich type) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Ullrich congenital muscular dystrophy 2 (UCMD2) [MIM:616470]: A form of Ullrich muscular dystrophy, a congenital myopathy characterized by muscle weakness and multiple joint contractures, generally noted at birth or early infancy. The clinical course is more severe than in Bethlem myopathy. (ECO:0000269	PubMed:24334604). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Ullrich Konjenital Müsküler Distrofi 2; Ucmd2 (Konjenital Müsküler Distrofi, Ullrich Tipi) (Prenatal) (COL12A1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	ULLRICH CONGENITAL MUSCULAR DYSTROPHY 2; UCMD2 (Congenital muscular dystrophy, Ullrich type) (COL12A1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL12A1	COL12A1, UCMD2, BTHLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ullrich congenital muscular dystrophy 2 (UCMD2) [MIM:616470]: A form of Ullrich muscular dystrophy, a congenital myopathy characterized by muscle weakness and multiple joint contractures, generally noted at birth or early infancy. The clinical course is more severe than in Bethlem myopathy. (ECO:0000269	PubMed:24334604). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ulna ve Fibula, Yokluğu, Ağır Estremite Eksikliği, 276820, Otozomal Resesif (Fokomeli, Schinzel Tipi) (WNT7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ulna and fibula, absence of, with severe limb deficiency, 276820, Autosomal recessive (Phocomelia, Schinzel type) (WNT7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WNT7A	WNT7A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fuhrmann syndrome (FUHRS) [MIM:228930]: Distinct limb- malformation disorder characterized also by various degrees of limb aplasia/hypoplasia and joint dysplasia. [ECO:0000269	PubMed:16826533]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ulna ve Fibula, Yokluğu, Ağır Estremite Eksikliği, 276820, Otozomal Resesif (Fokomeli, Schinzel Tipi) (Prenatal) (WNT7A geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ulna and fibula, absence of, with severe limb deficiency, 276820, Autosomal recessive (Phocomelia, Schinzel type) (WNT7A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WNT7A	WNT7A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Fuhrmann syndrome (FUHRS) [MIM:228930]: Distinct limb- malformation disorder characterized also by various degrees of limb aplasia/hypoplasia and joint dysplasia. [ECO:0000269	PubMed:16826533]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ulnar-Meme Sendromu, 181450, Otozomal Dominant; UMS (Ulnar-Meme Sendromu) (TBX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ulnar-mammary syndrome, 181450, Autosomal dominant; UMS (Ulnar-mammary syndrome) (TBX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBX3	TBX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ulnar-Meme Sendromu, 181450, Otozomal Dominant; UMS (Ulnar-Meme Sendromu) (Prenatal) (TBX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ulnar-mammary syndrome, 181450, Autosomal dominant; UMS (Ulnar-mammary syndrome) (TBX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBX3	TBX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Taranamaz Saç Sendromu 2, 617251, Otozomal Resesif; UHS2 (Taranamaz Saç Sendromu) (TGM3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Uncombable hair syndrome 2, 617251, Autosomal recessive; UHS2 (Uncombable hair syndrome) (TGM3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TGM3	TGM3, UHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Taranamaz Saç Sendromu 2, 617251, Otozomal Resesif; UHS2 (Taranamaz Saç Sendromu) (Prenatal) (TGM3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Uncombable hair syndrome 2, 617251, Autosomal recessive; UHS2 (Uncombable hair syndrome) (TGM3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TGM3	TGM3, UHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Taranamaz Saç Sendromu 3, 617252, Otozomal Resesif; UHS3 (Taranamaz Saç Sendromu) (TCHH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Uncombable hair syndrome 3, 617252, Autosomal recessive; UHS3 (Uncombable hair syndrome) (TCHH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TCHH	TCHH, THH, THL, TRHY, UHS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Taranamaz Saç Sendromu, 191480, Otozomal Resesif; UHS1 (Taranamaz Saç Sendromu) (PADI3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Uncombable hair syndrome, 191480, Autosomal recessive; UHS1 (Uncombable hair syndrome) (PADI3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PADI3	PADI3, UHS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Unipolar Depresyon, Yatkınlık, 608516 (TPH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Unipolar depression, susceptibility to, 608516 (TPH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TPH2	TPH2, NTPH, ADHD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Major depressive disorder (MDD) [MIM:608516]: A common psychiatric disorder. It is a complex trait characterized by one or more major depressive episodes without a history of manic, mixed, or hypomanic episodes. A major depressive episode is characterized by at least 2 weeks during which there is a new onset or clear worsening of either depressed mood or loss of interest or pleasure in nearly all activities. Four additional symptoms must also be present including changes in appetite, weight, sleep, and psychomotor activity; decreased energy; feelings of worthlessness or guilt; difficulty thinking, concentrating, or making decisions; or recurrent thoughts of death or suicidal ideation, plans, or attempts. The episode must be accompanied by distress or impairment in social, occupational, or other important areas of functioning. {ECO:0000269	PubMed:15629698}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample
Urbach-Wiethe Hastalığı, 247100, Otozomal Resesif (Lipoid Proteinozis (Urbach-Wiethe Hastalığı)) (ECM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Urbach-Wiethe disease, 247100, Autosomal recessive (Lipoid proteinosis) (ECM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ECM1	ECM1, URBWD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Urbach-Wiethe Hastalığı, 247100, Otozomal Resesif (Lipoid Proteinozis (Urbach-Wiethe Hastalığı)) (Prenatal) (ECM1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Urbach-Wiethe disease, 247100, Autosomal recessive (Lipoid proteinosis) (ECM1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ECM1	ECM1, URBWD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ürik Asit Konsantrasyonu, Serum, QTL 2, 612076, Otozomal Resesif, Otozomal Dominant (Kalıtsal Renal Hipo/üriserni) (SLC2A9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Uric acid concentration, serum, QTL 2, 612076, Autosomal recessive, Autosomal dominant (Hereditary renal hypouricemia) (SLC2A9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC2A9	SLC2A9, GLUT9, UAQTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Hyperuricemia renal 2 (RHUC2) [MIM:612076]: A disorder characterized by impaired uric acid reabsorption at the apical membrane of proximal renal tubule cells, and high urinary urate excretion. Patients often appear asymptomatic, but may be subject to exercise-induced acute renal failure, chronic renal dysfunction and nephrolithiasis. {ECO:0000269|PubMed:19026395, ECO:0000269|PubMed:19926891, ECO:0000269|PubMed:21810765}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Üridin 5' Monofosfat Hidroliz Eksikliğine Bağlı (Hemolitik Anemi, Pirimidine 5' Nükleotidaz Eksikliğine Bağlı) (NT5C3A geni) (Tüm Gen Dizi Analizi) (Postnatal)	URIDINE 5-PRIME MONOPHOSPHATE HYDROLASE DEFICIENCY, HEMOLYTIC ANEMIA DUE TO (Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency) (NT5C3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NT5C3A	NT5C3A, NT5C3, UMPH1, PSN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: PSN deficiency (PSND) [MIM:266120]: Autosomal recessive condition causing hemolytic anemia characterized by marked basophilic stippling and the accumulation of high concentrations of pyrimidine nucleotides within the erythrocyte. It is implicated in the anemia of lead poisoning and is possibly associated with learning difficulties. {ECO:0000269|PubMed:11369620, ECO:0000269|PubMed:12930399, ECO:0000269|PubMed:15238149, ECO:0000269|PubMed:15604219, ECO:0000269|PubMed:15968458, ECO:0000269|PubMed:16461318, ECO:0000269|PubMed:18499901, ECO:0000269|PubMed:25153905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Hemolitik Anemi, Üridin 5' Monofosfat Hidroliz Eksikliğine Bağlı (Hemolitik Anemi, Pirimidine 5' Nükleotidaz Eksikliğine Bağlı) (Prenatal) (NT5C3A geni) (Tüm Gen Dizi Analizi) (Prenatal)	URIDINE 5-PRIME MONOPHOSPHATE HYDROLASE DEFICIENCY, HEMOLYTIC ANEMIA DUE TO (Hemolytic anemia due to pyrimidine 5' nucleotidase deficiency) (NT5C3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NT5C3A	NT5C3A, NT5C3, UMPH1, PSN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: PSN deficiency (PSND) [MIM:266120]: Autosomal recessive condition causing hemolytic anemia characterized by marked basophilic stippling and the accumulation of high concentrations of pyrimidine nucleotides within the erythrocyte. It is implicated in the anemia of lead poisoning and is possibly associated with learning difficulties. {ECO:0000269|PubMed:11369620, ECO:0000269|PubMed:12930399, ECO:0000269|PubMed:15238149, ECO:0000269|PubMed:15604219, ECO:0000269|PubMed:15968458, ECO:0000269|PubMed:16461318, ECO:0000269|PubMed:18499901, ECO:0000269|PubMed:25153905}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ürokanaz Eksikliği, 276880, Otozomal Resesif; UROCD (Ürokanik Asidüri) (UROC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Urocanase deficiency, 276880, Autosomal recessive; UROCD (Urocanic aciduria) (UROC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UROC1	UROC1, UROCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Urocanase deficiency (UROCD) [MIM:276880]: An inborn error of histidine metabolism resulting in urocanic aciduria and neurological manifestations including mental retardation, ataxia, episodic aggressive behavior or exaggerated affection-seeking. {ECO:0000269|PubMed:19304569}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ürokanaz Eksikliği, 276880, Otozomal Resesif; UROCD (Ürokanik Asidüri) (Prenatal) (UROC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Urocanase deficiency, 276880, Autosomal recessive; UROCD (Urocanic aciduria) (UROC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	UROC1	UROC1, UROCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Urocanase deficiency (UROCD) [MIM:276880]: An inborn error of histidine metabolism resulting in urocanic aciduria and neurological manifestations including mental retardation, ataxia, episodic aggressive behavior or exaggerated affection-seeking. {ECO:0000269|PubMed:19304569}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ürofasiyal Sendrom 1, 236730, Otozomal Resesif; UFS1 (Ochoa Sendromu) (HPSE2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Urofacial syndrome 1, 236730, Autosomal recessive; UFS1 (Ochoa syndrome) (HPSE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HPSE2	HPSE2, HPA2, UFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Urofacial syndrome 1 (UFS1) [MIM:236730]: A rare autosomal recessive disorder characterized by facial grimacing when attempting to smile and failure of the urinary bladder to void completely despite a lack of anatomical bladder outflow obstruction or overt neurological damage. Affected individuals often have reflux of infected urine from the bladder to the upper renal tract, with a risk of kidney damage and renal failure. {ECO:0000269|PubMed:20560209, ECO:0000269|PubMed:20560210}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Ürofasiyal Sendrom 1, 236730, Otozomal Resesif; UFS1 (Ochoa Sendromu) (Prenatal) (HPSE2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Urofacial syndrome 1, 236730, Autosomal recessive; UFS1 (Ochoa syndrome) (HPSE2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HPSE2	HPSE2, HPA2, UFS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Urofacial syndrome 1 (UFS1) [MIM:236730]: A rare autosomal recessive disorder characterized by facial grimacing when attempting to smile and failure of the urinary bladder to void completely despite a lack of anatomical bladder outflow obstruction or overt neurological damage. Affected individuals often have reflux of infected urine from the bladder to the upper renal tract, with a risk of kidney damage and renal failure. {ECO:0000269|PubMed:20560209, ECO:0000269|PubMed:20560210}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ürofasiyal Sendrom 2, 615112, Otozomal Resesif; UFS2 (Ochoa Sendromu) (LRIG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Urofacial syndrome 2, 615112, Autosomal recessive; UFS2 (Ochoa syndrome) (LRIG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRIG2	LRIG2, LIG2, KIAA0806, UFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ürofasiyal Sendrom 2, 615112, Otozomal Resesif; UFS2 (Ochoa Sendromu) (Prenatal) (LRIG2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Urofacial syndrome 2, 615112, Autosomal recessive; UFS2 (Ochoa syndrome) (LRIG2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LRIG2	LRIG2, LIG2, KIAA0806, UFS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Usher Sendromu Tip 3B, 614504, Otozomal Resesif; USH3B (Usher Sendromu) (HARS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome type 3B, 614504, Autosomal recessive; USH3B (Usher syndrome) (HARS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HARS	HARS, USH3B, CMT2W	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Charcot-Marie-Tooth disease 2W (CMT2W) [MIM:616625]: An autosomal dominant, axonal form of Charcot-Marie-Tooth disease, a disorder of the peripheral nervous system, characterized by progressive weakness and atrophy, initially of the peroneal muscles and later of the distal muscles of the arms. Charcot-Marie-Tooth disease is classified in two main groups on the basis of electrophysiologic properties and histopathology: primary peripheral demyelinating neuropathies (designated CMT1 when they are dominantly inherited) and primary peripheral axonal neuropathies (CMT2). Neuropathies of the CMT2 group are characterized by signs of axonal degeneration in the absence of obvious myelin alterations, normal or slightly reduced nerve conduction velocities, and progressive distal muscle weakness and atrophy. CMT2W patients manifest a peripheral neuropathy mainly affecting the lower limbs and resulting in gait difficulties and distal sensory impairment. Most patients also have upper limb involvement. {ECO:0000269|PubMed:22930593, ECO:0000269|PubMed:26072516}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Usher Sendromu, Tip 1B, 276900, Otozomal Resesif; USH1 (Usher Sendromu) (MYO7A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1B, 276900, Autosomal recessive; USH1 (Usher syndrome) (MYO7A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MYO7A	MYO7A, USH1B, DFNB2, DFNA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1B (USH1B) [MIM:276900]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. {ECO:0000269|PubMed:10094549, ECO:0000269|PubMed:10364543, ECO:0000269|PubMed:10447383, ECO:0000269|PubMed:10930322, ECO:0000269|PubMed:12112664, ECO:0000269|PubMed:15660226, ECO:0000269|PubMed:16679490, ECO:0000269|PubMed:23559863, ECO:0000269|PubMed:24831256, ECO:0000269|PubMed:25798947, ECO:0000269|PubMed:7870171, ECO:0000269|PubMed:8900236, ECO:0000269|PubMed:9002678, ECO:0000269|PubMed:9382091, ECO:0000269|PubMed:9718356}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Usher Sendromu, Tip 1C, 276904, Otozomal Resesif; USH1C (Usher Sendromu) (USH1C geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1C, 276904, Autosomal recessive; USH1C (Usher syndrome) (USH1C gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USH1C	USH1C, DFNB18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1C (USH1C) [MIM:276904]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. {ECO:0000269|PubMed:10073247}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Usher Sendromu, Tip 1D, 601067, Otozomal Resesif, Diganik Resesif (Usher Sendromu) (CDH23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1D, 601067, Autosomal recessive, Digenic recessive (Usher syndrome) (CDH23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH23	CDH23, USH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1D (USH1D) [MIM:601067]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. (ECO:0000269|PubMed:11138009, ECO:0000269|PubMed:12075507, ECO:0000269|PubMed:15660226, ECO:0000269|PubMed:16679490, ECO:0000269|PubMed:18429043, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Usher Sendromu, Tip 1D / F Dijenik, 601067, Otozomal Resesif, Diganik Resesif (Usher Sendromu) (CDH23 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1D/F digenic, 601067, Autosomal recessive, Digenic recessive (Usher syndrome) (CDH23 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CDH23	CDH23, USH1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1D (USH1D) [MIM:601067]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. (ECO:0000269|PubMed:11138009, ECO:0000269|PubMed:12075507, ECO:0000269|PubMed:15660226, ECO:0000269|PubMed:16679490, ECO:0000269|PubMed:18429043, Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Usher Sendromu, Tip 1D / F Dijenik, 601067, Otozomal Resesif, Diganik Resesif; USH1D (Usher Sendromu) (PCDH15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1D/F digenic, 601067, Autosomal recessive, Digenic recessive; USH1D (Usher syndrome) (PCDH15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCDH15	PCDH15, DFNB23, USH1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1D/F (USH1DF) [MIM:601067]: USH1DF patients are heterozygous for mutations in CDH23 and PCDH15, indicating a digenic inheritance pattern. (ECO:0000269|PubMed:15537665, ECO:0000269|PubMed:18719945). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry.	EDTA Blood Sample
Usher Sendromu, Tip 1F, 602083, Otozomal Resesif; USH1F (Usher Sendromu) (PCDH15 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1F, 602083, Autosomal recessive; USH1F (Usher syndrome) (PCDH15 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PCDH15	PCDH15, DFNB23, USH1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1DF (USH1DF) [MIM:601067]: USH1DF patients are heterozygous for mutations in CDH23 and PCDH15, indicating a digenic inheritance pattern. (ECO:0000269|PubMed:15537665, ECO:0000269|PubMed:18719945). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene reanserted in this entry.	EDTA Blood Sample
Usher Sendromu, Tip 1G, 606943, Otozomal Resesif; USH1G (Usher Sendromu) (USH1G geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 1G, 606943, Autosomal recessive; USH1G (Usher syndrome) (USH1G gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USH1G	SANS, USH1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=The first cases with non-syndromic sensorineural hearing loss based on mutations in USH1G. The hearing loss has an onset during early childhood, is progressive, and has a downsloping audiogram configuration. Ophthalmic and vestibular abnormalities are absent. (ECO:0000269	PubMed:15253398).	EDTA Blood Sample
Usher Sendromu, Tip 2A, 276901, Otozomal Resesif (Usher Sendromu) (USH2A geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 2A, 276901, Autosomal recessive (Usher syndrome) (USH2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	USH2A	USH2A, RP39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 2A (USH2A) [MIM:276901]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. (ECO:0000269|PubMed:10729113, ECO:0000269|PubMed:10738000, ECO:0000269|PubMed:10909849, ECO:0000269|PubMed:11311042, ECO:0000269|PubMed:12112664, ECO:0000269|PubMed:12525556, ECO:0000269|PubMed:14970843, ECO:0000269|PubMed:15015129, ECO:0000269|PubMed:15025721, ECO:0000269|PubMed:15241801, ECO:0000269|PubMed:15325563, ECO:0000269|PubMed:17085681, ECO:0000269|PubMed:17405132, ECO:0000269|PubMed:18273898, ECO:0000269|PubMed:18452394, ECO:0000269|PubMed:19683999, ECO:0000269|PubMed:19737284, ECO:0000269|PubMed:20309401, ECO:0000269|PubMed:20440071, ECO:0000269|PubMed:20507924, ECO:0000269|PubMed:21593743, ECO:0000269|PubMed:21686329, ECO:0000269|PubMed:22004887, ECO:0000269|PubMed:23737954, ECO:0000269|PubMed:9624853). Note=The disease is caused	EDTA Blood Sample
Usher Sendromu, Tip 2C, 605472, Otozomal Resesif, Diganik Dominant (Usher Sendromu) (ADGRV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 2C, 605472, Autosomal recessive, Digenic dominant (Usher syndrome) (ADGRV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRV1	ADGRV1, GPR98, MASS1, VLGR1, KIAA0686, FEB4, USH2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Febrile seizures, familial, 4 (FEB4) [MIM:604352]: Seizures associated with febrile episodes in childhood without any evidence of intracranial infection or defined pathologic or traumatic cause. It is a common condition, affecting 2-5% of children aged 3 months to 5 years. The majority are simple febrile seizures (generally defined as generalized onset, single seizures with a duration of less than 30 minutes). Complex febrile seizures are characterized by focal onset, duration greater than 30 minutes, and/or more than one seizure in a 24 hour period. The likelihood of developing epilepsy following simple febrile seizures is low. Complex febrile seizures are associated with a moderately increased incidence of epilepsy. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Usher Sendromu, Tip 2C, GPR98 / PDZD7 Dijenik, 605472, Otozomal Resesif, Diganik Dominant; USH2C (Usher Sendromu) (ADGRV1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 2C, GPR98/PDZD7 digenic, 605472, Autosomal recessive, Digenic dominant; USH2C (Usher syndrome) (ADGRV1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRV1	ADGRV1, GPR98, MASS1, VLGR1, KIAA0686, FEB4, USH2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Febrile seizures, familial, 4 (FEB4) [MIM:604352]: Seizures associated with febrile episodes in childhood without any evidence of intracranial infection or defined pathologic or traumatic cause. It is a common condition, affecting 2-5% of children aged 3 months to 5 years. The majority are simple febrile seizures (generally defined as generalized onset, single seizures with a duration of less than 30 minutes). Complex febrile seizures are characterized by focal onset, duration greater than 30 minutes, and/or more than one seizure in a 24 hour period. The likelihood of developing epilepsy following simple febrile seizures is low. Complex febrile seizures are associated with a moderately increased incidence of epilepsy. Note=The disease may be caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Usher Sendromu, Tip 2D, 611383, Otozomal Resesif; USH2D (Usher Sendromu) (WHRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 2D, 611383, Autosomal recessive; USH2D (Usher syndrome) (WHRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WHRN	WHRN, CIP98, KIAA1526, DFNB31, USH2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Usher Sendromu, Tip 3A, 276902, Otozomal Resesif; USH3A (Usher Sendromu) (CLRN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type 3A, 276902, Autosomal recessive; USH3A (Usher syndrome) (CLRN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CLRN1	CLRN1, USH3A, USH3, RP61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinitis pigmentosa 61 (RP61) [MIM:614180]: A retinal dystrophy belonging to the group of pigmentary retinopathies. Retinitis pigmentosa is characterized by retinal pigment deposits visible on fundus examination and primary loss of rod photoreceptor cells followed by secondary loss of cone photoreceptors. Patients typically have night vision blindness and loss of midperipheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well. (ECO:0000269	PubMed:21310491). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Usher Sendromu, Tip 2C, GPR98 / PDZD7 Dijenik, 605472, Otozomal Resesif, Diganik Dominant (Usher Sendromu) (PDZD7 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type IIC, GPR98/PDZD7 digenic, 605472, Autosomal recessive, Digenic dominant; (Usher syndrome) (PDZD7 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PDZD7	PDZD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 2C (USH2C) [MIM:605472]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH2 is characterized by congenital mild hearing impairment with normal vestibular responses. (ECO:0000269|PubMed:20440071). Note=The disease is caused by mutations affecting distinct genetic loci, including the gene represented in this entry. PDZD7 mutations have been found in combination with mutations in USH2A and GPR98 in patients affected by Usher syndrome, suggesting a role as contributor to digenic Usher syndrome or a modifier of retinal disease expression. (ECO:0000269|PubMed:20440071).;	EDTA Blood Sample
Usher Sendromu, Tip 1J, 614869, Otozomal Resesif; USH1J (Usher Sendromu) (CIB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Usher syndrome, type IJ, 614869, Autosomal recessive; USH1J (Usher syndrome) (CIB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CIB2	CIB2, KIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Usher syndrome 1J (USH1J) [MIM:614869]: USH is a genetically heterogeneous condition characterized by the association of retinitis pigmentosa with sensorineural deafness. Age at onset and differences in auditory and vestibular function distinguish Usher syndrome type 1 (USH1, Usher syndrome type 2 (USH2) and Usher syndrome type 3 (USH3). USH1 is characterized by profound congenital sensorineural deafness, absent vestibular function and prepubertal onset of progressive retinitis pigmentosa leading to blindness. (ECO:0000269	PubMed:23023331). Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
UV Işınları İle Uyarılmış Deri Hasarı, 266300, Otozomal Resesif (MC1R geni) (Tüm Gen Dizi Analizi) (Postnatal)	UV-induced skin damage, 266300, Autosomal recessive (MC1R gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MC1R	MC1R, SHEP2, CMM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Melanoma, cutaneous malignant 5 (CMM5) [MIM:613099]: A malignant neoplasm of melanocytes, arising de novo or from a pre- existing benign nevus, which occurs most often in the skin but also may involve other sites. (ECO:0000269|PubMed:17434924, ECO:0000269|PubMed:19338054). Note=Disease susceptibility is associated with variations affecting the gene represented in this entry.	EDTA Blood Sample

UV Sensitivite Sendromu 1, 600630, Otozomal Resesif; UVSS1 (UV Hassasiyeti Sendromu) (ERCC6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	UV-sensitive syndrome 1, 600630, Autosomal recessive; UVSS1 (UV-sensitive syndrome) (ERCC6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC6	ERCC6, CKN2, COFS1, CSB, ARMDS, UVSS1, POF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome B (CSB) [MIM:133540]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:19894250, ECO:0000269|PubMed:9443879}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
UV Sensitivite Sendromu 2, 614621, Otozomal Resesif; UVSS2 (UV Hassasiyeti Sendromu) (ERCC8 geni) (Tüm Gen Dizi Analizi) (Postnatal)	UV-sensitive syndrome 2, 614621, Autosomal recessive; UVSS2 (UV-sensitive syndrome) (ERCC8 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC8	ERCC8, CKN1, CSA, UVSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cockayne syndrome A (CSA) [MIM:216400]: A rare disorder characterized by cutaneous sensitivity to sunlight, abnormal and slow growth, cachectic dwarfism, progeroid appearance, progressive pigmentary retinopathy and sensorineural deafness. There is delayed neural development and severe progressive neurologic degeneration resulting in mental retardation. Two clinical forms are recognized: in the classical form or Cockayne syndrome type 1, the symptoms are progressive and typically become apparent within the first few years or life; the less common Cockayne syndrome type 2 is characterized by more severe symptoms that manifest prenatally. Cockayne syndrome shows some overlap with certain forms of xeroderma pigmentosum. Unlike xeroderma pigmentosum, patients with Cockayne syndrome do not manifest increased freckling and other pigmentation abnormalities in the skin and have no significant increase in skin cancer. {ECO:0000269|PubMed:14661080, ECO:0000269|PubMed:15744458, ECO:0000269|PubMed:19894250}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
UV Sensitivite Sendromu 3, 614640, Otozomal Resesif; UVSS3 (UV Hassasiyeti Sendromu) (UVSSA geni) (Tüm Gen Dizi Analizi) (Postnatal)	UV-sensitive syndrome 3, 614640, Autosomal recessive; UVSS3 (UV-sensitive syndrome) (UVSSA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	UVSSA	UVSSA, KIAA1530, UVSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: UV-sensitive syndrome 3 (UVSS3) [MIM:614640]: An autosomal recessive disorder characterized by cutaneous photosensitivity and slight dyspigmentation, without an increased risk of skin tumors. {ECO:0000269|PubMed:22466610, ECO:0000269|PubMed:22466611, ECO:0000269|PubMed:22466612}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow-Sorauf Sendromu (Robinow-Sorauf Sendromu) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Robinow-Sorauf syndrome (Robinow-Sorauf syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
Robinow-Sorauf Sendromu (Robinow-Sorauf Sendromu) (Prenatal) (TWIST1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	v SYNDROME (Robinow-Sorauf syndrome) (TWIST1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TWIST1	TWIST1, ACS3, SCS, CRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Craniosynostosis 1 (CRS1) [MIM:123100]: A primary abnormality of skull growth involving premature fusion of one or more cranial sutures. The growth velocity of the skull often cannot match that of the developing brain resulting in an abnormal head shape and, in some cases, increased intracranial pressure, which must be treated promptly to avoid permanent neurodevelopmental disability. {ECO:0000269|PubMed:17343269}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
VACTERL Asosiasyonu-Hidrosefali (VACTERL-Hidrosefali) (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	VACTERL ASSOCIATION WITH HYDROCEPHALUS (VACTERL with hydrocephalus) (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
VACTERL Asosiasyonu-Hidrosefali (VACTERL-Hidrosefali) (MLPA)	VACTERL ASSOCIATION WITH HYDROCEPHALUS (VACTERL with hydrocephalus) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
VACTERL Asosiasyonu-Hidrosefali (VACTERL-Hidrosefali) (Prenatal) (PTEN geni) (Tüm Gen Dizi Analizi) (Prenatal)	VACTERL ASSOCIATION WITH HYDROCEPHALUS (VACTERL with hydrocephalus) (PTEN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
VACTERL Asosiasyonu-Hidrosefali (VACTERL-Hidrosefali) (Prenatal) (MLPA)	VACTERL ASSOCIATION WITH HYDROCEPHALUS (VACTERL with hydrocephalus) (Prenatal) (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241266, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
VACTERL Asosiasyonu, X'e Bağlı, 314390, X'e Bağlı Resesif; VACTERLX (VACTERL-Hidrosefali) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	VACTERL association, X-linked, 314390, X-linked recessive; VACTERLX (VACTERL with hydrocephalus) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
VACTERL Asosiasyonu, X'e Bağlı, 314390, X'e Bağlı Resesif; VACTERLX (VACTERL-Hidrosefali) (Prenatal) (ZIC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	VACTERL association, X-linked, 314390, X-linked recessive; VACTERLX (VACTERL with hydrocephalus) (ZIC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZIC3	ZIC3, HTX1, HTX, VACTERLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heterotaxy, visceral, 1, X-linked (HTX1) [MIM:306955]: A form of visceral heterotaxy, a complex disorder due to disruption of the normal left-right asymmetry of the thoracoabdominal organs. Visceral heterotaxy or situs ambiguus results in randomization of the placement of visceral organs, including the heart, lungs, liver, spleen, and stomach. The organs are oriented randomly with respect to the left-right axis and with respect to one another. It can been associated with variety of congenital defects including cardiac malformations. {ECO:0000269|PubMed:14681828, ECO:0000269|PubMed:17295247, ECO:0000269|PubMed:24123890, ECO:0000269|PubMed:9354794}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Van Buchem Hastalığı, 239100, Otozomal Resesif (Hiperostozis Kortikalis Jeneralizata (Van Buchem Hastalığı)) (SOST geni) (Tüm Gen Dizi Analizi) (Postnatal)	Van Buchem disease, 239100, Autosomal recessive (Hyperostosis corticalis generalisata) (SOST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOST	SOST, VBCH, CDD, SOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Craniodiaphyseal dysplasia autosomal dominant (CDD) [MIM:122860]: A severe bone dysplasia characterized by massive generalized hyperostosis and sclerosis, especially involving the skull and facial bones. The sclerosis is so severe that the resulting facial distortion is referred to as 'leontiasis ossea' (leonine faces) and the bone deposition results in progressive stenosis of craniofacial foramina. Respiratory obstruction due to choanal stenosis compromises the clinical outcomes of affected patients. {ECO:0000269|PubMed:21221996}. Note=The disease is caused by mutations affecting the gene represented in this entry. Heterozygous mutations located in the secretion signal of the SOST gene prevent sclerostin secretion and can be responsible for craniodiaphyseal dysplasia.	EDTA Blood Sample
Van Buchem Hastalığı, Tip 2, 607636, Otozomal Dominı (Hiperostozis Kortikalis Jeneralizata (Van Buchem Hastalığı)) (LRP5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	van Buchem disease, type 2, 607636, Autosomal domin (Hyperostosis corticalis generalisata) (LRP5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LRP5	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2, OPTA1, EVR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Endosteal hyperostosis, Worth type (WENHY) [MIM:144750]: An autosomal dominant sclerosing bone dysplasia clinically characterized by elongation of the mandible, increased gonial angle, flattened forehead, and the presence of a slowly enlarging osseous prominence of the hard palate (torus palatinus). Serum calcium, phosphorus and alkaline phosphatase levels are normal. Radiologically, it is characterized by early thickening of the endosteum of long bones, the skull and of the mandible. With advancing age, the trabeculae of the metaphysis become thickened. WENHY becomes clinically and radiologically evident by adolescence, does not cause deformity except in the skull and mandible, and is not associated with bone pain or fracture. Affected patients have normal height, proportion, intelligence and longevity. {ECO:0000269|PubMed:12579474}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Van Den Ende-Gupta Sendromu, 600920, Otozomal Resesif; VDEGS (Van Den Ende-Gupta Sendromu) (SCARF2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Van den Ende-Gupta syndrome, 600920, Autosomal recessive; VDEGS (Van den Ende-Gupta syndrome) (SCARF2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SCARF2	SCARF2, SREC2, VDEGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Van Den Ende-Gupta Sendromu, 600920, Otozomal Resesif; VDEGS (Van Den Ende-Gupta Sendromu) (Prenatal) (SCARF2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Van den Ende-Gupta syndrome, 600920, Autosomal recessive; VDEGS (Van den Ende-Gupta syndrome) (SCARF2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SCARF2	SCARF2, SREC2, VDEGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Van Der Woude Sendromu 2, 606713, Otozomal Dominant; VWS2 (Van Der Woude Sendromu) (GRHL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Van der Woude syndrome 2, 606713, Autosomal dominant; VWS2 (Van der Woude syndrome) (GRHL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRHL3	GRHL3, SOM, TFCP2L4, VWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Van Der Woude Sendromu 2, 606713, Otozomal Dominant; VWS2 (Van Der Woude Sendromu) (Prenatal) (GRHL3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Van der Woude syndrome 2, 606713, Autosomal dominant; VWS2 (Van der Woude syndrome) (GRHL3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GRHL3	GRHL3, SOM, TFCP2L4, VWS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Van Der Woude Sendromu, 119300, Otozomal Dominant; VWS1 (Van Der Woude Sendromu) (IRF6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	van der Woude syndrome, 119300, Autosomal dominant; VWS1 (Van der Woude syndrome) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. (ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Van Der Woude Sendromu, 119300, Otozomal Dominant; VWS1 (Van Der Woude Sendromu) (Prenatal) (IRF6 geni) (Tüm Gen Dizi Analizi) (Prenatal)	van der Woude syndrome, 119300, Autosomal dominant; VWS1 (Van der Woude syndrome) (IRF6 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	IRF6	IRF6, VWS, LPS, PIT, PPS1, OFC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Van der Woude syndrome 1 (VWS1) [MIM:119300]: An autosomal dominant developmental disorder characterized by lower lip pits, cleft lip and/or cleft palate. (ECO:0000269|PubMed:12219090, ECO:0000269|PubMed:12920575, ECO:0000269|PubMed:14618417, ECO:0000269|PubMed:14640121, ECO:0000269|PubMed:15300989, ECO:0000269|PubMed:17122170, ECO:0000269|PubMed:18478600). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Van Maldergem Sendromu 1, 601390, Otozomal Resesif; VMLDS1 (Serebrofasiyoartiküler Sendromu) (DCHS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Van Maldergem syndrome 1, 601390, Autosomal recessive; VMLDS1 (Cerebro-facio-articular syndrome) (DCHS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCHS1	DCHS1, PCDH16, FIB1, CDH19, VMLDS1, MVP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Mitral valve prolapse 2 (MVP2) [MIM:607829]: A form of mitral valve prolapse, a valvular hearth disease characterized by abnormally elongated and thickened mitral valve leaflets, that typically show myxomatous degeneration with increased leaflet compliance. It is associated with mitral regurgitation. Myxomatous mitral valves have an abnormal layered architecture characterized by loose collagen in fibrosa, expanded spongiosa strongly positive for proteoglycans, and disrupted elastin in atrialis. In classic mitral valve prolapse, leaflets are at least 5 mm thick, whereas in the non-classic form, they are less than 5 mm thick. Severe classic mitral valve prolapse is strongly associated with arrhythmias, endocarditis, heart failure, and need for valve surgery. (ECO:0000269	PubMed:26258302). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Van Maldergem Sendromu 1, 601390, Otozomal Resesif; VMLDS1 (Serebrofasiyoartiküler Sendromu) (Prenatal) (DCHS1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Van Maldergem syndrome 1, 601390, Autosomal recessive; VMLDS1 (Cerebro-facio-articular syndrome) (DCHS1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCHS1	DCHS1, PCDH16, FIB1, CDH19, VMLDS1, MVP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Mitral valve prolapse 2 (MVP2) [MIM:607829]: A form of mitral valve prolapse, a valvular hearth disease characterized by abnormally elongated and thickened mitral valve leaflets, that typically show myxomatous degeneration with increased leaflet compliance. It is associated with mitral regurgitation. Myxomatous mitral valves have an abnormal layered architecture characterized by loose collagen in fibrosa, expanded spongiosa strongly positive for proteoglycans, and disrupted elastin in atrialis. In classic mitral valve prolapse, leaflets are at least 5 mm thick, whereas in the non-classic form, they are less than 5 mm thick. Severe classic mitral valve prolapse is strongly associated with arrhythmias, endocarditis, heart failure, and need for valve surgery. (ECO:0000269	PubMed:26258302). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Van Maldergem Sendromu 2, 615546, Otozomal Resesif; VMLDS2 (Serebrofasiyoartiküler Sendromu) (FAT4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Van Maldergem syndrome 2, 615546, Autosomal recessive; VMLDS2 (Cerebro-facio-articular syndrome) (FAT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FAT4	FAT4, VMLDS2, HKLLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Van Maldergem syndrome 2 (VMLDS2) [MIM:615546]: An autosomal recessive disorder characterized by intellectual disability, typical craniofacial features, auditory malformations resulting in hearing loss, and skeletal and limb malformations. Some patients have renal hypoplasia. Brain MRI typically shows periventricular nodular heterotopia. (ECO:0000269	PubMed:24056717). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Van Maldergem Sendromu 2, 615546, Otozomal Resesif; VMLDS2 (Serebrofasiyoartiküler Sendromu) (Prenatal) (FAT4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Van Maldergem syndrome 2, 615546, Autosomal recessive; VMLDS2 (Cerebro-facio-articular syndrome) (FAT4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FAT4	FAT4, VMLDS2, HKLLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (KİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Van Maldergem syndrome 2 (VMLDS2) [MIM:615546]: An autosomal recessive disorder characterized by intellectual disability, typical craniofacial features, auditory malformations resulting in hearing loss, and skeletal and limb malformations. Some patients have renal hypoplasia. Brain MRI typically shows periventricular nodular heterotopia. (ECO:0000269	PubMed:24056717). Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çeşitli tümör tipleri, Epileptik ensefalopati (FHIT (3p14.2), WWOX (16q23.1)) (MLPA)	Various tumor types, Epileptic encephalopathy (FHIT (3p14.2), WWOX (16q23.1)) (MLPA)	FHIT (3p14.2), WWOX (16q23.1)	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	.	EDTA Blood Sample
Vas Deferens, Konjenital Bilateral Aplazisi; X'e Bağlı, 300985, X'e Bağlı; CBAVDX (Konjenital Bilateral Vaz Deferens Yokluğu) (ADGRG2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Vas deferens, congenital bilateral aplasia of, X-linked, 300985, X-linked; CBAVDX (Congenital bilateral absence of vas deferens) (ADGRG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRG2	ADGRG2, GPR64, HE6, CBAVDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vas Deferens, Konjenital Bilateral Aplazisi; CBAVD (Konjenital Bilateral Vaz Deferens Yokluğu) (CFTR geni) (Tüm Gen Dizi Analizi) (Postnatal)	VAS DEFERENS, CONGENITAL BILATERAL APLASIA OF; CBAVD (Congenital bilateral absence of vas deferens) (CFTR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CFTR	CFTR, ABCC7, CF, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	EDTA Blood Sample
Vas Deferens, Konjenital Bilateral Aplazisi; CBAVD (Konjenital Bilateral Vaz Deferens Yokluğu) (MLPA)	VAS DEFERENS, CONGENITAL BILATERAL APLASIA OF; CBAVD (Congenital bilateral absence of vas deferens) (MLPA)	CFTR	CFTR, ABCC7, CF, MRP7	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Cystic fibrosis (CF) [MIM:219700]: A common generalized disorder of the exocrine glands which impairs clearance of secretions in a variety of organs. It is characterized by the triad of chronic bronchopulmonary disease (with recurrent respiratory infections), pancreatic insufficiency (which leads to malabsorption and growth retardation) and elevated sweat electrolytes. It is the most common genetic disease in Caucasians, with a prevalence of about 1 in 2'000 live births. Inheritance is autosomal recessive. (ECO:0000269|PubMed:10094564, ECO:0000269|PubMed:1284466, ECO:0000269|PubMed:1284468, ECO:0000269|PubMed:1284529, ECO:0000269|PubMed:1284530, ECO:0000269|PubMed:1695717, ECO:0000269|PubMed:1710600, ECO:0000269|PubMed:2236053, ECO:0000269|PubMed:7504969, ECO:0000269|PubMed:7505694, ECO:0000269|PubMed:7513296, ECO:0000269|PubMed:7517264, ECO:0000269|PubMed:7520022, ECO:0000269|PubMed:7522211, ECO:0000269|PubMed:7524909, ECO:0000269|PubMed:7524913, ECO:0000269|PubMed:7525450, ECO:0000269|PubMed:7537150, ECO:0000269|PubMed:7541273, ECO:0000269|PubMed:7541510, ECO:0000269|PubMed:7543567, ECO:0000269|PubMed:7544319, ECO:0000269|PubMed:7581407.	EDTA Blood Sample
Vasküler Hastalık, Yatkınlık (MTHFR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Vascular disease, susceptibility to (MTHFR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MTHFR	MTHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. (ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030). Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample

Vasküler Hastalık, Yatkınlık (MLPA)	Vascular disease, susceptibility to (MLPA)	MTHFR	MTHFR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Methylenetetrahydrofolate reductase deficiency (MTHFRD) [MIM:236250]: Autosomal recessive disorder with a wide range of features including homocysteinuria, homocysteinemia [MIM:603174], developmental delay, severe mental retardation, perinatal death, psychiatric disturbances, and later-onset neurodegenerative disorders. {ECO:0000269|PubMed:10679944, ECO:0000269|PubMed:20236116, ECO:0000269|PubMed:25736335, ECO:0000269|PubMed:7726158, ECO:0000269|PubMed:8940272, ECO:0000269|PubMed:9781030}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Vaskiler Malformasyon, Primer Intraosseöz, 606893, Otozomal Resesif (Primer İntraosseöz Venöz Anomali) (ELMO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Vascular malformation, primary intraosseous, 606893, Autosomal recessive (Primary intraosseous venous malformation) (ELMO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ELMO2	ELMO2, CED12, VMPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vaskülopati, Retinal, Serebral Lökodistrofi, 192315, Otozomal Dominant, RVCL (Retina Vaskülopati ve Serebral Lökoensefalopati) (TREX1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Vasculopathy, retinal, with cerebral leukodystrophy, 192315, Autosomal dominant; RVCL (Retinal vasculopathy and cerebral leukoencephalopathy) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:17660818, ECO:0000269|PubMed:20131292}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Enhanced immune sensing of oxidized DNA may be involved in the phototoxicity experienced by SLE patients. Exposure to UV- light produces DNA oxidative damage. Oxidized DNA being a poor TREX1 substrate, it accumulates in skin, leading to enhanced auto- immune reactivity and eventually skin lesions (PubMed:23993650). {ECO:0000269|PubMed:23993650}.	EDTA Blood Sample
Vaskülopati, Retinal, Serebral Lökodistrofi, 192315, Otozomal Dominant, RVCL (Retina Vaskülopati ve Serebral Lökoensefalopati) (Prenatal) (TREX1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Vasculopathy, retinal, with cerebral leukodystrophy, 192315, Autosomal dominant; RVCL (Retinal vasculopathy and cerebral leukoencephalopathy) (TREX1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TREX1	TREX1, AGS1, CRV, HERNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Systemic lupus erythematosus (SLE) [MIM:152700]: A chronic, relapsing, inflammatory, and often febrile multisystemic disorder of connective tissue, characterized principally by involvement of the skin, joints, kidneys and serosal membranes. It is of unknown etiology, but is thought to represent a failure of the regulatory mechanisms of the autoimmune system. The disease is marked by a wide range of system dysfunctions, an elevated erythrocyte sedimentation rate, and the formation of LE cells in the blood or bone marrow. {ECO:0000269|PubMed:17660818, ECO:0000269|PubMed:20131292}. Note=Disease susceptibility is associated with variations affecting the gene represented in this entry. Enhanced immune sensing of oxidized DNA may be involved in the phototoxicity experienced by SLE patients. Exposure to UV- light produces DNA oxidative damage. Oxidized DNA being a poor TREX1 substrate, it accumulates in skin, leading to enhanced auto- immune reactivity and eventually skin lesions (PubMed:23993650). {ECO:0000269|PubMed:23993650}.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
VATER Asosiasyonu - Makrosefali ve Ventrikülomegali, 276950, Otozomal Resesif (VACTERL-Hidrosefali) - VACTERL-H Sendromu (PTEN geni) (Tüm Gen Dizi Analizi) (Postnatal)	VATER association with macrocephaly and ventriculomegaly, 276950, autosomal recessive (VACTERL with hydrocephalus) - VACTERL-H syndrome (PTEN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241366, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
VATER Asosiasyonu - Makrosefali ve Ventrikülomegali, 276950, Otozomal Resesif (VACTERL-Hidrosefali) - VACTERL-H Sendromu (MLPA)	VATER association with macrocephaly and ventriculomegaly, 276950, autosomal recessive (VACTERL with hydrocephalus) - VACTERL-H syndrome (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241366, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
VATER Asosiasyonu - Makrosefali ve Ventrikülomegali, 276950, Otozomal Resesif (VACTERL-Hidrosefali) - VACTERL-H Sendromu (Prenatal) (PTEN geni) (Tüm Gen Dizi Analizi) (Prenatal)	VATER association with macrocephaly and ventriculomegaly, 276950, autosomal recessive (VACTERL with hydrocephalus) - VACTERL-H syndrome (PTEN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PTEN	PTEN, MMAC1, GLM2, CWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Bannayan-Riley-Ruvalcaba syndrome (BRRS) [MIM:153480]: A rare hamartomatous disorder characterized by macrocephaly and multiple hemangiomas as well as subcutaneous and visceral lipomas. It belongs to the family of hamartomatous polyposis syndromes that includes Peutz Jeghers syndrome, juvenile polyposis, and Cowden syndrome. {ECO:0000269|PubMed:10400993, ECO:0000269|PubMed:11494117, ECO:0000269|PubMed:9241366, ECO:0000269|PubMed:9467011}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
VATER Asosiasyonu - Makrosefali ve Ventrikülomegali, 276950, Otozomal Resesif (VACTERL-Hidrosefali) - VACTERL-H Sendromu (Prenatal) (MLPA)	VATER association with macrocephaly and ventriculomegaly, 276950, autosomal recessive (VACTERL with hydrocephalus) - VACTERL-H syndrome (MLPA)	PTEN	PTEN, MMAC1, GLM2, CWS1	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Velokardiofasiyal Sendromu, 192430, Otozomal Dominant (22q11.2 Delesyon Sendromu) (FISH)	Velocardiofacial syndrome, 192430, Autosomal dominant (22q11.2 deletion syndrome) (FISH)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Velokardiofasiyal Sendrom, 192430, Otozomal Dominant (22q11.2 Delesyon Sendromu) (MLPA)	Velocardiofacial syndrome, 192430, Autosomal dominant (22q11.2 deletion syndrome) (MLPA)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Velokardiofasiyal Sendromu, 192430, Otozomal Dominant (22q11.2 Delesyon Sendromu) (Prenatal) (FISH)	Velocardiofacial syndrome, 192430, Autosomal dominant (22q11.2 deletion syndrome) (Prenatal) (FISH)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Velokardiofasiyal Sendrom, 192430, Otozomal Dominant (22q11.2 Delesyon Sendromu) (Prenatal) (MLPA)	Velocardiofacial syndrome, 192430, Autosomal dominant (22q11.2 deletion syndrome) (Prenatal) (MLPA)	TBX1	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kemik İliği Naklinden Sonra Venookluzif Hastalık (CPS1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Venoocclusive disease after bone marrow transplantation (CPS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CPS1	CPS1, PHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Venöz Malformasyonlar, Multipl Deri ve Mukoza, 600195, Otozomal Dominant; VMCM (Mukokutanöz Venöz Malformasyonlar) (TEK geni) (Tüm Gen Dizi Analizi) (Postnatal)	Venous malformations, multiple cutaneous and mucosal, 600195, Autosomal dominant; VMCM (Mucocutaneous venous malformations) (TEK gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TEK	TEK, TIE2, VMCM, GLC3E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Venöz Tromboembolizm, Yatkınlık, 188050, Otozomal Dominant (HABP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Venous thromboembolism, susceptibility to, 188050, Autosomal dominant (HABP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HABP2	HABP2, PHBP, HGFAL, FSAP, NMTC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Venöz Tromboz, Karşı Koruma, 188050, Otozomal Dominant (F13A1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Venous thrombosis, protection against, 188050, Autosomal dominant (F13A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F13A1	F13A1, F13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikuler Fibrilasyon, Paroksismal Allesel, 2, 612956, Otozomal Dominant; VF2 (Idiopatik Ventrikuler Fibrilasyon, Brugada Tipi Olmayan) (DPP6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular fibrillation, paroxysmal familial, 2, 612956, Autosomal dominant; VF2 (idiopathic ventricular fibrillation, not Brugada type) (DPP6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DPP6	DPP6, VF2, MRD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikuler Septal Defekt 1, 614429, Otozomal Dominant; VSD1 (GATA4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular septal defect 1, 614429, Autosomal dominant; VSD1 (GATA4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikuler Septal Defekt 1, 614429, Otozomal Dominant; VSD1 (MLPA)	Ventricular septal defect 1, 614429, Autosomal dominant; VSD1, MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Ventrikuler Septal Defekt 1, 614429, Otozomal Dominant; VSD1 (Prenatal) (GATA4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ventricular septal defect 1, 614429, Autosomal dominant; VSD1 (GATA4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ventrikuler Septal Defekt 1, 614429, Otozomal Dominant; VSD1 (Prenatal) (MLPA)	Ventricular septal defect 1, 614429, Autosomal dominant; VSD1 (Prenatal) (MLPA)	GATA4	GATA4, ASD2, VSD1, TACHD, TOF	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ventrikuler Septal Defekt 2, 614431, Otozomal Dominant; VSD2 (CITED2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular septal defect 2, 614431, Autosomal dominant; VSD2 (CITED2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CITED2	CITED2, MRG1, P35SRJ, VSD2, ASD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikuler Septal Defekt 2, 614431, Otozomal Dominant; VSD2 (Prenatal) (CITED2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ventricular septal defect 2, 614431, Autosomal dominant; VSD2 (CITED2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CITED2	CITED2, MRG1, P35SRJ, VSD2, ASD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ventrikuler Septal Defekt 3, 614432, Otozomal Dominant; VSD3 (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular septal defect 3, 614432, Autosomal dominant; VSD3 (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikuler Septal Defekt 3, 614432, Otozomal Dominant; VSD3 (Prenatal) (NKX2-5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Ventricular septal defect 3, 614432, Autosomal dominant; VSD3 (NKX2-5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NKX2-5	NKX2-5, NKX2E, CSX, CHNG5, VSD3, HLHS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ventrikiller Taşikardi, Katekolaminerjik Polimorfik, 1, 604772, Otozomal Dominant; CPVT1 (Katekolaminerjik Polimorfik Ventrikiller Taşikardi) (RYR2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, catecholaminergic polymorphic, 1, 604772, Autosomal dominant; CPVT1 (Catecholaminergic polymorphic ventricular tachycardia) (RYR2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RYR2	RYR2, VTSIP, ARVD2, ARVC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikiller Taşikardi, Katekolaminerjik Polimorfik, 2, 611938, Otozomal Resesif; CPVT2 (Katekolaminerjik Polimorfik Ventrikiller Taşikardi) (CASQ2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, catecholaminergic polymorphic, 2, 611938, Autosomal recessive; CPVT2 (Catecholaminergic polymorphic ventricular tachycardia) (CASQ2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CASQ2	CASQ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikiller Taşikardi, Katekolaminerjik Polimorfik, 3, 614021, Otozomal Resesif; CPVT3 (Katekolaminerjik Polimorfik Ventrikiller Taşikardi) (TECRL geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, catecholaminergic polymorphic, 3, 614021, Autosomal recessive; CPVT3 (Catecholaminergic polymorphic ventricular tachycardia) (TECRL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TECRL	TECRL, TERL, SRD5A2L2, GPSNZL, CPVT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikiller Taşikardi, Katekolaminerjik Polimorfik, 4, 614916, Otozomal Dominant; CPVT4 (Katekolaminerjik Polimorfik Ventrikiller Taşikardi) (CALM1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, catecholaminergic polymorphic, 4, 614916, Autosomal dominant; CPVT4 (Catecholaminergic polymorphic ventricular tachycardia) (CALM1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CALM1	CALM1, PHKD, CPVT4, LQT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Ventrikiler Taşikardi, Katekolaminerjik Polimorfik, 5, ± Kas Zayıflığı 615441, Otozomal Resesif; CPVT5 (Katekolaminerjik Polimorfik Ventrikiler Taşikardi) (TRDN gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, catecholaminergic polymorphic, 5, with or without muscle weakness, 615441, Autosomal recessive; CPVT5 (Catecholaminergic polymorphic ventricular tachycardia) (TRDN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TRDN	TRDN, TDN, CPVT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikiler Taşikardi, İdyopatik, 192605, Otozomal Dominant (GNAI2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ventricular tachycardia, idiopathic, 192605, Autosomal dominant (GNAI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GNAI2	GNAI2, GNAI2B, GIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikülomegali-Kistik Böbrek Hastalığı, 219730, Otozomal Resesif; VMCKD (Ventrikülomegali-Kistik Böbrek Hastalığı) (CRB2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Ventriculomegaly with cystic kidney disease, 219730, Autosomal recessive; VMCKD (Ventriculomegaly-cystic kidney disease) (CRB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CRB2	CRB2, FSGS9, VMCKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ventrikülomegali-Kistik Böbrek Hastalığı, 219730, Otozomal Resesif; VMCKD (Ventrikülomegali-Kistik Böbrek Hastalığı) (Prenatal) (CRB2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ventriculomegaly with cystic kidney disease, 219730, Autosomal recessive; VMCKD (Ventriculomegaly-cystic kidney disease) (CRB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CRB2	CRB2, FSGS9, VMCKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Verheij Sendromu, 615583, Otozomal Dominant; VRJS (PUF60 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Verheij syndrome, 615583, Autosomal dominant; VRJS (PUF60 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PUF60	PUF60, FIR, SIAHBP1, VRJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Verheij Sendromu, 615583, Otozomal Dominant; VRJS (PUF60 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Verheij syndrome, 615583, Autosomal dominant; VRJS (PUF60 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PUF60	PUF60, FIR, SIAHBP1, VRJS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Vertikal Talus, Konjenital, 192950, Otozomal Dominant; CVT (Konjenital Vertikal Talus) (HOXD10 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vertical talus, congenital, 192950, Autosomal dominant; CVT (Congenital vertical talus) (HOXD10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HOXD10	HOXD10, HOX4D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vezikoüreteral Reflü 2, 610878; VUR2 (Ailesel Vezikoüreteral Reflü) (ROBO2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vesicoureteral reflux 2, 610878; VUR2 (Familial vesicoureteral reflux) (ROBO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ROBO2	ROBO2, SAX3, KIAA1568	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vezikoüreteral Reflü 3, 613674, Otozomal Dominant; VUR3 (Ailesel Vezikoüreteral Reflü) (SOX17 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vesicoureteral reflux 3, 613674, Autosomal dominant; VUR3 (Familial vesicoureteral reflux) (SOX17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX17	SOX17, VUR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vezikoüreteral Reflü 8, 615963, Otozomal Dominant; VUR8 (Ailesel Vezikoüreteral Reflü) (TNXB gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vesicoureteral reflux 8, 615963, Autosomal dominant; VUR8 (Familial vesicoureteral reflux) (TNXB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TNXB	TNXB, TNX, TNXB1, TNXBS, TNXB2, EDS3, VUR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vezikoüreteral Reflü 8, 615963, Otozomal Dominant; VUR8 (Ailesel Vezikoüreteral Reflü) (MLPA)	Vesicoureteral reflux 8, 615963; Autosomal dominant; VUR8 (Familial vesicoureteral reflux) (MLPA)	TNXB	TNXB, TNX, TNXB1, TNXBS, TNXB2, EDS3, VUR8	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Vibrasyona Bağlı Ürtiker, 125630, Otozomal Dominant; VBU (ADGRE2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vibratory urticaria, 125630, Autosomal dominant; VBU (ADGRE2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADGRE2	ADGRE2, EMR2, VBU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vici Sendromu, 242840, Otozomal Resesif; VICIS (Vici Sendromu) (EPG5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vici syndrome, 242840, Autosomal recessive; VICIS (Vici syndrome) (EPG5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EPG5	EPG5, KIAA1632, HEEW1, VICIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vici Sendromu, 242840, Otozomal Resesif; VICIS (Vici Sendromu) (Prenatal) (EPG5 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Vici syndrome, 242840, Autosomal recessive; VICIS (Vici syndrome) (EPG5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EPG5	EPG5, KIAA1632, HEEW1, VICIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Virüs Enfeksiyonu, Yatkınlık (OAS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Viral infection, susceptibility to (OAS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	OAS1	OAS1, OIAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Visseral Miyopati, 155310, Otozomal Dominant; VSCM (Ailesel Visseral Miyopati) (ACTG2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Visceral myopathy, 155310, Autosomal dominant; VSCM (Familial visceral myopathy) (ACTG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ACTG2	ACTG2, ACTA3, VSCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B12 Plazma Seviyesi QTL1, 612542; B12QTL1 (FUT2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitamin B12 plasma level QTL1, 612542; B12QTL1 (FUT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FUT2	FUT2, SE, B12QTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
D Vitamini Bağımlı Raşitizm, Tip 1, 264700, Otozomal Resesif (Hipokalsemik D Vitamini Bağımlı Raşitizmi (CYP27B1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitamin D-dependent rickets, type I, 264700, Autosomal recessive (Hypocalcemic vitamin D-dependent rickets) (CYP27B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP27B1	CYP27B1, VDD1, PDDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
D Vitamini Bağımlı Raşitizm, Tip 1, 264700, Otozomal Resesif (Hipokalsemik D Vitamini Bağımlı Raşitizmi (Prenatal) (CYP27B1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Vitamin D-dependent rickets, type I, 264700, Autosomal recessive (Hypocalcemic vitamin D-dependent rickets) (CYP27B1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CYP27B1	CYP27B1, VDD1, PDDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Vitamin-K'ye Bağımlı Pıhtılaşma Faktörleri, 1, Kombine Eksikliği, 277450, Otozomal Resesif; VKCFD1 (K Vitamininе Bağlı Pıhtılaşma Faktörlerinin Kalıtsal Kombine Eksikliği) (GGCX gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450, Autosomal recessive; VKCFD1 (Hereditary combined deficiency of vitamin K-dependent clotting factors) (GGCX gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GGCX	GGCX, VKCFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitamin-K'ye Bağımlı Pıhtılaşma Faktörleri, 2, Kombine Eksikliği, 607473; VKCFD2 (K Vitamininе Bağlı Pıhtılaşma Faktörlerinin Kalıtsal Kombine Eksikliği) (VKORC1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473; VKCFD2 (Hereditary combined deficiency of vitamin K-dependent clotting factors) (VKORC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VKORC1	VKORC1, VKOR, VKCFD2, FLJ00289	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitiligo İle İlişkili Multipl Otoimmün Hastalık Yatkınlığı 1, 606579; VAMAS1 (NLRP1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitiligo-associated multiple autoimmune disease susceptibility 1, 606579; VAMAS1 (NLRP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NLRP1	NLRP1, NALP1, KIAA0926, DEFCAP, CARD7, SLEV1, VAMAS1, MSPC, AIADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitreoretinal Dejenerasyon, Kar Tanesi Tipi, Svd (Kar Tanesi Vitreoretinal Dejenerasyonu) (KCNJ13 gen) (Tüm Gen Dizi Analizi) (Postnatal)	VITREORETINAL DEGENERATION, SNOWFLAKE TYPE; SVD (Snowflake vitreoretinal degeneration) (KCNJ13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNJ13	KCNJ13, SVD, LCA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitreoretinokoroidopati, 193220, Otozomal Dominant; VRCP (Otozomal Dominant Vitreoretinokoroidopati) (BEST1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitreoretinochoroidopathy, 193220, Autosomal dominant; VRCP (Autosomal dominant vitreoretinochoroidopathy) (BEST1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BEST1	BEST1, VMD2, ARB, RP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitreoretinopati, Falangeal Epifiz Displazisi, Otozomal Dominant (COL2A1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitreoretinopathy with phalangeal epiphyseal dysplasia, autosomal dominant (COL2A1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL2A1	COL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vitreoretinopati, Falangeal Epifiz Displazisi, Otozomal Dominant (MLPA)	Vitreoretinopathy with phalangeal epiphyseal dysplasia, autosomal dominant (MLPA)	COL2A1	COL2A1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Vitreoretinopati, Neovasküler İnflamatuvar, 193235, Otozomal Dominant; VRNI (Otozomal Dominant Neovasküler Enflamatuar Vitreoretinopati) (CAPN5 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vitreoretinopathy, neovascular inflammatory, 193235, Autosomal dominant; VRNI (Autosomal dominant neovascular inflammatory vitreoretinopathy) (CAPN5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CAPN5	CAPN5, HTRA3, VRNI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vohwinkel Sendromu-İktiyozis, 604117, Otozomal Dominant (Ailesel Keratoderma Mutilans-İktiyozis (İktiyozisli Vohwinkel Sendromu)) (LOR gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vohwinkel syndrome with ichthyosis, 604117, Autosomal dominant (Keratoderma hereditarium mutilans with ichthyosis) (LOR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LOR	LOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vohwinkel Sendromu-İktiyozis, 604117, Otozomal Dominant (Ailesel Keratoderma Mutilans-İktiyozis (İktiyozisli Vohwinkel Sendromu)) (Prenatal) (LOR gen) (Tüm Gen Dizi Analizi) (Prenatal)	Vohwinkel syndrome with ichthyosis, 604117, Autosomal dominant (Keratoderma hereditarium mutilans with ichthyosis) (LOR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LOR	LOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Vohwinkel Sendromu, 124500, Otozomal Dominant; VOWNKL (Keratoderma Kalıtsalın Mutilans) (GJB2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Vohwinkel syndrome, 124500, Autosomal dominant; VOWNKL (Keratoderma hereditarium mutilans) (GJB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GJB2	GJB2, CX26, DFNB1A, PPK, DFNA3A, KID, HID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Vohwinkel Sendromu, Variant Form (Ailesel Keratoderma Mutilans-İktiyozis (İktiyozisli Vohwinkel Sendromu)) (LOR gen) (Tüm Gen Dizi Analizi) (Postnatal)	VOHWINKEL SYNDROME, VARIANT FORM (Keratoderma hereditarium mutilans with ichthyosis) (LOR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LOR	LOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Hippel Lindau Sendromu, 193300, Otozomal Dominant; VHL (Von Hippel-Lindau Hastalığı) (VHL gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Hippel-Lindau syndrome, 193300, Autosomal dominant; VHL (Von Hippel-Lindau disease) (VHL gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VHL	VHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Hippel Lindau Sendromu, 193300, Otozomal Dominant; VHL (Von Hippel-Lindau Hastalığı) (MLPA)	von Hippel-Lindau syndrome, 193300, Autosomal dominant; VHL (Von Hippel-Lindau disease) (MLPA)	VHL	VHL	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Von Hippel Lindau Sendromu, Modifiye Edici, 193300, Otozomal Dominant (Von Hippel-Lindau Hastalığı) (CCND1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Hippel-Lindau syndrome, modifier of, 193300, Autosomal dominant (Von Hippel-Lindau disease) (CCND1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCND1	CCND1, PRAD1, BCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Trombosit Tipi, 177820, Otozomal Dominant; VWDP (Psödo-Von Willebrand Hastalığı) (GP1BA gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Willebrand disease, platelet-type, 177820, Autosomal dominant; VWDP (Pseudo-von Willebrand disease) (GP1BA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GP1BA	GP1BA, BSS, BDPLT1, VWDP, BDPLT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 1, 193400, Otozomal Dominant; VWD1 (Von Willebrand Hastalığı Tip 1) (VWF gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Willebrand disease, type 1, 193400, Autosomal dominant; VWD1 (Von Willebrand disease type 1) (VWF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VWF	VWF, FBVWF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 1, 193400, Otozomal Dominant; VWD1 (Von Willebrand Hastalığı Tip 1) (MLPA)	von Willebrand disease, type 1, 193400, Autosomal dominant; VWD1 (Von Willebrand disease type 1) (MLPA)	VWF	VWF, FBVWF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, 1, 2A, 2B, 2M, 2N, 3 (VWF gen) (Tüm Gen - Dizi Analizi) (VWF gen) (Tüm Gen Dizi Analizi) (Postnatal)	Von Willebrand disease, type 1,2A, 2B, 2M, 2N, 3 (VWF gene) (VWF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VWF	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 2A, 2B, 2M ve 2N, 613554, Otozomal Resesif, Otozomal Dominant; VWD2 (Von Willebrand Hastalığı Tip 2A) (VWF gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554, Autosomal recessive, Autosomal dominant; VWD2 (Von Willebrand disease type 2A) (VWF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VWF	VWF, FBVWF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 2A, 2B, 2M ve 2N, 613554, Otozomal Resesif, Otozomal Dominant; VWD2 (Von Willebrand Hastalığı Tip 2A) (MLPA)	von Willebrand disease, types 2A, 2B, 2M, and 2N, 613554, Autosomal recessive, Autosomal dominant; VWD2 (Von Willebrand disease type 2A) (MLPA)	VWF	VWF, FBVWF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Von Willebrand hastalığı (vWD) (Miks 1) (vWF 12p13) (MLPA)/ Von Willebrand hastalığı (vWD) (Miks 2) (vWF 12p13) (MLPA)	von Willebrand's disease (vWD) (Mix 1) (vWF 12p13) (MLPA) / von Willebrand's disease (vWD) (Mix 2) (vWF 12p13) (MLPA)	vWF 12p13	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 3, 277480, Otozomal Resesif; VWD3 (Von Willebrand Hastalığı Tip 3) (VWF gen) (Tüm Gen Dizi Analizi) (Postnatal)	von Willebrand disease, type 3, 277480, Autosomal recessive; VWD3 (Von Willebrand disease type 3) (VWF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VWF	VWF, FBVWF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Von Willebrand Hastalığı, Tip 3, 277480, Otozomal Resesif; VWD3 (Von Willebrand Hastalığı Tip 3) (MLPA)	von Willebrand disease, type 3, 277480, Autosomal recessive; VWD3 (Von Willebrand disease type 3) (MLPA)	VWF	VWF, FBVWF	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waardenburg sendromu paneli (PAX3 2q35, MITF 3p14, SOX10 22q13.1) (MLPA)	Waardenburg syndrome panel (PAX3 2q35, MITF 3p14, SOX10 22q13.1) (MLPA)	PAX3 2q35, MITF 3p14, SOX10 22q13.1	.	MLPA	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waardenburg sendromu paneli (PAX3 2q35, MITF 3p14, SOX10 22q13.1) (MLPA) (Prenatal)	Waardenburg syndrome panel (PAX3 2q35, MITF 3p14, SOX10 22q13.1) (MLPA) (Prenatal)	PAX3 2q35, MITF 3p14, SOX10 22q13.1	.	MLPA	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

Waarderburg Sendromu, Tip 1, 193500, Otozomal Dominant; WS1 (Waarderburg Sendromu Tip 1) (PAX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 1, 193500, Autosomal dominant; WS1 (Waarderburg syndrome type 1) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 1, 193500, Otozomal Dominant; WS1 (Waarderburg Sendromu Tip 1) (MLPA)	Waarderburg syndrome, type 1, 193500, Autosomal dominant; WS1 (Waarderburg syndrome type 1) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 1, 193500, Otozomal Dominant; WS1 (Waarderburg Sendromu Tip 1) (Prenatal) (PAX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 1, 193500, Autosomal dominant; WS1 (Waarderburg syndrome type 1) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 1, 193500, Otozomal Dominant; WS1 (Waarderburg Sendromu Tip 1) (Prenatal) (MLPA)	Waarderburg syndrome, type 1, 193500, Autosomal dominant; WS1 (Waarderburg syndrome type 1) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 2A, 193510, Otozomal Dominant; WS2A (Waarderburg Sendromu Tip 2) (MITF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 2A, 193510, Autosomal dominant; WS2A (Waarderburg syndrome type 2) (MITF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 2A, 193510, Otozomal Dominant; WS2A (Waarderburg Sendromu Tip 2) (MLPA)	Waarderburg syndrome, type 2A, 193510, Autosomal dominant; WS2A (Waarderburg syndrome type 2) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 2A, 193510, Otozomal Dominant; WS2A (Waarderburg Sendromu Tip 2) (Prenatal) (MITF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 2A, 193510, Autosomal dominant; WS2A (Waarderburg syndrome type 2) (MITF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 2A, 193510, Otozomal Dominant; WS2A (Waarderburg Sendromu Tip 2) (Prenatal) (MLPA)	Waarderburg syndrome, type 2A, 193510, Autosomal dominant; WS2A (Waarderburg syndrome type 2) (Prenatal) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 2D, 608890, Otozomal Resesif; WS2D (Waarderburg Sendromu Tip 5) (SNAI2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 2D, 608890, Autosomal recessive; WS2D (Waarderburg syndrome type 2) (SNAI2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SNAI2	SNAI2, SLUG, WS2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 2D, 608890, Otozomal Resesif; WS2D (Waarderburg Sendromu Tip 5) (Prenatal) (SNAI2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 2D, 608890, Autosomal recessive; WS2D (Waarderburg syndrome type 2) (SNAI2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SNAI2	SNAI2, SLUG, WS2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 2E, ± Nörolojik Tutulum, 611584, Otozomal Dominant; WS2E (Waarderburg Sendromu Tip 6) (SOX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 2E, with or without neurologic involvement, 611584, Autosomal dominant; WS2E (Waarderburg syndrome type 2) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 2E, ± Nörolojik Tutulum, 611584, Otozomal Dominant; WS2E (Waarderburg Sendromu Tip 6) (Prenatal) (SOX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 2E, with or without neurologic involvement, 611584, Autosomal dominant; WS2E (Waarderburg syndrome type 2) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 3, 148820, Otozomal Resesif, Otozomal Dominant; WS3 (Waarderburg Sendromu Tip 3) (PAX3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 3, 148820, Autosomal recessive, Autosomal dominant; WS3 (Waarderburg syndrome type 3) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 3, 148820, Otozomal Resesif, Otozomal Dominant; WS3 (Waarderburg Sendromu Tip 3) (MLPA)	Waarderburg syndrome, type 3, 148820, Autosomal recessive, Autosomal dominant; WS3 (Waarderburg syndrome type 3) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 3, 148820, Otozomal Resesif, Otozomal Dominant; WS3 (Waarderburg Sendromu Tip 3) (Prenatal) (PAX3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 3, 148820, Autosomal recessive, Autosomal dominant; WS3 (Waarderburg syndrome type 3) (PAX3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 3, 148820, Otozomal Resesif, Otozomal Dominant; WS3 (Waarderburg Sendromu Tip 3) (Prenatal) (MLPA)	Waarderburg syndrome, type 3, 148820, Autosomal recessive, Autosomal dominant; WS3 (Waarderburg syndrome type 3) (Prenatal) (MLPA)	PAX3	PAX3, WS1, HUP2, CDHS, WS3	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 4A, 277580, Otozomal Resesif, Otozomal Dominant; WS4A (Waarderburg-Shah Sendromu) (EDNRB geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 4A, 277580, Autosomal recessive, Autosomal dominant; WS4A (Waarderburg-Shah syndrome) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 4A, 277580, Otozomal Resesif, Otozomal Dominant; WS4A (Waarderburg-Shah Sendromu) (Prenatal) (EDNRB geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 4A, 277580, Autosomal recessive, Autosomal dominant; WS4A (Waarderburg-Shah syndrome) (EDNRB gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDNRB	EDNRB, HSCR2, ABCDS, WS4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 4B, 613265, Otozomal Resesif, Otozomal Dominant; WS4B (Waarderburg-Shah Sendromu) (EDN3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 4B, 613265, Autosomal recessive, Autosomal dominant; WS4B (Waarderburg-Shah syndrome) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 4B, 613265, Otozomal Resesif, Otozomal Dominant; WS4B (Waarderburg-Shah Sendromu) (MLPA)	Waarderburg syndrome, type 4B, 613265, Autosomal recessive, Autosomal dominant; WS4B (Waarderburg-Shah syndrome) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 4B, 613265, Otozomal Resesif, Otozomal Dominant; WS4B (Waarderburg-Shah Sendromu) (Prenatal) (EDN3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 4B, 613265, Autosomal recessive, Autosomal dominant; WS4B (Waarderburg-Shah syndrome) (EDN3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EDN3	EDN3, WS4B, HSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 4B, 613265, Otozomal Resesif, Otozomal Dominant; WS4B (Waarderburg-Shah Sendromu) (Prenatal) (MLPA)	Waarderburg syndrome, type 4B, 613265, Autosomal recessive, Autosomal dominant; WS4B (Waarderburg-Shah syndrome) (Prenatal) (MLPA)	EDN3	EDN3, WS4B, HSCR4	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu, Tip 4C, 613266, Otozomal Dominant; WS4C (Waarderburg-Shah Sendromu) (SOX10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome, type 4C, 613266, Autosomal dominant; WS4C (Waarderburg-Shah syndrome) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu, Tip 4C, 613266, Otozomal Dominant; WS4C (Waarderburg-Shah Sendromu) (Prenatal) (SOX10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome, type 4C, 613266, Autosomal dominant; WS4C (Waarderburg-Shah syndrome) (SOX10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SOX10	SOX10, WS4, WS4C, PCWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu / Albinizm, Dijenik, 103470, Otozomal Dominant (TYR geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome/albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (TYR gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu / Albinizm, Dijenik, 103470, Otozomal Dominant (MLPA)	Waarderburg syndrome/albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu / Albinizm, Dijenik, 103470, Otozomal Dominant (Prenatal) (TYR geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome/albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (TYR gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu / Albinizm, Dijenik, 103470, Otozomal Dominant (Prenatal) (MLPA)	Waarderburg syndrome/albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (Prenatal) (MLPA)	TYR	TYR, SHEP3, CMM8, OCA1A, ATN	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu / Okiler Albinizm, Dijenik, 103470, Otozomal Dominant (Okiler Albinizm-Konjenital Sensorinöral Sağırlık) (MITF geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waarderburg syndrome/ocular albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (MITF gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu / Okiler Albinizm, Dijenik, 103470, Otozomal Dominant (Okiler Albinizm-Konjenital Sensorinöral Sağırlık) (MLPA)	Waarderburg syndrome/ocular albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Waarderburg Sendromu / Okiler Albinizm, Dijenik, 103470, Otozomal Dominant (Okiler Albinizm-Konjenital Sensorinöral Sağırlık) (Prenatal) (MITF geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waarderburg syndrome/ocular albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (MITF gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MITF	MITF, WS2A, CMM8, COMMAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waarderburg Sendromu / Okiler Albinizm, Dijenik, 103470, Otozomal Dominant (Okiler Albinizm-Konjenital Sensorinöral Sağırlık) (Prenatal) (MLPA)	Waarderburg syndrome/ocular albinism, digenic, 103470, Autosomal dominant (Ocular albinism with congenital sensorineural deafness) (Prenatal) (MLPA)	MITF	MITF, WS2A, CMM8, COMMAD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wagner Sendromu 1, 143200, Otozomal Dominant; WGVRP (Wagner Hastalığı 1) (VCAN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Wagner syndrome 1, 143200, Autosomal dominant; WGVRP (Wagner disease) (VCAN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VCAN	VCAN, CSPG2, WGN, WGN1, ERVR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wagner Sendromu 1, 143200, Otozomal Dominant; WGVRP (Wagner Hastalığı 1) (Prenatal) (VCAN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Wagner syndrome 1, 143200, Autosomal dominant; WGVRP (Wagner disease) (VCAN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	VCAN	VCAN, CSPG2, WGN, WGN1, ERVR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
WAGRO Sendromu, 612469 (WAGR Sendromu) (2200)	WAGRO syndrome, 612469 (WAGR syndrome) (440)	.	WAGRO, DEL11p14p12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
WAGRO Sendromu, 612469 (WAGR Sendromu) (Prenatal)	WAGRO syndrome, 612469 (WAGR syndrome) (Prenatal)	.	WAGRO, DEL11p14p12	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Waisman Sendromu, 311510, X'e Bağlı Resesif; WSMN (Erken Başlangıçlı Parkinsonizm-Entelektüel Yetersizlik Sendromu (Waisman Sendromu)) (RAB39B geni) (Tüm Gen Dizi Analizi) (Postnatal)	Waisman syndrome, 311510, X-linked recessive; WSMN (Early-onset parkinsonism-intellectual disability syndrome) (RAB39B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB39B	RAB39B, MRX72, WSMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Waisman Sendromu, 311510, X'e Bağlı Resesif; WSMN (Erken Başlangıçlı Parkinsonizm-Entelektüel Yetersizlik Sendromu (Waisman Sendromu)) (Prenatal) (RAB39B geni) (Tüm Gen Dizi Analizi) (Prenatal)	Waisman syndrome, 311510, X-linked recessive; WSMN (Early-onset parkinsonism-intellectual disability syndrome) (RAB39B gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB39B	RAB39B, MRX72, WSMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Warburg Mikro Sendromu 1, 600118, Otozomal Resesif; WARBM1 (Mikro Sendromu) (RAB3GAP1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warburg micro syndrome 1, 600118, Autosomal recessive; WARBM1 (Micro syndrome) (RAB3GAP1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB3GAP1	RAB3GAP1, WARBM1, P130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warburg Mikro Sendromu 1, 600118, Otozomal Resesif; WARBM1 (Mikro Sendromu) (Prenatal) (RAB3GAP1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Warburg micro syndrome 1, 600118, Autosomal recessive; WARBM1 (Micro syndrome) (RAB3GAP1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB3GAP1	RAB3GAP1, WARBM1, P130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Warburg Milero Sendromu 2, 614225, Otozomal Resesif; WARBM2 (Milero Sendromu) (RAB3GAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warburg micro syndrome 2, 614225, Autosomal recessive; WARBM2 (Micro syndrome) (RAB3GAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB3GAP2	RAB3GAP2, RAB3GAP150, p150, KIAA0839, WARBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warburg Milero Sendromu 2, 614225, Otozomal Resesif; WARBM2 (Milero Sendromu) (Prenatal) (RAB3GAP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Warburg micro syndrome 2, 614225, Autosomal recessive; WARBM2 (Micro syndrome) (RAB3GAP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB3GAP2	RAB3GAP2, RAB3GAP150, p150, KIAA0839, WARBM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Warburg Milero Sendromu 3, 614222, Otozomal Resesif; WARBM3 (Milero Sendromu) (RAB18 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warburg micro syndrome 3, 614222, Autosomal recessive; WARBM3 (Micro syndrome) (RAB18 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	RAB18	RAB18, WARBM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Warburg Mikro Sendromu 3, 614222, Otozomal Resesif; WARBM3 (Mikro Sendromu) (Prenatal) (RAB18 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Warburg micro syndrome 3, 614222, Autosomal recessive; WARBM3 (Micro syndrome) (RAB18 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	RAB18	RAB18, WARBM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Warburg Mikro Sendromu 4, 615663, Otozomal Resesif; WARBM4 (Mikro Sendromu) (TBC1D20 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warburg micro syndrome 4, 615663, Autosomal recessive; WARBM4 (Micro syndrome) (TBC1D20 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TBC1D20	TBC1D20, C20orf140, WARBM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warburg Mikro Sendromu 4, 615663, Otozomal Resesif; WARBM4 (Mikro Sendromu) (TBC1D20 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Warburg micro syndrome 4, 615663, Autosomal recessive; WARBM4 (Micro syndrome) (TBC1D20 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TBC1D20	TBC1D20, C20orf140, WARBM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Warfarin Direnci/ Duyarlılığı, Kumadin Direnci/ Duyarlılığı (VKORC1 1, 2, 3 ve 4 alelleri- SNAPSHOT Minisequencing ve CYP2C9 1, 2, 3, 4, 5, 6 ve 11 Alelleri - Dizi Analizi)	Warfarin - Coumadin resistance/sensitivity (VKORC1 1,2,3 and 4 alleles SNAPSHOT Minisequencing, CYP2C9 1,2,3,4,5,6 and 11 alleles All coding exons - Sequence analysis)	VCORC1, CYP2C9	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	45	Postnatal	.	EDTA Blood Sample
Warfarin Direnci, 122700, Otozomal Dominant (VKORC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warfarin resistance, 122700, Autosomal dominant (VKORC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	VKORC1	VKORC1, VKOR, WCFC02, FLJ00289	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warfarin Duyarlılığı (F9 geni) (Tüm Gen - Dizi Analizi) (F9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warfarin sensitivity (F9 gene) (All coding exons - Sequence analysis) (F9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F9	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warfarin Hassasiyeti, 122700, Otozomal Dominant (F9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warfarin sensitivity, 122700, Autosomal dominant (F9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	F9	F9, HEMB, THPHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warfarin Yatkınlık, 122700, Otozomal Dominant (CYP2C9 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warfarin sensitivity, 122700, Autosomal dominant (CYP2C9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C9	CYP2C9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Warfarin Hassasiyeti, 122700, Otozomal Dominant (MLPA)	Warfarin sensitivity, 122700, Autosomal dominant (MLPA)	F9	F9, HEMB, THPHB	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Warfarin Yatkınlık, 122700, Otozomal Dominant (MLPA)	Warfarin sensitivity, 122700, Autosomal dominant (MLPA)	CYP2C9	CYP2C9	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Varşova Kırık Sendromu, 613398, Otozomal Resesif; WABS (Varşova Kırık Sendromu) (DDX11 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Warsaw breakage syndrome, 613398, Autosomal recessive; WABS (Warsaw breakage syndrome) (DDX11 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDX11	DDX11, CHLR1, KRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Varşova Kırık Sendromu, 613398, Otozomal Resesif; WABS (Varşova Kırık Sendromu) (Prenatal) (DDX11 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Warsaw breakage syndrome, 613398, Autosomal recessive; WABS (Warsaw breakage syndrome) (DDX11 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDX11	DDX11, CHLR1, KRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Watson Sendromu, 193520, Otozomal Dominant; WTSN (Watson Sendromu) (NF1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Watson syndrome, 193520, Autosomal dominant; WTSN (Watson syndrome) (NF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Watson Sendromu, 193520, Otozomal Dominant; WTSN (Watson Sendromu) (MLPA)	Watson syndrome, 193520, Autosomal dominant; WTSN (Watson syndrome) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Watson Sendromu, 193520, Otozomal Dominant; WTSN (Watson Sendromu) (Prenatal) (NF1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Watson syndrome, 193520, Autosomal dominant; WTSN (Watson syndrome) (NF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NF1	NF1, VRNF, WSS, NFNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Watson Sendromu, 193520, Otozomal Dominant; WTSN (Watson Sendromu) (Prenatal) (MLPA)	Watson syndrome, 193520, Autosomal dominant; WTSN (Watson syndrome) (Prenatal) (MLPA)	NF1	NF1, VRNF, WSS, NFNS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weaver Sendromu, 277590, Otozomal Dominant; WVS (Weaver Sendromu) (EZH2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weaver syndrome, 277590, Autosomal dominant; WVS (Weaver syndrome) (EZH2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EZH2	EZH2, EZH1, WVS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weaver Sendromu, 277590, Otozomal Dominant; WVS (Weaver Sendromu) (Prenatal) (EZH2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weaver syndrome, 277590, Autosomal dominant; WVS (Weaver syndrome) (EZH2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EZH2	EZH2, EZH1, WVS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Webb-Dattani Sendromu, 615926, Otozomal Resesif; WEDAS (Hipotalamik Yetmezlik-Sekonder Mikrosefali-Görme Bozukluğu-Üriner Anomaliler Sendromu) (ARNT2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Webb-Dattani syndrome, 615926, Autosomal recessive; WEDAS (Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome) (ARNT2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ARNT2	ARNT2, WEDAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Webb-Dattani Sendromu, 615926, Otozomal Resesif; WEDAS (Hipotalamik Yetmezlik-Sekonder Mikrosefali-Görme Bozukluğu-Üriner Anomaliler Sendromu) (Prenatal) (ARNT2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Webb-Dattani syndrome, 615926, Autosomal recessive; WEDAS (Hypothalamic insufficiency-secondary microcephaly-visual impairment-urinary anomalies syndrome) (ARNT2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ARNT2	ARNT2, WEDAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wegener Benzeri Granülomatoz (İmmun Yetmezlik, HLA Sınıf 1 Defektif Ekspresyonuna Bağlı) (TAP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Wegener-like granulomatosis (Immunodeficiency by defective expression of HLA class 1) (TAP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TAP2	TAP2, ABCB3, PSF2, RING11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Sendromu 1, Resesif, 277600, Otozomal Resesif; WMS1 (Weill-Marchesani Sendromu) (ADAMTS10 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weill-Marchesani syndrome 1, recessive, 277600, Autosomal recessive; WMS1 (Weill-Marchesani syndrome) (ADAMTS10 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTS10	ADAMTS10, WMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Sendromu 1, Resesif, 277600, Otozomal Resesif; WMS1 (Weill-Marchesani Sendromu) (Prenatal) (ADAMTS10 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weill-Marchesani syndrome 1, recessive, 277600, Autosomal recessive; WMS1 (Weill-Marchesani syndrome) (ADAMTS10 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADAMTS10	ADAMTS10, WMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weill-Marchesani Sendromu 2, Dominant, 608328, Otozomal Dominant; WMS2 (Weill-Marchesani Sendromu) (FBN1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weill-Marchesani syndrome 2, dominant, 608328, Autosomal dominant; WMS2 (Weill-Marchesani syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)	*	30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Sendromu 2, Dominant, 608328, Otozomal Dominant; WMS2 (Weill-Marchesani Sendromu) (MLPA)	Weill-Marchesani syndrome 2, dominant, 608328, Autosomal dominant; WMS2 (Weill-Marchesani syndrome) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Sendromu 2, Dominant, 608328, Otozomal Dominant; WMS2 (Weill-Marchesani Sendromu) (Prenatal) (FBN1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weill-Marchesani syndrome 2, dominant, 608328, Autosomal dominant; WMS2 (Weill-Marchesani syndrome) (FBN1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN	*	.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weill-Marchesani Sendromu 2, Dominant, 608328, Otozomal Dominant; WMS2 (Weill-Marchesani Sendromu) (Prenatal) (MLPA)	Weill-Marchesani syndrome 2, dominant, 608328, Autosomal dominant; WMS2 (Weill-Marchesani syndrome) (Prenatal) (MLPA)	FBN1	FBN1, MFS1, WMS2, SSKS, GPHYSD2, ACMICD, ECTOL1, MFLS	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weill-Marchesani Sendromu 3, Resesif, 614819, Otozomal Resesif; WMS3 (Weill-Marchesani Sendromu) (LTBP2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weill-Marchesani syndrome 3, recessive, 614819, Autosomal recessive; WMS3 (Weill-Marchesani syndrome) (LTBP2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LTBP2	LTBP2, LTBP3, GLC3D, MSPKA, WMS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Sendromu 3, Resesif, 614819, Otozomal Resesif; WMS3 (Weill-Marchesani Sendromu) (Prenatal) (LTBP2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weill-Marchesani syndrome 3, recessive, 614819, Autosomal recessive; WMS3 (Weill-Marchesani syndrome) (LTBP2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LTBP2	LTBP2, LTBP3, GLC3D, MSPKA, WMS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weill-Marchesani Benzeri Sendrom, 613195, Otozomal Resesif (İdiyozis-Boy Kısalığı-Brakidaktili-Mikrosferofaki Sendromu) (ADAMTS17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weill-Marchesani-like syndrome, 613195, Autosomal recessive (ichthyosis-short stature-brachydactyly-microspherophakia syndrome) (ADAMTS17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ADAMTS17	ADAMTS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weill-Marchesani Benzeri Sendrom, 613195, Otozomal Resesif (İdiyozis-Boy Kısalığı-Brakidaktili-Mikrosferofaki Sendromu) (Prenatal) (ADAMTS17 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weill-Marchesani-like syndrome, 613195, Autosomal recessive (ichthyosis-short stature-brachydactyly-microspherophakia syndrome) (ADAMTS17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ADAMTS17	ADAMTS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weissenbacher-Zweymuller Sendromu, 277610, Otozomal Dominant; WZS (Weissenbacher- Zweymuller Sendromu) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weissenbacher-Zweymuller syndrome, 277610, Autosomal dominant; WZS (Weissenbacher-Zweymuller syndrome) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weissenbacher-Zweymuller Sendromu, 277610, Otozomal Dominant; WZS (Weissenbacher- Zweymuller Sendromu) (Prenatal) (COL11A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weissenbacher-Zweymuller syndrome, 277610, Autosomal dominant; WZS (Weissenbacher-Zweymuller syndrome) (COL11A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	COL11A2	COL11A2, STL3, DFNA13, DFNB53, FBCG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Welander Distal Miyopati, 604454, Otozomal Resesif, Otozomal Dominant; WDM (Distal Miyopati, Welander Tipi) (TIA1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Welander distal myopathy, 604454, Autosomal recessive, Autosomal dominant; WDM (Distal myopathy, Welander type) (TIA1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TIA1	TIA1, WDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Welander Distal Miyopati, 604454, Otozomal Resesif, Otozomal Dominant; WDM (Distal Miyopati, Welander Tipi) (Prenatal) (TIA1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Welander distal myopathy, 604454, Autosomal recessive, Autosomal dominant; WDM (Distal myopathy, Welander type) (TIA1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TIA1	TIA1, WDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Werner Sendromu, 277700, Otozomal Resesif; WRN (Werner Sendromu) (WRN geni) (Tüm Gen Dizi Analizi) (Postnatal)	Werner syndrome, 277700, Autosomal recessive; WRN (Werner syndrome) (WRN gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WRN	RECQL2, RECQ3, WRN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Werner Sendromu, 277700, Otozomal Resesif; WRN (Werner Sendromu) (Prenatal) (WRN geni) (Tüm Gen Dizi Analizi) (Prenatal)	Werner syndrome, 277700, Autosomal recessive; WRN (Werner syndrome) (WRN gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WRN	RECQL2, RECQ3, WRN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Batı Nil Virüsü, Yatkınlık, 610379 (Batı-Nil Ensefaliti) (CCR5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	West nile virus, susceptibility to, 610379 (West-Nile encephalitis) (CCR5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CCR5	CCR5, CMKBR5, CCCKR5, IDDM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weyers Akrodental Dysostozis, 193530, Otozomal Dominant (Akrofasiyal Disostoz, Weyers Tipi) (EVC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weyers acrodental dysostosis, 193530, Autosomal dominant (Acrofacial dysostosis, Weyers type) (EVC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EVC	EVC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weyers Akrodental Dysostosis, 193530, Otozomal Dominant (Akrofasiyal Disostoz, Weyers Tipi) (MLPA)	Weyers acrodental dysostosis, 193530, Autosomal dominant (Acrofacial dysostosis, Weyers type) (MLPA)	EVC	EVC	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Weyers Akrodental Dysostosis, 193530, Otozomal Dominant (Akrofasiyal Disostoz, Weyers Tipi) (Prenatal) (EVC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Weyers acrodental dysostosis, 193530, Autosomal dominant (Acrofacial dysostosis, Weyers type) (EVC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EVC	EVC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weyers Akrodental Dysostosis, 193530, Otozomal Dominant (Akrofasiyal Disostoz, Weyers Tipi) (Prenatal) (MLPA)	Weyers acrodental dysostosis, 193530, Autosomal dominant (Acrofacial dysostosis, Weyers type) (Prenatal) (MLPA)	EVC	EVC	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weyers Akrofasiyal Dysostosis, 193530, Otozomal Dominant; WAD (Akrofasiyal Disostoz, Weyers Tipi) (EVC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Weyers acrofacial dysostosis, 193530, Autosomal dominant; WAD (Acrofacial dysostosis, Weyers type) (EVC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EVC2	LBN, EVC2, WAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Weyers Akrofasiyal Dysostosis, 193530, Otozomal Dominant; WAD (Akrofasiyal Disostoz, Weyers Tipi) (MLPA)	Weyers acrofacial dysostosis, 193530, Autosomal dominant; WAD (Acrofacial dysostosis, Weyers type) (MLPA)	EVC2	LBN, EVC2, WAD	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample

Weyers Akrofasiyal Dysostozis, 193530, Otozomal Dominant; WAD (Akrofasiyal Disostoz Weyers Tipi) (Prenatal) (EVC2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Weyers acrofacial dysostosis, 193530, Autosomal dominant; WAD (Acrofacial dysostosis, Weyers type) (EVC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	EVC2	LBN, EVC2, WAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Weyers Akrofasiyal Dysostozis, 193530, Otozomal Dominant; WAD (Akrofasiyal Disostoz, Weyers Tipi) (Prenatal) (MLPA)	Weyers acrofacial dysostosis, 193530, Autosomal dominant; WAD (Acrofacial dysostosis, Weyers type) (Prenatal) (MLPA)	EVC2	LBN, EVC2, WAD	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
WHM Sendromu, 193670, Otozomal Dominant; WHMS (WHM Sendromu) (CXCR4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	WHM syndrome, 193670, Autosomal dominant; WHMS (WHM syndrome) (CXCR4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CXCR4	CXCR4, D2S201E, NPY3R, WHMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WHM Sendromu, 193670, Otozomal Dominant; WHMS (WHM Sendromu) (Prenatal) (CXCR4 gen) (Tüm Gen Dizi Analizi) (Prenatal)	WHM syndrome, 193670, Autosomal dominant; WHMS (WHM syndrome) (CXCR4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	CXCR4	CXCR4, D2S201E, NPY3R, WHMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Beyaz Sünger Nevus 1, 193900, Otozomal Dominant; WSN1 (Beyaz Sünger Nevus) (KRT4 gen) (Tüm Gen Dizi Analizi) (Postnatal)	White sponge nevus 1, 193900, Autosomal dominant; WSN1 (White sponge nevus) (KRT4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT4	KRT4, CYK4, WSN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Beyaz Sünger Nevus 2, 615785, Otozomal Dominant; WSN2 (Beyaz Sünger Nevus) (KRT13 gen) (Tüm Gen Dizi Analizi) (Postnatal)	White sponge nevus 2, 615785, Autosomal dominant; WSN2 (White sponge nevus) (KRT13 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT13	KRT13, WSN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
White Sutton Sendromu, 616364, Otozomal Dominant; WHSUS (Mental Retardasyon-Mikrosefali-Şaşılık-Davranışsal Anormallikler Sendromu) (POGZ gen) (Tüm Gen Dizi Analizi) (Postnatal)	White-Sutton syndrome, 616364, Autosomal dominant; WHSUS (Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome) (POGZ gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POGZ	POGZ, KIAA0461, MRD37, WHSUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
White Sutton Sendromu, 616364, Otozomal Dominant; WHSUS (Mental Retardasyon-Mikrosefali-Şaşılık-Davranışsal Anormallikler Sendromu) (Prenatal) (POGZ gen) (Tüm Gen Dizi Analizi) (Prenatal)	White-Sutton syndrome, 616364, Autosomal dominant; WHSUS (Intellectual disability-microcephaly-strabismus-behavioral abnormalities syndrome) (POGZ gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POGZ	POGZ, KIAA0461, MRD37, WHSUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Tüm Mitokondriyal Genom Analizi (Mitokondriyal Genom) (Dizi Analizi) (Postnatal)	Whole Mitochondrial Genome Analysis (Mitokondrial Genom) (Dizi Analizi) (Postnatal)	Mitokondrial Genom	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tüm Mitokondriyal Genom Analizi (Bilinen mutasyon) (Dizi Analizi) (Postnatal)	Whole Mitochondrial Genome Analysis (Known Familial Mutation) (Sequence Analysis) (Postnatal)	Mitokondrial Genom	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Tüm Mitokondriyal Delesyon Duplikasyon Analizi (Kearns - Sayre Sendromu (KSS) / Leigh Sendromu, Pearson Sendromu, Progresif Eksternal Oftalmopleji (PEO)) (Postnatal) (MLPA)	Whole Mitokondrial Deletion Duplication (Kearns-Sayre Syndrome (KSS), Leigh Syndrome, Pearson Syndrome, Progressive External Ophthalmoplegia (PEO)) (Postnatal) (MLPA)	Mitokondrial Genom	.	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Tüm Mitokondriyal Delesyon Duplikasyon Analizi (Kearns - Sayre Sendromu (KSS) / Leigh Sendromu, Pearson Sendromu, Progresif Eksternal Oftalmopleji (PEO)) (Prenatal) (MLPA)	Whole Mitokondrial Deletion Duplication (Kearns-Sayre Syndrome (KSS), Leigh Syndrome, Pearson Syndrome, Progressive External Ophthalmoplegia (PEO)) (Prenatal) (MLPA)	Mitokondrial Genom	.	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wieacker-Wolff Sendromu, 314580, X'e Bağlı; WRWF (İntelektüel Yetersizlik-Gelişimsel Gecikme-Kontraktür Sendromu (Wieacker-Wolff Sendromu)) (ZC4H2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wieacker-Wolff syndrome, 314580, X-linked recessive; WRWF (Intellectual disability-developmental delay-contractures syndrome) (ZC4H2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ZC4H2	ZC4H2, KIAA1166, WRWF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wieacker-Wolff Sendromu, 314580, X'e Bağlı; WRWF (İntelektüel Yetersizlik-Gelişimsel Gecikme-Kontraktür Sendromu (Wieacker-Wolff Sendromu)) (Prenatal) (ZC4H2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Wieacker-Wolff syndrome, 314580, X-linked recessive; WRWF (Intellectual disability-developmental delay-contractures syndrome) (ZC4H2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ZC4H2	ZC4H2, KIAA1166, WRWF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wiedemann-Steiner Sendromu, 605130, Otozomal Dominant; WDSTS (Wiedemann-Steiner Sendromu) (KMT2A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wiedemann-Steiner syndrome, 605130, Autosomal dominant; WDSTS (Wiedemann-Steiner syndrome) (KMT2A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KMT2A	KMT2A, MLL, HRX, HTRX1, WDSTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wiedemann-Steiner Sendromu, 605130, Otozomal Dominant; WDSTS (Wiedemann-Steiner Sendromu) (KMT2A gen) (Prenatal) (Tüm Gen Dizi Analizi) (Prenatal)	Wiedemann-Steiner syndrome, 605130, Autosomal dominant; WDSTS (Wiedemann-Steiner syndrome) (KMT2A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KMT2A	KMT2A, MLL, HRX, HTRX1, WDSTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Williams Sendromu (7q11.23 Mikrodelesyonu) (FISH)	Williams syndrome (7q11.23 microdeletion) (FISH)	7q11.23	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Williams Sendromu (7q11.23 Mikrodelesyonu) (FISH) (Prenatal)	Williams syndrome (7q11.23 microdeletion) (Prenatal) (FISH)	7q11.23	.	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Williams-Beuren Sendromu, 194050, Otozomal Dominant; WBS (Williams Sendromu) (FISH)	Williams-Beuren syndrome, 194050, Autosomal dominant; WBS (Williams syndrome) (FISH)	.	WBS, WMS, WS, DEL7q11, C7DELq11	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Williams-Beuren Sendromu, 194050, Otozomal Dominant; WBS (Williams Sendromu) (Prenatal) (FISH)	Williams-Beuren syndrome, 194050, Autosomal dominant; WBS (Williams syndrome) (Prenatal) (FISH)	.	WBS, WMS, WS, DEL7q11, C7DELq11	FISH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wilms Tümörü 2, 194071, Otozomal Dominant, Somatik Mutasyon; WT2 (Nefroblastomu) (H19 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor 2, 194071, Autosomal dominant, Somatic mutation; WT2 (Nephroblastoma) (H19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinize doku bloğu
Wilms Tümörü 2, 194071, Otozomal Dominant, Somatik Mutasyon; WT2 (Nefroblastomu) (Prenatal) (H19 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Wilms tumor 2, 194071, Autosomal dominant, Somatic mutation; WT2 (Nephroblastoma) (H19 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	H19	H19, D11S813E, ASM1, BWS, WT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wilms Tümör 6, Yatkınlık, 616806; WT6 (Nefroblastom) (REST gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor 6, susceptibility to, 616806; WT6 (Nephroblastoma) (REST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	REST	REST, NRSF, WT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wilms Tümör 6, Yatkınlık, 616806; WT6 (Nefroblastom) (Prenatal) (REST gen) (Tüm Gen Dizi Analizi) (Prenatal)	Wilms tumor 6, susceptibility to, 616806; WT6 (Nephroblastoma) (REST gene) (Sequence Analysis-All Coding Exons) (Prenatal)	REST	REST, NRSF, WT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wilms Tümörü Yatkınlık-5, 601583, Otozomal Dominant, Somatik Mutasyon; WT5 (Nefroblastom) (POU6F2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor susceptibility-5, 601583, Autosomal dominant, Somatic mutation; WT5 (Nephroblastoma) (POU6F2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POU6F2	POU6F2, WTSL, WT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Wilms Tümörü, 194070, Otozomal Dominant, Somatik Mutasyon (Nefroblastom) (BRCA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor, 194070, Autosomal dominant, Somatic mutation (Nephroblastoma) (BRCA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	BRCA2	BRCA2, FANCD1, BROVCA2, GLM3, PNCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	"		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Wilms Tümörü, Aniridi, Genitoüriner Anomaliler, Mental Retardasyon, ve Obezite Sendromu; Wagro (WAGR Sendromu) (2200)	WILMS TUMOR, ANIRIDIA, GENITOURINARY ANOMALIES, MENTAL RETARDATION, AND OBESITY SYNDROME; WAGRO (WAGR syndrome) (440)	.	WAGRO, DEL11p14p12	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Wilms Tümörü, Somatik, 194070; WT1 (Nefroblastom) (GPC3 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor, somatic, 194070; WT1 (Nephroblastoma) (GPC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GPC3	GPC3, SDYS, SGBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Wilms Tümörü, Tip 1, 194070, Otozomal Dominant, Somatik Mutasyon (Nefroblastom) (WT1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilms tumor, type 1, 194070, Autosomal dominant, Somatic mutation (Nephroblastoma) (WT1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WT1	WT1, NPHS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		30	120	.	.	60	Postnatal	.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Wilson Hastalığı, 277900, Otozomal Resesif (Wilson Hastalığı) (ATP7B gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilson disease, 277900, Autosomal recessive (Wilson disease) (ATP7B gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP7B	ATP7B, WND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wilson Hastalığı, 277900, Otozomal Resesif (Wilson Hastalığı) (MLPA)	Wilson disease, 277900, Autosomal recessive (Wilson disease) (MLPA)	ATP7B	ATP7B, WND	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Wilson-Turner Sendromu, 309585, X'e Bağlı; WTS (Wilson-Turner Sendromu) (LAS1L gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wilson-Turner syndrome, 309585, X-linked recessive; WTS (Wilson-Turner syndrome) (LAS1L gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LAS1L	LAS1L, WTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Winchester Sendromu, 277950; WNCHRS (Torg-Winchester Sendromu) (MMP14 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Winchester syndrome, 277950; WNCHRS (Torg-Winchester syndrome) (MMP14 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MMP14	MMP14, WNCHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Winchester Sendromu, 277950; WNCHRS (Torg-Winchester Sendromu) (Prenatal) (MMP14 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Winchester syndrome, 277950; WNCHRS (Torg-Winchester syndrome) (MMP14 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MMP14	MMP14, WNCHRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wiskott Aldrich Sendromu 2, 614493; WAS2 (Wiskott-Aldrich Sendromu) (WIPF1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wiskott-Aldrich syndrome 2, 614493; WAS2 (Wiskott-Aldrich syndrome) (WIPF1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WIPF1	WIPF1, WASPIP, WIP, WAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wiskott Aldrich Sendromu 2, 614493; WAS2 (Wiskott-Aldrich Sendromu) (Prenatal) (WIPF1 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Wiskott-Aldrich syndrome 2, 614493; WAS2 (Wiskott-Aldrich syndrome) (WIPF1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WIPF1	WIPF1, WASPIP, WIP, WAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wiskott Aldrich Sendromu, 301000, X'e Bağlı Resesif; WAS (Wiskott-Aldrich Sendromu) (WAS gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wiskott-Aldrich syndrome, 301000, X-linked recessive; WAS (Wiskott-Aldrich syndrome) (WAS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wiskott Aldrich Sendromu, 301000, X'e Bağlı Resesif; WAS (Wiskott-Aldrich Sendromu) (Prenatal) (WAS gen) (Tüm Gen Dizi Analizi) (Prenatal)	Wiskott-Aldrich syndrome, 301000, X-linked recessive; WAS (Wiskott-Aldrich syndrome) (WAS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	WAS	WAS, IMD2, THC1, SCNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Witkop Sendromu (Hipodontia - Tırnak Displazisi) (MSX1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	WITKOP SYNDROME (Hypodontia-dysplasia of nails syndrome) (MSX1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MSX1	MSX1, HOX7, HYD1, OFC5, STHAG1, ECTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Witteveen-Kolk Sendromu, 613406, Otozomal Dominant; WITKOS (15q24 Mikrodelesyon Sendromu) (SIN3A gen) (Tüm Gen Dizi Analizi) (Postnatal)	Witteveen-Kolk syndrome, 613406, Autosomal dominant; WITKOS (15q24 microdeletion syndrome) (SIN3A gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SIN3A	SIN3A, WITKOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Witteveen-Kolk Sendromu, 613406, Otozomal Dominant; WITKOS (15q24 Mikrodelesyon Sendromu) (Prenatal) (SIN3A gen) (Tüm Gen Dizi Analizi) (Prenatal)	Witteveen-Kolk syndrome, 613406, Autosomal dominant; WITKOS (15q24 microdeletion syndrome) (SIN3A gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SIN3A	SIN3A, WITKOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Wolff-Parkinson-White Sendromu, 194200, Otozomal Dominant (PRKAG2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wolff-Parkinson-White syndrome, 194200, Autosomal dominant (PRKAG2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	PRKAG2	PRKAG2, WPWS, CMH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wolfram Sendromu 2, 604928, Otozomal Resesif; WFS2 (Wolfram Sendromu) (CISD2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wolfram syndrome 2, 604928, Autosomal recessive; WFS2 (Wolfram syndrome) (CISD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CISD2	CISD2, WFS2, ZCD2, ERIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wolfram Sendromu, 222300, Otozomal Resesif; WFS1 (Wolfram Sendromu) (WFS1 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Wolfram syndrome, 222300, Autosomal recessive; WFS1 (Wolfram syndrome) (WFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wolfram Sendromu, 222300, Otozomal Resesif; WFS1 (Wolfram Sendromu) (MLPA)	Wolfram syndrome, 222300, Autosomal recessive; WFS1 (Wolfram syndrome) (MLPA)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Wolfram-Benzeri Sendrom, Otozomal Dominant, 614296, Otozomal Dominant; WFSL (Wolfram-like syndrome) (WFS1 gen) (Sequence Analysis-All Coding Exons) (Postnatal)	Wolfram-like syndrome, autosomal dominant, 614296, Autosomal dominant; WFSL (Wolfram-like syndrome) (WFS1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	WFS1	WFS1, WFRS, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

Wolfram-Benzeri Sendrom, Otozomal Dominant, 614296, Otozomal Dominant, WFSL (Wolfram Benzeri Sendromi) (MLPA)	Wolfram-like syndrome, autosomal dominant, 614296, Autosomal dominant; WFSL (Wolfram-like syndrome) (MLPA)	WFS1	WFS1, WFR5, WFS, DFNA6, DFNA14, DFNA38, WFSL, CTRCT41	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Wolman Hastalığı, 278000, Otozomal Resesif (Lizozomal Asit Lipaz Eksikliği) (LIPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Wolman disease, 278000, Autosomal recessive (Lysosomal acid lipase deficiency) (LIPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPA	LIPA, CESD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Wolman Hastalığı, 278000, Otozomal Resesif (Lizozomal Asit Lipaz Eksikliği) (Prenatal) (LIPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Wolman disease, 278000, Autosomal recessive (Lysosomal acid lipase deficiency) (LIPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	LIPA	LIPA, CESD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Woodhouse-Sakati Sendromu, 241080, Otozomal Resesif (Woodhouse-Sakati Sendromu) (DCAF17 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Woodhouse-Sakati syndrome, 241080, Autosomal recessive (Woodhouse-Sakati syndrome) (DCAF17 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DCAF17	DCAF17, C20orf37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Woodhouse-Sakati Sendromu, 241080, Otozomal Resesif (Woodhouse-Sakati Sendromu) (Prenatal) (DCAF17 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Woodhouse-Sakati syndrome, 241080, Autosomal recessive (Woodhouse-Sakati syndrome) (DCAF17 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DCAF17	DCAF17, C20orf37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yünsü Saç, Otozomal Dominant, 194300, Otozomal Dominant; ADWH (Yünsü Saçlar) (KRT74 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Woolly hair, autosomal dominant, 194300, Autosomal dominant; ADWH (Woolly hair) (KRT74 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT74	KRT74, K6IRS4, KRT6IRS4, HTSS2, HYPT3, ADWH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yünsü Saç, Otozomal Resesif 1, ± Hipotrikozis, 278150, Otozomal Resesif (Yünsü Saçlar) (LPAR6 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150, Autosomal recessive (Woolly hair) (LPAR6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LPAR6	LPAR6, P2RY5, P2Y5, LAH3, ARWH1, HYPT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yünsü Saç, Otozomal Resesif 2,± Hipotrikozis, 604379, Otozomal Resesif (Yünsü Saçlar) (LIPH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379, Autosomal recessive (Woolly hair) (LIPH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	LIPH	LIPH, LAH2, ARWH2, HYPT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yünsü Saç, Otozomal Resesif 3, 616760, Otozomal Resesif; ARWH3 (Yünsü Saçlar) (KRT25 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Woolly hair, autosomal recessive 3, 616760, Autosomal recessive; ARWH3 (Woolly hair) (KRT25 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KRT25	KRT25, K25, KRT24IRS1, ARWH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kırışık Deri Sendromu, 278250, Otozomal Resesif, WSS (Kırışık Deri Sendromu (Wrinkly Skin Syndrome)) (ATP6V0A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Wrinkly skin syndrome, 278250, Autosomal recessive; WSS (Wrinkly skin syndrome) (ATP6V0A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V0A2	ATP6V0A2, WSS, ARCL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kırışık Deri Sendromu, 278250, Otozomal Resesif, WSS (Kırışık Deri Sendromu (Wrinkly Skin Syndrome)) (Prenatal) (ATP6V0A2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Wrinkly skin syndrome, 278250, Autosomal recessive; WSS (Wrinkly skin syndrome) (ATP6V0A2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP6V0A2	ATP6V0A2, WSS, ARCL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
X alfa satellit/Y alfa satellit (FISH)	X alpha satellite/Y alpha satellite (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
X Inaktivasyonu, Aktesel Kayıtsız, 300087 (XIST geni) (Tüm Gen Dizi Analizi) (Postnatal)	X-inactivation, familial skewed, 300087 (XIST gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XIST	XIC, XCE, XIST, SXI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ksantinüri, Tip 1, 278300, Otozomal Resesif; XAN1 (Kalıtsal Ksantinüri) (XDH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xanthinuria, type I, 278300, Autosomal recessive; XAN1 (Hereditary xanthinuria) (XDH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XDH	XDH, XAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ksantinüri, Tip 1, 278300, Otozomal Resesif; XAN1 (Kalıtsal Ksantinüri) (Prenatal) (XDH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xanthinuria, type I, 278300, Autosomal recessive; XAN1 (Hereditary xanthinuria) (XDH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XDH	XDH, XAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Ksantinüri, Tip 2, 603592, Otozomal Resesif; XAN2 (Kalıtsal Ksantinüri) (MOCOS geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xanthinuria, type II, 603592, Autosomal recessive; XAN2 (Hereditary xanthinuria) (MOCOS gene) (Sequence Analysis-All Coding Exons) (Postnatal)	MOCOS	MOCOS, MCS, XAN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ksantinüri, Tip 2, 603592, Otozomal Resesif; XAN2 (Kalıtsal Ksantinüri) (Prenatal) (MOCOS geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xanthinuria, type II, 603592, Autosomal recessive; XAN2 (Hereditary xanthinuria) (MOCOS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	MOCOS	MOCOS, MCS, XAN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup A, 278700, Otozomal Resesif (Kseroderma Pigmentozum) (XPA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group A, 278700, Autosomal recessive (Xeroderma pigmentosum) (XPA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XPA	XPA, XPAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup A, 278700, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (XPA geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group A, 278700, Autosomal recessive (Xeroderma pigmentosum) (XPA gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XPA	XPA, XPAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup B, 610651, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group B, 610651, Autosomal recessive (Xeroderma pigmentosum) (ERCC3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC3	ERCC3, XPB, TTD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup B, 610651, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC3 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group B, 610651, Autosomal recessive (Xeroderma pigmentosum) (ERCC3 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC3	ERCC3, XPB, TTD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup C, 278720, Otozomal Resesif (Kseroderma Pigmentozum) (XPC geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group C, 278720, Autosomal recessive (Xeroderma pigmentosum) (XPC gene) (Sequence Analysis-All Coding Exons) (Postnatal)	XPC	XPC, XPCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup C, 278720, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (XPC geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group C, 278720, Autosomal recessive (Xeroderma pigmentosum) (XPC gene) (Sequence Analysis-All Coding Exons) (Prenatal)	XPC	XPC, XPCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup D, 278730, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group D, 278730, Autosomal recessive (Xeroderma pigmentosum) (ERCC2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC2	ERCC2, EM9, XPD, COFS2, TTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup D, 278730, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group D, 278730, Autosomal recessive (Xeroderma pigmentosum) (ERCC2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC2	ERCC2, EM9, XPD, COFS2, TTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup E, DDB Negatif Alt Tip, 278740, Otozomal Resesif (Kseroderma Pigmentozum) (DDB2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740, Autosomal recessive (Xeroderma pigmentosum) (DDB2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	DDB2	DDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup E, DDB Negatif Alt Tip, 278740, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (DDB2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740, Autosomal recessive (Xeroderma pigmentosum) (DDB2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	DDB2	DDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup F, 278760, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group F, 278760, Autosomal recessive (Xeroderma pigmentosum) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup F, 278760, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group F, 278760, Autosomal recessive (Xeroderma pigmentosum) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup G, 278780, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group G, 278780, Autosomal recessive (Xeroderma pigmentosum) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup G, 278780, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group G, 278780, Autosomal recessive (Xeroderma pigmentosum) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Grup G / Cockayne Sendromu, 278760, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC5 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780, Autosomal recessive (Xeroderma pigmentosum) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Grup G / Cockayne Sendromu, 278780, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC5 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, group G/Cockayne syndrome, 278780, Autosomal recessive (Xeroderma pigmentosum) (ERCC5 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC5	ERCC5, XPG, COFS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Tip F / Cockayne Sendromu, 278760, Otozomal Resesif (Kseroderma Pigmentozum) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760, Autosomal recessive (Xeroderma pigmentosum) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Tip F / Cockayne Sendromu, 278760, Otozomal Resesif (Kseroderma Pigmentozum) (Prenatal) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, type F/Cockayne syndrome, 278760, Autosomal recessive (Xeroderma pigmentosum) (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Kseroderma Pigmentozum, Varyant Tipi, 278750, Otozomal Resesif; XPV (Kseroderma Pigmentozum Varyant) (POLH geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xeroderma pigmentosum, variant type, 278750, Autosomal recessive; XPV (Xeroderma pigmentosum variant) (POLH gene) (Sequence Analysis-All Coding Exons) (Postnatal)	POLH	POLH, XPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Kseroderma Pigmentozum, Varyant Tipi, 278750, Otozomal Resesif; XPV (Kseroderma Pigmentozum Varyant) (Prenatal) (POLH geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xeroderma pigmentosum, variant type, 278750, Autosomal recessive; XPV (Xeroderma pigmentosum variant) (POLH gene) (Sequence Analysis-All Coding Exons) (Prenatal)	POLH	POLH, XPV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
XFE Progeroid Sendromu, 610965 (ERCC4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	XFE progeroid syndrome, 610965 (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XFE Progeroid Sendromu, 610965 (Prenatal) (ERCC4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	XFE progeroid syndrome, 610965 (ERCC4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ERCC4	ERCC4, XPF, FANCQ, XFEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Xia-Gibbs Sendromu, 615829, Otozomal Dominant (AHDC1 ile İlişkili Entelektüel Yetersizlik, Otistik/dif Uyku Apnesi-Hafif Dismorfizm Sendromu (Xia-Gibbs Sendromu)) (AHDC1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Xia-Gibbs syndrome, 615829, Autosomal dominant (AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome) (AHDC1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	AHDC1	AHDC1, MRD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Xia-Gibbs Sendromu, 615829, Otozomal Dominant (AHDC1 ile İlişkili Entelektüel Yetersizlik, Otistik/dif Uyku Apnesi-Hafif Dismorfizm Sendromu (Xia-Gibbs Sendromu)) (Prenatal) (AHDC1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Xia-Gibbs syndrome, 615829, Autosomal dominant (AHDC1-related intellectual disability-obstructive sleep apnea-mild dysmorphism syndrome) (AHDC1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	AHDC1	AHDC1, MRD25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Xp11.22 Mikroduplikasyon Sendromu, 300705 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Postnatal)	Xp11.22 microduplication syndrome, 300705 (X-linked non-syndromic intellectual disability) (440)	.	DUPXp11.22, MRX17, MRX31	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Xp11.22 Mikroduplikasyon Sendromu, 300705 (X'e Bağlı Sendromik Olmayan Mental Retardasyon) (Prenatal)	Xp11.22 microduplication syndrome, 300705 (X-linked non-syndromic intellectual disability) (Prenatal)	.	DUPXp11.22, MRX17, MRX31	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Xp subtel/Yp subtel (FISH)	Xp subtel/Yp subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Xq subtel/ Yq subtel (FISH)	Xq subtel/ Yq subtel (FISH)	.	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample
Xq13.2 (XIST) (FISH)	Xq13.2 (XIST) (FISH)	Xq13.2	.	FISH	Heparinli Kan (2-4 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample

Xq25 Duplikasyon Sendromu, 300979, X'e Bağlı (2300)	Xq25 duplication syndrome, 300979, X-linked (440)	.	DUPXq25	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Xq25 Duplikasyon Sendromu, 300979, X'e Bağlı (Prenatal)	Xq25 duplication syndrome, 300979, X-linked (Prenatal)	.	DUPXq25	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
XY Kromozomu (FISH)	XY chromosome (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Y Kromozomu Mikrodelesyonu (SRY, AZFa - AZFb - AZFc) (Delesyon Analizi)	Y chromosome microdeletions (SRY, AZFa-AZFb-AZFc genes) (Deletion analysis)	SRY, AZFa - AZFb - AZFc	.	PCR/ Jel elektroforezi	EDTA'lı Kan (2-4 ml)	*	20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Yao Sendromu, 617321, Multifaktöryel; YAOS (NOD2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Yao syndrome, 617321, Multifactorial; YAOS (NOD2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	NOD2	NOD2, CARD15, IBD1, CD, YAOS, BLAUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yao Sendromu, 617321, Multifaktöryel; YAOS (Prenatal) (NOD2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Yao syndrome, 617321, Multifactorial: YAOS (NOD2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	NOD2	NOD2, CARD15, IBD1, CD, YAOS, BLAUS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
You-Hoover-Fong Sendromu, 616954, Otozomal Resesif; YHFS (TELO2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	You-Hoover-Fong syndrome, 616954, Autosomal recessive; YHFS (TELO2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	TELO2	TELO2, TEL2, CLK2, KIAA0683, HCLK2, YHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
You-Hoover-Fong Sendromu, 616954, Otozomal Resesif; YHFS (Prenatal) (TELO2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	You-Hoover-Fong syndrome, 616954, Autosomal recessive; YHFS (TELO2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	TELO2	TELO2, TEL2, CLK2, KIAA0683, HCLK2, YHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yuan-Harel-Lupski Sendromu, 616652, Otozomal Dominant; YUHAL (PMP22-RAI1-17p12-p11.2 Komşu Gen Duplikasyon Sendromu) (2200)	Yuan-Harel-Lupski syndrome, 616652, Autosomal dominant; YUHAL (PMP22-RAI1-17p12-p11.2 contiguous gene duplication syndrome) (440)	.	YUHAL	Array-CGH	EDTA'lı Kan (2-4 ml) ve Heparinli Kan (2-4 ml)		20	120	.	.	60	Postnatal	.	EDTA Blood Sample and Heparinized blood sample
Yuan-Harel-Lupski Sendromu, 616652, Otozomal Dominant; YUHAL (PMP22-RAI1-17p12-p11.2 Komşu Gen Duplikasyon Sendromu) (Prenatal)	Yuan-Harel-Lupski syndrome, 616652, Autosomal dominant; YUHAL (PMP22-RAI1-17p12-p11.2 contiguous gene duplication syndrome) (Prenatal)	.	YUHAL	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Yunis-Varon Sendromu, 216340, Otozomal Resesif; YVS (Yunis-Varon Sendromu) (FIG4 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Yunis-Varon syndrome, 216340, Autosomal recessive; YVS (Yunis-Varon syndrome) (FIG4 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	FIG4	FIG4, KIAA0274, SAC3, ALS11, YVS, BTOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Yunis-Varon Sendromu, 216340, Otozomal Resesif; YVS (Yunis-Varon Sendromu) (Prenatal) (FIG4 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Yunis-Varon syndrome, 216340, Autosomal recessive; YVS (Yunis-Varon syndrome) (FIG4 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	FIG4	FIG4, KIAA0274, SAC3, ALS11, YVS, BTOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Zimmermann-Laband Sendromu 1, 135500, Otozomal Dominant; ZLS1 (Zimmermann-Laband Sendromu) (KCNH1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Zimmermann-Laband syndrome 1, 135500, Autosomal dominant; ZLS1 (Zimmermann-Laband syndrome) (KCNH1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	KCNH1	KCNH1, EAG, TMBTS, ZLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Zimmermann-Laband Sendromu 1, 135500; Otozomal Dominant; ZLS1 (Zimmermann-Laband Sendromu) (Prenatal) (KCNH1 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Zimmermann-Laband syndrome 1, 135500; Autosomal dominant; ZLS1 (Zimmermann-Laband syndrome) (KCNH1 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	KCNH1	KCNH1, EAG, TMBTS, ZLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Zimmermann-Laband Sendromu 2, 616455, Otozomal Dominant; ZLS2 (Zimmermann-Laband Sendromu) (ATP6V1B2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Zimmermann-Laband syndrome 2, 616455, Autosomal dominant; ZLS2 (Zimmermann-Laband syndrome) (ATP6V1B2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	ATP6V1B2	ATP6B1B2, ATP6B2, VPP3, DOOD, ZLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Zimmermann-Laband Sendromu 2, 616455, Otozomal Dominant; ZLS2 (Zimmermann-Laband Sendromu) (Prenatal) (ATP6V1B2 geni) (Tüm Gen Dizi Analizi) (Prenatal)	Zimmermann-Laband syndrome 2, 616455, Autosomal dominant; ZLS2 (Zimmermann-Laband syndrome) (ATP6V1B2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	ATP6V1B2	ATP6B1B2, ATP6B2, VPP3, DOOD, ZLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Çinko Eksikliği, Geçici Neonatal, 608118, Otozomal Dominant; TNZD (SLC30A2 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Zinc deficiency, transient neonatal, 608118, Autosomal dominant; TNZD (SLC30A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SLC30A2	SLC30A2, ZNT2, TNZD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZTTK Sendromu, 617140, Otozomal Dominant; ZTTKS (SON geni) (Tüm Gen Dizi Analizi) (Postnatal)	ZTTK syndrome, 617140, Autosomal dominant; ZTTKS (SON gene) (Sequence Analysis-All Coding Exons) (Postnatal)	SON	SON, TOKIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZTTK Sendromu, 617140, Otozomal Dominant; ZTTKS (Prenatal) (SON geni) (Tüm Gen Dizi Analizi) (Prenatal)	ZTTK syndrome, 617140, Autosomal dominant; ZTTKS (SON gene) (Sequence Analysis-All Coding Exons) (Prenatal)	SON	SON, TOKIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
ZYX geni (Tüm Gen Dizi Analizi)	ZYX gene (Sequence Analysis-All Coding Exons)	ZYX	ZYX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZXDC geni (Tüm Gen Dizi Analizi)	ZXDC gene (Sequence Analysis-All Coding Exons)	ZXDC	ZXDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZXDB geni (Tüm Gen Dizi Analizi)	ZXDB gene (Sequence Analysis-All Coding Exons)	ZXDB	ZXDB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZXDA geni (Tüm Gen Dizi Analizi)	ZXDA gene (Sequence Analysis-All Coding Exons)	ZXDA	ZXDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZWINT geni (Tüm Gen Dizi Analizi)	ZWINT gene (Sequence Analysis-All Coding Exons)	ZWINT	ZWINT, SIP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZWILCH geni (Tüm Gen Dizi Analizi)	ZWILCH gene (Sequence Analysis-All Coding Exons)	ZWILCH	ZWILCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZW10 geni (Tüm Gen Dizi Analizi)	ZW10 gene (Sequence Analysis-All Coding Exons)	ZW10	ZW10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSWIM7 geni (Tüm Gen Dizi Analizi)	ZSWIM7 gene (Sequence Analysis-All Coding Exons)	ZSWIM7	ZSWIM7, SWS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN9 geni (Tüm Gen Dizi Analizi)	ZSCAN9 gene (Sequence Analysis-All Coding Exons)	ZSCAN9	ZNF193	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN4 geni (Tüm Gen Dizi Analizi)	ZSCAN4 gene (Sequence Analysis-All Coding Exons)	ZSCAN4	ZSCAN4, ZNF494	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN31 geni (Tüm Gen Dizi Analizi)	ZSCAN31 gene (Sequence Analysis-All Coding Exons)	ZSCAN31	ZNF323	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN26 geni (Tüm Gen Dizi Analizi)	ZSCAN26 gene (Sequence Analysis-All Coding Exons)	ZSCAN26	ZSCAN26, SREZBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN22 geni (Tüm Gen Dizi Analizi)	ZSCAN22 gene (Sequence Analysis-All Coding Exons)	ZSCAN22	ZSCAN22, HKR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN21 geni (Tüm Gen Dizi Analizi)	ZSCAN21 gene (Sequence Analysis-All Coding Exons)	ZSCAN21	ZNF38, KOX25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN20 geni (Tüm Gen Dizi Analizi)	ZSCAN20 gene (Sequence Analysis-All Coding Exons)	ZSCAN20	ZSCAN20, ZNF31, KOX29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZSCAN12 geni (Tüm Gen Dizi Analizi)	ZSCAN12 gene (Sequence Analysis-All Coding Exons)	ZSCAN12	ZSCAN12, ZNF96, KIAA0426	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZRSR2 geni (Tüm Gen Dizi Analizi)	ZRSR2 gene (Sequence Analysis-All Coding Exons)	ZRSR2	ZRSR2, U2AF1RS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZRANB3 geni (Tüm Gen Dizi Analizi)	ZRANB3 gene (Sequence Analysis-All Coding Exons)	ZRANB3	ZRANB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZRANB2 geni (Tüm Gen Dizi Analizi)	ZRANB2 gene (Sequence Analysis-All Coding Exons)	ZRANB2	ZRANB2, ZNF265, ZIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZRANB1 geni (Tüm Gen Dizi Analizi)	ZRANB1 gene (Sequence Analysis-All Coding Exons)	ZRANB1	ZRANB1, TRABID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZPR1 geni (Tüm Gen Dizi Analizi)	ZPR1 gene (Sequence Analysis-All Coding Exons)	ZPR1	ZPR1, ZNF259	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZPLD1 geni (Tüm Gen Dizi Analizi)	ZPLD1 gene (Sequence Analysis-All Coding Exons)	ZPLD1	ZPLD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZPBP2 geni (Tüm Gen Dizi Analizi)	ZPBP2 gene (Sequence Analysis-All Coding Exons)	ZPBP2	ZPBP2, ZPBPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZPBP geni (Tüm Gen Dizi Analizi)	ZPBP gene (Sequence Analysis-All Coding Exons)	ZPBP	ZPBP, SP38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZP4 geni (Tüm Gen Dizi Analizi)	ZP4 gene (Sequence Analysis-All Coding Exons)	ZP4	ZP4, ZPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZP3 geni (Tüm Gen Dizi Analizi)	ZP3 gene (Sequence Analysis-All Coding Exons)	ZP3	ZP3, ZP3A, ZP3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZP2 geni (Tüm Gen Dizi Analizi)	ZP2 gene (Sequence Analysis-All Coding Exons)	ZP2	ZP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRF4 geni (Tüm Gen Dizi Analizi)	ZNRF4 gene (Sequence Analysis-All Coding Exons)	ZNRF4	ZNRF4, SPERIZIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRF3 geni (Tüm Gen Dizi Analizi)	ZNRF3 gene (Sequence Analysis-All Coding Exons)	ZNRF3	ZNRF3, KIAA1133	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRF2 geni (Tüm Gen Dizi Analizi)	ZNRF2 gene (Sequence Analysis-All Coding Exons)	ZNRF2	ZNRF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRF1 geni (Tüm Gen Dizi Analizi)	ZNRF1 gene (Sequence Analysis-All Coding Exons)	ZNRF1	ZNRF1, NIN283	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRD1ASP geni (Tüm Gen Dizi Analizi)	ZNRD1ASP gene (Sequence Analysis-All Coding Exons)	ZNRD1ASP	ZNRD1AS1, TCTEX4, HTEX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNRD1 geni (Tüm Gen Dizi Analizi)	ZNRD1 gene (Sequence Analysis-All Coding Exons)	ZNRD1	ZNRD1, TCTEX6, HTEX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNHIT3 geni (Tüm Gen Dizi Analizi)	ZNHIT3 gene (Sequence Analysis-All Coding Exons)	ZNHIT3	ZNHIT3, TRIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNHIT2 geni (Tüm Gen Dizi Analizi)	ZNHIT2 gene (Sequence Analysis-All Coding Exons)	ZNHIT2	ZNHIT2, C11orf5, FON	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF99 geni (Tüm Gen Dizi Analizi)	ZNF99 gene (Sequence Analysis-All Coding Exons)	ZNF99	ZNF99	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF98 geni (Tüm Gen Dizi Analizi)	ZNF98 gene (Sequence Analysis-All Coding Exons)	ZNF98	ZNF98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF93 geni (Tüm Gen Dizi Analizi)	ZNF93 gene (Sequence Analysis-All Coding Exons)	ZNF93	ZNF93	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF92 geni (Tüm Gen Dizi Analizi)	ZNF92 gene (Sequence Analysis-All Coding Exons)	ZNF92	ZNF92	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF91 geni (Tüm Gen Dizi Analizi)	ZNF91 gene (Sequence Analysis-All Coding Exons)	ZNF91	ZNF91	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF90 geni (Tüm Gen Dizi Analizi)	ZNF90 gene (Sequence Analysis-All Coding Exons)	ZNF90	ZNF90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF85 geni (Tüm Gen Dizi Analizi)	ZNF85 gene (Sequence Analysis-All Coding Exons)	ZNF85	ZNF85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF83 geni (Tüm Gen Dizi Analizi)	ZNF83 gene (Sequence Analysis-All Coding Exons)	ZNF83	ZNF83	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF81 geni (Tüm Gen Dizi Analizi)	ZNF81 gene (Sequence Analysis-All Coding Exons)	ZNF81	ZNF81	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF804A geni (Tüm Gen Dizi Analizi)	ZNF804A gene (Sequence Analysis-All Coding Exons)	ZNF804A	ZNF804A, C2orf10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF80 geni (Tüm Gen Dizi Analizi)	ZNF80 gene (Sequence Analysis-All Coding Exons)	ZNF80	ZNF80	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF8 geni (Tüm Gen Dizi Analizi)	ZNF8 gene (Sequence Analysis-All Coding Exons)	ZNF8	ZNF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF79 geni (Tüm Gen Dizi Analizi)	ZNF79 gene (Sequence Analysis-All Coding Exons)	ZNF79	ZNF79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF77 geni (Tüm Gen Dizi Analizi)	ZNF77 gene (Sequence Analysis-All Coding Exons)	ZNF77	ZNF77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF76 geni (Tüm Gen Dizi Analizi)	ZNF76 gene (Sequence Analysis-All Coding Exons)	ZNF76	ZNF76, D6S229E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF75D geni (Tüm Gen Dizi Analizi)	ZNF75D gene (Sequence Analysis-All Coding Exons)	ZNF75D	ZNF75D, ZNF75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF75A geni (Tüm Gen Dizi Analizi)	ZNF75A gene (Sequence Analysis-All Coding Exons)	ZNF75A	ZNF75A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF746 geni (Tüm Gen Dizi Analizi)	ZNF746 gene (Sequence Analysis-All Coding Exons)	ZNF746	ZNF746, PARIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF74 geni (Tüm Gen Dizi Analizi)	ZNF74 gene (Sequence Analysis-All Coding Exons)	ZNF74	ZNF74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF737 geni (Tüm Gen Dizi Analizi)	ZNF737 gene (Sequence Analysis-All Coding Exons)	ZNF737	ZNF737, ZNF102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF713 geni (Tüm Gen Dizi Analizi)	ZNF713 gene (Sequence Analysis-All Coding Exons)	ZNF713	ZNF713	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF71 geni (Tüm Gen Dizi Analizi)	ZNF71 gene (Sequence Analysis-All Coding Exons)	ZNF71	ZNF71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF703 geni (Tüm Gen Dizi Analizi)	ZNF703 gene (Sequence Analysis-All Coding Exons)	ZNF703	ZNF703, ZEPPO1, NLZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ZNF70 geni (Tüm Gen Dizi Analizi)	ZNF70 gene (Sequence Analysis-All Coding Exons)	ZNF70	ZNF70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF7 geni (Tüm Gen Dizi Analizi)	ZNF7 gene (Sequence Analysis-All Coding Exons)	ZNF7	ZNF7, KOX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF699 geni (Tüm Gen Dizi Analizi)	ZNF699 gene (Sequence Analysis-All Coding Exons)	ZNF699	ZNF699, FLJ38144	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF695 geni (Tüm Gen Dizi Analizi)	ZNF695 gene (Sequence Analysis-All Coding Exons)	ZNF695	ZNF695, SBZF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF69 geni (Tüm Gen Dizi Analizi)	ZNF69 gene (Sequence Analysis-All Coding Exons)	ZNF69	ZNF69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF683 geni (Tüm Gen Dizi Analizi)	ZNF683 gene (Sequence Analysis-All Coding Exons)	ZNF683	ZNF683, HOBIT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF674 geni (Tüm Gen Dizi Analizi)	ZNF674 gene (Sequence Analysis-All Coding Exons)	ZNF674	ZNF674	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF668 geni (Tüm Gen Dizi Analizi)	ZNF668 gene (Sequence Analysis-All Coding Exons)	ZNF668	ZNF668	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF667 geni (Tüm Gen Dizi Analizi)	ZNF667 gene (Sequence Analysis-All Coding Exons)	ZNF667	ZNF667, MIPU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF658 geni (Tüm Gen Dizi Analizi)	ZNF658 gene (Sequence Analysis-All Coding Exons)	ZNF658	ZNF658	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF653 geni (Tüm Gen Dizi Analizi)	ZNF653 gene (Sequence Analysis-All Coding Exons)	ZNF653	ZNF653, ZIF67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF652 geni (Tüm Gen Dizi Analizi)	ZNF652 gene (Sequence Analysis-All Coding Exons)	ZNF652	ZNF652, KIAA0924	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF649 geni (Tüm Gen Dizi Analizi)	ZNF649 gene (Sequence Analysis-All Coding Exons)	ZNF649	ZNF649	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF641 geni (Tüm Gen Dizi Analizi)	ZNF641 gene (Sequence Analysis-All Coding Exons)	ZNF641	ZNF641	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF638 geni (Tüm Gen Dizi Analizi)	ZNF638 gene (Sequence Analysis-All Coding Exons)	ZNF638	ZNF638, NP220	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF630 geni (Tüm Gen Dizi Analizi)	ZNF630 gene (Sequence Analysis-All Coding Exons)	ZNF630	ZNF630	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF628 geni (Tüm Gen Dizi Analizi)	ZNF628 gene (Sequence Analysis-All Coding Exons)	ZNF628	ZNF628, ZEC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF627 geni (Tüm Gen Dizi Analizi)	ZNF627 gene (Sequence Analysis-All Coding Exons)	ZNF627	ZNF627	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF618 geni (Tüm Gen Dizi Analizi)	ZNF618 gene (Sequence Analysis-All Coding Exons)	ZNF618	ZNF618, NEDD10, KIAA1952	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF606 geni (Tüm Gen Dizi Analizi)	ZNF606 gene (Sequence Analysis-All Coding Exons)	ZNF606	ZNF606, ZNF328, KIAA1852	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF597 geni (Tüm Gen Dizi Analizi)	ZNF597 gene (Sequence Analysis-All Coding Exons)	ZNF597	ZNF597	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF593 geni (Tüm Gen Dizi Analizi)	ZNF593 gene (Sequence Analysis-All Coding Exons)	ZNF593	ZNF593, ZT86	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF592 geni (Tüm Gen Dizi Analizi)	ZNF592 gene (Sequence Analysis-All Coding Exons)	ZNF592	ZNF592, KIAA0211	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZRSR1 geni (Tüm Gen Dizi Analizi)	ZRSR1 gene (Sequence Analysis-All Coding Exons)	ZRSR1	ZRSR1, U2AF1RS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF589 geni (Tüm Gen Dizi Analizi)	ZNF589 gene (Sequence Analysis-All Coding Exons)	ZNF589	ZNF589, SZF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF582 geni (Tüm Gen Dizi Analizi)	ZNF582 gene (Sequence Analysis-All Coding Exons)	ZNF582	ZNF582	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF569 geni (Tüm Gen Dizi Analizi)	ZNF569 gene (Sequence Analysis-All Coding Exons)	ZNF569	ZNF569	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF565 geni (Tüm Gen Dizi Analizi)	ZNF565 gene (Sequence Analysis-All Coding Exons)	ZNF565	ZNF565	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF543 geni (Tüm Gen Dizi Analizi)	ZNF543 gene (Sequence Analysis-All Coding Exons)	ZNF543	ZNF543	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF540 geni (Tüm Gen Dizi Analizi)	ZNF540 gene (Sequence Analysis-All Coding Exons)	ZNF540	ZNF540	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF528 geni (Tüm Gen Dizi Analizi)	ZNF528 gene (Sequence Analysis-All Coding Exons)	ZNF528	ZNF528, KIAA1827	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF526 geni (Tüm Gen Dizi Analizi)	ZNF526 gene (Sequence Analysis-All Coding Exons)	ZNF526	ZNF526, KIAA1951	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF521 geni (Tüm Gen Dizi Analizi)	ZNF521 gene (Sequence Analysis-All Coding Exons)	ZNF521	ZNF521, EHZF, EVI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF516 geni (Tüm Gen Dizi Analizi)	ZNF516 gene (Sequence Analysis-All Coding Exons)	ZNF516	ZNF516, KIAA0222	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF503 geni (Tüm Gen Dizi Analizi)	ZNF503 gene (Sequence Analysis-All Coding Exons)	ZNF503	ZNF503, NOLZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF496 geni (Tüm Gen Dizi Analizi)	ZNF496 gene (Sequence Analysis-All Coding Exons)	ZNF496	ZNF496, NZP1, ZFP496	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF480 geni (Tüm Gen Dizi Analizi)	ZNF480 gene (Sequence Analysis-All Coding Exons)	ZNF480	ZNF480	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF468 geni (Tüm Gen Dizi Analizi)	ZNF468 gene (Sequence Analysis-All Coding Exons)	ZNF468	ZNF468	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF467 geni (Tüm Gen Dizi Analizi)	ZNF467 gene (Sequence Analysis-All Coding Exons)	ZNF467	ZNF467, EZI, ZFP467	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF462 geni (Tüm Gen Dizi Analizi)	ZNF462 gene (Sequence Analysis-All Coding Exons)	ZNF462	ZNF462, KIAA1803	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF461 geni (Tüm Gen Dizi Analizi)	ZNF461 gene (Sequence Analysis-All Coding Exons)	ZNF461	ZNF461, GIOT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF460 geni (Tüm Gen Dizi Analizi)	ZNF460 gene (Sequence Analysis-All Coding Exons)	ZNF460	ZNF272, HZF8, ZNF460	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF451 geni (Tüm Gen Dizi Analizi)	ZNF451 gene (Sequence Analysis-All Coding Exons)	ZNF451	ZNF451, COASTER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF45 geni (Tüm Gen Dizi Analizi)	ZNF45 gene (Sequence Analysis-All Coding Exons)	ZNF45	ZNF45, ZNF13, KOX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF449 geni (Tüm Gen Dizi Analizi)	ZNF449 gene (Sequence Analysis-All Coding Exons)	ZNF449	ZNF449, ZSCAN19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF444 geni (Tüm Gen Dizi Analizi)	ZNF444 gene (Sequence Analysis-All Coding Exons)	ZNF444	ZNF444, EZF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF443 geni (Tüm Gen Dizi Analizi)	ZNF443 gene (Sequence Analysis-All Coding Exons)	ZNF443	ZK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF44 geni (Tüm Gen Dizi Analizi)	ZNF44 gene (Sequence Analysis-All Coding Exons)	ZNF44	ZNF44, KOX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF436 geni (Tüm Gen Dizi Analizi)	ZNF436 gene (Sequence Analysis-All Coding Exons)	ZNF436	ZNF436, KIAA1710	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF43 geni (Tüm Gen Dizi Analizi)	ZNF43 gene (Sequence Analysis-All Coding Exons)	ZNF43	ZNF43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF420 geni (Tüm Gen Dizi Analizi)	ZNF420 gene (Sequence Analysis-All Coding Exons)	ZNF420	ZNF420, APAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF41 geni (Tüm Gen Dizi Analizi)	ZNF41 gene (Sequence Analysis-All Coding Exons)	ZNF41	ZNF41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF407 geni (Tüm Gen Dizi Analizi)	ZNF407 gene (Sequence Analysis-All Coding Exons)	ZNF407	ZNF407	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF397 geni (Tüm Gen Dizi Analizi)	ZNF397 gene (Sequence Analysis-All Coding Exons)	ZNF397	ZNF397	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF396 geni (Tüm Gen Dizi Analizi)	ZNF396 gene (Sequence Analysis-All Coding Exons)	ZNF396	ZNF396	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF385B geni (Tüm Gen Dizi Analizi)	ZNF385B gene (Sequence Analysis-All Coding Exons)	ZNF385B	ZNF385B, ZFN533	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF385A geni (Tüm Gen Dizi Analizi)	ZNF385A gene (Sequence Analysis-All Coding Exons)	ZNF385A	ZNF385A, ZNF385, RZF, HZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF384 geni (Tüm Gen Dizi Analizi)	ZNF384 gene (Sequence Analysis-All Coding Exons)	ZNF384	ZNF384, CIZ, CAGH1, NMP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF382 geni (Tüm Gen Dizi Analizi)	ZNF382 gene (Sequence Analysis-All Coding Exons)	ZNF382	ZNF382, KS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF37A geni (Tüm Gen Dizi Analizi)	ZNF37A gene (Sequence Analysis-All Coding Exons)	ZNF37A	ZNF37A, KOX21, ZNF37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF367 geni (Tüm Gen Dizi Analizi)	ZNF367 gene (Sequence Analysis-All Coding Exons)	ZNF367	ZNF367, AFF29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF366 geni (Tüm Gen Dizi Analizi)	ZNF366 gene (Sequence Analysis-All Coding Exons)	ZNF366	ZNF366, DCSCRIPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF354A geni (Tüm Gen Dizi Analizi)	ZNF354A gene (Sequence Analysis-All Coding Exons)	ZNF354A	TCF17, KID1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF35 geni (Tüm Gen Dizi Analizi)	ZNF35 gene (Sequence Analysis-All Coding Exons)	ZNF35	ZNF35, HF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF34 geni (Tüm Gen Dizi Analizi)	ZNF34 gene (Sequence Analysis-All Coding Exons)	ZNF34	ZNF34, KOX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF338 geni (Tüm Gen Dizi Analizi)	ZNF338 gene (Sequence Analysis-All Coding Exons)	ZNF338	ZNF338, ZNF11B, KOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF33A geni (Tüm Gen Dizi Analizi)	ZNF33A gene (Sequence Analysis-All Coding Exons)	ZNF33A	ZNF33A, NF11A, KOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF333 geni (Tüm Gen Dizi Analizi)	ZNF333 gene (Sequence Analysis-All Coding Exons)	ZNF333	ZNF333, KIAA1806	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF331 geni (Tüm Gen Dizi Analizi)	ZNF331 gene (Sequence Analysis-All Coding Exons)	ZNF331	ZNF331, ZNF463	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF330 geni (Tüm Gen Dizi Analizi)	ZNF330 gene (Sequence Analysis-All Coding Exons)	ZNF330	ZNF330, NOA36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF326 geni (Tüm Gen Dizi Analizi)	ZNF326 gene (Sequence Analysis-All Coding Exons)	ZNF326	ZNF326, ZIRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF322 geni (Tüm Gen Dizi Analizi)	ZNF322 gene (Sequence Analysis-All Coding Exons)	ZNF322	ZNF322A, ZNF489, ZNF322	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF320 geni (Tüm Gen Dizi Analizi)	ZNF320 gene (Sequence Analysis-All Coding Exons)	ZNF320	ZNF320	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF32 geni (Tüm Gen Dizi Analizi)	ZNF32 gene (Sequence Analysis-All Coding Exons)	ZNF32	ZNF32, KOX30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF317 geni (Tüm Gen Dizi Analizi)	ZNF317 gene (Sequence Analysis-All Coding Exons)	ZNF317	ZNF317, KIAA1588	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF304 geni (Tüm Gen Dizi Analizi)	ZNF304 gene (Sequence Analysis-All Coding Exons)	ZNF304	ZNF304	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF300 geni (Tüm Gen Dizi Analizi)	ZNF300 gene (Sequence Analysis-All Coding Exons)	ZNF300	ZNF300	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF3 geni (Tüm Gen Dizi Analizi)	ZNF3 gene (Sequence Analysis-All Coding Exons)	ZNF3	ZNF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF296 geni (Tüm Gen Dizi Analizi)	ZNF296 gene (Sequence Analysis-All Coding Exons)	ZNF296	ZNF296, ZFP296, ZNF342	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF292 geni (Tüm Gen Dizi Analizi)	ZNF292 gene (Sequence Analysis-All Coding Exons)	ZNF292	ZNF292, ZFP292, KIAA0530	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF282 geni (Tüm Gen Dizi Analizi)	ZNF282 gene (Sequence Analysis-All Coding Exons)	ZNF282	ZNF282, HUB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF277 geni (Tüm Gen Dizi Analizi)	ZNF277 gene (Sequence Analysis-All Coding Exons)	ZNF277	ZNF277	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF276 geni (Tüm Gen Dizi Analizi)	ZNF276 gene (Sequence Analysis-All Coding Exons)	ZNF276	ZFP276	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF274 geni (Tüm Gen Dizi Analizi)	ZNF274 gene (Sequence Analysis-All Coding Exons)	ZNF274	ZNF274	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF271P geni (Tüm Gen Dizi Analizi)	ZNF271P gene (Sequence Analysis-All Coding Exons)	ZNF271P	ZNFEB, ZNF271	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF268 geni (Tüm Gen Dizi Analizi)	ZNF268 gene (Sequence Analysis-All Coding Exons)	ZNF268	ZNF268	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF267 geni (Tüm Gen Dizi Analizi)	ZNF267 gene (Sequence Analysis-All Coding Exons)	ZNF267	ZNF267, HZF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF266 geni (Tüm Gen Dizi Analizi)	ZNF266 gene (Sequence Analysis-All Coding Exons)	ZNF266	ZNF266, HZF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF622 geni (Tüm Gen Dizi Analizi)	ZNF622 gene (Sequence Analysis-All Coding Exons)	ZNF622		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF264 geni (Tüm Gen Dizi Analizi)	ZNF264 gene (Sequence Analysis-All Coding Exons)	ZNF264	ZNF264	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF263 geni (Tüm Gen Dizi Analizi)	ZNF263 gene (Sequence Analysis-All Coding Exons)	ZNF263	ZNF263	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF260 geni (Tüm Gen Dizi Analizi)	ZNF260 gene (Sequence Analysis-All Coding Exons)	ZNF260	ZNF260, PEX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF26 geni (Tüm Gen Dizi Analizi)	ZNF26 gene (Sequence Analysis-All Coding Exons)	ZNF26	ZNF26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ZNF257 geni (Tüm Gen Dizi Analizi)	ZNF257 gene (Sequence Analysis-All Coding Exons)	ZNF257	ZNF257	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF256 geni (Tüm Gen Dizi Analizi)	ZNF256 gene (Sequence Analysis-All Coding Exons)	ZNF256	ZNF256	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF254 geni (Tüm Gen Dizi Analizi)	ZNF254 gene (Sequence Analysis-All Coding Exons)	ZNF254	ZNF254, ZNF91L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF253 geni (Tüm Gen Dizi Analizi)	ZNF253 gene (Sequence Analysis-All Coding Exons)	ZNF253	ZNF253	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF25 geni (Tüm Gen Dizi Analizi)	ZNF25 gene (Sequence Analysis-All Coding Exons)	ZNF25	ZNF25, KOX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF24 geni (Tüm Gen Dizi Analizi)	ZNF24 gene (Sequence Analysis-All Coding Exons)	ZNF24	ZNF24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF239 geni (Tüm Gen Dizi Analizi)	ZNF239 gene (Sequence Analysis-All Coding Exons)	ZNF239	ZNF239, MOK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF236 geni (Tüm Gen Dizi Analizi)	ZNF236 gene (Sequence Analysis-All Coding Exons)	ZNF236	ZNF236	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF235 geni (Tüm Gen Dizi Analizi)	ZNF235 gene (Sequence Analysis-All Coding Exons)	ZNF235	ZFP93, ANF270, HZF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF232 geni (Tüm Gen Dizi Analizi)	ZNF232 gene (Sequence Analysis-All Coding Exons)	ZNF232	ZNF232, ZSCAN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF23 geni (Tüm Gen Dizi Analizi)	ZNF23 gene (Sequence Analysis-All Coding Exons)	ZNF23	ZNF23, KOX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF224 geni (Tüm Gen Dizi Analizi)	ZNF224 gene (Sequence Analysis-All Coding Exons)	ZNF224	ZNF224, ZNF255, BMZF2, KOX22, ZNF27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF222 geni (Tüm Gen Dizi Analizi)	ZNF222 gene (Sequence Analysis-All Coding Exons)	ZNF222	ZNF222	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF22 geni (Tüm Gen Dizi Analizi)	ZNF22 gene (Sequence Analysis-All Coding Exons)	ZNF22	ZNF22, KOX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF219 geni (Tüm Gen Dizi Analizi)	ZNF219 gene (Sequence Analysis-All Coding Exons)	ZNF219	ZNF219	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF217 geni (Tüm Gen Dizi Analizi)	ZNF217 gene (Sequence Analysis-All Coding Exons)	ZNF217	ZNF217	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF215 geni (Tüm Gen Dizi Analizi)	ZNF215 gene (Sequence Analysis-All Coding Exons)	ZNF215	ZNF215	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF214 geni (Tüm Gen Dizi Analizi)	ZNF214 gene (Sequence Analysis-All Coding Exons)	ZNF214	ZNF214	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF213 geni (Tüm Gen Dizi Analizi)	ZNF213 gene (Sequence Analysis-All Coding Exons)	ZNF213	ZNF213, CRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF212 geni (Tüm Gen Dizi Analizi)	ZNF212 gene (Sequence Analysis-All Coding Exons)	ZNF212	ZNF212, ZNFC150, ZNF182	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF211 geni (Tüm Gen Dizi Analizi)	ZNF211 gene (Sequence Analysis-All Coding Exons)	ZNF211	ZNFC25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF208 geni (Tüm Gen Dizi Analizi)	ZNF208 gene (Sequence Analysis-All Coding Exons)	ZNF208	ZNF208, ZNF95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF207 geni (Tüm Gen Dizi Analizi)	ZNF207 gene (Sequence Analysis-All Coding Exons)	ZNF207	ZNF207	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF205 geni (Tüm Gen Dizi Analizi)	ZNF205 gene (Sequence Analysis-All Coding Exons)	ZNF205	ZNF205, ZNF210	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF202 geni (Tüm Gen Dizi Analizi)	ZNF202 gene (Sequence Analysis-All Coding Exons)	ZNF202	ZNF202	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF200 geni (Tüm Gen Dizi Analizi)	ZNF200 gene (Sequence Analysis-All Coding Exons)	ZNF200	ZNF200	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF20 geni (Tüm Gen Dizi Analizi)	ZNF20 gene (Sequence Analysis-All Coding Exons)	ZNF20	ZNF20, KOX13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF2 geni (Tüm Gen Dizi Analizi)	ZNF2 gene (Sequence Analysis-All Coding Exons)	ZNF2	ZNF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF195 geni (Tüm Gen Dizi Analizi)	ZNF195 gene (Sequence Analysis-All Coding Exons)	ZNF195	ZNF195	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF19 geni (Tüm Gen Dizi Analizi)	ZNF19 gene (Sequence Analysis-All Coding Exons)	ZNF19	ZNF19, KOX12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF189 geni (Tüm Gen Dizi Analizi)	ZNF189 gene (Sequence Analysis-All Coding Exons)	ZNF189	ZNF189	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF185 geni (Tüm Gen Dizi Analizi)	ZNF185 gene (Sequence Analysis-All Coding Exons)	ZNF185	ZNF185	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF184 geni (Tüm Gen Dizi Analizi)	ZNF184 gene (Sequence Analysis-All Coding Exons)	ZNF184	ZNF184	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF182 geni (Tüm Gen Dizi Analizi)	ZNF182 gene (Sequence Analysis-All Coding Exons)	ZNF182	ZNF21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF181 geni (Tüm Gen Dizi Analizi)	ZNF181 gene (Sequence Analysis-All Coding Exons)	ZNF181	ZNF181	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF180 geni (Tüm Gen Dizi Analizi)	ZNF180 gene (Sequence Analysis-All Coding Exons)	ZNF180	ZNF180	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF18 geni (Tüm Gen Dizi Analizi)	ZNF18 gene (Sequence Analysis-All Coding Exons)	ZNF18	ZNF18, KOX11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF177 geni (Tüm Gen Dizi Analizi)	ZNF177 gene (Sequence Analysis-All Coding Exons)	ZNF177	ZNF177	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF395 geni (Tüm Gen Dizi Analizi)	ZNF395 gene (Sequence Analysis-All Coding Exons)	ZNF395		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF175 geni (Tüm Gen Dizi Analizi)	ZNF175 gene (Sequence Analysis-All Coding Exons)	ZNF175	ZNF175, OTK18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF174 geni (Tüm Gen Dizi Analizi)	ZNF174 gene (Sequence Analysis-All Coding Exons)	ZNF174	ZNF174	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF169 geni (Tüm Gen Dizi Analizi)	ZNF169 gene (Sequence Analysis-All Coding Exons)	ZNF169	ZNF169	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF165 geni (Tüm Gen Dizi Analizi)	ZNF165 gene (Sequence Analysis-All Coding Exons)	ZNF165	ZNF165, LD65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF160 geni (Tüm Gen Dizi Analizi)	ZNF160 gene (Sequence Analysis-All Coding Exons)	ZNF160	ZNF160	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF16 geni (Tüm Gen Dizi Analizi)	ZNF16 gene (Sequence Analysis-All Coding Exons)	ZNF16	ZNF16, KOX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF157 geni (Tüm Gen Dizi Analizi)	ZNF157 gene (Sequence Analysis-All Coding Exons)	ZNF157	ZNF157	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF155 geni (Tüm Gen Dizi Analizi)	ZNF155 gene (Sequence Analysis-All Coding Exons)	ZNF155	ZNF155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF154 geni (Tüm Gen Dizi Analizi)	ZNF154 gene (Sequence Analysis-All Coding Exons)	ZNF154	ZNF154	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF350 geni (Tüm Gen Dizi Analizi)	ZNF350 gene (Sequence Analysis-All Coding Exons)	ZNF350		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF146 geni (Tüm Gen Dizi Analizi)	ZNF146 gene (Sequence Analysis-All Coding Exons)	ZNF146	ZNF146, OZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF346 geni (Tüm Gen Dizi Analizi)	ZNF346 gene (Sequence Analysis-All Coding Exons)	ZNF346		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF143 geni (Tüm Gen Dizi Analizi)	ZNF143 gene (Sequence Analysis-All Coding Exons)	ZNF143	ZNF143, SBF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF142 geni (Tüm Gen Dizi Analizi)	ZNF142 gene (Sequence Analysis-All Coding Exons)	ZNF142	ZNF142	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF140 geni (Tüm Gen Dizi Analizi)	ZNF140 gene (Sequence Analysis-All Coding Exons)	ZNF140	ZNF140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF14 geni (Tüm Gen Dizi Analizi)	ZNF14 gene (Sequence Analysis-All Coding Exons)	ZNF14	ZNF14, KOX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF138 geni (Tüm Gen Dizi Analizi)	ZNF138 gene (Sequence Analysis-All Coding Exons)	ZNF138	ZNF138	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF136 geni (Tüm Gen Dizi Analizi)	ZNF136 gene (Sequence Analysis-All Coding Exons)	ZNF136	ZNF136	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF135 geni (Tüm Gen Dizi Analizi)	ZNF135 gene (Sequence Analysis-All Coding Exons)	ZNF135	ZNF135	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF134 geni (Tüm Gen Dizi Analizi)	ZNF134 gene (Sequence Analysis-All Coding Exons)	ZNF134	ZNF134	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF133 geni (Tüm Gen Dizi Analizi)	ZNF133 gene (Sequence Analysis-All Coding Exons)	ZNF133	ZNF133	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF132 geni (Tüm Gen Dizi Analizi)	ZNF132 gene (Sequence Analysis-All Coding Exons)	ZNF132	ZNF132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF131 geni (Tüm Gen Dizi Analizi)	ZNF131 gene (Sequence Analysis-All Coding Exons)	ZNF131	ZNF131	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF124 geni (Tüm Gen Dizi Analizi)	ZNF124 gene (Sequence Analysis-All Coding Exons)	ZNF124	ZNF124	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF121 geni (Tüm Gen Dizi Analizi)	ZNF121 gene (Sequence Analysis-All Coding Exons)	ZNF121	ZNF121, D19S204	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF12 geni (Tüm Gen Dizi Analizi)	ZNF12 gene (Sequence Analysis-All Coding Exons)	ZNF12	ZNF12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF117 geni (Tüm Gen Dizi Analizi)	ZNF117 gene (Sequence Analysis-All Coding Exons)	ZNF117	ZNF117	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF114 geni (Tüm Gen Dizi Analizi)	ZNF114 gene (Sequence Analysis-All Coding Exons)	ZNF114	ZNF114	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF112 geni (Tüm Gen Dizi Analizi)	ZNF112 gene (Sequence Analysis-All Coding Exons)	ZNF112	ZNF112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF107 geni (Tüm Gen Dizi Analizi)	ZNF107 gene (Sequence Analysis-All Coding Exons)	ZNF107	ZNF107	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF101 geni (Tüm Gen Dizi Analizi)	ZNF101 gene (Sequence Analysis-All Coding Exons)	ZNF101	ZNF101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF100 geni (Tüm Gen Dizi Analizi)	ZNF100 gene (Sequence Analysis-All Coding Exons)	ZNF100	ZNF100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF10 geni (Tüm Gen Dizi Analizi)	ZNF10 gene (Sequence Analysis-All Coding Exons)	ZNF10	ZNF10, KOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF273 geni (Tüm Gen Dizi Analizi)	ZNF273 gene (Sequence Analysis-All Coding Exons)	ZNF273		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYND8 geni (Tüm Gen Dizi Analizi)	ZMYND8 gene (Sequence Analysis-All Coding Exons)	ZMYND8	ZMYND8, PRKCBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYND19 geni (Tüm Gen Dizi Analizi)	ZMYND19 gene (Sequence Analysis-All Coding Exons)	ZMYND19	ZMYND19, MIZIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYM6 geni (Tüm Gen Dizi Analizi)	ZMYM6 gene (Sequence Analysis-All Coding Exons)	ZMYM6	ZMYM6, ZNF258	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYM5 geni (Tüm Gen Dizi Analizi)	ZMYM5 gene (Sequence Analysis-All Coding Exons)	ZMYM5	ZMYM5, ZNF237, ZNF198L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYM4 geni (Tüm Gen Dizi Analizi)	ZMYM4 gene (Sequence Analysis-All Coding Exons)	ZMYM4	ZMYM4, ZNF262, KIAA0425	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYM3 geni (Tüm Gen Dizi Analizi)	ZMYM3 gene (Sequence Analysis-All Coding Exons)	ZMYM3	ZMYM3, ZNF261, DXS6673E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMYM2 geni (Tüm Gen Dizi Analizi)	ZMYM2 gene (Sequence Analysis-All Coding Exons)	ZMYM2	ZMYM2, ZNF198, RAMP, FIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMIZ2 geni (Tüm Gen Dizi Analizi)	ZMIZ2 gene (Sequence Analysis-All Coding Exons)	ZMIZ2	ZMIZ2, KIAA1886, ZIMP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMIZ1 geni (Tüm Gen Dizi Analizi)	ZMIZ1 gene (Sequence Analysis-All Coding Exons)	ZMIZ1	RAI17, KIAA1224, ZIMP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZMAT3 geni (Tüm Gen Dizi Analizi)	ZMAT3 gene (Sequence Analysis-All Coding Exons)	ZMAT3	WIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZKSCAN8 geni (Tüm Gen Dizi Analizi)	ZKSCAN8 gene (Sequence Analysis-All Coding Exons)	ZKSCAN8	ZNF192	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZKSCAN5 geni (Tüm Gen Dizi Analizi)	ZKSCAN5 gene (Sequence Analysis-All Coding Exons)	ZKSCAN5	ZKSCAN5, ZFP95, KIAA1015	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZKSCAN4 geni (Tüm Gen Dizi Analizi)	ZKSCAN4 gene (Sequence Analysis-All Coding Exons)	ZKSCAN4	ZKSCAN4, ZNF307	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZKSCAN3 geni (Tüm Gen Dizi Analizi)	ZKSCAN3 gene (Sequence Analysis-All Coding Exons)	ZKSCAN3	ZKSCAN3, ZNF306	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZKSCAN1 geni (Tüm Gen Dizi Analizi)	ZKSCAN1 gene (Sequence Analysis-All Coding Exons)	ZKSCAN1	ZNF36, KOX18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZIC4 geni (Tüm Gen Dizi Analizi)	ZIC4 gene (Sequence Analysis-All Coding Exons)	ZIC4	ZIC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZIC4 geni (Tüm Gen Dizi Analizi)	ZIC4 gene (Sequence Analysis-All Coding Exons)	ZIC4	ZIC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZNF234 geni (Tüm Gen Dizi Analizi)	ZNF234 gene (Sequence Analysis-All Coding Exons)	ZNF234		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZHX3 geni (Tüm Gen Dizi Analizi)	ZHX3 gene (Sequence Analysis-All Coding Exons)	ZHX3	ZHX3, KIAA0395	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZHX1 geni (Tüm Gen Dizi Analizi)	ZHX1 gene (Sequence Analysis-All Coding Exons)	ZHX1	ZHX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ZGLP1 geni (Tüm Gen Dizi Analizi)	ZGLP1 gene (Sequence Analysis-All Coding Exons)	ZGLP1	ZGLP1, GLP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZG16 geni (Tüm Gen Dizi Analizi)	ZG16 gene (Sequence Analysis-All Coding Exons)	ZG16	ZG16, ZG16A, JCLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFYVE9 geni (Tüm Gen Dizi Analizi)	ZFYVE9 gene (Sequence Analysis-All Coding Exons)	ZFYVE9	ZFYVE9, MADHIP, SARA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFYVE28 geni (Tüm Gen Dizi Analizi)	ZFYVE28 gene (Sequence Analysis-All Coding Exons)	ZFYVE28	ZFYVE28, LST2, KIAA1643	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFYVE21 geni (Tüm Gen Dizi Analizi)	ZFYVE21 gene (Sequence Analysis-All Coding Exons)	ZFYVE21	ZFYVE21, ZF21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFYVE16 geni (Tüm Gen Dizi Analizi)	ZFYVE16 gene (Sequence Analysis-All Coding Exons)	ZFYVE16	ZFYVE16, ENDOFIN, KIAA0305	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFYVE1 geni (Tüm Gen Dizi Analizi)	ZFYVE1 gene (Sequence Analysis-All Coding Exons)	ZFYVE1	ZNFN2A1, DFCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFY geni (Tüm Gen Dizi Analizi)	ZFY gene (Sequence Analysis-All Coding Exons)	ZFY	ZFY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFX geni (Tüm Gen Dizi Analizi)	ZFX gene (Sequence Analysis-All Coding Exons)	ZFX	ZFX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFR geni (Tüm Gen Dizi Analizi)	ZFR gene (Sequence Analysis-All Coding Exons)	ZFR	ZFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFPM1 geni (Tüm Gen Dizi Analizi)	ZFPM1 gene (Sequence Analysis-All Coding Exons)	ZFPM1	ZFPM1, FOG, FOG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP90 geni (Tüm Gen Dizi Analizi)	ZFP90 gene (Sequence Analysis-All Coding Exons)	ZFP90	ZFP90, NK10, KIAA1954	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP62 geni (Tüm Gen Dizi Analizi)	ZFP62 gene (Sequence Analysis-All Coding Exons)	ZFP62	ZFP62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP42 geni (Tüm Gen Dizi Analizi)	ZFP42 gene (Sequence Analysis-All Coding Exons)	ZFP42	ZFP42, REX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP37 geni (Tüm Gen Dizi Analizi)	ZFP37 gene (Sequence Analysis-All Coding Exons)	ZFP37	ZFP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP36L2 geni (Tüm Gen Dizi Analizi)	ZFP36L2 gene (Sequence Analysis-All Coding Exons)	ZFP36L2	ZFP36L2, ERF2, TIS11D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP36L1 geni (Tüm Gen Dizi Analizi)	ZFP36L1 gene (Sequence Analysis-All Coding Exons)	ZFP36L1	ZFP36L1, BRF1, ERF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP36 geni (Tüm Gen Dizi Analizi)	ZFP36 gene (Sequence Analysis-All Coding Exons)	ZFP36	ZFP36, TTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP30 geni (Tüm Gen Dizi Analizi)	ZFP30 gene (Sequence Analysis-All Coding Exons)	ZFP30	ZFP30, ZNF745, KIAA0961	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP3 geni (Tüm Gen Dizi Analizi)	ZFP3 gene (Sequence Analysis-All Coding Exons)	ZFP3	ZFP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP28 geni (Tüm Gen Dizi Analizi)	ZFP28 gene (Sequence Analysis-All Coding Exons)	ZFP28	ZFP28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFP1 geni (Tüm Gen Dizi Analizi)	ZFP1 gene (Sequence Analysis-All Coding Exons)	ZFP1	ZFP1, ZNF475	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFAND6 geni (Tüm Gen Dizi Analizi)	ZFAND6 gene (Sequence Analysis-All Coding Exons)	ZFAND6	ZFAND6, ZA20D3, AWP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFAND5 geni (Tüm Gen Dizi Analizi)	ZFAND5 gene (Sequence Analysis-All Coding Exons)	ZFAND5	ZFAND5, ZNF216	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFAND3 geni (Tüm Gen Dizi Analizi)	ZFAND3 gene (Sequence Analysis-All Coding Exons)	ZFAND3	ZFAND3, TEX27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFAND2B geni (Tüm Gen Dizi Analizi)	ZFAND2B gene (Sequence Analysis-All Coding Exons)	ZFAND2B	ZFAND2B, AIRAPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZFAND2A geni (Tüm Gen Dizi Analizi)	ZFAND2A gene (Sequence Analysis-All Coding Exons)	ZFAND2A	ZFAND2A, AIRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC8 geni (Tüm Gen Dizi Analizi)	ZDHHC8 gene (Sequence Analysis-All Coding Exons)	ZDHHC8	ZDHHC8, ZNF378, KIAA1292	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC7 geni (Tüm Gen Dizi Analizi)	ZDHHC7 gene (Sequence Analysis-All Coding Exons)	ZDHHC7	ZDHHC7, DHHC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC5 geni (Tüm Gen Dizi Analizi)	ZDHHC5 gene (Sequence Analysis-All Coding Exons)	ZDHHC5	ZDHHC5, DHHC5, ZNF375, KIAA1748	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC3 geni (Tüm Gen Dizi Analizi)	ZDHHC3 gene (Sequence Analysis-All Coding Exons)	ZDHHC3	ZDHHC3, DHHC3, GODZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC23 geni (Tüm Gen Dizi Analizi)	ZDHHC23 gene (Sequence Analysis-All Coding Exons)	ZDHHC23	ZDHHC23, NIDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC21 geni (Tüm Gen Dizi Analizi)	ZDHHC21 gene (Sequence Analysis-All Coding Exons)	ZDHHC21	ZDHHC21, DHHC21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC17 geni (Tüm Gen Dizi Analizi)	ZDHHC17 gene (Sequence Analysis-All Coding Exons)	ZDHHC17	ZDHHC17, HIP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC16 geni (Tüm Gen Dizi Analizi)	ZDHHC16 gene (Sequence Analysis-All Coding Exons)	ZDHHC16	ZDHHC16, APH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDHHC13 geni (Tüm Gen Dizi Analizi)	ZDHHC13 gene (Sequence Analysis-All Coding Exons)	ZDHHC13	ZDHHC13, HIP14L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZDBF2 geni (Tüm Gen Dizi Analizi)	ZDBF2 gene (Sequence Analysis-All Coding Exons)	ZDBF2	ZDBF2, KIAA1571	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCRB1 geni (Tüm Gen Dizi Analizi)	ZCRB1 gene (Sequence Analysis-All Coding Exons)	ZCRB1	ZCRB1, MADP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCCHC8 geni (Tüm Gen Dizi Analizi)	ZCCHC8 gene (Sequence Analysis-All Coding Exons)	ZCCHC8	ZCCHC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCCHC6 geni (Tüm Gen Dizi Analizi)	ZCCHC6 gene (Sequence Analysis-All Coding Exons)	ZCCHC6	ZCCHC6, TUT7, KIAA1711	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCCHC4 geni (Tüm Gen Dizi Analizi)	ZCCHC4 gene (Sequence Analysis-All Coding Exons)	ZCCHC4	ZCCHC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCCHC12 geni (Tüm Gen Dizi Analizi)	ZCCHC12 gene (Sequence Analysis-All Coding Exons)	ZCCHC12	ZCCHC12, SIZN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZCCHC11 geni (Tüm Gen Dizi Analizi)	ZCCHC11 gene (Sequence Analysis-All Coding Exons)	ZCCHC11	ZCCHC11, KIAA0191	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3HAV1 geni (Tüm Gen Dizi Analizi)	ZC3HAV1 gene (Sequence Analysis-All Coding Exons)	ZC3HAV1	ZC3HAV1, ZAP, FLJ13288, FLJ58421	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H13 geni (Tüm Gen Dizi Analizi)	ZC3H13 gene (Sequence Analysis-All Coding Exons)	ZC3H13	ZC3H13, KIAA0853	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H12D geni (Tüm Gen Dizi Analizi)	ZC3H12D gene (Sequence Analysis-All Coding Exons)	ZC3H12D	ZC3H12D, p34, C6orf95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H12C geni (Tüm Gen Dizi Analizi)	ZC3H12C gene (Sequence Analysis-All Coding Exons)	ZC3H12C	ZC3H12C, MCPIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H12B geni (Tüm Gen Dizi Analizi)	ZC3H12B gene (Sequence Analysis-All Coding Exons)	ZC3H12B	ZC3H12B, MCPIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H12A geni (Tüm Gen Dizi Analizi)	ZC3H12A gene (Sequence Analysis-All Coding Exons)	ZC3H12A	ZC3H12A, MCPIP, MCPIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZC3H11A geni (Tüm Gen Dizi Analizi)	ZC3H11A gene (Sequence Analysis-All Coding Exons)	ZC3H11A	ZC3H11A, KIAA0663	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB8OS geni (Tüm Gen Dizi Analizi)	ZBTB8OS gene (Sequence Analysis-All Coding Exons)	ZBTB8OS	ZBTB8OS, ARCH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB7C geni (Tüm Gen Dizi Analizi)	ZBTB7C gene (Sequence Analysis-All Coding Exons)	ZBTB7C	ZBTB7C, APM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB7B geni (Tüm Gen Dizi Analizi)	ZBTB7B gene (Sequence Analysis-All Coding Exons)	ZBTB7B	ZBTB7B, ZFP67, CKROX, THPOK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB7A geni (Tüm Gen Dizi Analizi)	ZBTB7A gene (Sequence Analysis-All Coding Exons)	ZBTB7A	ZBTB7A, FBI1, LRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB5 geni (Tüm Gen Dizi Analizi)	ZBTB5 gene (Sequence Analysis-All Coding Exons)	ZBTB5	ZBTB5, KIAA0354	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB49 geni (Tüm Gen Dizi Analizi)	ZBTB49 gene (Sequence Analysis-All Coding Exons)	ZBTB49	ZBTB49, ZNF509	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB48 geni (Tüm Gen Dizi Analizi)	ZBTB48 gene (Sequence Analysis-All Coding Exons)	ZBTB48	ZBTB48, HKR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB46 geni (Tüm Gen Dizi Analizi)	ZBTB46 gene (Sequence Analysis-All Coding Exons)	ZBTB46	ZBTB46, BZEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB40 geni (Tüm Gen Dizi Analizi)	ZBTB40 gene (Sequence Analysis-All Coding Exons)	ZBTB40	ZBTB40, KIAA0478	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB4 geni (Tüm Gen Dizi Analizi)	ZBTB4 gene (Sequence Analysis-All Coding Exons)	ZBTB4	ZBTB4, KIAA1538	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB38 geni (Tüm Gen Dizi Analizi)	ZBTB38 gene (Sequence Analysis-All Coding Exons)	ZBTB38	ZBTB38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB34 geni (Tüm Gen Dizi Analizi)	ZBTB34 gene (Sequence Analysis-All Coding Exons)	ZBTB34	ZBTB34, KIAA1993	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB33 geni (Tüm Gen Dizi Analizi)	ZBTB33 gene (Sequence Analysis-All Coding Exons)	ZBTB33	ZBTB33, KAISO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB32 geni (Tüm Gen Dizi Analizi)	ZBTB32 gene (Sequence Analysis-All Coding Exons)	ZBTB32	ZBTB32, FAXF, TZFP, ROG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB25 geni (Tüm Gen Dizi Analizi)	ZBTB25 gene (Sequence Analysis-All Coding Exons)	ZBTB25	ZBTB25, ZNF46, KUP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB22 geni (Tüm Gen Dizi Analizi)	ZBTB22 gene (Sequence Analysis-All Coding Exons)	ZBTB22	ZBTB22, BING1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB21 geni (Tüm Gen Dizi Analizi)	ZBTB21 gene (Sequence Analysis-All Coding Exons)	ZBTB21	ZBTB21, ZNF295, KIAA1277	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB2 geni (Tüm Gen Dizi Analizi)	ZBTB2 gene (Sequence Analysis-All Coding Exons)	ZBTB2	ZBTB2, ZNF437, KIAA1483	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB17 geni (Tüm Gen Dizi Analizi)	ZBTB17 gene (Sequence Analysis-All Coding Exons)	ZBTB17	ZBTB17, ZNF151, MIZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB14 geni (Tüm Gen Dizi Analizi)	ZBTB14 gene (Sequence Analysis-All Coding Exons)	ZBTB14	ZFP161, ZF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB1 geni (Tüm Gen Dizi Analizi)	ZBTB1 gene (Sequence Analysis-All Coding Exons)	ZBTB1	ZBTB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBP1 geni (Tüm Gen Dizi Analizi)	ZBP1 gene (Sequence Analysis-All Coding Exons)	ZBP1	ZBP1, C20ORF183, DLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED9 geni (Tüm Gen Dizi Analizi)	ZBED9 gene (Sequence Analysis-All Coding Exons)	ZBED9	SCAND3, ZBED9, BUSTER4, KIAA1975	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED8 geni (Tüm Gen Dizi Analizi)	ZBED8 gene (Sequence Analysis-All Coding Exons)	ZBED8	C5orf54, ZBED8, BUSTER3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED6CL geni (Tüm Gen Dizi Analizi)	ZBED6CL gene (Sequence Analysis-All Coding Exons)	ZBED6CL	ZBED6CL, C7orf29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED6 geni (Tüm Gen Dizi Analizi)	ZBED6 gene (Sequence Analysis-All Coding Exons)	ZBED6	ZBED6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED5 geni (Tüm Gen Dizi Analizi)	ZBED5 gene (Sequence Analysis-All Coding Exons)	ZBED5	ZBED5, BUSTER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED4 geni (Tüm Gen Dizi Analizi)	ZBED4 gene (Sequence Analysis-All Coding Exons)	ZBED4	ZBED4, KIAA0637	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED3 geni (Tüm Gen Dizi Analizi)	ZBED3 gene (Sequence Analysis-All Coding Exons)	ZBED3	ZBED3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED2 geni (Tüm Gen Dizi Analizi)	ZBED2 gene (Sequence Analysis-All Coding Exons)	ZBED2	ZBED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBED1 geni (Tüm Gen Dizi Analizi)	ZBED1 gene (Sequence Analysis-All Coding Exons)	ZBED1	ZBED1, ALTE, KIAA0785, TRAMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZAR1 geni (Tüm Gen Dizi Analizi)	ZAR1 gene (Sequence Analysis-All Coding Exons)	ZAR1	ZAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZAN geni (Tüm Gen Dizi Analizi)	ZAN gene (Sequence Analysis-All Coding Exons)	ZAN	ZAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZACN geni (Tüm Gen Dizi Analizi)	ZACN gene (Sequence Analysis-All Coding Exons)	ZACN	LGICZ1, ZAC, L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YY2 geni (Tüm Gen Dizi Analizi)	YY2 gene (Sequence Analysis-All Coding Exons)	YY2	YY2, ZNF631	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YY1 geni (Tüm Gen Dizi Analizi)	YY1 gene (Sequence Analysis-All Coding Exons)	YY1	YY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAZ geni (Tüm Gen Dizi Analizi)	YWHAZ gene (Sequence Analysis-All Coding Exons)	YWHAZ	YWHAZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAQ geni (Tüm Gen Dizi Analizi)	YWHAQ gene (Sequence Analysis-All Coding Exons)	YWHAQ	TYHAQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZHX2 geni (Tüm Gen Dizi Analizi)	ZHX2 gene (Sequence Analysis-All Coding Exons)	ZHX2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAH geni (Tüm Gen Dizi Analizi)	YWHAH gene (Sequence Analysis-All Coding Exons)	YWHAH	YWHAH, YWHA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAG geni (Tüm Gen Dizi Analizi)	YWHAG gene (Sequence Analysis-All Coding Exons)	YWHAG	YWHAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAE geni (Tüm Gen Dizi Analizi)	YWHAE gene (Sequence Analysis-All Coding Exons)	YWHAE	YWHAE, MDCR, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAE geni (Tüm Gen Dizi Analizi)	YWHAE gene (Sequence Analysis-All Coding Exons)	YWHAE	YWHAE, MDCR, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

YWHAE geni (Tüm Gen Dizi Analizi)	YWHAE gene (Sequence Analysis-All Coding Exons)	YWHAE	YWHAE, MDCR, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAE geni (Tüm Gen Dizi Analizi)	YWHAE gene (Sequence Analysis-All Coding Exons)	YWHAE	YWHAE, MDCR, MDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YWHAB geni (Tüm Gen Dizi Analizi)	YWHAB gene (Sequence Analysis-All Coding Exons)	YWHAB	YWHAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YTHDF2 geni (Tüm Gen Dizi Analizi)	YTHDF2 gene (Sequence Analysis-All Coding Exons)	YTHDF2	YTHDF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YTHDF1 geni (Tüm Gen Dizi Analizi)	YTHDF1 gene (Sequence Analysis-All Coding Exons)	YTHDF1	YTHDF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YTHDC2 geni (Tüm Gen Dizi Analizi)	YTHDC2 gene (Sequence Analysis-All Coding Exons)	YTHDC2	YTHDC2, CAHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YTHDC1 geni (Tüm Gen Dizi Analizi)	YTHDC1 gene (Sequence Analysis-All Coding Exons)	YTHDC1	YTHDC1, KIAA1966	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YRDC geni (Tüm Gen Dizi Analizi)	YRDC gene (Sequence Analysis-All Coding Exons)	YRDC	YRDC, IRIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YPEL5 geni (Tüm Gen Dizi Analizi)	YPEL5 gene (Sequence Analysis-All Coding Exons)	YPEL5	YPEL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YPEL4 geni (Tüm Gen Dizi Analizi)	YPEL4 gene (Sequence Analysis-All Coding Exons)	YPEL4	YPEL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YPEL3 geni (Tüm Gen Dizi Analizi)	YPEL3 gene (Sequence Analysis-All Coding Exons)	YPEL3	YPEL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YPEL2 geni (Tüm Gen Dizi Analizi)	YPEL2 gene (Sequence Analysis-All Coding Exons)	YPEL2	YPEL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YPEL1 geni (Tüm Gen Dizi Analizi)	YPEL1 gene (Sequence Analysis-All Coding Exons)	YPEL1	YPEL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YOD1 geni (Tüm Gen Dizi Analizi)	YOD1 gene (Sequence Analysis-All Coding Exons)	YOD1	YOD1, OTUD2, DUBA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YIPF5 geni (Tüm Gen Dizi Analizi)	YIPF5 gene (Sequence Analysis-All Coding Exons)	YIPF5	YIPF5, SMAP5, YIP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YIPF3 geni (Tüm Gen Dizi Analizi)	YIPF3 gene (Sequence Analysis-All Coding Exons)	YIPF3	YIPF3, KLIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YIF1A geni (Tüm Gen Dizi Analizi)	YIF1A gene (Sequence Analysis-All Coding Exons)	YIF1A	YIF1A, YIF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YES1 geni (Tüm Gen Dizi Analizi)	YES1 gene (Sequence Analysis-All Coding Exons)	YES1	YES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YEATS4 geni (Tüm Gen Dizi Analizi)	YEATS4 gene (Sequence Analysis-All Coding Exons)	YEATS4	YEATS4, GAS41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YEATS2 geni (Tüm Gen Dizi Analizi)	YEATS2 gene (Sequence Analysis-All Coding Exons)	YEATS2	YEATS2, KIAA1197	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YBX3 geni (Tüm Gen Dizi Analizi)	YBX3 gene (Sequence Analysis-All Coding Exons)	YBX3	CSDA, DBPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YBX2 geni (Tüm Gen Dizi Analizi)	YBX2 gene (Sequence Analysis-All Coding Exons)	YBX2	YBX2, MSY2, CSDA3, DBPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YBX1 geni (Tüm Gen Dizi Analizi)	YBX1 gene (Sequence Analysis-All Coding Exons)	YBX1	YBX1, NSEP1, YB1, DBPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YAF2 geni (Tüm Gen Dizi Analizi)	YAF2 gene (Sequence Analysis-All Coding Exons)	YAF2	YAF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XYLB geni (Tüm Gen Dizi Analizi)	XYLB gene (Sequence Analysis-All Coding Exons)	XYLB	XYLB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XXYLT1 geni (Tüm Gen Dizi Analizi)	XXYLT1 gene (Sequence Analysis-All Coding Exons)	XXYLT1	XXYLT1, C3orf21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRRA1 geni (Tüm Gen Dizi Analizi)	XRRA1 gene (Sequence Analysis-All Coding Exons)	XRRA1	XRRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRN2 geni (Tüm Gen Dizi Analizi)	XRN2 gene (Sequence Analysis-All Coding Exons)	XRN2	XRN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRN1 geni (Tüm Gen Dizi Analizi)	XRN1 gene (Sequence Analysis-All Coding Exons)	XRN1	XRN1, SEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRCC6 geni (Tüm Gen Dizi Analizi)	XRCC6 gene (Sequence Analysis-All Coding Exons)	XRCC6	XRCC6, G22P1, TLAA, Ku70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRCC5 geni (Tüm Gen Dizi Analizi)	XRCC5 gene (Sequence Analysis-All Coding Exons)	XRCC5	XRCC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XRCC1 geni (Tüm Gen Dizi Analizi)	XRCC1 gene (Sequence Analysis-All Coding Exons)	XRCC1	XRCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPO7 geni (Tüm Gen Dizi Analizi)	XPO7 gene (Sequence Analysis-All Coding Exons)	XPO7	XPO7, RANBP16, KIAA0745	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPO6 geni (Tüm Gen Dizi Analizi)	XPO6 gene (Sequence Analysis-All Coding Exons)	XPO6	XPO6, EXP6, KIAA0370	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPO5 geni (Tüm Gen Dizi Analizi)	XPO5 gene (Sequence Analysis-All Coding Exons)	XPO5	XPO5, KIAA1291	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPO1 geni (Tüm Gen Dizi Analizi)	XPO1 gene (Sequence Analysis-All Coding Exons)	XPO1	XPO1, CRM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPNPEP1 geni (Tüm Gen Dizi Analizi)	XPNPEP1 gene (Sequence Analysis-All Coding Exons)	XPNPEP1	XPNPEP1, SAMP, XPNPEPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XKRX geni (Tüm Gen Dizi Analizi)	XKRX gene (Sequence Analysis-All Coding Exons)	XKRX	XKRX, XPLAC, XKR2, XRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XKR3 geni (Tüm Gen Dizi Analizi)	XKR3 gene (Sequence Analysis-All Coding Exons)	XKR3	XKR3, XTES, XRG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XIRP2 geni (Tüm Gen Dizi Analizi)	XIRP2 gene (Sequence Analysis-All Coding Exons)	XIRP2	CMYA3, XIRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XIRP1 geni (Tüm Gen Dizi Analizi)	XIRP1 gene (Sequence Analysis-All Coding Exons)	XIRP1	CMYA1, XIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XG geni (Tüm Gen Dizi Analizi)	XG gene (Sequence Analysis-All Coding Exons)	XG	XG, PBDX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XCR1 geni (Tüm Gen Dizi Analizi)	XCR1 gene (Sequence Analysis-All Coding Exons)	XCR1	XCR1, CCXCR1, GPR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XCL2 geni (Tüm Gen Dizi Analizi)	XCL2 gene (Sequence Analysis-All Coding Exons)	XCL2	XCL2, SCYC2, XCL2, SCM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XCL1 geni (Tüm Gen Dizi Analizi)	XCL1 gene (Sequence Analysis-All Coding Exons)	XCL1	XCL1, SCYC1, SCM1, LTN, LPTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE3 geni (Tüm Gen Dizi Analizi)	XAGE3 gene (Sequence Analysis-All Coding Exons)	XAGE3	XAGE3, PLAC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE2 geni (Tüm Gen Dizi Analizi)	XAGE2 gene (Sequence Analysis-All Coding Exons)	XAGE2	XAGE2, GAGED3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE1E geni (Tüm Gen Dizi Analizi)	XAGE1E gene (Sequence Analysis-All Coding Exons)	XAGE1E	GAGED2, XAGE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE1E geni (Tüm Gen Dizi Analizi)	XAGE1E gene (Sequence Analysis-All Coding Exons)	XAGE1C	XAGE1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE1E geni (Tüm Gen Dizi Analizi)	XAGE1E gene (Sequence Analysis-All Coding Exons)	XAGE1E	XAGE1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE1B geni (Tüm Gen Dizi Analizi)	XAGE1B gene (Sequence Analysis-All Coding Exons)	XAGE1B	XAGE1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAGE1B geni (Tüm Gen Dizi Analizi)	XAGE1B gene (Sequence Analysis-All Coding Exons)	XAGE1B	XAGE1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAF1 geni (Tüm Gen Dizi Analizi)	XAF1 gene (Sequence Analysis-All Coding Exons)	XAF1	XAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XACT geni (Tüm Gen Dizi Analizi)	XACT gene (Sequence Analysis-All Coding Exons)	XACT	XACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XAB2 geni (Tüm Gen Dizi Analizi)	XAB2 gene (Sequence Analysis-All Coding Exons)	XAB2	XAB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WWTR1 geni (Tüm Gen Dizi Analizi)	WWTR1 gene (Sequence Analysis-All Coding Exons)	WWTR1	WWTR1, TAZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WWP2 geni (Tüm Gen Dizi Analizi)	WWP2 gene (Sequence Analysis-All Coding Exons)	WWP2	WWP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WWP1 geni (Tüm Gen Dizi Analizi)	WWP1 gene (Sequence Analysis-All Coding Exons)	WWP1	WWP1, TIUL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WTIP geni (Tüm Gen Dizi Analizi)	WTIP gene (Sequence Analysis-All Coding Exons)	WTIP	WTIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WTAP geni (Tüm Gen Dizi Analizi)	WTAP gene (Sequence Analysis-All Coding Exons)	WTAP	WTAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WSPAR geni (Tüm Gen Dizi Analizi)	WSPAR gene (Sequence Analysis-All Coding Exons)	WSPAR	WSPAR, lncTCF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WSB1 geni (Tüm Gen Dizi Analizi)	WSB1 gene (Sequence Analysis-All Coding Exons)	WSB1	WSB1, SWIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WRNIP1 geni (Tüm Gen Dizi Analizi)	WRNIP1 gene (Sequence Analysis-All Coding Exons)	WRNIP1	WRNIP1, WHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WRB geni (Tüm Gen Dizi Analizi)	WRB gene (Sequence Analysis-All Coding Exons)	WRB	WRB, CHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WRAP73 geni (Tüm Gen Dizi Analizi)	WRAP73 gene (Sequence Analysis-All Coding Exons)	WRAP73	WDR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT9B geni (Tüm Gen Dizi Analizi)	WNT9B gene (Sequence Analysis-All Coding Exons)	WNT9B	WNT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT9A geni (Tüm Gen Dizi Analizi)	WNT9A gene (Sequence Analysis-All Coding Exons)	WNT9A	WNT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZBTB6 geni (Tüm Gen Dizi Analizi)	ZBTB6 gene (Sequence Analysis-All Coding Exons)	ZBTB6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT8B geni (Tüm Gen Dizi Analizi)	WNT8B gene (Sequence Analysis-All Coding Exons)	WNT8B	WNT8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT8A geni (Tüm Gen Dizi Analizi)	WNT8A gene (Sequence Analysis-All Coding Exons)	WNT8A	WNT8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT7B geni (Tüm Gen Dizi Analizi)	WNT7B gene (Sequence Analysis-All Coding Exons)	WNT7B	WNT7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT6 geni (Tüm Gen Dizi Analizi)	WNT6 gene (Sequence Analysis-All Coding Exons)	WNT6	WNT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT5B geni (Tüm Gen Dizi Analizi)	WNT5B gene (Sequence Analysis-All Coding Exons)	WNT5B	WNT5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT3A geni (Tüm Gen Dizi Analizi)	WNT3A gene (Sequence Analysis-All Coding Exons)	WNT3A	WNT3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT2B geni (Tüm Gen Dizi Analizi)	WNT2B gene (Sequence Analysis-All Coding Exons)	WNT2B	WNT13, XWNT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT2 geni (Tüm Gen Dizi Analizi)	WNT2 gene (Sequence Analysis-All Coding Exons)	WNT2	WNT2, INT1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT16 geni (Tüm Gen Dizi Analizi)	WNT16 gene (Sequence Analysis-All Coding Exons)	WNT16	WNT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNT11 geni (Tüm Gen Dizi Analizi)	WNT11 gene (Sequence Analysis-All Coding Exons)	WNT11	WNT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNK3 geni (Tüm Gen Dizi Analizi)	WNK3 gene (Sequence Analysis-All Coding Exons)	WNK3	WNK3, PRKWNK3, KIAA1566	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WNK2 geni (Tüm Gen Dizi Analizi)	WNK2 gene (Sequence Analysis-All Coding Exons)	WNK2	WNK2, PRKWNK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WLS geni (Tüm Gen Dizi Analizi)	WLS gene (Sequence Analysis-All Coding Exons)	WLS	WLS, GPR177, EVI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WISP2 geni (Tüm Gen Dizi Analizi)	WISP2 gene (Sequence Analysis-All Coding Exons)	WISP2	WISP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WISP1 geni (Tüm Gen Dizi Analizi)	WISP1 gene (Sequence Analysis-All Coding Exons)	WISP1	WISP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WIPI2 geni (Tüm Gen Dizi Analizi)	WIPI2 gene (Sequence Analysis-All Coding Exons)	WIPI2	WIPI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WIPF3 geni (Tüm Gen Dizi Analizi)	WIPF3 gene (Sequence Analysis-All Coding Exons)	WIPF3	WIPF3, CR16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WIPF2 geni (Tüm Gen Dizi Analizi)	WIPF2 gene (Sequence Analysis-All Coding Exons)	WIPF2	WIRE, WICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WIF1 geni (Tüm Gen Dizi Analizi)	WIF1 gene (Sequence Analysis-All Coding Exons)	WIF1	WIF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WHAMM geni (Tüm Gen Dizi Analizi)	WHAMM gene (Sequence Analysis-All Coding Exons)	WHAMM	WHAMM, WHAMM, KIAA1971	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WFIKKN2 geni (Tüm Gen Dizi Analizi)	WFIKKN2 gene (Sequence Analysis-All Coding Exons)	WFIKKN2	WFIKKN2, WFIKKNRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WFIKKN1 geni (Tüm Gen Dizi Analizi)	WFIKKN1 gene (Sequence Analysis-All Coding Exons)	WFIKKN1	WFIKKN1, WFIKKN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WFDC5 geni (Tüm Gen Dizi Analizi)	WFDC5 gene (Sequence Analysis-All Coding Exons)	WFDC5	WFDC5, PRG5, WAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WFDC12 geni (Tüm Gen Dizi Analizi)	WFDC12 gene (Sequence Analysis-All Coding Exons)	WFDC12	WFDC12, WAP2, SWAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WFDC1 geni (Tüm Gen Dizi Analizi)	WFDC1 gene (Sequence Analysis-All Coding Exons)	WFDC1	WFDC1, PS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

WEE2 geni (Tüm Gen Dizi Analizi)	WEE2 gene (Sequence Analysis-All Coding Exons)	WEE2	WEE2, WEE1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WEE1 geni (Tüm Gen Dizi Analizi)	WEE1 gene (Sequence Analysis-All Coding Exons)	WEE1	WEE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR92 geni (Tüm Gen Dizi Analizi)	WDR92 gene (Sequence Analysis-All Coding Exons)	WDR92	WDR92, LOC116143, MONAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR91 geni (Tüm Gen Dizi Analizi)	WDR91 gene (Sequence Analysis-All Coding Exons)	WDR91	WDR91, SORF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR83 geni (Tüm Gen Dizi Analizi)	WDR83 gene (Sequence Analysis-All Coding Exons)	WDR83	WDR83, MORG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR82 geni (Tüm Gen Dizi Analizi)	WDR82 gene (Sequence Analysis-All Coding Exons)	WDR82	WDR82, TMEM113	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR77 geni (Tüm Gen Dizi Analizi)	WDR77 gene (Sequence Analysis-All Coding Exons)	WDR77	WDR77, MEP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR70 geni (Tüm Gen Dizi Analizi)	WDR70 gene (Sequence Analysis-All Coding Exons)	WDR70	WDR70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR7 geni (Tüm Gen Dizi Analizi)	WDR7 gene (Sequence Analysis-All Coding Exons)	WDR7	WDR7, TRAG, KIAA0541	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR6 geni (Tüm Gen Dizi Analizi)	WDR6 gene (Sequence Analysis-All Coding Exons)	WDR6	WDR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR53 geni (Tüm Gen Dizi Analizi)	WDR53 gene (Sequence Analysis-All Coding Exons)	WDR53	WDR53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR48 geni (Tüm Gen Dizi Analizi)	WDR48 gene (Sequence Analysis-All Coding Exons)	WDR48	WDR48, KIAA1449	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR47 geni (Tüm Gen Dizi Analizi)	WDR47 gene (Sequence Analysis-All Coding Exons)	WDR47	WDR47, NEMITIN, KIAA0893	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR46 geni (Tüm Gen Dizi Analizi)	WDR46 gene (Sequence Analysis-All Coding Exons)	WDR46	WDR46, BING4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR45B geni (Tüm Gen Dizi Analizi)	WDR45B gene (Sequence Analysis-All Coding Exons)	WDR45B	WDR45B, WIPI3, WDR45L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR43 geni (Tüm Gen Dizi Analizi)	WDR43 gene (Sequence Analysis-All Coding Exons)	WDR43	WDR43, UTP5, KIAA0007	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR4 geni (Tüm Gen Dizi Analizi)	WDR4 gene (Sequence Analysis-All Coding Exons)	WDR4	WDR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR3 geni (Tüm Gen Dizi Analizi)	WDR3 gene (Sequence Analysis-All Coding Exons)	WDR3	WDR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR17 geni (Tüm Gen Dizi Analizi)	WDR17 gene (Sequence Analysis-All Coding Exons)	WDR17	WDR17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR13 geni (Tüm Gen Dizi Analizi)	WDR13 gene (Sequence Analysis-All Coding Exons)	WDR13	WDR13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR12 geni (Tüm Gen Dizi Analizi)	WDR12 gene (Sequence Analysis-All Coding Exons)	WDR12	WDR12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR1 geni (Tüm Gen Dizi Analizi)	WDR1 gene (Sequence Analysis-All Coding Exons)	WDR1	WDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDHD1 geni (Tüm Gen Dizi Analizi)	WDHD1 gene (Sequence Analysis-All Coding Exons)	WDHD1	WDHD1, AND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDFY2 geni (Tüm Gen Dizi Analizi)	WDFY2 gene (Sequence Analysis-All Coding Exons)	WDFY2	WDFY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDCP geni (Tüm Gen Dizi Analizi)	WDCP gene (Sequence Analysis-All Coding Exons)	WDCP	C2orf44, WDCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBSCR28 geni (Tüm Gen Dizi Analizi)	WBSCR28 gene (Sequence Analysis-All Coding Exons)	WBSCR28	WBSCR28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBSCR27 geni (Tüm Gen Dizi Analizi)	WBSCR27 gene (Sequence Analysis-All Coding Exons)	WBSCR27	WBSCR27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBSCR17 geni (Tüm Gen Dizi Analizi)	WBSCR17 gene (Sequence Analysis-All Coding Exons)	WBSCR17	WBSCR17, GALNTL3, GALNAT17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBP4 geni (Tüm Gen Dizi Analizi)	WBP4 gene (Sequence Analysis-All Coding Exons)	WBP4	WBP4, FBP21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBP2NL geni (Tüm Gen Dizi Analizi)	WBP2NL gene (Sequence Analysis-All Coding Exons)	WBP2NL	WBP2NL, PAWP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBP2 geni (Tüm Gen Dizi Analizi)	WBP2 gene (Sequence Analysis-All Coding Exons)	WBP2	WBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBP1L geni (Tüm Gen Dizi Analizi)	WBP1L gene (Sequence Analysis-All Coding Exons)	WBP1L	C10orf26, OPAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WBP1 geni (Tüm Gen Dizi Analizi)	WBP1 gene (Sequence Analysis-All Coding Exons)	WBP1	WBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASL geni (Tüm Gen Dizi Analizi)	WASL gene (Sequence Analysis-All Coding Exons)	WASL	WASL, NWASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASHC2C geni (Tüm Gen Dizi Analizi)	WASHC2C gene (Sequence Analysis-All Coding Exons)	WASHC2C	FAM21C, VPEF, KIAA0592	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASHC1 geni (Tüm Gen Dizi Analizi)	WASHC1 gene (Sequence Analysis-All Coding Exons)	WASHC1	WASH1, WASH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASF3 geni (Tüm Gen Dizi Analizi)	WASF3 gene (Sequence Analysis-All Coding Exons)	WASF3	WASF3, WAVE3, SCAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
YKT6 geni (Tüm Gen Dizi Analizi)	YKT6 gene (Sequence Analysis-All Coding Exons)	YKT6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASF2 geni (Tüm Gen Dizi Analizi)	WASF2 gene (Sequence Analysis-All Coding Exons)	WASF2	WASF2, WAVE2, SCAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WASF1 geni (Tüm Gen Dizi Analizi)	WASF1 gene (Sequence Analysis-All Coding Exons)	WASF1	WASF1, WAVE, WAVE1, SCAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WARS2 geni (Tüm Gen Dizi Analizi)	WARS2 gene (Sequence Analysis-All Coding Exons)	WARS2	WARS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WARS geni (Tüm Gen Dizi Analizi)	WARS gene (Sequence Analysis-All Coding Exons)	WARS	WARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WAPL geni (Tüm Gen Dizi Analizi)	WAPL gene (Sequence Analysis-All Coding Exons)	WAPL	WAPAL, KIAA0261, FOE, WAPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VWCE geni (Tüm Gen Dizi Analizi)	VWCE gene (Sequence Analysis-All Coding Exons)	VWCE	VWC1, URG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VWC2 geni (Tüm Gen Dizi Analizi)	VWC2 gene (Sequence Analysis-All Coding Exons)	VWC2	VWC2, BRORIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VWA7 geni (Tüm Gen Dizi Analizi)	VWA7 gene (Sequence Analysis-All Coding Exons)	VWA7	VWA7, C6orf27, G7C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VWA5A geni (Tüm Gen Dizi Analizi)	VWA5A gene (Sequence Analysis-All Coding Exons)	VWA5A	VWA5A, LOH11CR2A, BCSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VWA1 geni (Tüm Gen Dizi Analizi)	VWA1 gene (Sequence Analysis-All Coding Exons)	VWA1	VWA1, WARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTRNA1-3 geni (Tüm Gen Dizi Analizi)	VTRNA1-3 gene (Sequence Analysis-All Coding Exons)	VTRNA1-3	VTRNA1-3, HVG3, VAULTRC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTRNA1-2 geni (Tüm Gen Dizi Analizi)	VTRNA1-2 gene (Sequence Analysis-All Coding Exons)	VTRNA1-2	VTRNA1-2, HVG2, VAULTRC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTRNA1-1 geni (Tüm Gen Dizi Analizi)	VTRNA1-1 gene (Sequence Analysis-All Coding Exons)	VTRNA1-1	VTRNA1-1, HVG1, VAULTRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTN geni (Tüm Gen Dizi Analizi)	VTN gene (Sequence Analysis-All Coding Exons)	VTN	VTN, VNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTI1B geni (Tüm Gen Dizi Analizi)	VTI1B gene (Sequence Analysis-All Coding Exons)	VTI1B	VTI1B, VTI1, VTI1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTI1A geni (Tüm Gen Dizi Analizi)	VTI1A gene (Sequence Analysis-All Coding Exons)	VTI1A	VTI1A, VTI1RP2, MMDS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTCN1 geni (Tüm Gen Dizi Analizi)	VTCN1 gene (Sequence Analysis-All Coding Exons)	VTCN1	VCTN1, B7H4, B7X, B7S1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VTA1 geni (Tüm Gen Dizi Analizi)	VTA1 gene (Sequence Analysis-All Coding Exons)	VTA1	C6orf55, SBP1, LIP5, VTA1, DRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSTM2L geni (Tüm Gen Dizi Analizi)	VSTM2L gene (Sequence Analysis-All Coding Exons)	VSTM2L	VSTM2L, C20orf102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSTM1 geni (Tüm Gen Dizi Analizi)	VSTM1 gene (Sequence Analysis-All Coding Exons)	VSTM1	VSTM1, SIRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSNL1 geni (Tüm Gen Dizi Analizi)	VSNL1 gene (Sequence Analysis-All Coding Exons)	VSNL1	VSNL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSIR geni (Tüm Gen Dizi Analizi)	VSIR gene (Sequence Analysis-All Coding Exons)	VSIR	C10orf54, B7H5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSIG4 geni (Tüm Gen Dizi Analizi)	VSIG4 gene (Sequence Analysis-All Coding Exons)	VSIG4	Z39IG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSIG1 geni (Tüm Gen Dizi Analizi)	VSIG1 gene (Sequence Analysis-All Coding Exons)	VSIG1	VSIG1, GPA34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VRK2 geni (Tüm Gen Dizi Analizi)	VRK2 gene (Sequence Analysis-All Coding Exons)	VRK2	VRK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS72 geni (Tüm Gen Dizi Analizi)	VPS72 gene (Sequence Analysis-All Coding Exons)	VPS72	VPS72, CFL1, YL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS54 geni (Tüm Gen Dizi Analizi)	VPS54 gene (Sequence Analysis-All Coding Exons)	VPS54	VPS54, VPS54L, WR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS52 geni (Tüm Gen Dizi Analizi)	VPS52 gene (Sequence Analysis-All Coding Exons)	VPS52	VPS52, SACM2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS51 geni (Tüm Gen Dizi Analizi)	VPS51 gene (Sequence Analysis-All Coding Exons)	VPS51	VPS51, ANG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS50 geni (Tüm Gen Dizi Analizi)	VPS50 gene (Sequence Analysis-All Coding Exons)	VPS50	VPS50, CCDC132, KIAA1861	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS4B geni (Tüm Gen Dizi Analizi)	VPS4B gene (Sequence Analysis-All Coding Exons)	VPS4B	VPS4B, SKD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPOT geni (Tüm Gen Dizi Analizi)	XPOT gene (Sequence Analysis-All Coding Exons)	XPOT		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS4A geni (Tüm Gen Dizi Analizi)	VPS4A gene (Sequence Analysis-All Coding Exons)	VPS4A	VPS4A, VPS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS41 geni (Tüm Gen Dizi Analizi)	VPS41 gene (Sequence Analysis-All Coding Exons)	VPS41	VPS41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS39 geni (Tüm Gen Dizi Analizi)	VPS39 gene (Sequence Analysis-All Coding Exons)	VPS39	VPS39, VAM6, TLP, KIAA0720	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
XPO4 geni (Tüm Gen Dizi Analizi)	XPO4 gene (Sequence Analysis-All Coding Exons)	XPO4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS37D geni (Tüm Gen Dizi Analizi)	VPS37D gene (Sequence Analysis-All Coding Exons)	VPS37D	VPS37D, WBSCR24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS37C geni (Tüm Gen Dizi Analizi)	VPS37C gene (Sequence Analysis-All Coding Exons)	VPS37C	VPS37C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS37B geni (Tüm Gen Dizi Analizi)	VPS37B gene (Sequence Analysis-All Coding Exons)	VPS37B	VPS37B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS36 geni (Tüm Gen Dizi Analizi)	VPS36 gene (Sequence Analysis-All Coding Exons)	VPS36	VPS36, EAP45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS28 geni (Tüm Gen Dizi Analizi)	VPS28 gene (Sequence Analysis-All Coding Exons)	VPS28	VPS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS26B geni (Tüm Gen Dizi Analizi)	VPS26B gene (Sequence Analysis-All Coding Exons)	VPS26B	VPS26B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS26A geni (Tüm Gen Dizi Analizi)	VPS26A gene (Sequence Analysis-All Coding Exons)	VPS26A	VPS26A, VPS26, HBS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS25 geni (Tüm Gen Dizi Analizi)	VPS25 gene (Sequence Analysis-All Coding Exons)	VPS25	VPS25, FAP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS18 geni (Tüm Gen Dizi Analizi)	VPS18 gene (Sequence Analysis-All Coding Exons)	VPS18	VPS18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS16 geni (Tüm Gen Dizi Analizi)	VPS16 gene (Sequence Analysis-All Coding Exons)	VPS16	VPS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPREB3 geni (Tüm Gen Dizi Analizi)	VPREB3 gene (Sequence Analysis-All Coding Exons)	VPREB3	VPREB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPREB1 geni (Tüm Gen Dizi Analizi)	VPREB1 gene (Sequence Analysis-All Coding Exons)	VPREB1	VPREB1, IGI, VPREB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VOPP1 geni (Tüm Gen Dizi Analizi)	VOPP1 gene (Sequence Analysis-All Coding Exons)	VOPP1	VOPP1, ECOP, GASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VNN3 geni (Tüm Gen Dizi Analizi)	VNN3 gene (Sequence Analysis-All Coding Exons)	VNN3	VNN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VNN2 geni (Tüm Gen Dizi Analizi)	VNN2 gene (Sequence Analysis-All Coding Exons)	VNN2	VNN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VN1R1 geni (Tüm Gen Dizi Analizi)	VN1R1 gene (Sequence Analysis-All Coding Exons)	VN1R1	VN1R1, V1RL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VMP1 geni (Tüm Gen Dizi Analizi)	VMP1 gene (Sequence Analysis-All Coding Exons)	VMP1	VMP1, TMEM49, EPG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VMAC geni (Tüm Gen Dizi Analizi)	VMAC gene (Sequence Analysis-All Coding Exons)	VMAC	VMAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VKORC1L1 geni (Tüm Gen Dizi Analizi)	VKORC1L1 gene (Sequence Analysis-All Coding Exons)	VKORC1L1	VKORC1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

VIPR2 geni (Tüm Gen Dizi Analizi)	VIPR2 gene (Sequence Analysis-All Coding Exons)	VIPR2	VIPR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VIPR1 geni (Tüm Gen Dizi Analizi)	VIPR1 gene (Sequence Analysis-All Coding Exons)	VIPR1	VIPR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VIP geni (Tüm Gen Dizi Analizi)	VIP gene (Sequence Analysis-All Coding Exons)	VIP	VIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VGLL3 geni (Tüm Gen Dizi Analizi)	VGLL3 gene (Sequence Analysis-All Coding Exons)	VGLL3	VGLL3, VGL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VGLL2 geni (Tüm Gen Dizi Analizi)	VGLL2 gene (Sequence Analysis-All Coding Exons)	VGLL2	VGLL2, VGL2, VITO1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VGLL1 geni (Tüm Gen Dizi Analizi)	VGLL1 gene (Sequence Analysis-All Coding Exons)	VGLL1	VGLL1, VGL1, TDU	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VGF geni (Tüm Gen Dizi Analizi)	VGF gene (Sequence Analysis-All Coding Exons)	VGF	VGF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VEZF1 geni (Tüm Gen Dizi Analizi)	VEZF1 gene (Sequence Analysis-All Coding Exons)	VEZF1	VEZF1, ZNF161	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VEPH1 geni (Tüm Gen Dizi Analizi)	VEPH1 gene (Sequence Analysis-All Coding Exons)	VEPH1	VEPH1, MELT, KIAA1692	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VENTX geni (Tüm Gen Dizi Analizi)	VENTX gene (Sequence Analysis-All Coding Exons)	VENTX	VENTX, VENTX2, HPX42B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VEGFD geni (Tüm Gen Dizi Analizi)	VEGFD gene (Sequence Analysis-All Coding Exons)	VEGFD	FIGF, VEGFD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VEGFB geni (Tüm Gen Dizi Analizi)	VEGFB gene (Sequence Analysis-All Coding Exons)	VEGFB	VEGFB, VRF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VDAC3 geni (Tüm Gen Dizi Analizi)	VDAC3 gene (Sequence Analysis-All Coding Exons)	VDAC3	VDAC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VDAC2 geni (Tüm Gen Dizi Analizi)	VDAC2 gene (Sequence Analysis-All Coding Exons)	VDAC2	VDAC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VDAC1 geni (Tüm Gen Dizi Analizi)	VDAC1 gene (Sequence Analysis-All Coding Exons)	VDAC1	VDAC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCY geni (Tüm Gen Dizi Analizi)	VCY gene (Sequence Analysis-All Coding Exons)	VCY	VCY, BPY1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCX3B geni (Tüm Gen Dizi Analizi)	VCX3B gene (Sequence Analysis-All Coding Exons)	VCX3B	VCS3B, VCXC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCX3A geni (Tüm Gen Dizi Analizi)	VCX3A gene (Sequence Analysis-All Coding Exons)	VCX3A	VCX3A, VCX8R, VCXA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCX2 geni (Tüm Gen Dizi Analizi)	VCX2 gene (Sequence Analysis-All Coding Exons)	VCX2	VCX2, VCX2R, VCXB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCX geni (Tüm Gen Dizi Analizi)	VCX gene (Sequence Analysis-All Coding Exons)	VCX	VCX, VCXB1, VCX10R	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCPKMT geni (Tüm Gen Dizi Analizi)	VCPKMT gene (Sequence Analysis-All Coding Exons)	VCPKMT	METTL21D, C14orf138	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCPIP1 geni (Tüm Gen Dizi Analizi)	VCPIP1 gene (Sequence Analysis-All Coding Exons)	VCPIP1	VCPIP1 VCIP135, KIAA1850	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VCAM1 geni (Tüm Gen Dizi Analizi)	VCAM1 gene (Sequence Analysis-All Coding Exons)	VCAM1	VCAM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VBP1 geni (Tüm Gen Dizi Analizi)	VBP1 gene (Sequence Analysis-All Coding Exons)	VBP1	VBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAX2 geni (Tüm Gen Dizi Analizi)	VAX2 gene (Sequence Analysis-All Coding Exons)	VAX2	VAX2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAV3 geni (Tüm Gen Dizi Analizi)	VAV3 gene (Sequence Analysis-All Coding Exons)	VAV3	VAV3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAV2 geni (Tüm Gen Dizi Analizi)	VAV2 gene (Sequence Analysis-All Coding Exons)	VAV2	VAV2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAV1 geni (Tüm Gen Dizi Analizi)	VAV1 gene (Sequence Analysis-All Coding Exons)	VAV1	VAV1, VAV	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAT1 geni (Tüm Gen Dizi Analizi)	VAT1 gene (Sequence Analysis-All Coding Exons)	VAT1	VAT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VASP geni (Tüm Gen Dizi Analizi)	VASP gene (Sequence Analysis-All Coding Exons)	VASP	VASP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VASN geni (Tüm Gen Dizi Analizi)	VASN gene (Sequence Analysis-All Coding Exons)	VASN	VASN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VASH2 geni (Tüm Gen Dizi Analizi)	VASH2 gene (Sequence Analysis-All Coding Exons)	VASH2	VASH2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VARS geni (Tüm Gen Dizi Analizi)	VARS gene (Sequence Analysis-All Coding Exons)	VARS	VARS, VARS1, G7A, VARS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAPA geni (Tüm Gen Dizi Analizi)	VAPA gene (Sequence Analysis-All Coding Exons)	VAPA	VAPA, VAP33	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP8 geni (Tüm Gen Dizi Analizi)	VAMP8 gene (Sequence Analysis-All Coding Exons)	VAMP8	VAMP8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP7 geni (Tüm Gen Dizi Analizi)	VAMP7 gene (Sequence Analysis-All Coding Exons)	VAMP7	VAMP7, SYBL1, TIVAMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP5 geni (Tüm Gen Dizi Analizi)	VAMP5 gene (Sequence Analysis-All Coding Exons)	VAMP5	VAMP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP4 geni (Tüm Gen Dizi Analizi)	VAMP4 gene (Sequence Analysis-All Coding Exons)	VAMP4	VAMP24	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP3 geni (Tüm Gen Dizi Analizi)	VAMP3 gene (Sequence Analysis-All Coding Exons)	VAMP3	VAMP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAMP2 geni (Tüm Gen Dizi Analizi)	VAMP2 gene (Sequence Analysis-All Coding Exons)	VAMP2	VAMP2, SYB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VAC14 geni (Tüm Gen Dizi Analizi)	VAC14 gene (Sequence Analysis-All Coding Exons)	VAC14	VAC14, TAX1BP2, TRX, SNDC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UXT geni (Tüm Gen Dizi Analizi)	UXT gene (Sequence Analysis-All Coding Exons)	UXT	UXT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UXS1 geni (Tüm Gen Dizi Analizi)	UXS1 gene (Sequence Analysis-All Coding Exons)	UXS1	UXS1, UGD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UVRAG geni (Tüm Gen Dizi Analizi)	UVRAG gene (Sequence Analysis-All Coding Exons)	UVRAG	UVRAG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTY geni (Tüm Gen Dizi Analizi)	UTY gene (Sequence Analysis-All Coding Exons)	UTY	UTY	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTS2R geni (Tüm Gen Dizi Analizi)	UTS2R gene (Sequence Analysis-All Coding Exons)	UTS2R	GPR14, UTR2, UTR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTS2 geni (Tüm Gen Dizi Analizi)	UTS2 gene (Sequence Analysis-All Coding Exons)	UTS2	UTS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTRN geni (Tüm Gen Dizi Analizi)	UTRN gene (Sequence Analysis-All Coding Exons)	UTRN	UTRN, DMDL, DRP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP4 geni (Tüm Gen Dizi Analizi)	UTP4 gene (Sequence Analysis-All Coding Exons)	UTP4	UTP4, CIRH1A, TEX292, KIAA1008	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP3 geni (Tüm Gen Dizi Analizi)	UTP3 gene (Sequence Analysis-All Coding Exons)	UTP3	UTP3, CRL1, CRLZ1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP20 geni (Tüm Gen Dizi Analizi)	UTP20 gene (Sequence Analysis-All Coding Exons)	UTP20	UTP20, DRIM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP18 geni (Tüm Gen Dizi Analizi)	UTP18 gene (Sequence Analysis-All Coding Exons)	UTP18	UTP18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP15 geni (Tüm Gen Dizi Analizi)	UTP15 gene (Sequence Analysis-All Coding Exons)	UTP15	UTP15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP14A geni (Tüm Gen Dizi Analizi)	UTP14A gene (Sequence Analysis-All Coding Exons)	UTP14A	UTP14A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP11 geni (Tüm Gen Dizi Analizi)	UTP11 gene (Sequence Analysis-All Coding Exons)	UTP11	UTP11L, CGI94	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTF1 geni (Tüm Gen Dizi Analizi)	UTF1 gene (Sequence Analysis-All Coding Exons)	UTF1	UTF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UST geni (Tüm Gen Dizi Analizi)	UST gene (Sequence Analysis-All Coding Exons)	UST	UST	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP8 geni (Tüm Gen Dizi Analizi)	USP8 gene (Sequence Analysis-All Coding Exons)	USP8	USP8, HUMORF8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP8 geni (Tüm Gen Dizi Analizi)	USP8 gene (Sequence Analysis-All Coding Exons)	USP8	USP8, HUMORF8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP7 geni (Tüm Gen Dizi Analizi)	USP7 gene (Sequence Analysis-All Coding Exons)	USP7	USP7, HAUSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP6NL geni (Tüm Gen Dizi Analizi)	USP6NL gene (Sequence Analysis-All Coding Exons)	USP6NL	USP6NL, RNTRE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP6 geni (Tüm Gen Dizi Analizi)	USP6 gene (Sequence Analysis-All Coding Exons)	USP6	USP6, TRE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP5 geni (Tüm Gen Dizi Analizi)	USP5 gene (Sequence Analysis-All Coding Exons)	USP5	USP5, ISOT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP47 geni (Tüm Gen Dizi Analizi)	USP47 gene (Sequence Analysis-All Coding Exons)	USP47	USP47	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP46 geni (Tüm Gen Dizi Analizi)	USP46 gene (Sequence Analysis-All Coding Exons)	USP46	USP46	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP44 geni (Tüm Gen Dizi Analizi)	USP44 gene (Sequence Analysis-All Coding Exons)	USP44	USP44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP40 geni (Tüm Gen Dizi Analizi)	USP40 gene (Sequence Analysis-All Coding Exons)	USP40	USP40	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP4 geni (Tüm Gen Dizi Analizi)	USP4 gene (Sequence Analysis-All Coding Exons)	USP4	USP4, UNP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WIPI1 geni (Tüm Gen Dizi Analizi)	WIPI1 gene (Sequence Analysis-All Coding Exons)	WIPI1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP39 geni (Tüm Gen Dizi Analizi)	USP39 gene (Sequence Analysis-All Coding Exons)	USP39	USP39, SAD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP36 geni (Tüm Gen Dizi Analizi)	USP36 gene (Sequence Analysis-All Coding Exons)	USP36	USP36, KIAA1453	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP34 geni (Tüm Gen Dizi Analizi)	USP34 gene (Sequence Analysis-All Coding Exons)	USP34	USP34, KIAA0570	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP33 geni (Tüm Gen Dizi Analizi)	USP33 gene (Sequence Analysis-All Coding Exons)	USP33	USP33, VDU1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP32 geni (Tüm Gen Dizi Analizi)	USP32 gene (Sequence Analysis-All Coding Exons)	USP32	USP32, USP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP30 geni (Tüm Gen Dizi Analizi)	USP30 gene (Sequence Analysis-All Coding Exons)	USP30	USP30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP3 geni (Tüm Gen Dizi Analizi)	USP3 gene (Sequence Analysis-All Coding Exons)	USP3	USP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP28 geni (Tüm Gen Dizi Analizi)	USP28 gene (Sequence Analysis-All Coding Exons)	USP28	USP28, KIAA1515	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP26 geni (Tüm Gen Dizi Analizi)	USP26 gene (Sequence Analysis-All Coding Exons)	USP26	USP26	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP24 geni (Tüm Gen Dizi Analizi)	USP24 gene (Sequence Analysis-All Coding Exons)	USP24	USP24, KIAA1057	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP22 geni (Tüm Gen Dizi Analizi)	USP22 gene (Sequence Analysis-All Coding Exons)	USP22	USP22, KIAA1064	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP21 geni (Tüm Gen Dizi Analizi)	USP21 gene (Sequence Analysis-All Coding Exons)	USP21	USP21, USP23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP20 geni (Tüm Gen Dizi Analizi)	USP20 gene (Sequence Analysis-All Coding Exons)	USP20	USP20, VDU2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP19 geni (Tüm Gen Dizi Analizi)	USP19 gene (Sequence Analysis-All Coding Exons)	USP19	USP19, KIAA0891	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP17L9P geni (Tüm Gen Dizi Analizi)	USP17L9P gene (Sequence Analysis-All Coding Exons)	USP17L9P	USP17L9P, USP17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP17L2 geni (Tüm Gen Dizi Analizi)	USP17L2 gene (Sequence Analysis-All Coding Exons)	USP17L2	USP17L2, DUB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP16 geni (Tüm Gen Dizi Analizi)	USP16 gene (Sequence Analysis-All Coding Exons)	USP16	USP16, UBPM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP15 geni (Tüm Gen Dizi Analizi)	USP15 gene (Sequence Analysis-All Coding Exons)	USP15	USP15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP14 geni (Tüm Gen Dizi Analizi)	USP14 gene (Sequence Analysis-All Coding Exons)	USP14	USP14, TGT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP13 geni (Tüm Gen Dizi Analizi)	USP13 gene (Sequence Analysis-All Coding Exons)	USP13	USP13, ISOT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP11 geni (Tüm Gen Dizi Analizi)	USP11 gene (Sequence Analysis-All Coding Exons)	USP11	USP11, UHX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP10 geni (Tüm Gen Dizi Analizi)	USP10 gene (Sequence Analysis-All Coding Exons)	USP10	USP10, KIAA0190	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP1 geni (Tüm Gen Dizi Analizi)	USP1 gene (Sequence Analysis-All Coding Exons)	USP1	USP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USO1 geni (Tüm Gen Dizi Analizi)	USO1 gene (Sequence Analysis-All Coding Exons)	USO1	USO1, TAP, p115	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

USMG5 geni (Tüm Gen Dizi Analizi)	USMG5 gene (Sequence Analysis-All Coding Exons)	USMG5	USMG5, DAPIT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USHBP1 geni (Tüm Gen Dizi Analizi)	USHBP1 gene (Sequence Analysis-All Coding Exons)	USHBP1	USHBP1, MCC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USF2 geni (Tüm Gen Dizi Analizi)	USF2 gene (Sequence Analysis-All Coding Exons)	USF2	USF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URM1 geni (Tüm Gen Dizi Analizi)	URM1 gene (Sequence Analysis-All Coding Exons)	URM1	URM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URI1 geni (Tüm Gen Dizi Analizi)	URI1 gene (Sequence Analysis-All Coding Exons)	URI1	NNX3, RMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR61 geni (Tüm Gen Dizi Analizi)	WDR61 gene (Sequence Analysis-All Coding Exons)	WDR61		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URGCP geni (Tüm Gen Dizi Analizi)	URGCP gene (Sequence Analysis-All Coding Exons)	URGCP	URGCP, URG4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URB1 geni (Tüm Gen Dizi Analizi)	URB1 gene (Sequence Analysis-All Coding Exons)	URB1	URB1, NPA1, C21orf108, KIAA0539	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URAHP geni (Tüm Gen Dizi Analizi)	URAHP gene (Sequence Analysis-All Coding Exons)	URAHP	URAHP, URAH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDR5 geni (Tüm Gen Dizi Analizi)	WDR5 gene (Sequence Analysis-All Coding Exons)	WDR5		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
URAD geni (Tüm Gen Dizi Analizi)	URAD gene (Sequence Analysis-All Coding Exons)	URAD	URAD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCRH geni (Tüm Gen Dizi Analizi)	UQCRH gene (Sequence Analysis-All Coding Exons)	UQCRH	UQCRH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCRFS1 geni (Tüm Gen Dizi Analizi)	UQCRFS1 gene (Sequence Analysis-All Coding Exons)	UQCRFS1	UQCRFS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCRC1 geni (Tüm Gen Dizi Analizi)	UQCRC1 gene (Sequence Analysis-All Coding Exons)	UQCRC1	UQCRC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCR11 geni (Tüm Gen Dizi Analizi)	UQCR11 gene (Sequence Analysis-All Coding Exons)	UQCR11	UQCR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCR10 geni (Tüm Gen Dizi Analizi)	UQCR10 gene (Sequence Analysis-All Coding Exons)	UQCR10	UCRC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UQCC1 geni (Tüm Gen Dizi Analizi)	UQCC1 gene (Sequence Analysis-All Coding Exons)	UQCC1	UQCC, CBP3, BFZB, C20orf44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPRT geni (Tüm Gen Dizi Analizi)	UPRT gene (Sequence Analysis-All Coding Exons)	UPRT	UPRT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPP2 geni (Tüm Gen Dizi Analizi)	UPP2 gene (Sequence Analysis-All Coding Exons)	UPP2	UPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPP1 geni (Tüm Gen Dizi Analizi)	UPP1 gene (Sequence Analysis-All Coding Exons)	UPP1	UPP1, UP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPK3B geni (Tüm Gen Dizi Analizi)	UPK3B gene (Sequence Analysis-All Coding Exons)	UPK3B	UPK3B, UPIIIB, P3S	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPK3A geni (Tüm Gen Dizi Analizi)	UPK3A gene (Sequence Analysis-All Coding Exons)	UPK3A	UPK3A, UPK3, UPIII	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPK2 geni (Tüm Gen Dizi Analizi)	UPK2 gene (Sequence Analysis-All Coding Exons)	UPK2	UPK2, UP2, UPII	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPK1B geni (Tüm Gen Dizi Analizi)	UPK1B gene (Sequence Analysis-All Coding Exons)	UPK1B	UPK1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPK1A geni (Tüm Gen Dizi Analizi)	UPK1A gene (Sequence Analysis-All Coding Exons)	UPK1A	UPK1A, UPIA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPF3A geni (Tüm Gen Dizi Analizi)	UPF3A gene (Sequence Analysis-All Coding Exons)	UPF3A	UPF3A, RENT3A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPF2 geni (Tüm Gen Dizi Analizi)	UPF2 gene (Sequence Analysis-All Coding Exons)	UPF2	UPF2, RENT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UPF1 geni (Tüm Gen Dizi Analizi)	UPF1 gene (Sequence Analysis-All Coding Exons)	UPF1	UPF1, RENT1, HUPF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNK geni (Tüm Gen Dizi Analizi)	UNK gene (Sequence Analysis-All Coding Exons)	UNK	UNK, UNKEMPT, ZC3H5, KIAA1753	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC93A geni (Tüm Gen Dizi Analizi)	UNC93A gene (Sequence Analysis-All Coding Exons)	UNC93A	UNC93A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC79 geni (Tüm Gen Dizi Analizi)	UNC79 gene (Sequence Analysis-All Coding Exons)	UNC79	UNC79	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC5D geni (Tüm Gen Dizi Analizi)	UNC5D gene (Sequence Analysis-All Coding Exons)	UNC5D	UNC5D, UNC5H4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC5C geni (Tüm Gen Dizi Analizi)	UNC5C gene (Sequence Analysis-All Coding Exons)	UNC5C	UNC5C, UNC5H3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC5B geni (Tüm Gen Dizi Analizi)	UNC5B gene (Sequence Analysis-All Coding Exons)	UNC5B	UNC5B, UNC5H2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
WDFY4 geni (Tüm Gen Dizi Analizi)	WDFY4 gene (Sequence Analysis-All Coding Exons)	WDFY4	WDFY4, KIAA1607	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC5A geni (Tüm Gen Dizi Analizi)	UNC5A gene (Sequence Analysis-All Coding Exons)	UNC5A	UNC5A, UNC5H1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC45A geni (Tüm Gen Dizi Analizi)	UNC45A gene (Sequence Analysis-All Coding Exons)	UNC45A	UNC45A, SMAP1, GCUNC45	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC13C geni (Tüm Gen Dizi Analizi)	UNC13C gene (Sequence Analysis-All Coding Exons)	UNC13C	UNC13C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC13B geni (Tüm Gen Dizi Analizi)	UNC13B gene (Sequence Analysis-All Coding Exons)	UNC13B	UNC13, MUNC13	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UNC13A geni (Tüm Gen Dizi Analizi)	UNC13A gene (Sequence Analysis-All Coding Exons)	UNC13A	UNC13A, KIAA1032	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UMODL1 geni (Tüm Gen Dizi Analizi)	UMODL1 gene (Sequence Analysis-All Coding Exons)	UMODL1	UMODL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULK4 geni (Tüm Gen Dizi Analizi)	ULK4 gene (Sequence Analysis-All Coding Exons)	ULK4	ULK4, FAM7C1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULK3 geni (Tüm Gen Dizi Analizi)	ULK3 gene (Sequence Analysis-All Coding Exons)	ULK3	ULK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULK2 geni (Tüm Gen Dizi Analizi)	ULK2 gene (Sequence Analysis-All Coding Exons)	ULK2	ULK2, KIAA0623, UNC51.2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULK1 geni (Tüm Gen Dizi Analizi)	ULK1 gene (Sequence Analysis-All Coding Exons)	ULK1	ULK1, UNC51	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULBP3 geni (Tüm Gen Dizi Analizi)	ULBP3 gene (Sequence Analysis-All Coding Exons)	ULBP3	ULBP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULBP2 geni (Tüm Gen Dizi Analizi)	ULBP2 gene (Sequence Analysis-All Coding Exons)	ULBP2	ULBP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ULBP1 geni (Tüm Gen Dizi Analizi)	ULBP1 gene (Sequence Analysis-All Coding Exons)	ULBP1	ULBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UIMC1 geni (Tüm Gen Dizi Analizi)	UIMC1 gene (Sequence Analysis-All Coding Exons)	UIMC1	UIMC1, RAP80	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UHRF2 geni (Tüm Gen Dizi Analizi)	UHRF2 gene (Sequence Analysis-All Coding Exons)	UHRF2	UHRF2, NIRF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UHRF1 geni (Tüm Gen Dizi Analizi)	UHRF1 gene (Sequence Analysis-All Coding Exons)	UHRF1	UHRF1, ICBP90, NP95	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UHMK1 geni (Tüm Gen Dizi Analizi)	UHMK1 gene (Sequence Analysis-All Coding Exons)	UHMK1	KIS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT8 geni (Tüm Gen Dizi Analizi)	UGT8 gene (Sequence Analysis-All Coding Exons)	UGT8	UGT8, CGT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT3A2 geni (Tüm Gen Dizi Analizi)	UGT3A2 gene (Sequence Analysis-All Coding Exons)	UGT3A2	UGT3A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT3A1 geni (Tüm Gen Dizi Analizi)	UGT3A1 gene (Sequence Analysis-All Coding Exons)	UGT3A1	UGT3A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B7 geni (Tüm Gen Dizi Analizi)	UGT2B7 gene (Sequence Analysis-All Coding Exons)	UGT2B7	UGT2B7, UGT2B9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B4 geni (Tüm Gen Dizi Analizi)	UGT2B4 gene (Sequence Analysis-All Coding Exons)	UGT2B4	UGT2B4, UGT2B11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B28 geni (Tüm Gen Dizi Analizi)	UGT2B28 gene (Sequence Analysis-All Coding Exons)	UGT2B28	UGT2B28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B15 geni (Tüm Gen Dizi Analizi)	UGT2B15 gene (Sequence Analysis-All Coding Exons)	UGT2B15	UGT2B15, UGT2B8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B11 geni (Tüm Gen Dizi Analizi)	UGT2B11 gene (Sequence Analysis-All Coding Exons)	UGT2B11	UGT2B11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2B10 geni (Tüm Gen Dizi Analizi)	UGT2B10 gene (Sequence Analysis-All Coding Exons)	UGT2B10	UGT2B10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2A3 geni (Tüm Gen Dizi Analizi)	UGT2A3 gene (Sequence Analysis-All Coding Exons)	UGT2A3	UGT2A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT2A1 geni (Tüm Gen Dizi Analizi)	UGT2A1 gene (Sequence Analysis-All Coding Exons)	UGT2A1	UGT2A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A9 geni (Tüm Gen Dizi Analizi)	UGT1A9 gene (Sequence Analysis-All Coding Exons)	UGT1A9	UGT1A9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A8 geni (Tüm Gen Dizi Analizi)	UGT1A8 gene (Sequence Analysis-All Coding Exons)	UGT1A8	UGT1A8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A7 geni (Tüm Gen Dizi Analizi)	UGT1A7 gene (Sequence Analysis-All Coding Exons)	UGT1A7	UGT1A7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A6 geni (Tüm Gen Dizi Analizi)	UGT1A6 gene (Sequence Analysis-All Coding Exons)	UGT1A6	UGT1A6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A5 geni (Tüm Gen Dizi Analizi)	UGT1A5 gene (Sequence Analysis-All Coding Exons)	UGT1A5	UGT1A5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A4 geni (Tüm Gen Dizi Analizi)	UGT1A4 gene (Sequence Analysis-All Coding Exons)	UGT1A4	UGT1A4, UGT1D	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A3 geni (Tüm Gen Dizi Analizi)	UGT1A3 gene (Sequence Analysis-All Coding Exons)	UGT1A3	UGT1A3, UGT1C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGT1A10 geni (Tüm Gen Dizi Analizi)	UGT1A10 gene (Sequence Analysis-All Coding Exons)	UGT1A10	UGT1A10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGP2 geni (Tüm Gen Dizi Analizi)	UGP2 gene (Sequence Analysis-All Coding Exons)	UGP2	UGP2, UGPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGGT2 geni (Tüm Gen Dizi Analizi)	UGGT2 gene (Sequence Analysis-All Coding Exons)	UGGT2	UGGT2, HUGT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGGT1 geni (Tüm Gen Dizi Analizi)	UGGT1 gene (Sequence Analysis-All Coding Exons)	UGGT1	UGGT1, HUGT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGDH geni (Tüm Gen Dizi Analizi)	UGDH gene (Sequence Analysis-All Coding Exons)	UGDH	UGDH, UDPGDH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UGCG geni (Tüm Gen Dizi Analizi)	UGCG gene (Sequence Analysis-All Coding Exons)	UGCG	UGCG, GCS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UFSP1 geni (Tüm Gen Dizi Analizi)	UFSP1 gene (Sequence Analysis-All Coding Exons)	UFSP1	UFSP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UFM1 geni (Tüm Gen Dizi Analizi)	UFM1 gene (Sequence Analysis-All Coding Exons)	UFM1	UFM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UFL1 geni (Tüm Gen Dizi Analizi)	UFL1 gene (Sequence Analysis-All Coding Exons)	UFL1	UFL1, KIAA0776, NLBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UFD1 geni (Tüm Gen Dizi Analizi)	UFD1 gene (Sequence Analysis-All Coding Exons)	UFD1	UFD1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UFC1 geni (Tüm Gen Dizi Analizi)	UFC1 gene (Sequence Analysis-All Coding Exons)	UFC1	UFC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UEVLD geni (Tüm Gen Dizi Analizi)	UEVLD gene (Sequence Analysis-All Coding Exons)	UEVLD	UEVLD, UEV3, ATTP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCN2 geni (Tüm Gen Dizi Analizi)	UCN2 gene (Sequence Analysis-All Coding Exons)	UCN2	UCN2, SRP, UR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCN geni (Tüm Gen Dizi Analizi)	UCN gene (Sequence Analysis-All Coding Exons)	UCN	UCN, UROC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCKL1 geni (Tüm Gen Dizi Analizi)	UCKL1 gene (Sequence Analysis-All Coding Exons)	UCKL1	UCKL1, UCK1L, URKL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCK2 geni (Tüm Gen Dizi Analizi)	UCK2 gene (Sequence Analysis-All Coding Exons)	UCK2	UCK2, TSA903	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCK1 geni (Tüm Gen Dizi Analizi)	UCK1 gene (Sequence Analysis-All Coding Exons)	UCK1	UCK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VSIG2 geni (Tüm Gen Dizi Analizi)	VSIG2 gene (Sequence Analysis-All Coding Exons)	VSIG2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCHL5 geni (Tüm Gen Dizi Analizi)	UCHL5 gene (Sequence Analysis-All Coding Exons)	UCHL5	UCHL5, UCH37	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCHL3 geni (Tüm Gen Dizi Analizi)	UCHL3 gene (Sequence Analysis-All Coding Exons)	UCHL3	UCHL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN8 geni (Tüm Gen Dizi Analizi)	UBXN8 gene (Sequence Analysis-All Coding Exons)	UBXN8	UBXN8, D8S2298E, REP8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN7 geni (Tüm Gen Dizi Analizi)	UBXN7 gene (Sequence Analysis-All Coding Exons)	UBXN7	UBXN7, KIAA0794	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN6 geni (Tüm Gen Dizi Analizi)	UBXN6 gene (Sequence Analysis-All Coding Exons)	UBXN6	UBXN6, UBXD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

UBXN4 geni (Tüm Gen Dizi Analizi)	UBXN4 gene (Sequence Analysis-All Coding Exons)	UBXN4	UBXN4, UBXD2, KIAA2042, PRAJA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN2B geni (Tüm Gen Dizi Analizi)	UBXN2B gene (Sequence Analysis-All Coding Exons)	UBXN2B	UBXN2B, LOC137886, p37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN11 geni (Tüm Gen Dizi Analizi)	UBXN11 gene (Sequence Analysis-All Coding Exons)	UBXN11	UBXN11, SOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN10 geni (Tüm Gen Dizi Analizi)	UBXN10 gene (Sequence Analysis-All Coding Exons)	UBXN10	UBXN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBXN1 geni (Tüm Gen Dizi Analizi)	UBXN1 gene (Sequence Analysis-All Coding Exons)	UBXN1	UBXN1, SAKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBTFL1 geni (Tüm Gen Dizi Analizi)	UBTFL1 gene (Sequence Analysis-All Coding Exons)	UBTFL1	UBTFL1, HMGPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBTF geni (Tüm Gen Dizi Analizi)	UBTF gene (Sequence Analysis-All Coding Exons)	UBTF	UBTF, UBF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBTD2 geni (Tüm Gen Dizi Analizi)	UBTD2 gene (Sequence Analysis-All Coding Exons)	UBTD2	UBTD2, DCUBP, MGC30022	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBTD1 geni (Tüm Gen Dizi Analizi)	UBTD1 gene (Sequence Analysis-All Coding Exons)	UBTD1	UBTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBR7 geni (Tüm Gen Dizi Analizi)	UBR7 gene (Sequence Analysis-All Coding Exons)	UBR7	UBR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBR5 geni (Tüm Gen Dizi Analizi)	UBR5 gene (Sequence Analysis-All Coding Exons)	UBR5	UBR5, EDD1, HYD, KIAA0896	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBR4 geni (Tüm Gen Dizi Analizi)	UBR4 gene (Sequence Analysis-All Coding Exons)	UBR4	UBR4, ZUBR1, RBAF600, KIAA1307	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBR3 geni (Tüm Gen Dizi Analizi)	UBR3 gene (Sequence Analysis-All Coding Exons)	UBR3	UBR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBR2 geni (Tüm Gen Dizi Analizi)	UBR2 gene (Sequence Analysis-All Coding Exons)	UBR2	UBR2, C6orf133, KIAA0349	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBQLN4 geni (Tüm Gen Dizi Analizi)	UBQLN4 gene (Sequence Analysis-All Coding Exons)	UBQLN4	UBQLN4, A1U, C1orf6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBQLN3 geni (Tüm Gen Dizi Analizi)	UBQLN3 gene (Sequence Analysis-All Coding Exons)	UBQLN3	UBQLN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBQLN1 geni (Tüm Gen Dizi Analizi)	UBQLN1 gene (Sequence Analysis-All Coding Exons)	UBQLN1	UBQLN1, DA41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS29 geni (Tüm Gen Dizi Analizi)	VPS29 gene (Sequence Analysis-All Coding Exons)	VPS29		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBP1 geni (Tüm Gen Dizi Analizi)	UBP1 gene (Sequence Analysis-All Coding Exons)	UBP1	UBP1, LBP1A, LBP1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBN2 geni (Tüm Gen Dizi Analizi)	UBN2 gene (Sequence Analysis-All Coding Exons)	UBN2	UBN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBN1 geni (Tüm Gen Dizi Analizi)	UBN1 gene (Sequence Analysis-All Coding Exons)	UBN1	UBN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBLCP1 geni (Tüm Gen Dizi Analizi)	UBLCP1 gene (Sequence Analysis-All Coding Exons)	UBLCP1	UBLCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBL7 geni (Tüm Gen Dizi Analizi)	UBL7 gene (Sequence Analysis-All Coding Exons)	UBL7	UBL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBL5 geni (Tüm Gen Dizi Analizi)	UBL5 gene (Sequence Analysis-All Coding Exons)	UBL5	UBL5, HUB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VPS13D geni (Tüm Gen Dizi Analizi)	VPS13D gene (Sequence Analysis-All Coding Exons)	VPS13D		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBL4B geni (Tüm Gen Dizi Analizi)	UBL4B gene (Sequence Analysis-All Coding Exons)	UBL4B	UBL4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBL4A geni (Tüm Gen Dizi Analizi)	UBL4A gene (Sequence Analysis-All Coding Exons)	UBL4A	UBL4A, GDX, UBL4, DX254E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBL3 geni (Tüm Gen Dizi Analizi)	UBL3 gene (Sequence Analysis-All Coding Exons)	UBL3	UBL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE4B geni (Tüm Gen Dizi Analizi)	UBE4B gene (Sequence Analysis-All Coding Exons)	UBE4B	UBE4B, UFD2A, KIAA0684	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE4A geni (Tüm Gen Dizi Analizi)	UBE4A gene (Sequence Analysis-All Coding Exons)	UBE4A	UBE4A, UFD2, E4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE3D geni (Tüm Gen Dizi Analizi)	UBE3D gene (Sequence Analysis-All Coding Exons)	UBE3D	UBE2CBP, H10BH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE3C geni (Tüm Gen Dizi Analizi)	UBE3C gene (Sequence Analysis-All Coding Exons)	UBE3C	UBE3C, HECTH2, KIAA0010, KIAA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2Z geni (Tüm Gen Dizi Analizi)	UBE2Z gene (Sequence Analysis-All Coding Exons)	UBE2Z	UBE2Z, USE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2W geni (Tüm Gen Dizi Analizi)	UBE2W gene (Sequence Analysis-All Coding Exons)	UBE2W	UBE2W, UBC16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2V2 geni (Tüm Gen Dizi Analizi)	UBE2V2 gene (Sequence Analysis-All Coding Exons)	UBE2V2	UBE2V2, UEV2, DDVT1, EDPF1, MMS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2V1 geni (Tüm Gen Dizi Analizi)	UBE2V1 gene (Sequence Analysis-All Coding Exons)	UBE2V1	UBE2V1, UEV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2S geni (Tüm Gen Dizi Analizi)	UBE2S gene (Sequence Analysis-All Coding Exons)	UBE2S	UBE2S, EPF5, E2EPF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2R2 geni (Tüm Gen Dizi Analizi)	UBE2R2 gene (Sequence Analysis-All Coding Exons)	UBE2R2	UBE2R2, UBC3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2QL1 geni (Tüm Gen Dizi Analizi)	UBE2QL1 gene (Sequence Analysis-All Coding Exons)	UBE2QL1	UBE2QL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2Q2 geni (Tüm Gen Dizi Analizi)	UBE2Q2 gene (Sequence Analysis-All Coding Exons)	UBE2Q2	UBE2Q2, LOC92912	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2N geni (Tüm Gen Dizi Analizi)	UBE2N gene (Sequence Analysis-All Coding Exons)	UBE2N	UBE2N, UBCHBEN, UBC13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2M geni (Tüm Gen Dizi Analizi)	UBE2M gene (Sequence Analysis-All Coding Exons)	UBE2M	UBE2M, UBC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2L6 geni (Tüm Gen Dizi Analizi)	UBE2L6 gene (Sequence Analysis-All Coding Exons)	UBE2L6	UBE2L6, UBCH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2L3 geni (Tüm Gen Dizi Analizi)	UBE2L3 gene (Sequence Analysis-All Coding Exons)	UBE2L3	UBE2L3, UBCH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2K geni (Tüm Gen Dizi Analizi)	UBE2K gene (Sequence Analysis-All Coding Exons)	UBE2K	HIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2J1 geni (Tüm Gen Dizi Analizi)	UBE2J1 gene (Sequence Analysis-All Coding Exons)	UBE2J1	UBE2J1, UBC6E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2I geni (Tüm Gen Dizi Analizi)	UBE2I gene (Sequence Analysis-All Coding Exons)	UBE2I	UBE2I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2H geni (Tüm Gen Dizi Analizi)	UBE2H gene (Sequence Analysis-All Coding Exons)	UBE2H	UBE2H, UBCH2, UBC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2G2 geni (Tüm Gen Dizi Analizi)	UBE2G2 gene (Sequence Analysis-All Coding Exons)	UBE2G2	UBE2G2, UBC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2G1 geni (Tüm Gen Dizi Analizi)	UBE2G1 gene (Sequence Analysis-All Coding Exons)	UBE2G1	UBE2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2E3 geni (Tüm Gen Dizi Analizi)	UBE2E3 gene (Sequence Analysis-All Coding Exons)	UBE2E3	UBE2E3, UBCH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2E2 geni (Tüm Gen Dizi Analizi)	UBE2E2 gene (Sequence Analysis-All Coding Exons)	UBE2E2	UBE2E2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2E1 geni (Tüm Gen Dizi Analizi)	UBE2E1 gene (Sequence Analysis-All Coding Exons)	UBE2E1	UBE2E1, UBCH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2D3 geni (Tüm Gen Dizi Analizi)	UBE2D3 gene (Sequence Analysis-All Coding Exons)	UBE2D3	UBE2D3, UBCH5C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2D2 geni (Tüm Gen Dizi Analizi)	UBE2D2 gene (Sequence Analysis-All Coding Exons)	UBE2D2	UBE2D2, UBCH5B, UBC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2D1 geni (Tüm Gen Dizi Analizi)	UBE2D1 gene (Sequence Analysis-All Coding Exons)	UBE2D1	UBE2D1, UBCH5A, UBCH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBE2C geni (Tüm Gen Dizi Analizi)	UBE2C gene (Sequence Analysis-All Coding Exons)	UBE2C	UBE2C, UBCH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBD geni (Tüm Gen Dizi Analizi)	UBD gene (Sequence Analysis-All Coding Exons)	UBD	UBD, FAT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBC geni (Tüm Gen Dizi Analizi)	UBC gene (Sequence Analysis-All Coding Exons)	UBC	UBC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBASH3B geni (Tüm Gen Dizi Analizi)	UBASH3B gene (Sequence Analysis-All Coding Exons)	UBASH3B	UBASH3B, STS1, KIAA1959, p70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBASH3A geni (Tüm Gen Dizi Analizi)	UBASH3A gene (Sequence Analysis-All Coding Exons)	UBASH3A	UBASH3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBAP2L geni (Tüm Gen Dizi Analizi)	UBAP2L gene (Sequence Analysis-All Coding Exons)	UBAP2L	UBAP2L, NICE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBAP1 geni (Tüm Gen Dizi Analizi)	UBAP1 gene (Sequence Analysis-All Coding Exons)	UBAP1	UBAP1, UBAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBAC1 geni (Tüm Gen Dizi Analizi)	UBAC1 gene (Sequence Analysis-All Coding Exons)	UBAC1	UBAC1, GBDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBA7 geni (Tüm Gen Dizi Analizi)	UBA7 gene (Sequence Analysis-All Coding Exons)	UBA7	UBE7, UBE1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBA6 geni (Tüm Gen Dizi Analizi)	UBA6 gene (Sequence Analysis-All Coding Exons)	UBA6	UBA6, UBE1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBAS2 geni (Tüm Gen Dizi Analizi)	UBAS2 gene (Sequence Analysis-All Coding Exons)	UBAS2	UBAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBA2 geni (Tüm Gen Dizi Analizi)	UBA2 gene (Sequence Analysis-All Coding Exons)	UBA2	UBA2, SAE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UAP1 geni (Tüm Gen Dizi Analizi)	UAP1 gene (Sequence Analysis-All Coding Exons)	UAP1	UAP1, SPAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UACA geni (Tüm Gen Dizi Analizi)	UACA gene (Sequence Analysis-All Coding Exons)	UACA	UACA, NUCLING	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
U2AF2 geni (Tüm Gen Dizi Analizi)	U2AF2 gene (Sequence Analysis-All Coding Exons)	U2AF2	U2AF2, U2AF65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
U2AF1L4 geni (Tüm Gen Dizi Analizi)	U2AF1L4 gene (Sequence Analysis-All Coding Exons)	U2AF1L4	U2AF1L4, U2AF1RS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
U2AF1 geni (Tüm Gen Dizi Analizi)	U2AF1 gene (Sequence Analysis-All Coding Exons)	U2AF1	U2AF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TYW3 geni (Tüm Gen Dizi Analizi)	TYW3 gene (Sequence Analysis-All Coding Exons)	TYW3	TYW3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TYW1 geni (Tüm Gen Dizi Analizi)	TYW1 gene (Sequence Analysis-All Coding Exons)	TYW1	TYW1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TYSND1 geni (Tüm Gen Dizi Analizi)	TYSND1 gene (Sequence Analysis-All Coding Exons)	TYSND1	TYSND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TYRO3 geni (Tüm Gen Dizi Analizi)	TYRO3 gene (Sequence Analysis-All Coding Exons)	TYRO3	TYRO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TYMS geni (Tüm Gen Dizi Analizi)	TYMS gene (Sequence Analysis-All Coding Exons)	TYMS	TYMS, TS, TMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNRD3 geni (Tüm Gen Dizi Analizi)	TXNRD3 gene (Sequence Analysis-All Coding Exons)	TXNRD3	TXNRD3, TRXR3, TR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNRD2 geni (Tüm Gen Dizi Analizi)	TXNRD2 gene (Sequence Analysis-All Coding Exons)	TXNRD2	TXNRD2, TRXR2, SELZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNRD2 geni (Tüm Gen Dizi Analizi)	TXNRD2 gene (Sequence Analysis-All Coding Exons)	TXNRD2	TXNRD2, TRXR2, SELZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNRD1 geni (Tüm Gen Dizi Analizi)	TXNRD1 gene (Sequence Analysis-All Coding Exons)	TXNRD1	TXNRD1, TXNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNL1 geni (Tüm Gen Dizi Analizi)	TXNL1 gene (Sequence Analysis-All Coding Exons)	TXNL1	TXNL1, TXNL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNIP geni (Tüm Gen Dizi Analizi)	TXNIP gene (Sequence Analysis-All Coding Exons)	TXNIP	TXNIP, VDUP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNDC9 geni (Tüm Gen Dizi Analizi)	TXNDC9 gene (Sequence Analysis-All Coding Exons)	TXNDC9	TXNDC9, PHLP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNDC5 geni (Tüm Gen Dizi Analizi)	TXNDC5 gene (Sequence Analysis-All Coding Exons)	TXNDC5	TXNDC5, ERP46, ENDOPDI, HCC-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNDC17 geni (Tüm Gen Dizi Analizi)	TXNDC17 gene (Sequence Analysis-All Coding Exons)	TXNDC17	TXNDC17, TRP14, TXNL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNDC16 geni (Tüm Gen Dizi Analizi)	TXNDC16 gene (Sequence Analysis-All Coding Exons)	TXNDC16	TXNDC16, KIAA1344	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXNDC12 geni (Tüm Gen Dizi Analizi)	TXNDC12 gene (Sequence Analysis-All Coding Exons)	TXNDC12	TXNDC12, TLP19, ERP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXN geni (Tüm Gen Dizi Analizi)	TXN gene (Sequence Analysis-All Coding Exons)	TXN	TXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXLNGY geni (Tüm Gen Dizi Analizi)	TXLNGY gene (Sequence Analysis-All Coding Exons)	TXLNGY	CYorf15A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXLNG geni (Tüm Gen Dizi Analizi)	TXLNG gene (Sequence Analysis-All Coding Exons)	TXLNG	CXorf15, FIAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXK geni (Tüm Gen Dizi Analizi)	TXK gene (Sequence Analysis-All Coding Exons)	TXK	TXK, BTKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TWSG1 geni (Tüm Gen Dizi Analizi)	TWSG1 gene (Sequence Analysis-All Coding Exons)	TWSG1	TWSG1, TSG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
VASH1 geni (Tüm Gen Dizi Analizi)	VASH1 gene (Sequence Analysis-All Coding Exons)	VASH1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TWISTNB geni (Tüm Gen Dizi Analizi)	TWISTNB gene (Sequence Analysis-All Coding Exons)	TWISTNB	TWISTNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TWF2 geni (Tüm Gen Dizi Analizi)	TWF2 gene (Sequence Analysis-All Coding Exons)	TWF2	TWF2, PTK9L, A6RP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TWF1 geni (Tüm Gen Dizi Analizi)	TWF1 gene (Sequence Analysis-All Coding Exons)	TWF1	TWF1, PTK9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUSC5 geni (Tüm Gen Dizi Analizi)	TUSC5 gene (Sequence Analysis-All Coding Exons)	TUSC5	TUSC5, LOST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUSC2 geni (Tüm Gen Dizi Analizi)	TUSC2 gene (Sequence Analysis-All Coding Exons)	TUSC2	FUS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUSC1 geni (Tüm Gen Dizi Analizi)	TUSC1 gene (Sequence Analysis-All Coding Exons)	TUSC1	TUSC1, TSG9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUNAR geni (Tüm Gen Dizi Analizi)	TUNAR gene (Sequence Analysis-All Coding Exons)	TUNAR	TUNAR, TUNA, LINC00617	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TULP3 geni (Tüm Gen Dizi Analizi)	TULP3 gene (Sequence Analysis-All Coding Exons)	TULP3	TULP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TULP2 geni (Tüm Gen Dizi Analizi)	TULP2 gene (Sequence Analysis-All Coding Exons)	TULP2	TULP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUFT1 geni (Tüm Gen Dizi Analizi)	TUFT1 gene (Sequence Analysis-All Coding Exons)	TUFT1	TUFT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBGCP5 geni (Tüm Gen Dizi Analizi)	TUBGCP5 gene (Sequence Analysis-All Coding Exons)	TUBGCP5	TUBGCP5, GCP5, KIAA1899	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBG2 geni (Tüm Gen Dizi Analizi)	TUBG2 gene (Sequence Analysis-All Coding Exons)	TUBG2	TUBG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBE1 geni (Tüm Gen Dizi Analizi)	TUBE1 gene (Sequence Analysis-All Coding Exons)	TUBE1	TUBE1, TUBE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBD1 geni (Tüm Gen Dizi Analizi)	TUBD1 gene (Sequence Analysis-All Coding Exons)	TUBD1	TUBD1, TUBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBB6 geni (Tüm Gen Dizi Analizi)	TUBB6 gene (Sequence Analysis-All Coding Exons)	TUBB6	TUBB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBB4B geni (Tüm Gen Dizi Analizi)	TUBB4B gene (Sequence Analysis-All Coding Exons)	TUBB4B	TUBB4B, TUBB2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBA3C geni (Tüm Gen Dizi Analizi)	TUBA3C gene (Sequence Analysis-All Coding Exons)	TUBA3C	TUBA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTYH2 geni (Tüm Gen Dizi Analizi)	TTYH2 gene (Sequence Analysis-All Coding Exons)	TTYH2	TTYH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTYH1 geni (Tüm Gen Dizi Analizi)	TTYH1 gene (Sequence Analysis-All Coding Exons)	TTYH1	TTYH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTTY6 geni (Tüm Gen Dizi Analizi)	TTTY6 gene (Sequence Analysis-All Coding Exons)	TTTY6	TTTY6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTTY5 geni (Tüm Gen Dizi Analizi)	TTTY5 gene (Sequence Analysis-All Coding Exons)	TTTY5	TTTY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTTY4 geni (Tüm Gen Dizi Analizi)	TTTY4 gene (Sequence Analysis-All Coding Exons)	TTTY4	TTTY4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTTY3 geni (Tüm Gen Dizi Analizi)	TTTY3 gene (Sequence Analysis-All Coding Exons)	TTTY3	TTTY3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTTY17A geni (Tüm Gen Dizi Analizi)	TTTY17A gene (Sequence Analysis-All Coding Exons)	TTTY17A	TTY17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTLL6 geni (Tüm Gen Dizi Analizi)	TTLL6 gene (Sequence Analysis-All Coding Exons)	TTLL6	TTLL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTLL1 geni (Tüm Gen Dizi Analizi)	TTLL1 gene (Sequence Analysis-All Coding Exons)	TTLL1	TTLL1, KIAA0173	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTL geni (Tüm Gen Dizi Analizi)	TTL gene (Sequence Analysis-All Coding Exons)	TTL	TTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTK geni (Tüm Gen Dizi Analizi)	TTK gene (Sequence Analysis-All Coding Exons)	TTK	TTK, MPS1L1, PYT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTI1 geni (Tüm Gen Dizi Analizi)	TTI1 gene (Sequence Analysis-All Coding Exons)	TTI1	TTI1, KIAA0406	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTF2 geni (Tüm Gen Dizi Analizi)	TTF2 gene (Sequence Analysis-All Coding Exons)	TTF2	TTF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UTP14C geni (Tüm Gen Dizi Analizi)	UTP14C gene (Sequence Analysis-All Coding Exons)	UTP14C		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC9 geni (Tüm Gen Dizi Analizi)	TTC9 gene (Sequence Analysis-All Coding Exons)	TTC9	TTC9, KIAA0227	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC4 geni (Tüm Gen Dizi Analizi)	TTC4 gene (Sequence Analysis-All Coding Exons)	TTC4	TTC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC39B geni (Tüm Gen Dizi Analizi)	TTC39B gene (Sequence Analysis-All Coding Exons)	TTC39B	TTC39B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC3 geni (Tüm Gen Dizi Analizi)	TTC3 gene (Sequence Analysis-All Coding Exons)	TTC3	TTC3, TPRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC28 geni (Tüm Gen Dizi Analizi)	TTC28 gene (Sequence Analysis-All Coding Exons)	TTC28	TTC28, TPRBK, KIAA1043	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC23L geni (Tüm Gen Dizi Analizi)	TTC23L gene (Sequence Analysis-All Coding Exons)	TTC23L	TTC23L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC12 geni (Tüm Gen Dizi Analizi)	TTC12 gene (Sequence Analysis-All Coding Exons)	TTC12	TTC12, TPARM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC1 geni (Tüm Gen Dizi Analizi)	TTC1 gene (Sequence Analysis-All Coding Exons)	TTC1	TTC1, TPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSTD1 geni (Tüm Gen Dizi Analizi)	TSTD1 gene (Sequence Analysis-All Coding Exons)	TSTD1	TSTD1, KAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSTA3 geni (Tüm Gen Dizi Analizi)	TSTA3 gene (Sequence Analysis-All Coding Exons)	TSTA3	TSTA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TST geni (Tüm Gen Dizi Analizi)	TST gene (Sequence Analysis-All Coding Exons)	TST	TST, RDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSK6 geni (Tüm Gen Dizi Analizi)	TSSK6 gene (Sequence Analysis-All Coding Exons)	TSSK6	TSSK6, SSTK, TSSK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSK4 geni (Tüm Gen Dizi Analizi)	TSSK4 gene (Sequence Analysis-All Coding Exons)	TSSK4	TSSK4, TSSK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSK3 geni (Tüm Gen Dizi Analizi)	TSSK3 gene (Sequence Analysis-All Coding Exons)	TSSK3	TSSK3, STK22C, STK22D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSK2 geni (Tüm Gen Dizi Analizi)	TSSK2 gene (Sequence Analysis-All Coding Exons)	TSSK2	TSSK2, STK22B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSK1B geni (Tüm Gen Dizi Analizi)	TSSK1B gene (Sequence Analysis-All Coding Exons)	TSSK1B	TSSK1, STK22D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSC4 geni (Tüm Gen Dizi Analizi)	TSSC4 gene (Sequence Analysis-All Coding Exons)	TSSC4	TSSC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSSC1 geni (Tüm Gen Dizi Analizi)	TSSC1 gene (Sequence Analysis-All Coding Exons)	TSSC1	TSSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSR3 geni (Tüm Gen Dizi Analizi)	TSR3 gene (Sequence Analysis-All Coding Exons)	TSR3	TSR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSR1 geni (Tüm Gen Dizi Analizi)	TSR1 gene (Sequence Analysis-All Coding Exons)	TSR1	TSR1, KIAA1401	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPYL5 geni (Tüm Gen Dizi Analizi)	TSPYL5 gene (Sequence Analysis-All Coding Exons)	TSPYL5	TSPYL5, KIAA1750	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPYL2 geni (Tüm Gen Dizi Analizi)	TSPYL2 gene (Sequence Analysis-All Coding Exons)	TSPYL2	TSPYL2, CDA1, DENTT, ONAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPY1 geni (Tüm Gen Dizi Analizi)	TSPY1 gene (Sequence Analysis-All Coding Exons)	TSPY1	TSPY1, TSPY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPOAP1 geni (Tüm Gen Dizi Analizi)	TSPOAP1 gene (Sequence Analysis-All Coding Exons)	TSPOAP1	BZRAP1, PRAX1, RIMBP1, KIAA0612	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP29 geni (Tüm Gen Dizi Analizi)	USP29 gene (Sequence Analysis-All Coding Exons)	USP29		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPO geni (Tüm Gen Dizi Analizi)	TSPO gene (Sequence Analysis-All Coding Exons)	TSPO	TSPO, BZRP, PBR, PBS, BPBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN9 geni (Tüm Gen Dizi Analizi)	TSPAN9 gene (Sequence Analysis-All Coding Exons)	TSPAN9	TSPAN9, NET5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN8 geni (Tüm Gen Dizi Analizi)	TSPAN8 gene (Sequence Analysis-All Coding Exons)	TSPAN8	TSPAN8, TM4SF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP25 geni (Tüm Gen Dizi Analizi)	USP25 gene (Sequence Analysis-All Coding Exons)	USP25		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN6 geni (Tüm Gen Dizi Analizi)	TSPAN6 gene (Sequence Analysis-All Coding Exons)	TSPAN6	TSPAN6, TM4SF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN5 geni (Tüm Gen Dizi Analizi)	TSPAN5 gene (Sequence Analysis-All Coding Exons)	TSPAN5	TSPAN5, NET4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN4 geni (Tüm Gen Dizi Analizi)	TSPAN4 gene (Sequence Analysis-All Coding Exons)	TSPAN4	TSPAN4, TM4SF7, NAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN33 geni (Tüm Gen Dizi Analizi)	TSPAN33 gene (Sequence Analysis-All Coding Exons)	TSPAN33	TSPAN33, PEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USP2 geni (Tüm Gen Dizi Analizi)	USP2 gene (Sequence Analysis-All Coding Exons)	USP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN32 geni (Tüm Gen Dizi Analizi)	TSPAN32 gene (Sequence Analysis-All Coding Exons)	TSPAN32	TSPAN32, PHEMX, TSSC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN31 geni (Tüm Gen Dizi Analizi)	TSPAN31 gene (Sequence Analysis-All Coding Exons)	TSPAN31	TSPAN31, SAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN3 geni (Tüm Gen Dizi Analizi)	TSPAN3 gene (Sequence Analysis-All Coding Exons)	TSPAN3	TSPAN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN2 geni (Tüm Gen Dizi Analizi)	TSPAN2 gene (Sequence Analysis-All Coding Exons)	TSPAN2	TSPAN2, NET3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN15 geni (Tüm Gen Dizi Analizi)	TSPAN15 gene (Sequence Analysis-All Coding Exons)	TSPAN15	TSPAN15, NET7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN13 geni (Tüm Gen Dizi Analizi)	TSPAN13 gene (Sequence Analysis-All Coding Exons)	TSPAN13	TSPAN13, NET6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSPAN1 geni (Tüm Gen Dizi Analizi)	TSPAN1 gene (Sequence Analysis-All Coding Exons)	TSPAN1	TSPAN1, NET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSNAXIP1 geni (Tüm Gen Dizi Analizi)	TSNAXIP1 gene (Sequence Analysis-All Coding Exons)	TSNAXIP1	TSNAXIP1, TXI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSNAX geni (Tüm Gen Dizi Analizi)	TSNAX gene (Sequence Analysis-All Coding Exons)	TSNAX	TSNAX, TRAX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSN geni (Tüm Gen Dizi Analizi)	TSN gene (Sequence Analysis-All Coding Exons)	TSN	TSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSLP geni (Tüm Gen Dizi Analizi)	TSLP gene (Sequence Analysis-All Coding Exons)	TSLP	TSLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSKU geni (Tüm Gen Dizi Analizi)	TSKU gene (Sequence Analysis-All Coding Exons)	TSKU	TSKU, E2IG4, TSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSIX geni (Tüm Gen Dizi Analizi)	TSIX gene (Sequence Analysis-All Coding Exons)	TSIX	TSIX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSHZ3 geni (Tüm Gen Dizi Analizi)	TSHZ3 gene (Sequence Analysis-All Coding Exons)	TSHZ3	TSHZ3, KIAA1474	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSHZ2 geni (Tüm Gen Dizi Analizi)	TSHZ2 gene (Sequence Analysis-All Coding Exons)	TSHZ2	TSHZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSGA10 geni (Tüm Gen Dizi Analizi)	TSGA10 gene (Sequence Analysis-All Coding Exons)	TSGA10	TSGA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSC22D4 geni (Tüm Gen Dizi Analizi)	TSC22D4 gene (Sequence Analysis-All Coding Exons)	TSC22D4	TSC22D4, THG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSC22D3 geni (Tüm Gen Dizi Analizi)	TSC22D3 gene (Sequence Analysis-All Coding Exons)	TSC22D3	TSC22D3, DSIPI, GILZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSC22D1 geni (Tüm Gen Dizi Analizi)	TSC22D1 gene (Sequence Analysis-All Coding Exons)	TSC22D1	TSC22D1, TSC22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRUB2 geni (Tüm Gen Dizi Analizi)	TRUB2 gene (Sequence Analysis-All Coding Exons)	TRUB2	TRUB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRUB1 geni (Tüm Gen Dizi Analizi)	TRUB1 gene (Sequence Analysis-All Coding Exons)	TRUB1	TRUB1, PUS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
USE1 geni (Tüm Gen Dizi Analizi)	USE1 gene (Sequence Analysis-All Coding Exons)	USE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRRAP geni (Tüm Gen Dizi Analizi)	TRRAP gene (Sequence Analysis-All Coding Exons)	TRRAP	TRRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPV6 geni (Tüm Gen Dizi Analizi)	TRPV6 gene (Sequence Analysis-All Coding Exons)	TRPV6	TRPV6, ECAC2, CAT1, CATL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPV5 geni (Tüm Gen Dizi Analizi)	TRPV5 gene (Sequence Analysis-All Coding Exons)	TRPV5	TRPV5, ECAC1, CAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPV2 geni (Tüm Gen Dizi Analizi)	TRPV2 gene (Sequence Analysis-All Coding Exons)	TRPV2	TRPV2, VRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TRPV1 geni (Tüm Gen Dizi Analizi)	TRPV1 gene (Sequence Analysis-All Coding Exons)	TRPV1	TRPV1, VR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPT1 geni (Tüm Gen Dizi Analizi)	TRPT1 gene (Sequence Analysis-All Coding Exons)	TRPT1	TRPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPM8 geni (Tüm Gen Dizi Analizi)	TRPM8 gene (Sequence Analysis-All Coding Exons)	TRPM8	TRPM8, TRPP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPM5 geni (Tüm Gen Dizi Analizi)	TRPM5 gene (Sequence Analysis-All Coding Exons)	TRPM5	TRPM5, MTR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPM3 geni (Tüm Gen Dizi Analizi)	TRPM3 gene (Sequence Analysis-All Coding Exons)	TRPM3	TRPM3, MLSN2, LTRPC3, KIAA1616	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPM2 geni (Tüm Gen Dizi Analizi)	TRPM2 gene (Sequence Analysis-All Coding Exons)	TRPM2	TRPM2, TRPC7, KNP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPC5 geni (Tüm Gen Dizi Analizi)	TRPC5 gene (Sequence Analysis-All Coding Exons)	TRPC5	TRPC5, TRP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPC4AP geni (Tüm Gen Dizi Analizi)	TRPC4AP gene (Sequence Analysis-All Coding Exons)	TRPC4AP	TRPC4AP, TRUSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPC4 geni (Tüm Gen Dizi Analizi)	TRPC4 gene (Sequence Analysis-All Coding Exons)	TRPC4	TRPC4, TRP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRPC1 geni (Tüm Gen Dizi Analizi)	TRPC1 gene (Sequence Analysis-All Coding Exons)	TRPC1	TRPC1, TRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TROVE2 geni (Tüm Gen Dizi Analizi)	TROVE2 gene (Sequence Analysis-All Coding Exons)	TROVE2	TROVE2, RO60, SSA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRO geni (Tüm Gen Dizi Analizi)	TRO gene (Sequence Analysis-All Coding Exons)	TRO	TRO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRNT1 geni (Tüm Gen Dizi Analizi)	TRNT1 gene (Sequence Analysis-All Coding Exons)	TRNT1	TRNT1, SIFD, RPEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Sideroblastic anemia with B-cell immunodeficiency, periodic fevers, and developmental delay (SIFD) [MIM:616084]: An autosomal recessive disease characterized by severe sideroblastic anemia with onset in the neonatal period or infancy, recurrent periodic fevers without an infectious etiology, B-cell lymphopenia and hypogammaglobulinemia. Affected individuals show delayed psychomotor development with variable neurodegeneration. Additional variable features include sensorineural hearing loss, retinitis pigmentosa, nephrocalcinosis, and cardiomyopathy. [ECO:0000269|PubMed:25193871]. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
TRNP1 geni (Tüm Gen Dizi Analizi)	TRNP1 gene (Sequence Analysis-All Coding Exons)	TRNP1	TRNP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRMT44 geni (Tüm Gen Dizi Analizi)	TRMT44 gene (Sequence Analysis-All Coding Exons)	TRMT44	METTL19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRMT2A geni (Tüm Gen Dizi Analizi)	TRMT2A gene (Sequence Analysis-All Coding Exons)	TRMT2A	TRMT2A, HTF9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRMT1L geni (Tüm Gen Dizi Analizi)	TRMT1L gene (Sequence Analysis-All Coding Exons)	TRMT1L	C1orf25, TRM1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRMT12 geni (Tüm Gen Dizi Analizi)	TRMT12 gene (Sequence Analysis-All Coding Exons)	TRMT12	TRMT12, TRM12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRMT1 geni (Tüm Gen Dizi Analizi)	TRMT1 gene (Sequence Analysis-All Coding Exons)	TRMT1	TRMT1, TRM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIP6 geni (Tüm Gen Dizi Analizi)	TRIP6 gene (Sequence Analysis-All Coding Exons)	TRIP6	TRIP6, CIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIP13 geni (Tüm Gen Dizi Analizi)	TRIP13 gene (Sequence Analysis-All Coding Exons)	TRIP13	TRIP13, 16E1BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIP12 geni (Tüm Gen Dizi Analizi)	TRIP12 gene (Sequence Analysis-All Coding Exons)	TRIP12	TRIP12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIP10 geni (Tüm Gen Dizi Analizi)	TRIP10 gene (Sequence Analysis-All Coding Exons)	TRIP10	TRIP10, CIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM9 geni (Tüm Gen Dizi Analizi)	TRIM9 gene (Sequence Analysis-All Coding Exons)	TRIM9	TRIM9, SPRING, KIAA0282	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM8 geni (Tüm Gen Dizi Analizi)	TRIM8 gene (Sequence Analysis-All Coding Exons)	TRIM8	TRIM8, RNF27, GERP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM74 geni (Tüm Gen Dizi Analizi)	TRIM74 gene (Sequence Analysis-All Coding Exons)	TRIM74	TRIM74, TRIM50C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM73 geni (Tüm Gen Dizi Analizi)	TRIM73 gene (Sequence Analysis-All Coding Exons)	TRIM73	TRIM73, TRIM50B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM72 geni (Tüm Gen Dizi Analizi)	TRIM72 gene (Sequence Analysis-All Coding Exons)	TRIM72	TRIM72, MG53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM7 geni (Tüm Gen Dizi Analizi)	TRIM7 gene (Sequence Analysis-All Coding Exons)	TRIM7	TRIM7, GNIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM69 geni (Tüm Gen Dizi Analizi)	TRIM69 gene (Sequence Analysis-All Coding Exons)	TRIM69	TRIM69, TRIF, RNF36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM68 geni (Tüm Gen Dizi Analizi)	TRIM68 gene (Sequence Analysis-All Coding Exons)	TRIM68	TRIM68, SS56, GC109	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM67 geni (Tüm Gen Dizi Analizi)	TRIM67 gene (Sequence Analysis-All Coding Exons)	TRIM67	TRIM67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM66 geni (Tüm Gen Dizi Analizi)	TRIM66 gene (Sequence Analysis-All Coding Exons)	TRIM66	TRIM66, TIF1D, KIAA0298	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM63 geni (Tüm Gen Dizi Analizi)	TRIM63 gene (Sequence Analysis-All Coding Exons)	TRIM63	TRIM63, RNF28, SMRZ, MURF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM62 geni (Tüm Gen Dizi Analizi)	TRIM62 gene (Sequence Analysis-All Coding Exons)	TRIM62	TRIM62, DEAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM6 geni (Tüm Gen Dizi Analizi)	TRIM6 gene (Sequence Analysis-All Coding Exons)	TRIM6	TRIM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM59 geni (Tüm Gen Dizi Analizi)	TRIM59 gene (Sequence Analysis-All Coding Exons)	TRIM59	TRIM59, RNF104, MRF1, IFT80L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM56 geni (Tüm Gen Dizi Analizi)	TRIM56 gene (Sequence Analysis-All Coding Exons)	TRIM56	TRIM56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM55 geni (Tüm Gen Dizi Analizi)	TRIM55 gene (Sequence Analysis-All Coding Exons)	TRIM55	TRIM55, RNF29, MURF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM54 geni (Tüm Gen Dizi Analizi)	TRIM54 gene (Sequence Analysis-All Coding Exons)	TRIM54	TRIM54, RNF30, MURF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM50 geni (Tüm Gen Dizi Analizi)	TRIM50 gene (Sequence Analysis-All Coding Exons)	TRIM50	TRIM50, TRIM50A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM5 geni (Tüm Gen Dizi Analizi)	TRIM5 gene (Sequence Analysis-All Coding Exons)	TRIM5	TRIM5, RNF88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM49 geni (Tüm Gen Dizi Analizi)	TRIM49 gene (Sequence Analysis-All Coding Exons)	TRIM49	RNF18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM47 geni (Tüm Gen Dizi Analizi)	TRIM47 gene (Sequence Analysis-All Coding Exons)	TRIM47	TRIM47, GOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM46 geni (Tüm Gen Dizi Analizi)	TRIM46 gene (Sequence Analysis-All Coding Exons)	TRIM46	GENEY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM45 geni (Tüm Gen Dizi Analizi)	TRIM45 gene (Sequence Analysis-All Coding Exons)	TRIM45	TRIM45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM41 geni (Tüm Gen Dizi Analizi)	TRIM41 gene (Sequence Analysis-All Coding Exons)	TRIM41	TRIM41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM40 geni (Tüm Gen Dizi Analizi)	TRIM40 gene (Sequence Analysis-All Coding Exons)	TRIM40	TRIM40, RNF35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM39 geni (Tüm Gen Dizi Analizi)	TRIM39 gene (Sequence Analysis-All Coding Exons)	TRIM39	TRIM39, RNF23, TFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM36 geni (Tüm Gen Dizi Analizi)	TRIM36 gene (Sequence Analysis-All Coding Exons)	TRIM36	TRIM36, RBCC728	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM35 geni (Tüm Gen Dizi Analizi)	TRIM35 gene (Sequence Analysis-All Coding Exons)	TRIM35	TRIM35, MAIR, HLS5, KIAA1098	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM34 geni (Tüm Gen Dizi Analizi)	TRIM34 gene (Sequence Analysis-All Coding Exons)	TRIM34	TRIM34, RNF21, IFP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM33 geni (Tüm Gen Dizi Analizi)	TRIM33 gene (Sequence Analysis-All Coding Exons)	TRIM33	TRIM33, TIF1G, RFG7, PTC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM31 geni (Tüm Gen Dizi Analizi)	TRIM31 gene (Sequence Analysis-All Coding Exons)	TRIM31	TRIM31, HCGI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM3 geni (Tüm Gen Dizi Analizi)	TRIM3 gene (Sequence Analysis-All Coding Exons)	TRIM3	TRIM3, RNF22, BERP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM27 geni (Tüm Gen Dizi Analizi)	TRIM27 gene (Sequence Analysis-All Coding Exons)	TRIM27	TRIM27, RFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM26 geni (Tüm Gen Dizi Analizi)	TRIM26 gene (Sequence Analysis-All Coding Exons)	TRIM26	ZNF173	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM25 geni (Tüm Gen Dizi Analizi)	TRIM25 gene (Sequence Analysis-All Coding Exons)	TRIM25	TRIM25, ZNF147, EFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM24 geni (Tüm Gen Dizi Analizi)	TRIM24 gene (Sequence Analysis-All Coding Exons)	TRIM24	TRIM24, TIF1, TIF1A, PTC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM23 geni (Tüm Gen Dizi Analizi)	TRIM23 gene (Sequence Analysis-All Coding Exons)	TRIM23	TRIM23, ARD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM22 geni (Tüm Gen Dizi Analizi)	TRIM22 gene (Sequence Analysis-All Coding Exons)	TRIM22	TRIM22, STAF50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM21 geni (Tüm Gen Dizi Analizi)	TRIM21 gene (Sequence Analysis-All Coding Exons)	TRIM21	TRIM21, SSA1, RO52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM17 geni (Tüm Gen Dizi Analizi)	TRIM17 gene (Sequence Analysis-All Coding Exons)	TRIM17	RNF16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM16 geni (Tüm Gen Dizi Analizi)	TRIM16 gene (Sequence Analysis-All Coding Exons)	TRIM16	TRIM16, EBBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM14 geni (Tüm Gen Dizi Analizi)	TRIM14 gene (Sequence Analysis-All Coding Exons)	TRIM14	TRIM14, KIAA0129	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM13 geni (Tüm Gen Dizi Analizi)	TRIM13 gene (Sequence Analysis-All Coding Exons)	TRIM13	TRIM13, RFP2, LEU5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM11 geni (Tüm Gen Dizi Analizi)	TRIM11 gene (Sequence Analysis-All Coding Exons)	TRIM11	TRIM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM10 geni (Tüm Gen Dizi Analizi)	TRIM10 gene (Sequence Analysis-All Coding Exons)	TRIM10	RNF9, RFB30, HERF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIL geni (Tüm Gen Dizi Analizi)	TRIL gene (Sequence Analysis-All Coding Exons)	TRIL	TRIL, KIAA0644	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIB3 geni (Tüm Gen Dizi Analizi)	TRIB3 gene (Sequence Analysis-All Coding Exons)	TRIB3	TRIB3, NIPK, SINK, C20orf97, SKIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIB1 geni (Tüm Gen Dizi Analizi)	TRIB1 gene (Sequence Analysis-All Coding Exons)	TRIB1	TRIB1, TRB1, SKIP1, C8FW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIAP1 geni (Tüm Gen Dizi Analizi)	TRIAP1 gene (Sequence Analysis-All Coding Exons)	TRIAP1	TRIAP1, HSPC132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRHDE geni (Tüm Gen Dizi Analizi)	TRHDE gene (Sequence Analysis-All Coding Exons)	TRHDE	TRHDE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TREX2 geni (Tüm Gen Dizi Analizi)	TREX2 gene (Sequence Analysis-All Coding Exons)	TREX2	TREX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRERF1 geni (Tüm Gen Dizi Analizi)	TRERF1 gene (Sequence Analysis-All Coding Exons)	TRERF1	TRERF1, TREP132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TREML4 geni (Tüm Gen Dizi Analizi)	TREML4 gene (Sequence Analysis-All Coding Exons)	TREML4	TREML4, TLT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TREML2 geni (Tüm Gen Dizi Analizi)	TREML2 gene (Sequence Analysis-All Coding Exons)	TREML2	TREML2, TLT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TREML1 geni (Tüm Gen Dizi Analizi)	TREML1 gene (Sequence Analysis-All Coding Exons)	TREML1	TREML1, TLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TREM1 geni (Tüm Gen Dizi Analizi)	TREM1 gene (Sequence Analysis-All Coding Exons)	TREM1	TREM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRDMT1 geni (Tüm Gen Dizi Analizi)	TRDMT1 gene (Sequence Analysis-All Coding Exons)	TRDMT1	TRDMT1, DMNT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAT1 geni (Tüm Gen Dizi Analizi)	TRAT1 gene (Sequence Analysis-All Coding Exons)	TRAT1	TRAT1, TRIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC8 geni (Tüm Gen Dizi Analizi)	TRAPPC8 gene (Sequence Analysis-All Coding Exons)	TRAPPC8	TRAPPC8, KIAA1012	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC6B geni (Tüm Gen Dizi Analizi)	TRAPPC6B gene (Sequence Analysis-All Coding Exons)	TRAPPC6B	TRAPPC6B, TPC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC6A geni (Tüm Gen Dizi Analizi)	TRAPPC6A gene (Sequence Analysis-All Coding Exons)	TRAPPC6A	TRAPPC6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC4 geni (Tüm Gen Dizi Analizi)	TRAPPC4 gene (Sequence Analysis-All Coding Exons)	TRAPPC4	TRAPPC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC3L geni (Tüm Gen Dizi Analizi)	TRAPPC3L gene (Sequence Analysis-All Coding Exons)	TRAPPC3L	BET3L, TRAPPC3L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC3 geni (Tüm Gen Dizi Analizi)	TRAPPC3 gene (Sequence Analysis-All Coding Exons)	TRAPPC3	TRAPPC3, BET3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TRAPPC2L geni (Tüm Gen Dizi Analizi)	TRAPPC2L gene (Sequence Analysis-All Coding Exons)	TRAPPC2L	TRAPPC2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC12 geni (Tüm Gen Dizi Analizi)	TRAPPC12 gene (Sequence Analysis-All Coding Exons)	TRAPPC12	TRAPPC12, TTC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC10 geni (Tüm Gen Dizi Analizi)	TRAPPC10 gene (Sequence Analysis-All Coding Exons)	TRAPPC10	TMEM1, EHOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAPPC1 geni (Tüm Gen Dizi Analizi)	TRAPPC1 gene (Sequence Analysis-All Coding Exons)	TRAPPC1	TRAPPC1, MUM2, BET5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAP1 geni (Tüm Gen Dizi Analizi)	TRAP1 gene (Sequence Analysis-All Coding Exons)	TRAP1	TRAP1, HSP75, HSP90L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAM2 geni (Tüm Gen Dizi Analizi)	TRAM2 gene (Sequence Analysis-All Coding Exons)	TRAM2	TRAM2, KIAA0057	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAM1 geni (Tüm Gen Dizi Analizi)	TRAM1 gene (Sequence Analysis-All Coding Exons)	TRAM1	TRAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAK2 geni (Tüm Gen Dizi Analizi)	TRAK2 gene (Sequence Analysis-All Coding Exons)	TRAK2	TRAK2, GRIF1, AL32CR3, KIAA0549, OIP98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAK1 geni (Tüm Gen Dizi Analizi)	TRAK1 gene (Sequence Analysis-All Coding Exons)	TRAK1	TRAK1, OIP106, KIAA1042	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAFD1 geni (Tüm Gen Dizi Analizi)	TRAFD1 gene (Sequence Analysis-All Coding Exons)	TRAFD1	TRAFD1, FLN29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF7 geni (Tüm Gen Dizi Analizi)	TRAF7 gene (Sequence Analysis-All Coding Exons)	TRAF7	TRAF7, RFWD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF6 geni (Tüm Gen Dizi Analizi)	TRAF6 gene (Sequence Analysis-All Coding Exons)	TRAF6	TRAF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF5 geni (Tüm Gen Dizi Analizi)	TRAF5 gene (Sequence Analysis-All Coding Exons)	TRAF5	TRAF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF3IP3 geni (Tüm Gen Dizi Analizi)	TRAF3IP3 gene (Sequence Analysis-All Coding Exons)	TRAF3IP3	TRAF3IP3, T3JAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF2 geni (Tüm Gen Dizi Analizi)	TRAF2 gene (Sequence Analysis-All Coding Exons)	TRAF2	TRAF2, TRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF1 geni (Tüm Gen Dizi Analizi)	TRAF1 gene (Sequence Analysis-All Coding Exons)	TRAF1	TRAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRADD geni (Tüm Gen Dizi Analizi)	TRADD gene (Sequence Analysis-All Coding Exons)	TRADD	TRADD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRABD2B geni (Tüm Gen Dizi Analizi)	TRABD2B gene (Sequence Analysis-All Coding Exons)	TRABD2B	TRABD2B, TIKI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UCN3 geni (Tüm Gen Dizi Analizi)	UCN3 gene (Sequence Analysis-All Coding Exons)	UCN3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRABD2A geni (Tüm Gen Dizi Analizi)	TRABD2A gene (Sequence Analysis-All Coding Exons)	TRABD2A	TRABD2A, TIKI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRA2B geni (Tüm Gen Dizi Analizi)	TRA2B gene (Sequence Analysis-All Coding Exons)	TRA2B	TRA2B, SFRS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRA2A geni (Tüm Gen Dizi Analizi)	TRA2A gene (Sequence Analysis-All Coding Exons)	TRA2A	TRA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPX2 geni (Tüm Gen Dizi Analizi)	TPX2 gene (Sequence Analysis-All Coding Exons)	TPX2	C20orf1, P100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPTE2 geni (Tüm Gen Dizi Analizi)	TPTE2 gene (Sequence Analysis-All Coding Exons)	TPTE2	TPTE2, TPIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPTE geni (Tüm Gen Dizi Analizi)	TPTE gene (Sequence Analysis-All Coding Exons)	TPTE	TPTE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPT1 geni (Tüm Gen Dizi Analizi)	TPT1 gene (Sequence Analysis-All Coding Exons)	TPT1	TPT1, HRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPST2 geni (Tüm Gen Dizi Analizi)	TPST2 gene (Sequence Analysis-All Coding Exons)	TPST2	TPST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPST1 geni (Tüm Gen Dizi Analizi)	TPST1 gene (Sequence Analysis-All Coding Exons)	TPST1	TPST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPSG1 geni (Tüm Gen Dizi Analizi)	TPSG1 gene (Sequence Analysis-All Coding Exons)	TPSG1	TPSG1, TMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPSD1 geni (Tüm Gen Dizi Analizi)	TPSD1 gene (Sequence Analysis-All Coding Exons)	TPSD1	TPSD1, MCP7L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPSB2 geni (Tüm Gen Dizi Analizi)	TPSB2 gene (Sequence Analysis-All Coding Exons)	TPSB2	TPSB2, TPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPSAB1 geni (Tüm Gen Dizi Analizi)	TPSAB1 gene (Sequence Analysis-All Coding Exons)	TPSAB1	TPSAB1, TPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPRXL geni (Tüm Gen Dizi Analizi)	TPRXL gene (Sequence Analysis-All Coding Exons)	TPRXL	TPRXL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPRX1 geni (Tüm Gen Dizi Analizi)	TPRX1 gene (Sequence Analysis-All Coding Exons)	TPRX1	TPRX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPRG1L geni (Tüm Gen Dizi Analizi)	TPRG1L gene (Sequence Analysis-All Coding Exons)	TPRG1L	TPRG1L, FAM79A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPR geni (Tüm Gen Dizi Analizi)	TPR gene (Sequence Analysis-All Coding Exons)	TPR	TPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPPP3 geni (Tüm Gen Dizi Analizi)	TPPP3 gene (Sequence Analysis-All Coding Exons)	TPPP3	TPPP3, p20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPPP2 geni (Tüm Gen Dizi Analizi)	TPPP2 gene (Sequence Analysis-All Coding Exons)	TPPP2	TPPP2, p18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPPP geni (Tüm Gen Dizi Analizi)	TPPP gene (Sequence Analysis-All Coding Exons)	TPPP	TPPP, P25, P24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPP2 geni (Tüm Gen Dizi Analizi)	TPP2 gene (Sequence Analysis-All Coding Exons)	TPP2	TPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPM4 geni (Tüm Gen Dizi Analizi)	TPM4 gene (Sequence Analysis-All Coding Exons)	TPM4	TPM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPH1 geni (Tüm Gen Dizi Analizi)	TPH1 gene (Sequence Analysis-All Coding Exons)	TPH1	TPH1, TPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPD52L2 geni (Tüm Gen Dizi Analizi)	TPD52L2 gene (Sequence Analysis-All Coding Exons)	TPD52L2	TPD52L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPD52L1 geni (Tüm Gen Dizi Analizi)	TPD52L1 gene (Sequence Analysis-All Coding Exons)	TPD52L1	TPD52L1, D53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPD52 geni (Tüm Gen Dizi Analizi)	TPD52 gene (Sequence Analysis-All Coding Exons)	TPD52	TPD52, D52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPCN1 geni (Tüm Gen Dizi Analizi)	TPCN1 gene (Sequence Analysis-All Coding Exons)	TPCN1	TPCN1, TPC1, KIAA1169	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPBG geni (Tüm Gen Dizi Analizi)	TPBG gene (Sequence Analysis-All Coding Exons)	TPBG	TPBG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53TG5 geni (Tüm Gen Dizi Analizi)	TP53TG5 gene (Sequence Analysis-All Coding Exons)	TP53TG5	TP53TG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53INP1 geni (Tüm Gen Dizi Analizi)	TP53INP1 gene (Sequence Analysis-All Coding Exons)	TP53INP1	TP53DINP1, P53DINP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53I3 geni (Tüm Gen Dizi Analizi)	TP53I3 gene (Sequence Analysis-All Coding Exons)	TP53I3	TP53I3, PIG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53BP2 geni (Tüm Gen Dizi Analizi)	TP53BP2 gene (Sequence Analysis-All Coding Exons)	TP53BP2	TP53BP2, ASPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53BP1 geni (Tüm Gen Dizi Analizi)	TP53BP1 gene (Sequence Analysis-All Coding Exons)	TP53BP1	TP53BP1, 53BP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53AIP1 geni (Tüm Gen Dizi Analizi)	TP53AIP1 gene (Sequence Analysis-All Coding Exons)	TP53AIP1	TP53AIP1, P53AIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOX4 geni (Tüm Gen Dizi Analizi)	TOX4 gene (Sequence Analysis-All Coding Exons)	TOX4	TOX4, LCP1, KIAA0737	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOX2 geni (Tüm Gen Dizi Analizi)	TOX2 gene (Sequence Analysis-All Coding Exons)	TOX2	TOX2, GCX1, C20orf100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOX geni (Tüm Gen Dizi Analizi)	TOX gene (Sequence Analysis-All Coding Exons)	TOX	TOX, KIAA0808	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOR3A geni (Tüm Gen Dizi Analizi)	TOR3A gene (Sequence Analysis-All Coding Exons)	TOR3A	TOR3A, ADIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOR2A geni (Tüm Gen Dizi Analizi)	TOR2A gene (Sequence Analysis-All Coding Exons)	TOR2A	TOR2A, TORP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOR1B geni (Tüm Gen Dizi Analizi)	TOR1B gene (Sequence Analysis-All Coding Exons)	TOR1B	TOR1B, DQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOR1AIP2 geni (Tüm Gen Dizi Analizi)	TOR1AIP2 gene (Sequence Analysis-All Coding Exons)	TOR1AIP2	TOR1AIP2, LULL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOPBP1 geni (Tüm Gen Dizi Analizi)	TOPBP1 gene (Sequence Analysis-All Coding Exons)	TOPBP1	TOPBP1, KIAA0259	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOPAZ1 geni (Tüm Gen Dizi Analizi)	TOPAZ1 gene (Sequence Analysis-All Coding Exons)	TOPAZ1	C3orf77, TOPAZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOP3B geni (Tüm Gen Dizi Analizi)	TOP3B gene (Sequence Analysis-All Coding Exons)	TOP3B	TOP3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOP3A geni (Tüm Gen Dizi Analizi)	TOP3A gene (Sequence Analysis-All Coding Exons)	TOP3A	TOP3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOP2B geni (Tüm Gen Dizi Analizi)	TOP2B gene (Sequence Analysis-All Coding Exons)	TOP2B	TOP2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOP1MT geni (Tüm Gen Dizi Analizi)	TOP1MT gene (Sequence Analysis-All Coding Exons)	TOP1MT	TOP1MT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TONSL geni (Tüm Gen Dizi Analizi)	TONSL gene (Sequence Analysis-All Coding Exons)	TONSL	NFKBIL2, IKBR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM7 geni (Tüm Gen Dizi Analizi)	TOMM7 gene (Sequence Analysis-All Coding Exons)	TOMM7	TOMM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM6 geni (Tüm Gen Dizi Analizi)	TOMM6 gene (Sequence Analysis-All Coding Exons)	TOMM6	TOMM6, TOM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM5 geni (Tüm Gen Dizi Analizi)	TOMM5 gene (Sequence Analysis-All Coding Exons)	TOMM5	TOMM5, TOM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM40 geni (Tüm Gen Dizi Analizi)	TOMM40 gene (Sequence Analysis-All Coding Exons)	TOMM40	TOMM40, TOM40, PEREC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM34 geni (Tüm Gen Dizi Analizi)	TOMM34 gene (Sequence Analysis-All Coding Exons)	TOMM34	TOMM34, TOM34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM22 geni (Tüm Gen Dizi Analizi)	TOMM22 gene (Sequence Analysis-All Coding Exons)	TOMM22	TOMM22, TOM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOM1L2 geni (Tüm Gen Dizi Analizi)	TOM1L2 gene (Sequence Analysis-All Coding Exons)	TOM1L2	TOM1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOM1L1 geni (Tüm Gen Dizi Analizi)	TOM1L1 gene (Sequence Analysis-All Coding Exons)	TOM1L1	TOM1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOM1 geni (Tüm Gen Dizi Analizi)	TOM1 gene (Sequence Analysis-All Coding Exons)	TOM1	TOM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOLLIP geni (Tüm Gen Dizi Analizi)	TOLLIP gene (Sequence Analysis-All Coding Exons)	TOLLIP	TOLLIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOE1 geni (Tüm Gen Dizi Analizi)	TOE1 gene (Sequence Analysis-All Coding Exons)	TOE1	TOE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOB2 geni (Tüm Gen Dizi Analizi)	TOB2 gene (Sequence Analysis-All Coding Exons)	TOB2	TOB2, KIAA1663	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOB1 geni (Tüm Gen Dizi Analizi)	TOB1 gene (Sequence Analysis-All Coding Exons)	TOB1	TOB1, TOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNS4 geni (Tüm Gen Dizi Analizi)	TNS4 gene (Sequence Analysis-All Coding Exons)	TNS4	TNS4, CTEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNS3 geni (Tüm Gen Dizi Analizi)	TNS3 gene (Sequence Analysis-All Coding Exons)	TNS3	TNS3, TEM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNS2 geni (Tüm Gen Dizi Analizi)	TNS2 gene (Sequence Analysis-All Coding Exons)	TNS2	TNS2, TENC1, C1TEN, KIAA1075	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNS1 geni (Tüm Gen Dizi Analizi)	TNS1 gene (Sequence Analysis-All Coding Exons)	TNS1	TNS1, TNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNRC8C geni (Tüm Gen Dizi Analizi)	TNRC8C gene (Sequence Analysis-All Coding Exons)	TNRC8C	TNRC8C, KIAA1582	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNRC6B geni (Tüm Gen Dizi Analizi)	TNRC6B gene (Sequence Analysis-All Coding Exons)	TNRC6B	TNRC6B, KIAA1093	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNRC6A geni (Tüm Gen Dizi Analizi)	TNRC6A gene (Sequence Analysis-All Coding Exons)	TNRC6A	TNRC6A, GW182, KIAA1460	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNR geni (Tüm Gen Dizi Analizi)	TNR gene (Sequence Analysis-All Coding Exons)	TNR	TNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNPO2 geni (Tüm Gen Dizi Analizi)	TNPO2 gene (Sequence Analysis-All Coding Exons)	TNPO2	TNPO2, TRN2, KPNB2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNPO1 geni (Tüm Gen Dizi Analizi)	TNPO1 gene (Sequence Analysis-All Coding Exons)	TNPO1	TNPO1, KPNB2, MIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNP2 geni (Tüm Gen Dizi Analizi)	TNP2 gene (Sequence Analysis-All Coding Exons)	TNP2	TNP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNP1 geni (Tüm Gen Dizi Analizi)	TNP1 gene (Sequence Analysis-All Coding Exons)	TNP1	TNP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNNI1 geni (Tüm Gen Dizi Analizi)	TNNI1 gene (Sequence Analysis-All Coding Exons)	TNNI1	TNNI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TNNC2 geni (Tüm Gen Dizi Analizi)	TNNC2 gene (Sequence Analysis-All Coding Exons)	TNNC2	TNNC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNMD geni (Tüm Gen Dizi Analizi)	TNMD gene (Sequence Analysis-All Coding Exons)	TNMD	TNMD, TEM, CHM1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNKS1BP1 geni (Tüm Gen Dizi Analizi)	TNKS1BP1 gene (Sequence Analysis-All Coding Exons)	TNKS1BP1	TNKS1BP1, TAB182, KIAA1741	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNKS geni (Tüm Gen Dizi Analizi)	TNKS gene (Sequence Analysis-All Coding Exons)	TNKS	TNKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNK2 geni (Tüm Gen Dizi Analizi)	TNK2 gene (Sequence Analysis-All Coding Exons)	TNK2	TNK2, ACK, ACK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNK1 geni (Tüm Gen Dizi Analizi)	TNK1 gene (Sequence Analysis-All Coding Exons)	TNK1	TNK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNIP2 geni (Tüm Gen Dizi Analizi)	TNIP2 gene (Sequence Analysis-All Coding Exons)	TNIP2	TNIP2, ABIN2, FLIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNIP1 geni (Tüm Gen Dizi Analizi)	TNIP1 gene (Sequence Analysis-All Coding Exons)	TNIP1	TNIP1, NAF1, KIAA0113	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF9 geni (Tüm Gen Dizi Analizi)	TNFSF9 gene (Sequence Analysis-All Coding Exons)	TNFSF9	TNFSF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF8 geni (Tüm Gen Dizi Analizi)	TNFSF8 gene (Sequence Analysis-All Coding Exons)	TNFSF8	TNFSF8, CD30L, CD30LG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF18 geni (Tüm Gen Dizi Analizi)	TNFSF18 gene (Sequence Analysis-All Coding Exons)	TNFSF18	TNFSF18, AITRL, GITRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF15 geni (Tüm Gen Dizi Analizi)	TNFSF15 gene (Sequence Analysis-All Coding Exons)	TNFSF15	TNFSF15, TL1, VEGI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF14 geni (Tüm Gen Dizi Analizi)	TNFSF14 gene (Sequence Analysis-All Coding Exons)	TNFSF14	TNFSF14, HVEML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF13B geni (Tüm Gen Dizi Analizi)	TNFSF13B gene (Sequence Analysis-All Coding Exons)	TNFSF13B	TNFSF13B, BLYS, BAFF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
UBA3 geni (Tüm Gen Dizi Analizi)	UBA3 gene (Sequence Analysis-All Coding Exons)	UBA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF13 geni (Tüm Gen Dizi Analizi)	TNFSF13 gene (Sequence Analysis-All Coding Exons)	TNFSF13	TNFSF13, APRIL, TALL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF12 geni (Tüm Gen Dizi Analizi)	TNFSF12 gene (Sequence Analysis-All Coding Exons)	TNFSF12	TNFSF12, TWEAK, APO3L, DR3LG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF12 geni (Tüm Gen Dizi Analizi)	TNFSF12 gene (Sequence Analysis-All Coding Exons)	TNFSF12	TNFSF12, TWEAK, APO3L, DR3LG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFSF10 geni (Tüm Gen Dizi Analizi)	TNFSF10 gene (Sequence Analysis-All Coding Exons)	TNFSF10	TNFSF10, TRAIL, APO2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF9 geni (Tüm Gen Dizi Analizi)	TNFRSF9 gene (Sequence Analysis-All Coding Exons)	TNFRSF9	TNFRSF9, ILA, CD137	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF8 geni (Tüm Gen Dizi Analizi)	TNFRSF8 gene (Sequence Analysis-All Coding Exons)	TNFRSF8	TNFRSF8, CD30, D1S166E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF6B geni (Tüm Gen Dizi Analizi)	TNFRSF6B gene (Sequence Analysis-All Coding Exons)	TNFRSF6B	TNFRSF6B, DCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF25 geni (Tüm Gen Dizi Analizi)	TNFRSF25 gene (Sequence Analysis-All Coding Exons)	TNFRSF25	TNFRSF25, TNFRSF12, DR3, LARD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF1B geni (Tüm Gen Dizi Analizi)	TNFRSF1B gene (Sequence Analysis-All Coding Exons)	TNFRSF1B	TNFRSF1B, TNFR2, TNFBR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF1B geni (Tüm Gen Dizi Analizi)	TNFRSF1B gene (Sequence Analysis-All Coding Exons)	TNFRSF1B	TNFRSF1B, TNFR2, TNFBR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF19 geni (Tüm Gen Dizi Analizi)	TNFRSF19 gene (Sequence Analysis-All Coding Exons)	TNFRSF19	TAJ, TROY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF18 geni (Tüm Gen Dizi Analizi)	TNFRSF18 gene (Sequence Analysis-All Coding Exons)	TNFRSF18	TNFRSF18, AITR, GITR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF17 geni (Tüm Gen Dizi Analizi)	TNFRSF17 gene (Sequence Analysis-All Coding Exons)	TNFRSF17	TNFRSF17, BCMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF14 geni (Tüm Gen Dizi Analizi)	TNFRSF14 gene (Sequence Analysis-All Coding Exons)	TNFRSF14	TNFRSF14, HVEM, TR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF12A geni (Tüm Gen Dizi Analizi)	TNFRSF12A gene (Sequence Analysis-All Coding Exons)	TNFRSF12A	TNFRSF12A, FN14, TWEAKR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF10D geni (Tüm Gen Dizi Analizi)	TNFRSF10D gene (Sequence Analysis-All Coding Exons)	TNFRSF10D	TNFRSF10D, DCR2, TRAILR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF10C geni (Tüm Gen Dizi Analizi)	TNFRSF10C gene (Sequence Analysis-All Coding Exons)	TNFRSF10C	TNFRSF10C, DCR1, TRAILR3, TRID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF10A geni (Tüm Gen Dizi Analizi)	TNFRSF10A gene (Sequence Analysis-All Coding Exons)	TNFRSF10A	TNFRSF10A, DR4, TRAILR1, APO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP8L3 geni (Tüm Gen Dizi Analizi)	TNFAIP8L3 gene (Sequence Analysis-All Coding Exons)	TNFAIP8L3	TNFAIP8L3, TIPE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP8L2 geni (Tüm Gen Dizi Analizi)	TNFAIP8L2 gene (Sequence Analysis-All Coding Exons)	TNFAIP8L2	TNFAIP8L2, TIPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP8 geni (Tüm Gen Dizi Analizi)	TNFAIP8 gene (Sequence Analysis-All Coding Exons)	TNFAIP8	TNFAIP8, SCCS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP6 geni (Tüm Gen Dizi Analizi)	TNFAIP6 gene (Sequence Analysis-All Coding Exons)	TNFAIP6	TNFAIP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP2 geni (Tüm Gen Dizi Analizi)	TNFAIP2 gene (Sequence Analysis-All Coding Exons)	TNFAIP2	TNFAIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP1 geni (Tüm Gen Dizi Analizi)	TNFAIP1 gene (Sequence Analysis-All Coding Exons)	TNFAIP1	TNFAIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMX4 geni (Tüm Gen Dizi Analizi)	TMX4 gene (Sequence Analysis-All Coding Exons)	TMX4	TMX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMX3 geni (Tüm Gen Dizi Analizi)	TMX3 gene (Sequence Analysis-All Coding Exons)	TMX3	TMX3, TXNDC10, KIAA1830	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMX2 geni (Tüm Gen Dizi Analizi)	TMX2 gene (Sequence Analysis-All Coding Exons)	TMX2	TMX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMX1 geni (Tüm Gen Dizi Analizi)	TMX1 gene (Sequence Analysis-All Coding Exons)	TMX1	TXNDC1, TMX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMUB1 geni (Tüm Gen Dizi Analizi)	TMUB1 gene (Sequence Analysis-All Coding Exons)	TMUB1	TMUB1, DULP, SB144, C7orf21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMTC2 geni (Tüm Gen Dizi Analizi)	TMTC2 gene (Sequence Analysis-All Coding Exons)	TMTC2	TMTC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMTC1 geni (Tüm Gen Dizi Analizi)	TMTC1 gene (Sequence Analysis-All Coding Exons)	TMTC1	TMTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMSB4Y geni (Tüm Gen Dizi Analizi)	TMSB4Y gene (Sequence Analysis-All Coding Exons)	TMSB4Y	TMSB4Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMSB4X geni (Tüm Gen Dizi Analizi)	TMSB4X gene (Sequence Analysis-All Coding Exons)	TMSB4X	TMSB4X, TMSB4, TB4X, PTMB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMSB15A geni (Tüm Gen Dizi Analizi)	TMSB15A gene (Sequence Analysis-All Coding Exons)	TMSB15A	TMSB15A, TMSB15, TMSNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMSB10 geni (Tüm Gen Dizi Analizi)	TMSB10 gene (Sequence Analysis-All Coding Exons)	TMSB10	TMSB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS9 geni (Tüm Gen Dizi Analizi)	TMPRSS9 gene (Sequence Analysis-All Coding Exons)	TMPRSS9	TMPRSS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS5 geni (Tüm Gen Dizi Analizi)	TMPRSS5 gene (Sequence Analysis-All Coding Exons)	TMPRSS5	TMPRSS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS4 geni (Tüm Gen Dizi Analizi)	TMPRSS4 gene (Sequence Analysis-All Coding Exons)	TMPRSS4	TMPRSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS2 geni (Tüm Gen Dizi Analizi)	TMPRSS2 gene (Sequence Analysis-All Coding Exons)	TMPRSS2	TMPRSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXLNB geni (Tüm Gen Dizi Analizi)	TXLNB gene (Sequence Analysis-All Coding Exons)	TXLNB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TXLNA geni (Tüm Gen Dizi Analizi)	TXLNA gene (Sequence Analysis-All Coding Exons)	TXLNA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS13 geni (Tüm Gen Dizi Analizi)	TMPRSS13 gene (Sequence Analysis-All Coding Exons)	TMPRSS13	TMPRSS13, MSPL, MSPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS11E geni (Tüm Gen Dizi Analizi)	TMPRSS11E gene (Sequence Analysis-All Coding Exons)	TMPRSS11E	TMPRSS11E, DESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS11A geni (Tüm Gen Dizi Analizi)	TMPRSS11A gene (Sequence Analysis-All Coding Exons)	TMPRSS11A	TMPRSS11A, ECRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPO geni (Tüm Gen Dizi Analizi)	TMPO gene (Sequence Analysis-All Coding Exons)	TMPO	TMPO, LAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMOD4 geni (Tüm Gen Dizi Analizi)	TMOD4 gene (Sequence Analysis-All Coding Exons)	TMOD4	TMOD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMOD3 geni (Tüm Gen Dizi Analizi)	TMOD3 gene (Sequence Analysis-All Coding Exons)	TMOD3	TMOD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMOD2 geni (Tüm Gen Dizi Analizi)	TMOD2 gene (Sequence Analysis-All Coding Exons)	TMOD2	TMOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMOD1 geni (Tüm Gen Dizi Analizi)	TMOD1 gene (Sequence Analysis-All Coding Exons)	TMOD1	TMOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMIGD2 geni (Tüm Gen Dizi Analizi)	TMIGD2 gene (Sequence Analysis-All Coding Exons)	TMIGD2	TMIGD2, IGPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMF1 geni (Tüm Gen Dizi Analizi)	TMF1 gene (Sequence Analysis-All Coding Exons)	TMF1	TMF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM97 geni (Tüm Gen Dizi Analizi)	TMEM97 gene (Sequence Analysis-All Coding Exons)	TMEM97	TMEM97, MAC30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM9 geni (Tüm Gen Dizi Analizi)	TMEM9 gene (Sequence Analysis-All Coding Exons)	TMEM9	TMEM9, TMEM9A, DERM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM8C geni (Tüm Gen Dizi Analizi)	TMEM8C gene (Sequence Analysis-All Coding Exons)	TMEM8C	TMEM8C, MYOMAKER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM8B geni (Tüm Gen Dizi Analizi)	TMEM8B gene (Sequence Analysis-All Coding Exons)	TMEM8B	TMEM8B, NGX6, NAG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM87B geni (Tüm Gen Dizi Analizi)	TMEM87B gene (Sequence Analysis-All Coding Exons)	TMEM87B	TMEM87B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUT1 geni (Tüm Gen Dizi Analizi)	TUT1 gene (Sequence Analysis-All Coding Exons)	TUT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM79 geni (Tüm Gen Dizi Analizi)	TMEM79 gene (Sequence Analysis-All Coding Exons)	TMEM79	TMEM79, MATT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM74 geni (Tüm Gen Dizi Analizi)	TMEM74 gene (Sequence Analysis-All Coding Exons)	TMEM74	TMEM74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM65 geni (Tüm Gen Dizi Analizi)	TMEM65 gene (Sequence Analysis-All Coding Exons)	TMEM65	TMEM65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM59L geni (Tüm Gen Dizi Analizi)	TMEM59L gene (Sequence Analysis-All Coding Exons)	TMEM59L	TMEM59L, BSMAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM59 geni (Tüm Gen Dizi Analizi)	TMEM59 gene (Sequence Analysis-All Coding Exons)	TMEM59	TMEM59, DCF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM57 geni (Tüm Gen Dizi Analizi)	TMEM57 gene (Sequence Analysis-All Coding Exons)	TMEM57	TMEM57, FLJ10747	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM55A geni (Tüm Gen Dizi Analizi)	TMEM55A gene (Sequence Analysis-All Coding Exons)	TMEM55A	TMEM55A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM50A geni (Tüm Gen Dizi Analizi)	TMEM50A gene (Sequence Analysis-All Coding Exons)	TMEM50A	TMEM50A, SMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM47 geni (Tüm Gen Dizi Analizi)	TMEM47 gene (Sequence Analysis-All Coding Exons)	TMEM47	TMEM47, BCMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM45A geni (Tüm Gen Dizi Analizi)	TMEM45A gene (Sequence Analysis-All Coding Exons)	TMEM45A	TMEM45A, DERP7, DNAPTP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM38A geni (Tüm Gen Dizi Analizi)	TMEM38A gene (Sequence Analysis-All Coding Exons)	TMEM38A	TMEM38A, TRICA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM30CP geni (Tüm Gen Dizi Analizi)	TMEM30CP gene (Sequence Analysis-All Coding Exons)	TMEM30CP	TMEM30C, CDC50C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM30B geni (Tüm Gen Dizi Analizi)	TMEM30B gene (Sequence Analysis-All Coding Exons)	TMEM30B	TMEM30B, CDC50B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM30A geni (Tüm Gen Dizi Analizi)	TMEM30A gene (Sequence Analysis-All Coding Exons)	TMEM30A	TMEM30A, CDC50A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM27 geni (Tüm Gen Dizi Analizi)	TMEM27 gene (Sequence Analysis-All Coding Exons)	TMEM27	TMEM27, NX17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM261 geni (Tüm Gen Dizi Analizi)	TMEM261 gene (Sequence Analysis-All Coding Exons)	TMEM261	TMEM261, DMAC1, C9orf123	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM259 geni (Tüm Gen Dizi Analizi)	TMEM259 gene (Sequence Analysis-All Coding Exons)	TMEM259	C19orf6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM257 geni (Tüm Gen Dizi Analizi)	TMEM257 gene (Sequence Analysis-All Coding Exons)	TMEM257	CXorf1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM25 geni (Tüm Gen Dizi Analizi)	TMEM25 gene (Sequence Analysis-All Coding Exons)	TMEM25	TMEM25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM243 geni (Tüm Gen Dizi Analizi)	TMEM243 gene (Sequence Analysis-All Coding Exons)	TMEM243	TMEM243, C7orf23, MMTRAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM241 geni (Tüm Gen Dizi Analizi)	TMEM241 gene (Sequence Analysis-All Coding Exons)	TMEM241	TMEM241, C18orf45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TMEM230 geni (Tüm Gen Dizi Analizi)	TMEM230 gene (Sequence Analysis-All Coding Exons)	TMEM230	TMEM230, C20orf30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM214 geni (Tüm Gen Dizi Analizi)	TMEM214 gene (Sequence Analysis-All Coding Exons)	TMEM214	TMEM214	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM207 geni (Tüm Gen Dizi Analizi)	TMEM207 gene (Sequence Analysis-All Coding Exons)	TMEM207	TMEM207	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM205 geni (Tüm Gen Dizi Analizi)	TMEM205 gene (Sequence Analysis-All Coding Exons)	TMEM205	TMEM205	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM204 geni (Tüm Gen Dizi Analizi)	TMEM204 gene (Sequence Analysis-All Coding Exons)	TMEM204	TMEM204, C16orf30, CLP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM203 geni (Tüm Gen Dizi Analizi)	TMEM203 gene (Sequence Analysis-All Coding Exons)	TMEM203	TMEM203	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM2 geni (Tüm Gen Dizi Analizi)	TMEM2 gene (Sequence Analysis-All Coding Exons)	TMEM2	TMEM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM189 geni (Tüm Gen Dizi Analizi)	TMEM189 gene (Sequence Analysis-All Coding Exons)	TMEM189	TMEM189, KUA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM187 geni (Tüm Gen Dizi Analizi)	TMEM187 gene (Sequence Analysis-All Coding Exons)	TMEM187	TMEM187, CXorf12, DXS9878E, ITBA4, gene	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM185A geni (Tüm Gen Dizi Analizi)	TMEM185A gene (Sequence Analysis-All Coding Exons)	TMEM185A	FAM11A, FRAXF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM184C geni (Tüm Gen Dizi Analizi)	TMEM184C gene (Sequence Analysis-All Coding Exons)	TMEM184C	TMEM184C, TMEM34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM183B geni (Tüm Gen Dizi Analizi)	TMEM183B gene (Sequence Analysis-All Coding Exons)	TMEM183B	TMEM183B, C1ORF37DUP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TUBA1B geni (Tüm Gen Dizi Analizi)	TUBA1B gene (Sequence Analysis-All Coding Exons)	TUBA1B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM181 geni (Tüm Gen Dizi Analizi)	TMEM181 gene (Sequence Analysis-All Coding Exons)	TMEM181	TMEM181, GPR178, KIAA1423	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM18 geni (Tüm Gen Dizi Analizi)	TMEM18 gene (Sequence Analysis-All Coding Exons)	TMEM18	TMEM18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTYH3 geni (Tüm Gen Dizi Analizi)	TTYH3 gene (Sequence Analysis-All Coding Exons)	TTYH3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM176B geni (Tüm Gen Dizi Analizi)	TMEM176B gene (Sequence Analysis-All Coding Exons)	TMEM176B	LR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM176A geni (Tüm Gen Dizi Analizi)	TMEM176A gene (Sequence Analysis-All Coding Exons)	TMEM176A	TEM176A, HCA112, GS188	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM175 geni (Tüm Gen Dizi Analizi)	TMEM175 gene (Sequence Analysis-All Coding Exons)	TMEM175	TMEM175	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM174 geni (Tüm Gen Dizi Analizi)	TMEM174 gene (Sequence Analysis-All Coding Exons)	TMEM174	TMEM174	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM17 geni (Tüm Gen Dizi Analizi)	TMEM17 gene (Sequence Analysis-All Coding Exons)	TMEM17	TMEM17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM159 geni (Tüm Gen Dizi Analizi)	TMEM159 gene (Sequence Analysis-All Coding Exons)	TMEM159	TMEM159	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM150C geni (Tüm Gen Dizi Analizi)	TMEM150C gene (Sequence Analysis-All Coding Exons)	TMEM150C	TMEM150C, TTN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM150B geni (Tüm Gen Dizi Analizi)	TMEM150B gene (Sequence Analysis-All Coding Exons)	TMEM150B	TMEM150B, DRAM3, TTN2, TMEM224	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM150A geni (Tüm Gen Dizi Analizi)	TMEM150A gene (Sequence Analysis-All Coding Exons)	TMEM150A	TMEM150A, TMEM150	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM14C geni (Tüm Gen Dizi Analizi)	TMEM14C gene (Sequence Analysis-All Coding Exons)	TMEM14C	TMEM14C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM14A geni (Tüm Gen Dizi Analizi)	TMEM14A gene (Sequence Analysis-All Coding Exons)	TMEM14A	TMEM14A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM139 geni (Tüm Gen Dizi Analizi)	TMEM139 gene (Sequence Analysis-All Coding Exons)	TMEM139	TMEM139	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM135 geni (Tüm Gen Dizi Analizi)	TMEM135 gene (Sequence Analysis-All Coding Exons)	TMEM135	TMEM135, PMP52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM132E geni (Tüm Gen Dizi Analizi)	TMEM132E gene (Sequence Analysis-All Coding Exons)	TMEM132E	TMEM132E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM132D geni (Tüm Gen Dizi Analizi)	TMEM132D gene (Sequence Analysis-All Coding Exons)	TMEM132D	TMEM132D, KIAA1944, MOLT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM132A geni (Tüm Gen Dizi Analizi)	TMEM132A gene (Sequence Analysis-All Coding Exons)	TMEM132A	TMEM132A, KIAA15B3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM131 geni (Tüm Gen Dizi Analizi)	TMEM131 gene (Sequence Analysis-All Coding Exons)	TMEM131	TMEM131, KIAA0257, CC28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM129 geni (Tüm Gen Dizi Analizi)	TMEM129 gene (Sequence Analysis-All Coding Exons)	TMEM129	TMEM129	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM123 geni (Tüm Gen Dizi Analizi)	TMEM123 gene (Sequence Analysis-All Coding Exons)	TMEM123	TMEM123, PORIMIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM120B geni (Tüm Gen Dizi Analizi)	TMEM120B gene (Sequence Analysis-All Coding Exons)	TMEM120B	TMEM120B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM120A geni (Tüm Gen Dizi Analizi)	TMEM120A gene (Sequence Analysis-All Coding Exons)	TMEM120A	TMEM120A, TMPIT, NET29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM115 geni (Tüm Gen Dizi Analizi)	TMEM115 gene (Sequence Analysis-All Coding Exons)	TMEM115	TMEM115, PL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM114 geni (Tüm Gen Dizi Analizi)	TMEM114 gene (Sequence Analysis-All Coding Exons)	TMEM114	TMEM114	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM110 geni (Tüm Gen Dizi Analizi)	TMEM110 gene (Sequence Analysis-All Coding Exons)	TMEM110	TMEM110, STIMATE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM108 geni (Tüm Gen Dizi Analizi)	TMEM108 gene (Sequence Analysis-All Coding Exons)	TMEM108	TMEM108, RTLN, KIAA1690	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTF1 geni (Tüm Gen Dizi Analizi)	TTF1 gene (Sequence Analysis-All Coding Exons)	TTF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM107 geni (Tüm Gen Dizi Analizi)	TMEM107 gene (Sequence Analysis-All Coding Exons)	TMEM107	TMEM107	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM106B geni (Tüm Gen Dizi Analizi)	TMEM106B gene (Sequence Analysis-All Coding Exons)	TMEM106B	TMEM106B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM102 geni (Tüm Gen Dizi Analizi)	TMEM102 gene (Sequence Analysis-All Coding Exons)	TMEM102	TMEM102, CBAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM100 geni (Tüm Gen Dizi Analizi)	TMEM100 gene (Sequence Analysis-All Coding Exons)	TMEM100	TMEM100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEFF2 geni (Tüm Gen Dizi Analizi)	TMEFF2 gene (Sequence Analysis-All Coding Exons)	TMEFF2	TMEFF2, HPP1, TR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEFF1 geni (Tüm Gen Dizi Analizi)	TMEFF1 gene (Sequence Analysis-All Coding Exons)	TMEFF1	TMEFF1, C9ORF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMED5 geni (Tüm Gen Dizi Analizi)	TMED5 gene (Sequence Analysis-All Coding Exons)	TMED5	TMED5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMED4 geni (Tüm Gen Dizi Analizi)	TMED4 gene (Sequence Analysis-All Coding Exons)	TMED4	TMED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMED10 geni (Tüm Gen Dizi Analizi)	TMED10 gene (Sequence Analysis-All Coding Exons)	TMED10	TMED10, TMP21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMED1 geni (Tüm Gen Dizi Analizi)	TMED1 gene (Sequence Analysis-All Coding Exons)	TMED1	TMED1, IL1RL1LG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMCO3 geni (Tüm Gen Dizi Analizi)	TMCO3 gene (Sequence Analysis-All Coding Exons)	TMCO3	TMCO3, C13orf11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMCC1 geni (Tüm Gen Dizi Analizi)	TMCC1 gene (Sequence Analysis-All Coding Exons)	TMCC1	TMCC1, KIAA0779	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMC7 geni (Tüm Gen Dizi Analizi)	TMC7 gene (Sequence Analysis-All Coding Exons)	TMC7	TMC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TTC21A geni (Tüm Gen Dizi Analizi)	TTC21A gene (Sequence Analysis-All Coding Exons)	TTC21A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMC5 geni (Tüm Gen Dizi Analizi)	TMC5 gene (Sequence Analysis-All Coding Exons)	TMC5	TMC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMC4 geni (Tüm Gen Dizi Analizi)	TMC4 gene (Sequence Analysis-All Coding Exons)	TMC4	TMC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMC3 geni (Tüm Gen Dizi Analizi)	TMC3 gene (Sequence Analysis-All Coding Exons)	TMC3	TMC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMC2 geni (Tüm Gen Dizi Analizi)	TMC2 gene (Sequence Analysis-All Coding Exons)	TMC2	TMC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMBIM6 geni (Tüm Gen Dizi Analizi)	TMBIM6 gene (Sequence Analysis-All Coding Exons)	TMBIM6	TMBIM6, TEGT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMBIM4 geni (Tüm Gen Dizi Analizi)	TMBIM4 gene (Sequence Analysis-All Coding Exons)	TMBIM4	TMBIM4, GAAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMBIM1 geni (Tüm Gen Dizi Analizi)	TMBIM1 gene (Sequence Analysis-All Coding Exons)	TMBIM1	TMBIM1, RECS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMA7 geni (Tüm Gen Dizi Analizi)	TMA7 gene (Sequence Analysis-All Coding Exons)	TMA7	TMA7, HSPC016	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM9SF3 geni (Tüm Gen Dizi Analizi)	TM9SF3 gene (Sequence Analysis-All Coding Exons)	TM9SF3	TM9SF3, SMBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM9SF2 geni (Tüm Gen Dizi Analizi)	TM9SF2 gene (Sequence Analysis-All Coding Exons)	TM9SF2	TM9SF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM7SF3 geni (Tüm Gen Dizi Analizi)	TM7SF3 gene (Sequence Analysis-All Coding Exons)	TM7SF3	TM7SF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM7SF2 geni (Tüm Gen Dizi Analizi)	TM7SF2 gene (Sequence Analysis-All Coding Exons)	TM7SF2	TM7SF2, ANG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM6SF2 geni (Tüm Gen Dizi Analizi)	TM6SF2 gene (Sequence Analysis-All Coding Exons)	TM6SF2	TM6SF2, KIAA1926	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM6SF1 geni (Tüm Gen Dizi Analizi)	TM6SF1 gene (Sequence Analysis-All Coding Exons)	TM6SF1	TM6SF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM4SF5 geni (Tüm Gen Dizi Analizi)	TM4SF5 gene (Sequence Analysis-All Coding Exons)	TM4SF5	TM4SF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM4SF4 geni (Tüm Gen Dizi Analizi)	TM4SF4 gene (Sequence Analysis-All Coding Exons)	TM4SF4	TM4SF4, IL1MP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM4SF1 geni (Tüm Gen Dizi Analizi)	TM4SF1 gene (Sequence Analysis-All Coding Exons)	TM4SF1	TM4SF1, M3S1, TAAL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM2D3 geni (Tüm Gen Dizi Analizi)	TM2D3 gene (Sequence Analysis-All Coding Exons)	TM2D3	TM2D3, BLP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM2D2 geni (Tüm Gen Dizi Analizi)	TM2D2 gene (Sequence Analysis-All Coding Exons)	TM2D2	TM2D2, BLP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TM2D1 geni (Tüm Gen Dizi Analizi)	TM2D1 gene (Sequence Analysis-All Coding Exons)	TM2D1	TM2D1, BBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLX3 geni (Tüm Gen Dizi Analizi)	TLX3 gene (Sequence Analysis-All Coding Exons)	TLX3	TLX3, HOX11L2, RNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLX2 geni (Tüm Gen Dizi Analizi)	TLX2 gene (Sequence Analysis-All Coding Exons)	TLX2	TLX2, HOX11L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLX1NB geni (Tüm Gen Dizi Analizi)	TLX1NB gene (Sequence Analysis-All Coding Exons)	TLX1NB	TD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLX1 geni (Tüm Gen Dizi Analizi)	TLX1 gene (Sequence Analysis-All Coding Exons)	TLX1	TLX1, HOX11, TCL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR9 geni (Tüm Gen Dizi Analizi)	TLR9 gene (Sequence Analysis-All Coding Exons)	TLR9	TLR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR8 geni (Tüm Gen Dizi Analizi)	TLR8 gene (Sequence Analysis-All Coding Exons)	TLR8	TLR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR7 geni (Tüm Gen Dizi Analizi)	TLR7 gene (Sequence Analysis-All Coding Exons)	TLR7	TLR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR6 geni (Tüm Gen Dizi Analizi)	TLR6 gene (Sequence Analysis-All Coding Exons)	TLR6	TLR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR4 geni (Tüm Gen Dizi Analizi)	TLR4 gene (Sequence Analysis-All Coding Exons)	TLR4	TLR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLN2 geni (Tüm Gen Dizi Analizi)	TLN2 gene (Sequence Analysis-All Coding Exons)	TLN2	TLN2, KIAA0320	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLN1 geni (Tüm Gen Dizi Analizi)	TLN1 gene (Sequence Analysis-All Coding Exons)	TLN1	TLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLL2 geni (Tüm Gen Dizi Analizi)	TLL2 gene (Sequence Analysis-All Coding Exons)	TLL2	TLL2, KIAA0932	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLK2 geni (Tüm Gen Dizi Analizi)	TLK2 gene (Sequence Analysis-All Coding Exons)	TLK2	TLK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLE3 geni (Tüm Gen Dizi Analizi)	TLE3 gene (Sequence Analysis-All Coding Exons)	TLE3	TLE3, ESG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLE2 geni (Tüm Gen Dizi Analizi)	TLE2 gene (Sequence Analysis-All Coding Exons)	TLE2	TLE2, ESG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLE1 geni (Tüm Gen Dizi Analizi)	TLE1 gene (Sequence Analysis-All Coding Exons)	TLE1	TLE1, ESG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TKTL1 geni (Tüm Gen Dizi Analizi)	TKTL1 gene (Sequence Analysis-All Coding Exons)	TKTL1	TKTL1, TKT2, TKR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TKT geni (Tüm Gen Dizi Analizi)	TKT gene (Sequence Analysis-All Coding Exons)	TKT	TKT, SDDHD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TKFC geni (Tüm Gen Dizi Analizi)	TKFC gene (Sequence Analysis-All Coding Exons)	TKFC	DAK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TK1 geni (Tüm Gen Dizi Analizi)	TK1 gene (Sequence Analysis-All Coding Exons)	TK1	TK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TJP3 geni (Tüm Gen Dizi Analizi)	TJP3 gene (Sequence Analysis-All Coding Exons)	TJP3	TJP3, ZO3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TJP1 geni (Tüm Gen Dizi Analizi)	TJP1 gene (Sequence Analysis-All Coding Exons)	TJP1	TJP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TJAP1 geni (Tüm Gen Dizi Analizi)	TJAP1 gene (Sequence Analysis-All Coding Exons)	TJAP1	TJAP1, PILT, TJP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIPRL geni (Tüm Gen Dizi Analizi)	TIPRL gene (Sequence Analysis-All Coding Exons)	TIPRL	TIPRL, TIP41, TIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TSKS geni (Tüm Gen Dizi Analizi)	TSKS gene (Sequence Analysis-All Coding Exons)	TSKS		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIPIN geni (Tüm Gen Dizi Analizi)	TIPIN gene (Sequence Analysis-All Coding Exons)	TIPIN	TIPIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIPARP geni (Tüm Gen Dizi Analizi)	TIPARP gene (Sequence Analysis-All Coding Exons)	TIPARP	TIPARP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TINAGL1 geni (Tüm Gen Dizi Analizi)	TINAGL1 gene (Sequence Analysis-All Coding Exons)	TINAGL1	TINAGL1, TINAGRP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TINAG geni (Tüm Gen Dizi Analizi)	TINAG gene (Sequence Analysis-All Coding Exons)	TINAG	TINAG, TIN1, TIN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMP4 geni (Tüm Gen Dizi Analizi)	TIMP4 gene (Sequence Analysis-All Coding Exons)	TIMP4	TIMP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMP2 geni (Tüm Gen Dizi Analizi)	TIMP2 gene (Sequence Analysis-All Coding Exons)	TIMP2	TIMP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMP1 geni (Tüm Gen Dizi Analizi)	TIMP1 gene (Sequence Analysis-All Coding Exons)	TIMP1	TIMP1, TIMP, EPA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMMDC1 geni (Tüm Gen Dizi Analizi)	TIMMDC1 gene (Sequence Analysis-All Coding Exons)	TIMMDC1	TIMMDC1, C3orf1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM9 geni (Tüm Gen Dizi Analizi)	TIMM9 gene (Sequence Analysis-All Coding Exons)	TIMM9	TIMM9, TIM9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM50 geni (Tüm Gen Dizi Analizi)	TIMM50 gene (Sequence Analysis-All Coding Exons)	TIMM50	TIMM50, TIM50	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM44 geni (Tüm Gen Dizi Analizi)	TIMM44 gene (Sequence Analysis-All Coding Exons)	TIMM44	TIMM44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM29 geni (Tüm Gen Dizi Analizi)	TIMM29 gene (Sequence Analysis-All Coding Exons)	TIMM29	TIMM29, C19orf52	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM23 geni (Tüm Gen Dizi Analizi)	TIMM23 gene (Sequence Analysis-All Coding Exons)	TIMM23	TIMM23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM22 geni (Tüm Gen Dizi Analizi)	TIMM22 gene (Sequence Analysis-All Coding Exons)	TIMM22	TIMM22, TIM22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM21 geni (Tüm Gen Dizi Analizi)	TIMM21 gene (Sequence Analysis-All Coding Exons)	TIMM21	TIMM21, TIM21	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM17B geni (Tüm Gen Dizi Analizi)	TIMM17B gene (Sequence Analysis-All Coding Exons)	TIMM17B	TIMM17B, TIM17B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM17A geni (Tüm Gen Dizi Analizi)	TIMM17A gene (Sequence Analysis-All Coding Exons)	TIMM17A	TIMM17A, TIM17A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM13 geni (Tüm Gen Dizi Analizi)	TIMM13 gene (Sequence Analysis-All Coding Exons)	TIMM13	TIMM13	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM10B geni (Tüm Gen Dizi Analizi)	TIMM10B gene (Sequence Analysis-All Coding Exons)	TIMM10B	FXC1, TIMM10B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM10 geni (Tüm Gen Dizi Analizi)	TIMM10 gene (Sequence Analysis-All Coding Exons)	TIMM10	TIMM10, TIM10A, TIM10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMELESS geni (Tüm Gen Dizi Analizi)	TIMELESS gene (Sequence Analysis-All Coding Exons)	TIMELESS	TIMELESS, TIM, TIM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMD4 geni (Tüm Gen Dizi Analizi)	TIMD4 gene (Sequence Analysis-All Coding Exons)	TIMD4	TIMED4, TIM4, SMUCKLER	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIGIT geni (Tüm Gen Dizi Analizi)	TIGIT gene (Sequence Analysis-All Coding Exons)	TIGIT	TIGIT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIGD7 geni (Tüm Gen Dizi Analizi)	TIGD7 gene (Sequence Analysis-All Coding Exons)	TIGD7	TIGD7, SANCHO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIGD2 geni (Tüm Gen Dizi Analizi)	TIGD2 gene (Sequence Analysis-All Coding Exons)	TIGD2	TIGD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIGD1 geni (Tüm Gen Dizi Analizi)	TIGD1 gene (Sequence Analysis-All Coding Exons)	TIGD1	TIGD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIGAR geni (Tüm Gen Dizi Analizi)	TIGAR gene (Sequence Analysis-All Coding Exons)	TIGAR	TIGAR, C12orf5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIFAB geni (Tüm Gen Dizi Analizi)	TIFAB gene (Sequence Analysis-All Coding Exons)	TIFAB	TIFAB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIE1 geni (Tüm Gen Dizi Analizi)	TIE1 gene (Sequence Analysis-All Coding Exons)	TIE1	TIE, JTK14, TIE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TICRR geni (Tüm Gen Dizi Analizi)	TICRR gene (Sequence Analysis-All Coding Exons)	TICRR	C15orf42, TRESLIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TICAM2 geni (Tüm Gen Dizi Analizi)	TICAM2 gene (Sequence Analysis-All Coding Exons)	TICAM2	TIRP, TRAM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIAM2 geni (Tüm Gen Dizi Analizi)	TIAM2 gene (Sequence Analysis-All Coding Exons)	TIAM2	TIAM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIAM1 geni (Tüm Gen Dizi Analizi)	TIAM1 gene (Sequence Analysis-All Coding Exons)	TIAM1	TIAM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIAL1 geni (Tüm Gen Dizi Analizi)	TIAL1 gene (Sequence Analysis-All Coding Exons)	TIAL1	TIAL1, TIAR, TCBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIAF1 geni (Tüm Gen Dizi Analizi)	TIAF1 gene (Sequence Analysis-All Coding Exons)	TIAF1	TIAF1, MYO18A, MYSPDZ, M43N, SPR210, KIAA0216	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THYN1 geni (Tüm Gen Dizi Analizi)	THYN1 gene (Sequence Analysis-All Coding Exons)	THYN1	THYN1, HSPC144, THY28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THY1 geni (Tüm Gen Dizi Analizi)	THY1 gene (Sequence Analysis-All Coding Exons)	THY1	THY1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THUMPD2 geni (Tüm Gen Dizi Analizi)	THUMPD2 gene (Sequence Analysis-All Coding Exons)	THUMPD2	THUMPD2, C2orf8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THUMPD1 geni (Tüm Gen Dizi Analizi)	THUMPD1 gene (Sequence Analysis-All Coding Exons)	THUMPD1	THUMPD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THTPA geni (Tüm Gen Dizi Analizi)	THTPA gene (Sequence Analysis-All Coding Exons)	THTPA	THTPA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THSD7A geni (Tüm Gen Dizi Analizi)	THSD7A gene (Sequence Analysis-All Coding Exons)	THSD7A	THSD7A, KIAA0960	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THSD4 geni (Tüm Gen Dizi Analizi)	THSD4 gene (Sequence Analysis-All Coding Exons)	THSD4	THSD4, ADAMTSL6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THSD1 geni (Tüm Gen Dizi Analizi)	THSD1 gene (Sequence Analysis-All Coding Exons)	THSD1	THSD1, TMTSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THRSP geni (Tüm Gen Dizi Analizi)	THRSP gene (Sequence Analysis-All Coding Exons)	THRSP	THRSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THRIL geni (Tüm Gen Dizi Analizi)	THRIL gene (Sequence Analysis-All Coding Exons)	THRIL	THRIL, LINC1992, BRIP1AS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THRAP3 geni (Tüm Gen Dizi Analizi)	THRAP3 gene (Sequence Analysis-All Coding Exons)	THRAP3	THRAP3, TRAP150	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THOP1 geni (Tüm Gen Dizi Analizi)	THOP1 gene (Sequence Analysis-All Coding Exons)	THOP1	THOP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THOC7 geni (Tüm Gen Dizi Analizi)	THOC7 gene (Sequence Analysis-All Coding Exons)	THOC7	THOC7, NIF3L1BP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THOC5 geni (Tüm Gen Dizi Analizi)	THOC5 gene (Sequence Analysis-All Coding Exons)	THOC5	THOC5, FMIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THOC3 geni (Tüm Gen Dizi Analizi)	THOC3 gene (Sequence Analysis-All Coding Exons)	THOC3	THOC3, TEX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THOC1 geni (Tüm Gen Dizi Analizi)	THOC1 gene (Sequence Analysis-All Coding Exons)	THOC1	THOC1, HPR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THNSL2 geni (Tüm Gen Dizi Analizi)	THNSL2 gene (Sequence Analysis-All Coding Exons)	THNSL2	THNSL2, THS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THNSL1 geni (Tüm Gen Dizi Analizi)	THNSL1 gene (Sequence Analysis-All Coding Exons)	THNSL1	THNSL1, TSH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THEMIS geni (Tüm Gen Dizi Analizi)	THEMIS gene (Sequence Analysis-All Coding Exons)	THEMIS	THEMIS, GASP, SPOT, C6ORF190	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THEM5 geni (Tüm Gen Dizi Analizi)	THEM5 gene (Sequence Analysis-All Coding Exons)	THEM5	THEM5, ACOT15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THEM4 geni (Tüm Gen Dizi Analizi)	THEM4 gene (Sequence Analysis-All Coding Exons)	THEM4	CTMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THEG geni (Tüm Gen Dizi Analizi)	THEG gene (Sequence Analysis-All Coding Exons)	THEG	THEG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THBS3 geni (Tüm Gen Dizi Analizi)	THBS3 gene (Sequence Analysis-All Coding Exons)	THBS3	THBS3, TSP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THBS1 geni (Tüm Gen Dizi Analizi)	THBS1 gene (Sequence Analysis-All Coding Exons)	THBS1	THBS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP9 geni (Tüm Gen Dizi Analizi)	THAP9 gene (Sequence Analysis-All Coding Exons)	THAP9	THAP9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP8 geni (Tüm Gen Dizi Analizi)	THAP8 gene (Sequence Analysis-All Coding Exons)	THAP8	THAP8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP6 geni (Tüm Gen Dizi Analizi)	THAP6 gene (Sequence Analysis-All Coding Exons)	THAP6	THAP6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TROAP geni (Tüm Gen Dizi Analizi)	TROAP gene (Sequence Analysis-All Coding Exons)	TROAP		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP5 geni (Tüm Gen Dizi Analizi)	THAP5 gene (Sequence Analysis-All Coding Exons)	THAP5	THAP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP4 geni (Tüm Gen Dizi Analizi)	THAP4 gene (Sequence Analysis-All Coding Exons)	THAP4	THAP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP3 geni (Tüm Gen Dizi Analizi)	THAP3 gene (Sequence Analysis-All Coding Exons)	THAP3	THAP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP2 geni (Tüm Gen Dizi Analizi)	THAP2 gene (Sequence Analysis-All Coding Exons)	THAP2	THAP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP12 geni (Tüm Gen Dizi Analizi)	THAP12 gene (Sequence Analysis-All Coding Exons)	THAP12	PRKRIR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP11 geni (Tüm Gen Dizi Analizi)	THAP11 gene (Sequence Analysis-All Coding Exons)	THAP11	THAP11, RONIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP10 geni (Tüm Gen Dizi Analizi)	THAP10 gene (Sequence Analysis-All Coding Exons)	THAP10	THAP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THADA geni (Tüm Gen Dizi Analizi)	THADA gene (Sequence Analysis-All Coding Exons)	THADA	THADA, KIAA1767	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGS1 geni (Tüm Gen Dizi Analizi)	TGS1 gene (Sequence Analysis-All Coding Exons)	TGS1	TGS1, NCOABP, PIMT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGOLN2 geni (Tüm Gen Dizi Analizi)	TGOLN2 gene (Sequence Analysis-All Coding Exons)	TGOLN2	TGOLN2, TGN46, TGN38	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGM7 geni (Tüm Gen Dizi Analizi)	TGM7 gene (Sequence Analysis-All Coding Exons)	TGM7	TGM7, TGMZ	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGM4 geni (Tüm Gen Dizi Analizi)	TGM4 gene (Sequence Analysis-All Coding Exons)	TGM4	TGM4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGM2 geni (Tüm Gen Dizi Analizi)	TGM2 gene (Sequence Analysis-All Coding Exons)	TGM2	TGM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGIF2LY geni (Tüm Gen Dizi Analizi)	TGIF2LY gene (Sequence Analysis-All Coding Exons)	TGIF2LY	TGIF2LY, TGIFLY	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGIF2LX geni (Tüm Gen Dizi Analizi)	TGIF2LX gene (Sequence Analysis-All Coding Exons)	TGIF2LX	TGIF2LX, TGIFLX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGIF2 geni (Tüm Gen Dizi Analizi)	TGIF2 gene (Sequence Analysis-All Coding Exons)	TGIF2	TGIF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGFBRAP1 geni (Tüm Gen Dizi Analizi)	TGFBRAP1 gene (Sequence Analysis-All Coding Exons)	TGFBRAP1	TGFBRAP1, TRAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGFBR3 geni (Tüm Gen Dizi Analizi)	TGFBR3 gene (Sequence Analysis-All Coding Exons)	TGFBR3	TGFBR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGFB1I1 geni (Tüm Gen Dizi Analizi)	TGFB1I1 gene (Sequence Analysis-All Coding Exons)	TGFB1I1	TGFB1I1, ARA55, HIC5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TGFA geni (Tüm Gen Dizi Analizi)	TGFA gene (Sequence Analysis-All Coding Exons)	TGFA	TGFA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFPI2 geni (Tüm Gen Dizi Analizi)	TFPI2 gene (Sequence Analysis-All Coding Exons)	TFPI2	TFPI2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFPI geni (Tüm Gen Dizi Analizi)	TFPI gene (Sequence Analysis-All Coding Exons)	TFPI	TFPI, LACI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TFIP11 geni (Tüm Gen Dizi Analizi)	TFIP11 gene (Sequence Analysis-All Coding Exons)	TFIP11	TFIP11, TIP39, NTR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFF3 geni (Tüm Gen Dizi Analizi)	TFF3 gene (Sequence Analysis-All Coding Exons)	TFF3	TFF3, ITF	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFF2 geni (Tüm Gen Dizi Analizi)	TFF2 gene (Sequence Analysis-All Coding Exons)	TFF2	TFF2, SML1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFF1 geni (Tüm Gen Dizi Analizi)	TFF1 gene (Sequence Analysis-All Coding Exons)	TFF1	TFF1, BCEI	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFEC geni (Tüm Gen Dizi Analizi)	TFEC gene (Sequence Analysis-All Coding Exons)	TFEC	TFEC, TFECL	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFEB geni (Tüm Gen Dizi Analizi)	TFEB gene (Sequence Analysis-All Coding Exons)	TFEB	TFEB, TCFEB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFDP3 geni (Tüm Gen Dizi Analizi)	TFDP3 gene (Sequence Analysis-All Coding Exons)	TFDP3	TFDP3, HCA661, CT30, DP4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFDP2 geni (Tüm Gen Dizi Analizi)	TFDP2 gene (Sequence Analysis-All Coding Exons)	TFDP2	TFDP2, DP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFDP1 geni (Tüm Gen Dizi Analizi)	TFDP1 gene (Sequence Analysis-All Coding Exons)	TFDP1	TFDP1, DP1, DRTF1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFCP2L1 geni (Tüm Gen Dizi Analizi)	TFCP2L1 gene (Sequence Analysis-All Coding Exons)	TFCP2L1	TFCP2L1, CRTR1, LBP9	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFCP2 geni (Tüm Gen Dizi Analizi)	TFCP2 gene (Sequence Analysis-All Coding Exons)	TFCP2	TFCP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFB2M geni (Tüm Gen Dizi Analizi)	TFB2M gene (Sequence Analysis-All Coding Exons)	TFB2M	TFB2M	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFB1M geni (Tüm Gen Dizi Analizi)	TFB1M gene (Sequence Analysis-All Coding Exons)	TFB1M	TFB1M, CGI75	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFAP4 geni (Tüm Gen Dizi Analizi)	TFAP4 gene (Sequence Analysis-All Coding Exons)	TFAP4	TFAP4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFAP2E geni (Tüm Gen Dizi Analizi)	TFAP2E gene (Sequence Analysis-All Coding Exons)	TFAP2E	TFAP2E	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFAP2D geni (Tüm Gen Dizi Analizi)	TFAP2D gene (Sequence Analysis-All Coding Exons)	TFAP2D	TFAP2D, TFAP2BL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFAP2C geni (Tüm Gen Dizi Analizi)	TFAP2C gene (Sequence Analysis-All Coding Exons)	TFAP2C	TFAP2C	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX28 geni (Tüm Gen Dizi Analizi)	TEX28 gene (Sequence Analysis-All Coding Exons)	TEX28	TEX28	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX19 geni (Tüm Gen Dizi Analizi)	TEX19 gene (Sequence Analysis-All Coding Exons)	TEX19	TEX19	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX15 geni (Tüm Gen Dizi Analizi)	TEX15 gene (Sequence Analysis-All Coding Exons)	TEX15	TEX15	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX14 geni (Tüm Gen Dizi Analizi)	TEX14 gene (Sequence Analysis-All Coding Exons)	TEX14	TEX14	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX13B geni (Tüm Gen Dizi Analizi)	TEX13B gene (Sequence Analysis-All Coding Exons)	TEX13B	TEX13B	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX13A geni (Tüm Gen Dizi Analizi)	TEX13A gene (Sequence Analysis-All Coding Exons)	TEX13A	TEX13A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX12 geni (Tüm Gen Dizi Analizi)	TEX12 gene (Sequence Analysis-All Coding Exons)	TEX12	TEX12	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX101 geni (Tüm Gen Dizi Analizi)	TEX101 gene (Sequence Analysis-All Coding Exons)	TEX101	TEX101, SGRG, TES101RP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEX10 geni (Tüm Gen Dizi Analizi)	TEX10 gene (Sequence Analysis-All Coding Exons)	TEX10	TEX10	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TET3 geni (Tüm Gen Dizi Analizi)	TET3 gene (Sequence Analysis-All Coding Exons)	TET3	TET3, KIAA0401	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TET1 geni (Tüm Gen Dizi Analizi)	TET1 gene (Sequence Analysis-All Coding Exons)	TET1	TET1, CXXC6, LCX, KIAA1676	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TESPA1 geni (Tüm Gen Dizi Analizi)	TESPA1 gene (Sequence Analysis-All Coding Exons)	TESPA1	TESPA1, KIAA0748	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TESMIN geni (Tüm Gen Dizi Analizi)	TESMIN gene (Sequence Analysis-All Coding Exons)	TESMIN	MTLS, TESMIN	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TESK2 geni (Tüm Gen Dizi Analizi)	TESK2 gene (Sequence Analysis-All Coding Exons)	TESK2	TESK2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TESK1 geni (Tüm Gen Dizi Analizi)	TESK1 gene (Sequence Analysis-All Coding Exons)	TESK1	TESK1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TESC geni (Tüm Gen Dizi Analizi)	TESC gene (Sequence Analysis-All Coding Exons)	TESC	TESC, TSC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TES geni (Tüm Gen Dizi Analizi)	TES gene (Sequence Analysis-All Coding Exons)	TES	TES	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TERF2IP geni (Tüm Gen Dizi Analizi)	TERF2IP gene (Sequence Analysis-All Coding Exons)	TERF2IP	TERF2IP, RAP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TERF2 geni (Tüm Gen Dizi Analizi)	TERF2 gene (Sequence Analysis-All Coding Exons)	TERF2	TERF2, TRF2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TERF1 geni (Tüm Gen Dizi Analizi)	TERF1 gene (Sequence Analysis-All Coding Exons)	TERF1	TERF1, TRF1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TERB2 geni (Tüm Gen Dizi Analizi)	TERB2 gene (Sequence Analysis-All Coding Exons)	TERB2	TERB2, C15orf43	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM29 geni (Tüm Gen Dizi Analizi)	TRIM29 gene (Sequence Analysis-All Coding Exons)	TRIM29		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIM28 geni (Tüm Gen Dizi Analizi)	TRIM28 gene (Sequence Analysis-All Coding Exons)	TRIM28		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TERB1 geni (Tüm Gen Dizi Analizi)	TERB1 gene (Sequence Analysis-All Coding Exons)	TERB1	TERB1, CCDC79	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEPP geni (Tüm Gen Dizi Analizi)	TEPP gene (Sequence Analysis-All Coding Exons)	TEPP	TEPP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEP1 geni (Tüm Gen Dizi Analizi)	TEP1 gene (Sequence Analysis-All Coding Exons)	TEP1	TEP1, TP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TENM4 geni (Tüm Gen Dizi Analizi)	TENM4 gene (Sequence Analysis-All Coding Exons)	TENM4	TENM4, ODZ4, TNM4, DOC4, KIAA1302, ETM5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TENM2 geni (Tüm Gen Dizi Analizi)	TENM2 gene (Sequence Analysis-All Coding Exons)	TENM2	TENM2, ODZ2, TNM2, KIAA1127	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TENM1 geni (Tüm Gen Dizi Analizi)	TENM1 gene (Sequence Analysis-All Coding Exons)	TENM1	TENM1, ODZ1, TNM1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEN1 geni (Tüm Gen Dizi Analizi)	TEN1 gene (Sequence Analysis-All Coding Exons)	TEN1	TEN1, C17orf106	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TELO2 geni (Tüm Gen Dizi Analizi)	TELO2 gene (Sequence Analysis-All Coding Exons)	TELO2	TELO2, TEL2, CLK2, KIAA0683, HCLK2, YHFS	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEKT3 geni (Tüm Gen Dizi Analizi)	TEKT3 gene (Sequence Analysis-All Coding Exons)	TEKT3	TEKT3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEKT1 geni (Tüm Gen Dizi Analizi)	TEKT1 gene (Sequence Analysis-All Coding Exons)	TEKT1	TEKT1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEFM geni (Tüm Gen Dizi Analizi)	TEFM gene (Sequence Analysis-All Coding Exons)	TEFM	TEFM, C17orf42	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEF geni (Tüm Gen Dizi Analizi)	TEF gene (Sequence Analysis-All Coding Exons)	TEF	TEF	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TECPR1 geni (Tüm Gen Dizi Analizi)	TECPR1 gene (Sequence Analysis-All Coding Exons)	TECPR1	TECPR1, KIAA1358	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEC geni (Tüm Gen Dizi Analizi)	TEC gene (Sequence Analysis-All Coding Exons)	TEC	TEC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEAD4 geni (Tüm Gen Dizi Analizi)	TEAD4 gene (Sequence Analysis-All Coding Exons)	TEAD4	TEAD4, TCF13L1, RTEF1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEAD3 geni (Tüm Gen Dizi Analizi)	TEAD3 gene (Sequence Analysis-All Coding Exons)	TEAD3	TEAD3, TEF5, TEAD5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRIB2 geni (Tüm Gen Dizi Analizi)	TRIB2 gene (Sequence Analysis-All Coding Exons)	TRIB2		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEAD2 geni (Tüm Gen Dizi Analizi)	TEAD2 gene (Sequence Analysis-All Coding Exons)	TEAD2	TEAD2, TEF4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDRG1 geni (Tüm Gen Dizi Analizi)	TDRG1 gene (Sequence Analysis-All Coding Exons)	TDRG1	TDRG1, LINC00532	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDRD6 geni (Tüm Gen Dizi Analizi)	TDRD6 gene (Sequence Analysis-All Coding Exons)	TDRD6	TDRD6	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDRD3 geni (Tüm Gen Dizi Analizi)	TDRD3 gene (Sequence Analysis-All Coding Exons)	TDRD3	TDRD3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDRD1 geni (Tüm Gen Dizi Analizi)	TDRD1 gene (Sequence Analysis-All Coding Exons)	TDRD1	TDRD1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDO2 geni (Tüm Gen Dizi Analizi)	TDO2 gene (Sequence Analysis-All Coding Exons)	TDO2	TDO2, TPH2, TRPO	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRGC2 geni (Tüm Gen Dizi Analizi)	TRGC2 gene (Sequence Analysis-All Coding Exons)	TRGC2	TRGC2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRGC1 geni (Tüm Gen Dizi Analizi)	TRGC1 gene (Sequence Analysis-All Coding Exons)	TRGC1	TRGC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDG geni (Tüm Gen Dizi Analizi)	TDG gene (Sequence Analysis-All Coding Exons)	TDG	TDG	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCTEX1D4 geni (Tüm Gen Dizi Analizi)	TCTEX1D4 gene (Sequence Analysis-All Coding Exons)	TCTEX1D4	TCTEX1D4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCTEX1D2 geni (Tüm Gen Dizi Analizi)	TCTEX1D2 gene (Sequence Analysis-All Coding Exons)	TCTEX1D2	TCTEX1D2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCTE3 geni (Tüm Gen Dizi Analizi)	TCTE3 gene (Sequence Analysis-All Coding Exons)	TCTE3	TCTE3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCTE1 geni (Tüm Gen Dizi Analizi)	TCTE1 gene (Sequence Analysis-All Coding Exons)	TCTE1	TCTE1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCTA geni (Tüm Gen Dizi Analizi)	TCTA gene (Sequence Analysis-All Coding Exons)	TCTA	TCTA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCP11 geni (Tüm Gen Dizi Analizi)	TCP11 gene (Sequence Analysis-All Coding Exons)	TCP11	TCP11, 06S230E	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCP10L geni (Tüm Gen Dizi Analizi)	TCP10L gene (Sequence Analysis-All Coding Exons)	TCP10L	TCP10L	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCP10 geni (Tüm Gen Dizi Analizi)	TCP10 gene (Sequence Analysis-All Coding Exons)	TCP10	TCP10	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCP1 geni (Tüm Gen Dizi Analizi)	TCP1 gene (Sequence Analysis-All Coding Exons)	TCP1	TCP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCN1 geni (Tüm Gen Dizi Analizi)	TCN1 gene (Sequence Analysis-All Coding Exons)	TCN1	TCN1, TC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCL6 geni (Tüm Gen Dizi Analizi)	TCL6 gene (Sequence Analysis-All Coding Exons)	TCL6	TCL6, TNG1, TNG2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCHP geni (Tüm Gen Dizi Analizi)	TCHP gene (Sequence Analysis-All Coding Exons)	TCHP	TCHP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF7L1 geni (Tüm Gen Dizi Analizi)	TCF7L1 gene (Sequence Analysis-All Coding Exons)	TCF7L1	TCF7L1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRDC geni (Tüm Gen Dizi Analizi)	TRDC gene (Sequence Analysis-All Coding Exons)	TRDC	TRDC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRBD1 geni (Tüm Gen Dizi Analizi)	TRBD1 gene (Sequence Analysis-All Coding Exons)	TRBD1	TRBD1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRBC2 geni (Tüm Gen Dizi Analizi)	TRBC2 gene (Sequence Analysis-All Coding Exons)	TRBC2	TRBC2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRBC1 geni (Tüm Gen Dizi Analizi)	TRBC1 gene (Sequence Analysis-All Coding Exons)	TRBC1	TRBC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF7 geni (Tüm Gen Dizi Analizi)	TCF7 gene (Sequence Analysis-All Coding Exons)	TCF7	TCF7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF25 geni (Tüm Gen Dizi Analizi)	TCF25 gene (Sequence Analysis-All Coding Exons)	TCF25	TCF25, NULP1, KIAA1049	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF23 geni (Tüm Gen Dizi Analizi)	TCF23 gene (Sequence Analysis-All Coding Exons)	TCF23	TCF23	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF21 geni (Tüm Gen Dizi Analizi)	TCF21 gene (Sequence Analysis-All Coding Exons)	TCF21	TCF21, POD1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF20 geni (Tüm Gen Dizi Analizi)	TCF20 gene (Sequence Analysis-All Coding Exons)	TCF20	TCF20, SPBP, AR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF19 geni (Tüm Gen Dizi Analizi)	TCF19 gene (Sequence Analysis-All Coding Exons)	TCF19	TCF19, SC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCF15 geni (Tüm Gen Dizi Analizi)	TCF15 gene (Sequence Analysis-All Coding Exons)	TCF15	TCF15	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCERG1 geni (Tüm Gen Dizi Analizi)	TCERG1 gene (Sequence Analysis-All Coding Exons)	TCERG1	TCERG1, TAF2S, CA150	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCEAL7 geni (Tüm Gen Dizi Analizi)	TCEAL7 gene (Sequence Analysis-All Coding Exons)	TCEAL7	TCEAL7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCEAL1 geni (Tüm Gen Dizi Analizi)	TCEAL1 gene (Sequence Analysis-All Coding Exons)	TCEAL1	TCEAL1, P21, SIIR	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TCEA3 geni (Tüm Gen Dizi Analizi)	TCEA3 gene (Sequence Analysis-All Coding Exons)	TCEA3	TCEA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCEA1 geni (Tüm Gen Dizi Analizi)	TCEA1 gene (Sequence Analysis-All Coding Exons)	TCEA1	TCEA1, TF2S, GTF2S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCAM1P geni (Tüm Gen Dizi Analizi)	TCAM1P gene (Sequence Analysis-All Coding Exons)	TCAM1P	TCAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCAF2 geni (Tüm Gen Dizi Analizi)	TCAF2 gene (Sequence Analysis-All Coding Exons)	TCAF2	TCAF2, FAM115C, FAM139A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCAF1 geni (Tüm Gen Dizi Analizi)	TCAF1 gene (Sequence Analysis-All Coding Exons)	TCAF1	TCAF1, FAM115A, KIAA0738	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBX2 geni (Tüm Gen Dizi Analizi)	TBX2 gene (Sequence Analysis-All Coding Exons)	TBX2	TBX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBX10 geni (Tüm Gen Dizi Analizi)	TBX10 gene (Sequence Analysis-All Coding Exons)	TBX10	TBX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBRG4 geni (Tüm Gen Dizi Analizi)	TBRG4 gene (Sequence Analysis-All Coding Exons)	TBRG4	TBRG4, KIAA0948, CPR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBRG1 geni (Tüm Gen Dizi Analizi)	TBRG1 gene (Sequence Analysis-All Coding Exons)	TBRG1	TBRG1, NIAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBR1 geni (Tüm Gen Dizi Analizi)	TBR1 gene (Sequence Analysis-All Coding Exons)	TBR1	TBR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBL3 geni (Tüm Gen Dizi Analizi)	TBL3 gene (Sequence Analysis-All Coding Exons)	TBL3	TBL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBL2 geni (Tüm Gen Dizi Analizi)	TBL2 gene (Sequence Analysis-All Coding Exons)	TBL2	TBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBL1Y geni (Tüm Gen Dizi Analizi)	TBL1Y gene (Sequence Analysis-All Coding Exons)	TBL1Y	TBL1Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBL1X geni (Tüm Gen Dizi Analizi)	TBL1X gene (Sequence Analysis-All Coding Exons)	TBL1X	TBL1X, TBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TRAF4 geni (Tüm Gen Dizi Analizi)	TRAF4 gene (Sequence Analysis-All Coding Exons)	TRAF4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBKBP1 geni (Tüm Gen Dizi Analizi)	TBKBP1 gene (Sequence Analysis-All Coding Exons)	TBKBP1	TBKBP1, PROSAPIP2, KIAA0726	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBCEL geni (Tüm Gen Dizi Analizi)	TBCEL gene (Sequence Analysis-All Coding Exons)	TBCEL	LRRC35, EL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBCC geni (Tüm Gen Dizi Analizi)	TBCC gene (Sequence Analysis-All Coding Exons)	TBCC	TBCC, CFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBCB geni (Tüm Gen Dizi Analizi)	TBCB gene (Sequence Analysis-All Coding Exons)	TBCB	CKAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBCA geni (Tüm Gen Dizi Analizi)	TBCA gene (Sequence Analysis-All Coding Exons)	TBCA	TBCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D5 geni (Tüm Gen Dizi Analizi)	TBC1D5 gene (Sequence Analysis-All Coding Exons)	TBC1D5	TBC1D5, KIAA0210	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3H geni (Tüm Gen Dizi Analizi)	TBC1D3H gene (Sequence Analysis-All Coding Exons)	TBC1D3H	TBC1D3H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3G geni (Tüm Gen Dizi Analizi)	TBC1D3G gene (Sequence Analysis-All Coding Exons)	TBC1D3G	TBC1D3G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3F geni (Tüm Gen Dizi Analizi)	TBC1D3F gene (Sequence Analysis-All Coding Exons)	TBC1D3F	TBC1D3F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3E geni (Tüm Gen Dizi Analizi)	TBC1D3E gene (Sequence Analysis-All Coding Exons)	TBC1D3E	TBC1D3E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3D geni (Tüm Gen Dizi Analizi)	TBC1D3D gene (Sequence Analysis-All Coding Exons)	TBC1D3D	TBC1D3D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3C geni (Tüm Gen Dizi Analizi)	TBC1D3C gene (Sequence Analysis-All Coding Exons)	TBC1D3C	TBC1D3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3B geni (Tüm Gen Dizi Analizi)	TBC1D3B gene (Sequence Analysis-All Coding Exons)	TBC1D3B	TBC1D3B, PRC17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D32 geni (Tüm Gen Dizi Analizi)	TBC1D32 gene (Sequence Analysis-All Coding Exons)	TBC1D32	TBC1D32, BROM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D30 geni (Tüm Gen Dizi Analizi)	TBC1D30 gene (Sequence Analysis-All Coding Exons)	TBC1D30	TBC1D30, KIAA0984	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D3 geni (Tüm Gen Dizi Analizi)	TBC1D3 gene (Sequence Analysis-All Coding Exons)	TBC1D3	TBC1D3, PRC17, TBC1D3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D25 geni (Tüm Gen Dizi Analizi)	TBC1D25 gene (Sequence Analysis-All Coding Exons)	TBC1D25	TBC1D25, OATL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D22B geni (Tüm Gen Dizi Analizi)	TBC1D22B gene (Sequence Analysis-All Coding Exons)	TBC1D22B	TBC1D22B, C6orf197	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D22A geni (Tüm Gen Dizi Analizi)	TBC1D22A gene (Sequence Analysis-All Coding Exons)	TBC1D22A	TBC1D22A, C22orf4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D2 geni (Tüm Gen Dizi Analizi)	TBC1D2 gene (Sequence Analysis-All Coding Exons)	TBC1D2	TBC1D2, PARIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D17 geni (Tüm Gen Dizi Analizi)	TBC1D17 gene (Sequence Analysis-All Coding Exons)	TBC1D17	TBC1D17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D16 geni (Tüm Gen Dizi Analizi)	TBC1D16 gene (Sequence Analysis-All Coding Exons)	TBC1D16	TBC1D16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D15 geni (Tüm Gen Dizi Analizi)	TBC1D15 gene (Sequence Analysis-All Coding Exons)	TBC1D15	TBC1D15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D14 geni (Tüm Gen Dizi Analizi)	TBC1D14 gene (Sequence Analysis-All Coding Exons)	TBC1D14	TBC1D14, KIAA1322	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D13 geni (Tüm Gen Dizi Analizi)	TBC1D13 gene (Sequence Analysis-All Coding Exons)	TBC1D13	TBC1D13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPRKB geni (Tüm Gen Dizi Analizi)	TPRKB gene (Sequence Analysis-All Coding Exons)	TPRKB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D10C geni (Tüm Gen Dizi Analizi)	TBC1D10C gene (Sequence Analysis-All Coding Exons)	TBC1D10C	TBC1D10C, CARABN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TPRA1 geni (Tüm Gen Dizi Analizi)	TPRA1 gene (Sequence Analysis-All Coding Exons)	TPRA1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D10B geni (Tüm Gen Dizi Analizi)	TBC1D10B gene (Sequence Analysis-All Coding Exons)	TBC1D10B	TBC1D10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D10A geni (Tüm Gen Dizi Analizi)	TBC1D10A gene (Sequence Analysis-All Coding Exons)	TBC1D10A	TBC1D10A, EPI64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBC1D1 geni (Tüm Gen Dizi Analizi)	TBC1D1 gene (Sequence Analysis-All Coding Exons)	TBC1D1	TBC1D1, KIAA1108	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBATA geni (Tüm Gen Dizi Analizi)	TBATA gene (Sequence Analysis-All Coding Exons)	TBATA	C10orf27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAX1BP3 geni (Tüm Gen Dizi Analizi)	TAX1BP3 gene (Sequence Analysis-All Coding Exons)	TAX1BP3	TAX1BP3, TIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAX1BP1 geni (Tüm Gen Dizi Analizi)	TAX1BP1 gene (Sequence Analysis-All Coding Exons)	TAX1BP1	TAX1BP1, TXBP151	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TASP1 geni (Tüm Gen Dizi Analizi)	TASP1 gene (Sequence Analysis-All Coding Exons)	TASP1	TASP1, C20orf13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R9 geni (Tüm Gen Dizi Analizi)	TAS2R9 gene (Sequence Analysis-All Coding Exons)	TAS2R9	TRB6, T2R9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R8 geni (Tüm Gen Dizi Analizi)	TAS2R8 gene (Sequence Analysis-All Coding Exons)	TAS2R8	TRB5, T2R8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R7 geni (Tüm Gen Dizi Analizi)	TAS2R7 gene (Sequence Analysis-All Coding Exons)	TAS2R7	TRB4, T2R7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R60 geni (Tüm Gen Dizi Analizi)	TAS2R60 gene (Sequence Analysis-All Coding Exons)	TAS2R60	TAS2R60, T2R60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R50 geni (Tüm Gen Dizi Analizi)	TAS2R50 gene (Sequence Analysis-All Coding Exons)	TAS2R50	TAS2R50, T2R50, TAS2R51, T2R51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R5 geni (Tüm Gen Dizi Analizi)	TAS2R5 gene (Sequence Analysis-All Coding Exons)	TAS2R5	TAS2R5, T2R5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R46 geni (Tüm Gen Dizi Analizi)	TAS2R46 gene (Sequence Analysis-All Coding Exons)	TAS2R46	TAS2R46, T2R46, T2R54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R45 geni (Tüm Gen Dizi Analizi)	TAS2R45 gene (Sequence Analysis-All Coding Exons)	TAS2R45	TAS2R45, T2R45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R43 geni (Tüm Gen Dizi Analizi)	TAS2R43 gene (Sequence Analysis-All Coding Exons)	TAS2R43	TAS2R43, T2R52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R42 geni (Tüm Gen Dizi Analizi)	TAS2R42 gene (Sequence Analysis-All Coding Exons)	TAS2R42	TAS2R42, T2R42, T2R55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R41 geni (Tüm Gen Dizi Analizi)	TAS2R41 gene (Sequence Analysis-All Coding Exons)	TAS2R41	TAS2R41, T2R59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R40 geni (Tüm Gen Dizi Analizi)	TAS2R40 gene (Sequence Analysis-All Coding Exons)	TAS2R40	TAS2R40, T2R40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R4 geni (Tüm Gen Dizi Analizi)	TAS2R4 gene (Sequence Analysis-All Coding Exons)	TAS2R4	TAS2R4, T2R4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R31 geni (Tüm Gen Dizi Analizi)	TAS2R31 gene (Sequence Analysis-All Coding Exons)	TAS2R31	TAS2R31, TAS2R44, T2R53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R30 geni (Tüm Gen Dizi Analizi)	TAS2R30 gene (Sequence Analysis-All Coding Exons)	TAS2R30	TAS2R30, T2R30, T2R47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R3 geni (Tüm Gen Dizi Analizi)	TAS2R3 gene (Sequence Analysis-All Coding Exons)	TAS2R3	TAS2R3, T2R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R20 geni (Tüm Gen Dizi Analizi)	TAS2R20 gene (Sequence Analysis-All Coding Exons)	TAS2R20	TAS2R20, T2R56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R19 geni (Tüm Gen Dizi Analizi)	TAS2R19 gene (Sequence Analysis-All Coding Exons)	TAS2R19	TAS2R19, T2R19, T2R48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R14 geni (Tüm Gen Dizi Analizi)	TAS2R14 gene (Sequence Analysis-All Coding Exons)	TAS2R14	TRB1, T2R14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R13 geni (Tüm Gen Dizi Analizi)	TAS2R13 gene (Sequence Analysis-All Coding Exons)	TAS2R13	TRB3, T2R13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R10 geni (Tüm Gen Dizi Analizi)	TAS2R10 gene (Sequence Analysis-All Coding Exons)	TAS2R10	TRB2, T2R10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS2R1 geni (Tüm Gen Dizi Analizi)	TAS2R1 gene (Sequence Analysis-All Coding Exons)	TAS2R1	TRB7, T2R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS1R3 geni (Tüm Gen Dizi Analizi)	TAS1R3 gene (Sequence Analysis-All Coding Exons)	TAS1R3	TAS1R3, T1R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS1R2 geni (Tüm Gen Dizi Analizi)	TAS1R2 gene (Sequence Analysis-All Coding Exons)	TAS1R2	TAS1R2, T1R2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAS1R1 geni (Tüm Gen Dizi Analizi)	TAS1R1 gene (Sequence Analysis-All Coding Exons)	TAS1R1	TAS1R1, T1R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TARS geni (Tüm Gen Dizi Analizi)	TARS gene (Sequence Analysis-All Coding Exons)	TARS	TARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TARM1 geni (Tüm Gen Dizi Analizi)	TARM1 gene (Sequence Analysis-All Coding Exons)	TARM1	TARM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TARID geni (Tüm Gen Dizi Analizi)	TARID gene (Sequence Analysis-All Coding Exons)	TARID	TARID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TARBP2 geni (Tüm Gen Dizi Analizi)	TARBP2 gene (Sequence Analysis-All Coding Exons)	TARBP2	TARBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TARBP1 geni (Tüm Gen Dizi Analizi)	TARBP1 gene (Sequence Analysis-All Coding Exons)	TARBP1	TARBP1, TRP185	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAPBPL geni (Tüm Gen Dizi Analizi)	TAPBPL gene (Sequence Analysis-All Coding Exons)	TAPBPL	TAPBPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAOK3 geni (Tüm Gen Dizi Analizi)	TAOK3 gene (Sequence Analysis-All Coding Exons)	TAOK3	TAOK3, JIK, DPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAOK2 geni (Tüm Gen Dizi Analizi)	TAOK2 gene (Sequence Analysis-All Coding Exons)	TAOK2	TAOK2, TAO2, KIAA0881, PSK, PSK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TP53RK geni (Tüm Gen Dizi Analizi)	TP53RK gene (Sequence Analysis-All Coding Exons)	TP53RK		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAOK1 geni (Tüm Gen Dizi Analizi)	TAOK1 gene (Sequence Analysis-All Coding Exons)	TAOK1	TAOK1, PSK2, TAO1, MARKK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TANK geni (Tüm Gen Dizi Analizi)	TANK gene (Sequence Analysis-All Coding Exons)	TANK	TANK, ITRAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TANC2 geni (Tüm Gen Dizi Analizi)	TANC2 gene (Sequence Analysis-All Coding Exons)	TANC2	TANC2, KIAA1636	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TANC1 geni (Tüm Gen Dizi Analizi)	TANC1 gene (Sequence Analysis-All Coding Exons)	TANC1	TANC1, KIAA1728	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAMM41 geni (Tüm Gen Dizi Analizi)	TAMM41 gene (Sequence Analysis-All Coding Exons)	TAMM41	TAMM41, RAM41, C3orf31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAGLN3 geni (Tüm Gen Dizi Analizi)	TAGLN3 gene (Sequence Analysis-All Coding Exons)	TAGLN3	TAGLN3, NP24, NP22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAGLN2 geni (Tüm Gen Dizi Analizi)	TAGLN2 gene (Sequence Analysis-All Coding Exons)	TAGLN2	TAGLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAGLN geni (Tüm Gen Dizi Analizi)	TAGLN gene (Sequence Analysis-All Coding Exons)	TAGLN	TAGLN, SMCC, SM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAGAP geni (Tüm Gen Dizi Analizi)	TAGAP gene (Sequence Analysis-All Coding Exons)	TAGAP	TAGAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

TAF9B geni (Tüm Gen Dizi Analizi)	TAF9B gene (Sequence Analysis-All Coding Exons)	TAF9B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF9 geni (Tüm Gen Dizi Analizi)	TAF9 gene (Sequence Analysis-All Coding Exons)	TAF9	TAF9, TAF2G, TAFII32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF7L geni (Tüm Gen Dizi Analizi)	TAF7L gene (Sequence Analysis-All Coding Exons)	TAF7L	TAF7L, TAF2Q	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF7 geni (Tüm Gen Dizi Analizi)	TAF7 gene (Sequence Analysis-All Coding Exons)	TAF7	TAF7, TAF2F, TAFII55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF5 geni (Tüm Gen Dizi Analizi)	TAF5 gene (Sequence Analysis-All Coding Exons)	TAF5	TAF5, TAF2D, TAFII100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF4 geni (Tüm Gen Dizi Analizi)	TAF4 gene (Sequence Analysis-All Coding Exons)	TAF4	TAF4, TAF2C1, TAFII130, TAF2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF3 geni (Tüm Gen Dizi Analizi)	TAF3 gene (Sequence Analysis-All Coding Exons)	TAF3	TAF3, TAFII140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF1D geni (Tüm Gen Dizi Analizi)	TAF1D gene (Sequence Analysis-All Coding Exons)	TAF1D	TAF1D, RAFI41, JOSD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOX3 geni (Tüm Gen Dizi Analizi)	TOX3 gene (Sequence Analysis-All Coding Exons)	TOX3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF1C geni (Tüm Gen Dizi Analizi)	TAF1C gene (Sequence Analysis-All Coding Exons)	TAF1C	TAF1C, TAFI110, SL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF1B geni (Tüm Gen Dizi Analizi)	TAF1B gene (Sequence Analysis-All Coding Exons)	TAF1B	RAF1B, RAFI63, SL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF1A geni (Tüm Gen Dizi Analizi)	TAF1A gene (Sequence Analysis-All Coding Exons)	TAF1A	TAF1A, RAFI48, SL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF12 geni (Tüm Gen Dizi Analizi)	TAF12 gene (Sequence Analysis-All Coding Exons)	TAF12	TAF12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF11 geni (Tüm Gen Dizi Analizi)	TAF11 gene (Sequence Analysis-All Coding Exons)	TAF11	TAF11, TAF2I, TAFII28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF10 geni (Tüm Gen Dizi Analizi)	TAF10 gene (Sequence Analysis-All Coding Exons)	TAF10	TAF10, TAF2H, TAF2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TADA3 geni (Tüm Gen Dizi Analizi)	TADA3 gene (Sequence Analysis-All Coding Exons)	TADA3	TADA3, TADA3L, ADA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TADA2B geni (Tüm Gen Dizi Analizi)	TADA2B gene (Sequence Analysis-All Coding Exons)	TADA2B	TADA2B, ADA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TADA2A geni (Tüm Gen Dizi Analizi)	TADA2A gene (Sequence Analysis-All Coding Exons)	TADA2A	TADA2L, TADA2A, ADA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TADA1 geni (Tüm Gen Dizi Analizi)	TADA1 gene (Sequence Analysis-All Coding Exons)	TADA1	TADA1L, STAF42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACR2 geni (Tüm Gen Dizi Analizi)	TACR2 gene (Sequence Analysis-All Coding Exons)	TACR2	TACR2, TAC2R, NKNAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACR1 geni (Tüm Gen Dizi Analizi)	TACR1 gene (Sequence Analysis-All Coding Exons)	TACR1	TACR1, TAC1R, NK1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACC3 geni (Tüm Gen Dizi Analizi)	TACC3 gene (Sequence Analysis-All Coding Exons)	TACC3	TACC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACC3 geni (Tüm Gen Dizi Analizi)	TACC3 gene (Sequence Analysis-All Coding Exons)	TACC3	TACC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACC2 geni (Tüm Gen Dizi Analizi)	TACC2 gene (Sequence Analysis-All Coding Exons)	TACC2	TACC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACC1 geni (Tüm Gen Dizi Analizi)	TACC1 gene (Sequence Analysis-All Coding Exons)	TACC1	TACC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TACC1 geni (Tüm Gen Dizi Analizi)	TACC1 gene (Sequence Analysis-All Coding Exons)	TACC1	TACC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAC4 geni (Tüm Gen Dizi Analizi)	TAC4 gene (Sequence Analysis-All Coding Exons)	TAC4	RAC4, PPTC, HK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM70 geni (Tüm Gen Dizi Analizi)	TOMM70 gene (Sequence Analysis-All Coding Exons)	TOMM70		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAC1 geni (Tüm Gen Dizi Analizi)	TAC1 gene (Sequence Analysis-All Coding Exons)	TAC1	TAC1, TAC2, NKNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAB3 geni (Tüm Gen Dizi Analizi)	TAB3 gene (Sequence Analysis-All Coding Exons)	TAB3	TAB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAB1 geni (Tüm Gen Dizi Analizi)	TAB1 gene (Sequence Analysis-All Coding Exons)	TAB1	TAB1, MAP3K7IP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR9 geni (Tüm Gen Dizi Analizi)	TAAR9 gene (Sequence Analysis-All Coding Exons)	TAAR9	TAR3, TA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR8 geni (Tüm Gen Dizi Analizi)	TAAR8 gene (Sequence Analysis-All Coding Exons)	TAAR8	GPR102, TA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR6 geni (Tüm Gen Dizi Analizi)	TAAR6 gene (Sequence Analysis-All Coding Exons)	TAAR6	TAAR6, TRAR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TOMM20 geni (Tüm Gen Dizi Analizi)	TOMM20 gene (Sequence Analysis-All Coding Exons)	TOMM20		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR5 geni (Tüm Gen Dizi Analizi)	TAAR5 gene (Sequence Analysis-All Coding Exons)	TAAR5	PNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR2 geni (Tüm Gen Dizi Analizi)	TAAR2 gene (Sequence Analysis-All Coding Exons)	TAAR2	TAAR2, GPR58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAAR1 geni (Tüm Gen Dizi Analizi)	TAAR1 gene (Sequence Analysis-All Coding Exons)	TAAR1	TAAR1, TA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYVN1 geni (Tüm Gen Dizi Analizi)	SYVN1 gene (Sequence Analysis-All Coding Exons)	SYVN1	SYVN1, HRD1, KIAA1810	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYTL4 geni (Tüm Gen Dizi Analizi)	SYTL4 gene (Sequence Analysis-All Coding Exons)	SYTL4	SYTL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYTL2 geni (Tüm Gen Dizi Analizi)	SYTL2 gene (Sequence Analysis-All Coding Exons)	SYTL2	SYTL2, SLP2, SLP2A, EXO4, KIAA1597	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYTL1 geni (Tüm Gen Dizi Analizi)	SYTL1 gene (Sequence Analysis-All Coding Exons)	SYTL1	SYTL1, SLP1, JFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT9 geni (Tüm Gen Dizi Analizi)	SYT9 gene (Sequence Analysis-All Coding Exons)	SYT9	SYT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT8 geni (Tüm Gen Dizi Analizi)	SYT8 gene (Sequence Analysis-All Coding Exons)	SYT8	SYT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT7 geni (Tüm Gen Dizi Analizi)	SYT7 gene (Sequence Analysis-All Coding Exons)	SYT7	SYT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT6 geni (Tüm Gen Dizi Analizi)	SYT6 gene (Sequence Analysis-All Coding Exons)	SYT6	SYT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT5 geni (Tüm Gen Dizi Analizi)	SYT5 gene (Sequence Analysis-All Coding Exons)	SYT5	SYT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT4 geni (Tüm Gen Dizi Analizi)	SYT4 gene (Sequence Analysis-All Coding Exons)	SYT4	SYT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT3 geni (Tüm Gen Dizi Analizi)	SYT3 gene (Sequence Analysis-All Coding Exons)	SYT3	SYT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT16 geni (Tüm Gen Dizi Analizi)	SYT16 gene (Sequence Analysis-All Coding Exons)	SYT16	SYT16, STREP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT15 geni (Tüm Gen Dizi Analizi)	SYT15 gene (Sequence Analysis-All Coding Exons)	SYT15	SYT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT13 geni (Tüm Gen Dizi Analizi)	SYT13 gene (Sequence Analysis-All Coding Exons)	SYT13	SYT13, KIAA1427	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT12 geni (Tüm Gen Dizi Analizi)	SYT12 gene (Sequence Analysis-All Coding Exons)	SYT12	SYT12, SRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT1 geni (Tüm Gen Dizi Analizi)	SYT1 gene (Sequence Analysis-All Coding Exons)	SYT1	SYT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYS1 geni (Tüm Gen Dizi Analizi)	SYS1 gene (Sequence Analysis-All Coding Exons)	SYS1	SYS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYPL1 geni (Tüm Gen Dizi Analizi)	SYPL1 gene (Sequence Analysis-All Coding Exons)	SYPL1	SYPL1, SYPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNRG geni (Tüm Gen Dizi Analizi)	SYNRG gene (Sequence Analysis-All Coding Exons)	SYNRG	AP1GBP1, SYNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNPO geni (Tüm Gen Dizi Analizi)	SYNPO gene (Sequence Analysis-All Coding Exons)	SYNPO	SYNPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNM geni (Tüm Gen Dizi Analizi)	SYNM gene (Sequence Analysis-All Coding Exons)	SYNM	DMN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNJ2BP geni (Tüm Gen Dizi Analizi)	SYNJ2BP gene (Sequence Analysis-All Coding Exons)	SYNJ2BP	SYNJ2BP, OMP25, ARIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNJ2 geni (Tüm Gen Dizi Analizi)	SYNJ2 gene (Sequence Analysis-All Coding Exons)	SYNJ2	SYNJ2, KIAA0348	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNGR4 geni (Tüm Gen Dizi Analizi)	SYNGR4 gene (Sequence Analysis-All Coding Exons)	SYNGR4	SYNGR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNGR3 geni (Tüm Gen Dizi Analizi)	SYNGR3 gene (Sequence Analysis-All Coding Exons)	SYNGR3	SYNGR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNGR2 geni (Tüm Gen Dizi Analizi)	SYNGR2 gene (Sequence Analysis-All Coding Exons)	SYNGR2	SYNGR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNGR1 geni (Tüm Gen Dizi Analizi)	SYNGR1 gene (Sequence Analysis-All Coding Exons)	SYNGR1	SYNGR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNE3 geni (Tüm Gen Dizi Analizi)	SYNE3 gene (Sequence Analysis-All Coding Exons)	SYNE3	SYNE3, C14orf49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNDIG1L geni (Tüm Gen Dizi Analizi)	SYNDIG1L gene (Sequence Analysis-All Coding Exons)	SYNDIG1L	SYNDIG1L, TMEM90A, C14orf34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNDIG1 geni (Tüm Gen Dizi Analizi)	SYNDIG1 gene (Sequence Analysis-All Coding Exons)	SYNDIG1	SYNDIG1, TMEM90B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNCRIP geni (Tüm Gen Dizi Analizi)	SYNCRIP gene (Sequence Analysis-All Coding Exons)	SYNCRIP	SYNCRIP, NSAP1, GRYRBP, HNRNPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYNC geni (Tüm Gen Dizi Analizi)	SYNC gene (Sequence Analysis-All Coding Exons)	SYNC	SYNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYN3 geni (Tüm Gen Dizi Analizi)	SYN3 gene (Sequence Analysis-All Coding Exons)	SYN3	SYN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYMPK geni (Tüm Gen Dizi Analizi)	SYMPK gene (Sequence Analysis-All Coding Exons)	SYMPK	SPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYK geni (Tüm Gen Dizi Analizi)	SYK gene (Sequence Analysis-All Coding Exons)	SYK	SYK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYF2 geni (Tüm Gen Dizi Analizi)	SYF2 gene (Sequence Analysis-All Coding Exons)	SYF2	SYF2, P29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNKS2 geni (Tüm Gen Dizi Analizi)	TNKS2 gene (Sequence Analysis-All Coding Exons)	TNKS2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYDE1 geni (Tüm Gen Dizi Analizi)	SYDE1 gene (Sequence Analysis-All Coding Exons)	SYDE1	SYDE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYCP2L geni (Tüm Gen Dizi Analizi)	SYCP2L gene (Sequence Analysis-All Coding Exons)	SYCP2L	SYCP2L, C5orf177	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYCP2 geni (Tüm Gen Dizi Analizi)	SYCP2 gene (Sequence Analysis-All Coding Exons)	SYCP2	SYCP2, SCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYCP1 geni (Tüm Gen Dizi Analizi)	SYCP1 gene (Sequence Analysis-All Coding Exons)	SYCP1	SYCP1, SCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNIP3 geni (Tüm Gen Dizi Analizi)	TNIP3 gene (Sequence Analysis-All Coding Exons)	TNIP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYCE3 geni (Tüm Gen Dizi Analizi)	SYCE3 gene (Sequence Analysis-All Coding Exons)	SYCE3	SYCE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYCE2 geni (Tüm Gen Dizi Analizi)	SYCE2 gene (Sequence Analysis-All Coding Exons)	SYCE2	SYCE2, CESC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYBU geni (Tüm Gen Dizi Analizi)	SYBU gene (Sequence Analysis-All Coding Exons)	SYBU	GOLSYN, SYBU, KIAA1472	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SWSAP1 geni (Tüm Gen Dizi Analizi)	SWSAP1 gene (Sequence Analysis-All Coding Exons)	SWSAP1	SWSAP1, ZSWIM7AP1, C10orf39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SWI5 geni (Tüm Gen Dizi Analizi)	SWI5 gene (Sequence Analysis-All Coding Exons)	SWI5	SWI5, SAE3, C9orf119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SWAP70 geni (Tüm Gen Dizi Analizi)	SWAP70 gene (Sequence Analysis-All Coding Exons)	SWAP70	SWAP70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SVOPL geni (Tüm Gen Dizi Analizi)	SVOPL gene (Sequence Analysis-All Coding Exons)	SVOPL	SVOPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SVOP geni (Tüm Gen Dizi Analizi)	SVOP gene (Sequence Analysis-All Coding Exons)	SVOP	SVOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SVIL geni (Tüm Gen Dizi Analizi)	SVIL gene (Sequence Analysis-All Coding Exons)	SVIL	SVIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SVEP1 geni (Tüm Gen Dizi Analizi)	SVEP1 gene (Sequence Analysis-All Coding Exons)	SVEP1	SVEP1, POLYDOM, SELOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SV2C geni (Tüm Gen Dizi Analizi)	SV2C gene (Sequence Analysis-All Coding Exons)	SV2C	SV2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SV2B geni (Tüm Gen Dizi Analizi)	SV2B gene (Sequence Analysis-All Coding Exons)	SV2B	SV2B, KIAA0735	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SV2A geni (Tüm Gen Dizi Analizi)	SV2A gene (Sequence Analysis-All Coding Exons)	SV2A	SV2A, SV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SUZ12 geni (Tüm Gen Dizi Analizi)	SUZ12 gene (Sequence Analysis-All Coding Exons)	SUZ12	JJAZ1, KIAA0160	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUV39H2 geni (Tüm Gen Dizi Analizi)	SUV39H2 gene (Sequence Analysis-All Coding Exons)	SUV39H2	SUV39H2, FLJ23414	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUV39H1 geni (Tüm Gen Dizi Analizi)	SUV39H1 gene (Sequence Analysis-All Coding Exons)	SUV39H1	SUV39H1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUSD6 geni (Tüm Gen Dizi Analizi)	SUSD6 gene (Sequence Analysis-All Coding Exons)	SUSD6	SUSD6, DRAGO, KIAA0247	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUSD4 geni (Tüm Gen Dizi Analizi)	SUSD4 gene (Sequence Analysis-All Coding Exons)	SUSD4	SUSD4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUSD3 geni (Tüm Gen Dizi Analizi)	SUSD3 gene (Sequence Analysis-All Coding Exons)	SUSD3	SUSD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUSD2 geni (Tüm Gen Dizi Analizi)	SUSD2 gene (Sequence Analysis-All Coding Exons)	SUSD2	SUSD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFRSF21 geni (Tüm Gen Dizi Analizi)	TNFRSF21 gene (Sequence Analysis-All Coding Exons)	TNFRSF21		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SURF6 geni (Tüm Gen Dizi Analizi)	SURF6 gene (Sequence Analysis-All Coding Exons)	SURF6	SURF6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SURF4 geni (Tüm Gen Dizi Analizi)	SURF4 gene (Sequence Analysis-All Coding Exons)	SURF4	SURF4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SURF2 geni (Tüm Gen Dizi Analizi)	SURF2 gene (Sequence Analysis-All Coding Exons)	SURF2	SURF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPV3L1 geni (Tüm Gen Dizi Analizi)	SUPV3L1 gene (Sequence Analysis-All Coding Exons)	SUPV3L1	SUPV3L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT7L geni (Tüm Gen Dizi Analizi)	SUPT7L gene (Sequence Analysis-All Coding Exons)	SUPT7L	SUPT7L, SUPT7H, SPT7l, KIAA0764, STAF65G	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT6H geni (Tüm Gen Dizi Analizi)	SUPT6H gene (Sequence Analysis-All Coding Exons)	SUPT6H	SUPT6H	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT5H geni (Tüm Gen Dizi Analizi)	SUPT5H gene (Sequence Analysis-All Coding Exons)	SUPT5H	SUPT5H, SPT5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT3H geni (Tüm Gen Dizi Analizi)	SUPT3H gene (Sequence Analysis-All Coding Exons)	SUPT3H	SUPT3H, SPT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT20H geni (Tüm Gen Dizi Analizi)	SUPT20H gene (Sequence Analysis-All Coding Exons)	SUPT20H	FAM48A, P38IP, C13orf19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT16H geni (Tüm Gen Dizi Analizi)	SUPT16H gene (Sequence Analysis-All Coding Exons)	SUPT16H	SUPT16H, SPT16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUN5 geni (Tüm Gen Dizi Analizi)	SUN5 gene (Sequence Analysis-All Coding Exons)	SUN5	SUN5, TSARG4, SPAG4L, SPGF16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUN2 geni (Tüm Gen Dizi Analizi)	SUN2 gene (Sequence Analysis-All Coding Exons)	SUN2	SUN2, UNC84B, KIAA0668	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUN1 geni (Tüm Gen Dizi Analizi)	SUN1 gene (Sequence Analysis-All Coding Exons)	SUN1	UNC84A, KIAA0810, SUN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUMO3 geni (Tüm Gen Dizi Analizi)	SUMO3 gene (Sequence Analysis-All Coding Exons)	SUMO3	SUMO3, SMT3H1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUMF2 geni (Tüm Gen Dizi Analizi)	SUMF2 gene (Sequence Analysis-All Coding Exons)	SUMF2	SUMF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT6B1 geni (Tüm Gen Dizi Analizi)	SULT6B1 gene (Sequence Analysis-All Coding Exons)	SULT6B1	SULT6B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT4A1 geni (Tüm Gen Dizi Analizi)	SULT4A1 gene (Sequence Analysis-All Coding Exons)	SULT4A1	SULT4A1, BRSTL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT2B1 geni (Tüm Gen Dizi Analizi)	SULT2B1 gene (Sequence Analysis-All Coding Exons)	SULT2B1	SULT2B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT2A1 geni (Tüm Gen Dizi Analizi)	SULT2A1 gene (Sequence Analysis-All Coding Exons)	SULT2A1	SULT2A1, STD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1E1 geni (Tüm Gen Dizi Analizi)	SULT1E1 gene (Sequence Analysis-All Coding Exons)	SULT1E1	STE, EST	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1C4 geni (Tüm Gen Dizi Analizi)	SULT1C4 gene (Sequence Analysis-All Coding Exons)	SULT1C4	SULT1C2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1C3 geni (Tüm Gen Dizi Analizi)	SULT1C3 gene (Sequence Analysis-All Coding Exons)	SULT1C3	SULT1C3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TNFAIP8L1 geni (Tüm Gen Dizi Analizi)	TNFAIP8L1 gene (Sequence Analysis-All Coding Exons)	TNFAIP8L1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1C2 geni (Tüm Gen Dizi Analizi)	SULT1C2 gene (Sequence Analysis-All Coding Exons)	SULT1C2	SULT1C1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1B1 geni (Tüm Gen Dizi Analizi)	SULT1B1 gene (Sequence Analysis-All Coding Exons)	SULT1B1	SULT1B1, ST1B2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1A4 geni (Tüm Gen Dizi Analizi)	SULT1A4 gene (Sequence Analysis-All Coding Exons)	SULT1A4	SULT1A4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1A3 geni (Tüm Gen Dizi Analizi)	SULT1A3 gene (Sequence Analysis-All Coding Exons)	SULT1A3	SULT1A3, STM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1A2 geni (Tüm Gen Dizi Analizi)	SULT1A2 gene (Sequence Analysis-All Coding Exons)	SULT1A2	SULT1A2, STP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULT1A1 geni (Tüm Gen Dizi Analizi)	SULT1A1 gene (Sequence Analysis-All Coding Exons)	SULT1A1	SULT1A1, STP1, STP, PST	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULF2 geni (Tüm Gen Dizi Analizi)	SULF2 gene (Sequence Analysis-All Coding Exons)	SULF2	SULF2, KIAA1247	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SULF1 geni (Tüm Gen Dizi Analizi)	SULF1 gene (Sequence Analysis-All Coding Exons)	SULF1	SULF1, KIAA1077	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUGT1 geni (Tüm Gen Dizi Analizi)	SUGT1 gene (Sequence Analysis-All Coding Exons)	SUGT1	SUGT1, SGT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUGP2 geni (Tüm Gen Dizi Analizi)	SUGP2 gene (Sequence Analysis-All Coding Exons)	SUGP2	SRFS14, KIAA0365	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUGP1 geni (Tüm Gen Dizi Analizi)	SUGP1 gene (Sequence Analysis-All Coding Exons)	SUGP1	SF4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUCNR1 geni (Tüm Gen Dizi Analizi)	SUCNR1 gene (Sequence Analysis-All Coding Exons)	SUCNR1	SUCNR1, GPR91	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUCLG2 geni (Tüm Gen Dizi Analizi)	SUCLG2 gene (Sequence Analysis-All Coding Exons)	SUCLG2	SUCLG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUB1 geni (Tüm Gen Dizi Analizi)	SUB1 gene (Sequence Analysis-All Coding Exons)	SUB1	SUB1, PC4, p15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STYXL1 geni (Tüm Gen Dizi Analizi)	STYXL1 gene (Sequence Analysis-All Coding Exons)	STYXL1	STYXL1, MKSTYX, DUSP24	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STYX geni (Tüm Gen Dizi Analizi)	STYX gene (Sequence Analysis-All Coding Exons)	STYX	STYX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STYK1 geni (Tüm Gen Dizi Analizi)	STYK1 gene (Sequence Analysis-All Coding Exons)	STYK1	STYK1, NOK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STXBP6 geni (Tüm Gen Dizi Analizi)	STXBP6 gene (Sequence Analysis-All Coding Exons)	STXBP6	STXBP6, AMSYN, HSPC156	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STXBP5L geni (Tüm Gen Dizi Analizi)	STXBP5L gene (Sequence Analysis-All Coding Exons)	STXBP5L	STXBP5L, LLGL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STXBP5 geni (Tüm Gen Dizi Analizi)	STXBP5 gene (Sequence Analysis-All Coding Exons)	STXBP5	STXBP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STXBP4 geni (Tüm Gen Dizi Analizi)	STXBP4 gene (Sequence Analysis-All Coding Exons)	STXBP4	STXBP4, SYNIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX8 geni (Tüm Gen Dizi Analizi)	STX8 gene (Sequence Analysis-All Coding Exons)	STX8	STX8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX7 geni (Tüm Gen Dizi Analizi)	STX7 gene (Sequence Analysis-All Coding Exons)	STX7	STX7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX6 geni (Tüm Gen Dizi Analizi)	STX6 gene (Sequence Analysis-All Coding Exons)	STX6	STX6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX5 geni (Tüm Gen Dizi Analizi)	STX5 gene (Sequence Analysis-All Coding Exons)	STX5	STX5, STX5A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX4 geni (Tüm Gen Dizi Analizi)	STX4 gene (Sequence Analysis-All Coding Exons)	STX4	STX4, STX4A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX3 geni (Tüm Gen Dizi Analizi)	STX3 gene (Sequence Analysis-All Coding Exons)	STX3	STX3, STX3A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX2 geni (Tüm Gen Dizi Analizi)	STX2 gene (Sequence Analysis-All Coding Exons)	STX2	STX2, EPIM, STX2C, STX2B, STX2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX1A geni (Tüm Gen Dizi Analizi)	STX1A gene (Sequence Analysis-All Coding Exons)	STX1A	STX1A, STX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX17 geni (Tüm Gen Dizi Analizi)	STX17 gene (Sequence Analysis-All Coding Exons)	STX17	STX17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX10 geni (Tüm Gen Dizi Analizi)	STX10 gene (Sequence Analysis-All Coding Exons)	STX10	STX10, SYN10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRN4 geni (Tüm Gen Dizi Analizi)	STRN4 gene (Sequence Analysis-All Coding Exons)	STRN4	STRN4, ZIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMPRSS11D geni (Tüm Gen Dizi Analizi)	TMPRSS11D gene (Sequence Analysis-All Coding Exons)	TMPRSS11D		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRN3 geni (Tüm Gen Dizi Analizi)	STRN3 gene (Sequence Analysis-All Coding Exons)	STRN3	STRN3, SG2NA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRN geni (Tüm Gen Dizi Analizi)	STRN gene (Sequence Analysis-All Coding Exons)	STRN	STRN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRBP geni (Tüm Gen Dizi Analizi)	STRBP gene (Sequence Analysis-All Coding Exons)	STRBP	STRBP, SPNR, p74	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRAP geni (Tüm Gen Dizi Analizi)	STRAP gene (Sequence Analysis-All Coding Exons)	STRAP	STRAP, UNRIP, MAWD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRADB geni (Tüm Gen Dizi Analizi)	STRADB gene (Sequence Analysis-All Coding Exons)	STRADB	STRADB, ILPIP, ALS2CR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STRA8 geni (Tüm Gen Dizi Analizi)	STRA8 gene (Sequence Analysis-All Coding Exons)	STRA8	STRA8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STPG1 geni (Tüm Gen Dizi Analizi)	STPG1 gene (Sequence Analysis-All Coding Exons)	STPG1	STPG1, MAPO2, C1orf201	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STOX2 geni (Tüm Gen Dizi Analizi)	STOX2 gene (Sequence Analysis-All Coding Exons)	STOX2	STOX2, KIAA1392	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STON2 geni (Tüm Gen Dizi Analizi)	STON2 gene (Sequence Analysis-All Coding Exons)	STON2	STON2, STN2, STN8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STON1 geni (Tüm Gen Dizi Analizi)	STON1 gene (Sequence Analysis-All Coding Exons)	STON1	STON1, STN1, SBLF, SALF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STOML3 geni (Tüm Gen Dizi Analizi)	STOML3 gene (Sequence Analysis-All Coding Exons)	STOML3	STOML3, SRO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STOML2 geni (Tüm Gen Dizi Analizi)	STOML2 gene (Sequence Analysis-All Coding Exons)	STOML2	STOML2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STOML1 geni (Tüm Gen Dizi Analizi)	STOML1 gene (Sequence Analysis-All Coding Exons)	STOML1	STOML1, STORP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STOM geni (Tüm Gen Dizi Analizi)	STOM gene (Sequence Analysis-All Coding Exons)	STOM	STOM, EPB72, BND7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STMN3 geni (Tüm Gen Dizi Analizi)	STMN3 gene (Sequence Analysis-All Coding Exons)	STMN3	STMN3, SCLIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STMN2 geni (Tüm Gen Dizi Analizi)	STMN2 gene (Sequence Analysis-All Coding Exons)	STMN2	STMN2, SCG N10, SCG10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STMN1 geni (Tüm Gen Dizi Analizi)	STMN1 gene (Sequence Analysis-All Coding Exons)	STMN1	STMN1, LAP18, SMN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK40 geni (Tüm Gen Dizi Analizi)	STK40 gene (Sequence Analysis-All Coding Exons)	STK40	STK40, SHIK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK39 geni (Tüm Gen Dizi Analizi)	STK39 gene (Sequence Analysis-All Coding Exons)	STK39	STK39, SPAK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK38L geni (Tüm Gen Dizi Analizi)	STK38L gene (Sequence Analysis-All Coding Exons)	STK38L	STK38L, NDR2, KIAA0965	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK38 geni (Tüm Gen Dizi Analizi)	STK38 gene (Sequence Analysis-All Coding Exons)	STK38	STK38, NDR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK36 geni (Tüm Gen Dizi Analizi)	STK36 gene (Sequence Analysis-All Coding Exons)	STK36	STK36, FU, KIAA1278	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK35 geni (Tüm Gen Dizi Analizi)	STK35 gene (Sequence Analysis-All Coding Exons)	STK35	STK35, CLIK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK33 geni (Tüm Gen Dizi Analizi)	STK33 gene (Sequence Analysis-All Coding Exons)	STK33	STK33	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK31 geni (Tüm Gen Dizi Analizi)	STK31 gene (Sequence Analysis-All Coding Exons)	STK31	STK31	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK3 geni (Tüm Gen Dizi Analizi)	STK3 gene (Sequence Analysis-All Coding Exons)	STK3	STK3, MST2, KRS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK26 geni (Tüm Gen Dizi Analizi)	STK26 gene (Sequence Analysis-All Coding Exons)	STK26	MASK, MST4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK25 geni (Tüm Gen Dizi Analizi)	STK25 gene (Sequence Analysis-All Coding Exons)	STK25	STK25, SOK1, YSK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM55B geni (Tüm Gen Dizi Analizi)	TMEM55B gene (Sequence Analysis-All Coding Exons)	TMEM55B		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

STK24 geni (Tüm Gen Dizi Analizi)	STK24 gene (Sequence Analysis-All Coding Exons)	STK24	STK24, MST3, MST3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK19 geni (Tüm Gen Dizi Analizi)	STK19 gene (Sequence Analysis-All Coding Exons)	STK19	STK19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK17B geni (Tüm Gen Dizi Analizi)	STK17B gene (Sequence Analysis-All Coding Exons)	STK17B	STK17B, DRAK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK17A geni (Tüm Gen Dizi Analizi)	STK17A gene (Sequence Analysis-All Coding Exons)	STK17A	STK17A, DRAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK11IP geni (Tüm Gen Dizi Analizi)	STK11IP gene (Sequence Analysis-All Coding Exons)	STK11IP	STK11IP, KIAA1898, LIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK10 geni (Tüm Gen Dizi Analizi)	STK10 gene (Sequence Analysis-All Coding Exons)	STK10	STK10, LOK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STIP1 geni (Tüm Gen Dizi Analizi)	STIP1 gene (Sequence Analysis-All Coding Exons)	STIP1	STIP1, HOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STIM2 geni (Tüm Gen Dizi Analizi)	STIM2 gene (Sequence Analysis-All Coding Exons)	STIM2	STIM2, KIAA1482	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STH geni (Tüm Gen Dizi Analizi)	STH gene (Sequence Analysis-All Coding Exons)	STH	STH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STEAP4 geni (Tüm Gen Dizi Analizi)	STEAP4 gene (Sequence Analysis-All Coding Exons)	STEAP4	STEAP4, STAMP2, TIARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STEAP2 geni (Tüm Gen Dizi Analizi)	STEAP2 gene (Sequence Analysis-All Coding Exons)	STEAP2	STEAP2, STAMP1, IPCA1, PCANAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STEAP1 geni (Tüm Gen Dizi Analizi)	STEAP1 gene (Sequence Analysis-All Coding Exons)	STEAP1	STEAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STC2 geni (Tüm Gen Dizi Analizi)	STC2 gene (Sequence Analysis-All Coding Exons)	STC2	STC2, STCRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STC1 geni (Tüm Gen Dizi Analizi)	STC1 gene (Sequence Analysis-All Coding Exons)	STC1	STC1, STC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STBD1 geni (Tüm Gen Dizi Analizi)	STBD1 gene (Sequence Analysis-All Coding Exons)	STBD1	GENEX3414	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAU2 geni (Tüm Gen Dizi Analizi)	STAU2 gene (Sequence Analysis-All Coding Exons)	STAU2	STAU2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAU1 geni (Tüm Gen Dizi Analizi)	STAU1 gene (Sequence Analysis-All Coding Exons)	STAU1	STAU1, STAU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STATH geni (Tüm Gen Dizi Analizi)	STATH gene (Sequence Analysis-All Coding Exons)	STATH	STATH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAT6 geni (Tüm Gen Dizi Analizi)	STAT6 gene (Sequence Analysis-All Coding Exons)	STAT6	STAT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD9 geni (Tüm Gen Dizi Analizi)	STARD9 gene (Sequence Analysis-All Coding Exons)	STARD9	STARD9, KIAA1300	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD8 geni (Tüm Gen Dizi Analizi)	STARD8 gene (Sequence Analysis-All Coding Exons)	STARD8	STARD8, DLC3, START-GAP3, KIAA0189	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD7 geni (Tüm Gen Dizi Analizi)	STARD7 gene (Sequence Analysis-All Coding Exons)	STARD7	STARD7, GTT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD6 geni (Tüm Gen Dizi Analizi)	STARD6 gene (Sequence Analysis-All Coding Exons)	STARD6	STARD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD5 geni (Tüm Gen Dizi Analizi)	STARD5 gene (Sequence Analysis-All Coding Exons)	STARD5	STARD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD4 geni (Tüm Gen Dizi Analizi)	STARD4 gene (Sequence Analysis-All Coding Exons)	STARD4	STARD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD3NL geni (Tüm Gen Dizi Analizi)	STARD3NL gene (Sequence Analysis-All Coding Exons)	STARD3NL	STARD3NL, MENTHO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD3 geni (Tüm Gen Dizi Analizi)	STARD3 gene (Sequence Analysis-All Coding Exons)	STARD3	STARD3, MLN64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD13 geni (Tüm Gen Dizi Analizi)	STARD13 gene (Sequence Analysis-All Coding Exons)	STARD13	STARD13, DLC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STARD10 geni (Tüm Gen Dizi Analizi)	STARD10 gene (Sequence Analysis-All Coding Exons)	STARD10	STARD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAP2 geni (Tüm Gen Dizi Analizi)	STAP2 gene (Sequence Analysis-All Coding Exons)	STAP2	STAP2, BKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAP1 geni (Tüm Gen Dizi Analizi)	STAP1 gene (Sequence Analysis-All Coding Exons)	STAP1	STAP1, BRDG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAMBPL1 geni (Tüm Gen Dizi Analizi)	STAMBPL1 gene (Sequence Analysis-All Coding Exons)	STAMBPL1	STAMBPL1, KIAA1373	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAM2 geni (Tüm Gen Dizi Analizi)	STAM2 gene (Sequence Analysis-All Coding Exons)	STAM2	STAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAM geni (Tüm Gen Dizi Analizi)	STAM gene (Sequence Analysis-All Coding Exons)	STAM	STAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAG2 geni (Tüm Gen Dizi Analizi)	STAG2 gene (Sequence Analysis-All Coding Exons)	STAG2	STAG2, SA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAC geni (Tüm Gen Dizi Analizi)	STAC gene (Sequence Analysis-All Coding Exons)	STAC	STAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA6 geni (Tüm Gen Dizi Analizi)	ST8SIA6 gene (Sequence Analysis-All Coding Exons)	ST8SIA6	ST8SIA6, SIAT8F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA5 geni (Tüm Gen Dizi Analizi)	ST8SIA5 gene (Sequence Analysis-All Coding Exons)	ST8SIA5	SIAT8E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA4 geni (Tüm Gen Dizi Analizi)	ST8SIA4 gene (Sequence Analysis-All Coding Exons)	ST8SIA4	PST, PST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA2 geni (Tüm Gen Dizi Analizi)	ST8SIA2 gene (Sequence Analysis-All Coding Exons)	ST8SIA2	STX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA1 geni (Tüm Gen Dizi Analizi)	ST8SIA1 gene (Sequence Analysis-All Coding Exons)	ST8SIA1	SIAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST7 geni (Tüm Gen Dizi Analizi)	ST7 gene (Sequence Analysis-All Coding Exons)	ST7	ST7, TSG7, RAY1, FAM4A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC6 geni (Tüm Gen Dizi Analizi)	ST6GALNAC6 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC6	ST6GALNAC6, SIAT7F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC5 geni (Tüm Gen Dizi Analizi)	ST6GALNAC5 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC5	ST6GALNAC5, SIAT7E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC4 geni (Tüm Gen Dizi Analizi)	ST6GALNAC4 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC4	SIAT7D, SIAT3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC3 geni (Tüm Gen Dizi Analizi)	ST6GALNAC3 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC3	ST6GALNAC3, SIAT7C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC2 geni (Tüm Gen Dizi Analizi)	ST6GALNAC2 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC2	ST6GALNAC2, SIAT7B, SIAT6.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GALNAC1 geni (Tüm Gen Dizi Analizi)	ST6GALNAC1 gene (Sequence Analysis-All Coding Exons)	ST6GALNAC1	ST6GALNAC1, SIAT7A, STY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GAL2 geni (Tüm Gen Dizi Analizi)	ST6GAL2 gene (Sequence Analysis-All Coding Exons)	ST6GAL2	ST6GALII, KIAA1877	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST6GAL1 geni (Tüm Gen Dizi Analizi)	ST6GAL1 gene (Sequence Analysis-All Coding Exons)	ST6GAL1	SIAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST5 geni (Tüm Gen Dizi Analizi)	ST5 gene (Sequence Analysis-All Coding Exons)	ST5	ST5, HTS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST3GAL6 geni (Tüm Gen Dizi Analizi)	ST3GAL6 gene (Sequence Analysis-All Coding Exons)	ST3GAL6	ST3GAL6, ST3GALVI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST3GAL4 geni (Tüm Gen Dizi Analizi)	ST3GAL4 gene (Sequence Analysis-All Coding Exons)	ST3GAL4	ST3GAL4, SIAT4C, SIAT4, CGS23, NANTA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TMEM147 geni (Tüm Gen Dizi Analizi)	TMEM147 gene (Sequence Analysis-All Coding Exons)	TMEM147		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST18 geni (Tüm Gen Dizi Analizi)	ST18 gene (Sequence Analysis-All Coding Exons)	ST18	ST18, ZNF387, KIAA0535	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST13 geni (Tüm Gen Dizi Analizi)	ST13 gene (Sequence Analysis-All Coding Exons)	ST13	ST13, P48, HOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX9 geni (Tüm Gen Dizi Analizi)	SSX9 gene (Sequence Analysis-All Coding Exons)	SSX9	SSX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX7 geni (Tüm Gen Dizi Analizi)	SSX7 gene (Sequence Analysis-All Coding Exons)	SSX7	SSX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX5 geni (Tüm Gen Dizi Analizi)	SSX5 gene (Sequence Analysis-All Coding Exons)	SSX5	SSX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX4 geni (Tüm Gen Dizi Analizi)	SSX4 gene (Sequence Analysis-All Coding Exons)	SSX4	SSX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX3 geni (Tüm Gen Dizi Analizi)	SSX3 gene (Sequence Analysis-All Coding Exons)	SSX3	SSX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSTR4 geni (Tüm Gen Dizi Analizi)	SSTR4 gene (Sequence Analysis-All Coding Exons)	SSTR4	SSTR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSTR3 geni (Tüm Gen Dizi Analizi)	SSTR3 gene (Sequence Analysis-All Coding Exons)	SSTR3	SSTR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSTR2 geni (Tüm Gen Dizi Analizi)	SSTR2 gene (Sequence Analysis-All Coding Exons)	SSTR2	SSTR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSTR1 geni (Tüm Gen Dizi Analizi)	SSTR1 gene (Sequence Analysis-All Coding Exons)	SSTR1	SSTR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SST geni (Tüm Gen Dizi Analizi)	SST gene (Sequence Analysis-All Coding Exons)	SST	SST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSRP1 geni (Tüm Gen Dizi Analizi)	SSRP1 gene (Sequence Analysis-All Coding Exons)	SSRP1	SSRP1, FACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSR3 geni (Tüm Gen Dizi Analizi)	SSR3 gene (Sequence Analysis-All Coding Exons)	SSR3	SSR3, TRAPG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSR2 geni (Tüm Gen Dizi Analizi)	SSR2 gene (Sequence Analysis-All Coding Exons)	SSR2	SSR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSR1 geni (Tüm Gen Dizi Analizi)	SSR1 gene (Sequence Analysis-All Coding Exons)	SSR1	SSR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSPO geni (Tüm Gen Dizi Analizi)	SSPO gene (Sequence Analysis-All Coding Exons)	SSPO	SSPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSPN geni (Tüm Gen Dizi Analizi)	SSPN gene (Sequence Analysis-All Coding Exons)	SSPN	SSPN, KRAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSNA1 geni (Tüm Gen Dizi Analizi)	SSNA1 gene (Sequence Analysis-All Coding Exons)	SSNA1	SSNA1, NA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSH3 geni (Tüm Gen Dizi Analizi)	SSH3 gene (Sequence Analysis-All Coding Exons)	SSH3	SSH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSH2 geni (Tüm Gen Dizi Analizi)	SSH2 gene (Sequence Analysis-All Coding Exons)	SSH2	SSH2, KIAA1725	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSH1 geni (Tüm Gen Dizi Analizi)	SSH1 gene (Sequence Analysis-All Coding Exons)	SSH1	SSH1, KIAA1298	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSFA2 geni (Tüm Gen Dizi Analizi)	SSFA2 gene (Sequence Analysis-All Coding Exons)	SSFA2	SSFA2, CS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSC4D geni (Tüm Gen Dizi Analizi)	SSC4D gene (Sequence Analysis-All Coding Exons)	SSC4D	SRCRB4D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSBP4 geni (Tüm Gen Dizi Analizi)	SSBP4 gene (Sequence Analysis-All Coding Exons)	SSBP4	SSBP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSBP3 geni (Tüm Gen Dizi Analizi)	SSBP3 gene (Sequence Analysis-All Coding Exons)	SSBP3	SSBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSBP2 geni (Tüm Gen Dizi Analizi)	SSBP2 gene (Sequence Analysis-All Coding Exons)	SSBP2	SSBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSBP1 geni (Tüm Gen Dizi Analizi)	SSBP1 gene (Sequence Analysis-All Coding Exons)	SSBP1	SSBP1, SSBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSB geni (Tüm Gen Dizi Analizi)	SSB gene (Sequence Analysis-All Coding Exons)	SSB	SSB, LARP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SS18L2 geni (Tüm Gen Dizi Analizi)	SS18L2 gene (Sequence Analysis-All Coding Exons)	SS18L2	SS18L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SS18L1 geni (Tüm Gen Dizi Analizi)	SS18L1 gene (Sequence Analysis-All Coding Exons)	SS18L1	SS18L1, KIAA0693, CREST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SS18 geni (Tüm Gen Dizi Analizi)	SS18 gene (Sequence Analysis-All Coding Exons)	SS18	SS18, SSXT, SYT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF9 geni (Tüm Gen Dizi Analizi)	SRSF9 gene (Sequence Analysis-All Coding Exons)	SRSF9	SRSF9, SFRS9, SRp30c	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF8 geni (Tüm Gen Dizi Analizi)	SRSF8 gene (Sequence Analysis-All Coding Exons)	SRSF8	SRSF8, SFRS2B, SRp46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF7 geni (Tüm Gen Dizi Analizi)	SRSF7 gene (Sequence Analysis-All Coding Exons)	SRSF7	SRSF7, SFRS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF6 geni (Tüm Gen Dizi Analizi)	SRSF6 gene (Sequence Analysis-All Coding Exons)	SRSF6	SRSF6, SFRS6, SRp55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF5 geni (Tüm Gen Dizi Analizi)	SRSF5 gene (Sequence Analysis-All Coding Exons)	SRSF5	SRSF5, SFRS5, SRp40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF4 geni (Tüm Gen Dizi Analizi)	SRSF4 gene (Sequence Analysis-All Coding Exons)	SRSF4	SRSF4, SFRS4, SRp75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF3 geni (Tüm Gen Dizi Analizi)	SRSF3 gene (Sequence Analysis-All Coding Exons)	SRSF3	SRSF3, SFRS3, SRP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SRSF2 geni (Tüm Gen Dizi Analizi)	SRSF2 gene (Sequence Analysis-All Coding Exons)	SRSF2	SRSF2, SFRS2, SC35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF11 geni (Tüm Gen Dizi Analizi)	SRSF11 gene (Sequence Analysis-All Coding Exons)	SRSF11	SRSF11, SFRS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF10 geni (Tüm Gen Dizi Analizi)	SRSF10 gene (Sequence Analysis-All Coding Exons)	SRSF10	SRSF10, FUSIP1, TASR, TASR1, TASR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRSF1 geni (Tüm Gen Dizi Analizi)	SRSF1 gene (Sequence Analysis-All Coding Exons)	SRSF1	SRSF1, SFRS1, ASF, SF2, Sf2p33a	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRRT geni (Tüm Gen Dizi Analizi)	SRRT gene (Sequence Analysis-All Coding Exons)	SRRT	SRRT, ARS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRRM4 geni (Tüm Gen Dizi Analizi)	SRRM4 gene (Sequence Analysis-All Coding Exons)	SRRM4	SRRM4, KIAA1853, NSR100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRRM2 geni (Tüm Gen Dizi Analizi)	SRRM2 gene (Sequence Analysis-All Coding Exons)	SRRM2	SRRM2, SRM300, KIAA0324	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRRM1 geni (Tüm Gen Dizi Analizi)	SRRM1 gene (Sequence Analysis-All Coding Exons)	SRRM1	SRRM1, SRM160	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRRD geni (Tüm Gen Dizi Analizi)	SRRD gene (Sequence Analysis-All Coding Exons)	SRRD	SRRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRR geni (Tüm Gen Dizi Analizi)	SRR gene (Sequence Analysis-All Coding Exons)	SRR	SRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRPX geni (Tüm Gen Dizi Analizi)	SRPX gene (Sequence Analysis-All Coding Exons)	SRPX	SRPX, ETX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRPRB geni (Tüm Gen Dizi Analizi)	SRPRB gene (Sequence Analysis-All Coding Exons)	SRPRB	SRPRB, APMCF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRPRA geni (Tüm Gen Dizi Analizi)	SRPRA gene (Sequence Analysis-All Coding Exons)	SRPRA	SRPRA, SRPR, DP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRP9 geni (Tüm Gen Dizi Analizi)	SRP9 gene (Sequence Analysis-All Coding Exons)	SRP9	SRP9, ALURBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRP68 geni (Tüm Gen Dizi Analizi)	SRP68 gene (Sequence Analysis-All Coding Exons)	SRP68	SRP68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRP54 geni (Tüm Gen Dizi Analizi)	SRP54 gene (Sequence Analysis-All Coding Exons)	SRP54	SRP54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRP19 geni (Tüm Gen Dizi Analizi)	SRP19 gene (Sequence Analysis-All Coding Exons)	SRP19	SRP19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRP14 geni (Tüm Gen Dizi Analizi)	SRP14 gene (Sequence Analysis-All Coding Exons)	SRP14	SRP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRM geni (Tüm Gen Dizi Analizi)	SRM gene (Sequence Analysis-All Coding Exons)	SRM	SRM, SPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRL geni (Tüm Gen Dizi Analizi)	SRL gene (Sequence Analysis-All Coding Exons)	SRL	SRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRI geni (Tüm Gen Dizi Analizi)	SRI gene (Sequence Analysis-All Coding Exons)	SRI	SRI, SCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRGN geni (Tüm Gen Dizi Analizi)	SRGN gene (Sequence Analysis-All Coding Exons)	SRGN	SRGN, PRG1, PRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRGAP3 geni (Tüm Gen Dizi Analizi)	SRGAP3 gene (Sequence Analysis-All Coding Exons)	SRGAP3	SRGAP3, KIAA0411, MEGAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRGAP2C geni (Tüm Gen Dizi Analizi)	SRGAP2C gene (Sequence Analysis-All Coding Exons)	SRGAP2C	SRGAP2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRGAP2B geni (Tüm Gen Dizi Analizi)	SRGAP2B gene (Sequence Analysis-All Coding Exons)	SRGAP2B	SRGAP2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRGAP2 geni (Tüm Gen Dizi Analizi)	SRGAP2 gene (Sequence Analysis-All Coding Exons)	SRGAP2	SRGAP2, KIAA0456	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRFBP1 geni (Tüm Gen Dizi Analizi)	SRFBP1 gene (Sequence Analysis-All Coding Exons)	SRFBP1	SRFBP1, P49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRF geni (Tüm Gen Dizi Analizi)	SRF gene (Sequence Analysis-All Coding Exons)	SRF	SRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SREK1 geni (Tüm Gen Dizi Analizi)	SREK1 gene (Sequence Analysis-All Coding Exons)	SREK1	SREK1, SFRS12, SRRp508	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SREBF2 geni (Tüm Gen Dizi Analizi)	SREBF2 gene (Sequence Analysis-All Coding Exons)	SREBF2	SREBF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SREBF1 geni (Tüm Gen Dizi Analizi)	SREBF1 gene (Sequence Analysis-All Coding Exons)	SREBF1	SREBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRD5A1 geni (Tüm Gen Dizi Analizi)	SRD5A1 gene (Sequence Analysis-All Coding Exons)	SRD5A1	SRD5A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRCIN1 geni (Tüm Gen Dizi Analizi)	SRCIN1 gene (Sequence Analysis-All Coding Exons)	SRCIN1	SRCIN1, SNIP, KIAA1684	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRA1 geni (Tüm Gen Dizi Analizi)	SRA1 gene (Sequence Analysis-All Coding Exons)	SRA1	SRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SQLE geni (Tüm Gen Dizi Analizi)	SQLE gene (Sequence Analysis-All Coding Exons)	SQLE	SQLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTSSB geni (Tüm Gen Dizi Analizi)	SPTSSB gene (Sequence Analysis-All Coding Exons)	SPTSSB	SPTSSB, C3orf57, SSSPTB, ADMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLR10 geni (Tüm Gen Dizi Analizi)	TLR10 gene (Sequence Analysis-All Coding Exons)	TLR10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTSSA geni (Tüm Gen Dizi Analizi)	SPTSSA gene (Sequence Analysis-All Coding Exons)	SPTSSA	SPTSSA, C14orf147, SSSPTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTLC3 geni (Tüm Gen Dizi Analizi)	SPTLC3 gene (Sequence Analysis-All Coding Exons)	SPTLC3	SPTLC3, SPTLC2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTBN5 geni (Tüm Gen Dizi Analizi)	SPTBN5 gene (Sequence Analysis-All Coding Exons)	SPTBN5	SPTBN5, BSPECV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTBN4 geni (Tüm Gen Dizi Analizi)	SPTBN4 gene (Sequence Analysis-All Coding Exons)	SPTBN4	SPTBN4, QV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPTBN1 geni (Tüm Gen Dizi Analizi)	SPTBN1 gene (Sequence Analysis-All Coding Exons)	SPTBN1	SPTBN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPSB4 geni (Tüm Gen Dizi Analizi)	SPSB4 gene (Sequence Analysis-All Coding Exons)	SPSB4	SPSB4, SSB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPSB3 geni (Tüm Gen Dizi Analizi)	SPSB3 gene (Sequence Analysis-All Coding Exons)	SPSB3	SPSB3, SSB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLK1 geni (Tüm Gen Dizi Analizi)	TLK1 gene (Sequence Analysis-All Coding Exons)	TLK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPSB2 geni (Tüm Gen Dizi Analizi)	SPSB2 gene (Sequence Analysis-All Coding Exons)	SPSB2	SPSB2, SSB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TLE4 geni (Tüm Gen Dizi Analizi)	TLE4 gene (Sequence Analysis-All Coding Exons)	TLE4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPSB1 geni (Tüm Gen Dizi Analizi)	SPSB1 gene (Sequence Analysis-All Coding Exons)	SPSB1	SPSB, SSB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRYD7 geni (Tüm Gen Dizi Analizi)	SPRYD7 gene (Sequence Analysis-All Coding Exons)	SPRYD7	CLLD6, C13orf1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRY3 geni (Tüm Gen Dizi Analizi)	SPRY3 gene (Sequence Analysis-All Coding Exons)	SPRY3	SPRY3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRY1 geni (Tüm Gen Dizi Analizi)	SPRY1 gene (Sequence Analysis-All Coding Exons)	SPRY1	SPRY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR4 geni (Tüm Gen Dizi Analizi)	SPRR4 gene (Sequence Analysis-All Coding Exons)	SPRR4	SPRR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR3 geni (Tüm Gen Dizi Analizi)	SPRR3 gene (Sequence Analysis-All Coding Exons)	SPRR3	SPRR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR2B geni (Tüm Gen Dizi Analizi)	SPRR2B gene (Sequence Analysis-All Coding Exons)	SPRR2B	SPRR2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR2A geni (Tüm Gen Dizi Analizi)	SPRR2A gene (Sequence Analysis-All Coding Exons)	SPRR2A	SPRR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR1B geni (Tüm Gen Dizi Analizi)	SPRR1B gene (Sequence Analysis-All Coding Exons)	SPRR1B	SPRR1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRR1A geni (Tüm Gen Dizi Analizi)	SPRR1A gene (Sequence Analysis-All Coding Exons)	SPRR1A	SPRR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRN geni (Tüm Gen Dizi Analizi)	SPRN gene (Sequence Analysis-All Coding Exons)	SPRN	SPRN, SHADOO, SHO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRED3 geni (Tüm Gen Dizi Analizi)	SPRED3 gene (Sequence Analysis-All Coding Exons)	SPRED3	SPRED3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPRED2 geni (Tüm Gen Dizi Analizi)	SPRED2 gene (Sequence Analysis-All Coding Exons)	SPRED2	SPRED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPPL3 geni (Tüm Gen Dizi Analizi)	SPPL3 gene (Sequence Analysis-All Coding Exons)	SPPL3	SPPL3, IMP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPPL2C geni (Tüm Gen Dizi Analizi)	SPPL2C gene (Sequence Analysis-All Coding Exons)	SPPL2C	IMP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPPL2B geni (Tüm Gen Dizi Analizi)	SPPL2B gene (Sequence Analysis-All Coding Exons)	SPPL2B	SNPPL2B, IMP4, KIAA1532	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPPL2A geni (Tüm Gen Dizi Analizi)	SPPL2A gene (Sequence Analysis-All Coding Exons)	SPPL2A	SPPL2A, IMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPP2 geni (Tüm Gen Dizi Analizi)	SPP2 gene (Sequence Analysis-All Coding Exons)	SPP2	SPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPP1 geni (Tüm Gen Dizi Analizi)	SPP1 gene (Sequence Analysis-All Coding Exons)	SPP1	SPP1, OPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPOP geni (Tüm Gen Dizi Analizi)	SPOP gene (Sequence Analysis-All Coding Exons)	SPOP	SPOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPON2 geni (Tüm Gen Dizi Analizi)	SPON2 gene (Sequence Analysis-All Coding Exons)	SPON2	SPON2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPON1 geni (Tüm Gen Dizi Analizi)	SPON1 gene (Sequence Analysis-All Coding Exons)	SPON1	SPON1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPOCK3 geni (Tüm Gen Dizi Analizi)	SPOCK3 gene (Sequence Analysis-All Coding Exons)	SPOCK3	SPOCK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPOCK2 geni (Tüm Gen Dizi Analizi)	SPOCK2 gene (Sequence Analysis-All Coding Exons)	SPOCK2	SPOCK2, KIAA0275	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TINCR geni (Tüm Gen Dizi Analizi)	TINCR gene (Sequence Analysis-All Coding Exons)	TINCR	TINCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPOCK1 geni (Tüm Gen Dizi Analizi)	SPOCK1 gene (Sequence Analysis-All Coding Exons)	SPOCK1	SPOCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPO11 geni (Tüm Gen Dizi Analizi)	SPO11 gene (Sequence Analysis-All Coding Exons)	SPO11	SPO11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPNS3 geni (Tüm Gen Dizi Analizi)	SPNS3 gene (Sequence Analysis-All Coding Exons)	SPNS3	SPNS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPNS2 geni (Tüm Gen Dizi Analizi)	SPNS2 gene (Sequence Analysis-All Coding Exons)	SPNS2	SPNS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPNS1 geni (Tüm Gen Dizi Analizi)	SPNS1 gene (Sequence Analysis-All Coding Exons)	SPNS1	SPNS1, SPIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPN geni (Tüm Gen Dizi Analizi)	SPN gene (Sequence Analysis-All Coding Exons)	SPN	SPN, LSN, CD43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINT3 geni (Tüm Gen Dizi Analizi)	SPINT3 gene (Sequence Analysis-All Coding Exons)	SPINT3	SPINT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK9 geni (Tüm Gen Dizi Analizi)	SPINK9 gene (Sequence Analysis-All Coding Exons)	SPINK9	SPINK9, LEKTI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIMM8B geni (Tüm Gen Dizi Analizi)	TIMM8B gene (Sequence Analysis-All Coding Exons)	TIMM8B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK7 geni (Tüm Gen Dizi Analizi)	SPINK7 gene (Sequence Analysis-All Coding Exons)	SPINK7	SPINK7, ECRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK6 geni (Tüm Gen Dizi Analizi)	SPINK6 gene (Sequence Analysis-All Coding Exons)	SPINK6	SPINK6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK4 geni (Tüm Gen Dizi Analizi)	SPINK4 gene (Sequence Analysis-All Coding Exons)	SPINK4	SPINK4, PEC60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK2 geni (Tüm Gen Dizi Analizi)	SPINK2 gene (Sequence Analysis-All Coding Exons)	SPINK2	SPINK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINK13 geni (Tüm Gen Dizi Analizi)	SPINK13 gene (Sequence Analysis-All Coding Exons)	SPINK13	SPINK13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIN2B geni (Tüm Gen Dizi Analizi)	SPIN2B gene (Sequence Analysis-All Coding Exons)	SPIN2B	SPIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIN2A geni (Tüm Gen Dizi Analizi)	SPIN2A gene (Sequence Analysis-All Coding Exons)	SPIN2A	DSF34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIN1 geni (Tüm Gen Dizi Analizi)	SPIN1 gene (Sequence Analysis-All Coding Exons)	SPIN1	SPIN, SPIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIDR geni (Tüm Gen Dizi Analizi)	SPIDR gene (Sequence Analysis-All Coding Exons)	SPIDR	SPIDR, KIAA0146	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPICE1 geni (Tüm Gen Dizi Analizi)	SPICE1 gene (Sequence Analysis-All Coding Exons)	SPICE1	SPICE1, CCDC52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIC geni (Tüm Gen Dizi Analizi)	SPIC gene (Sequence Analysis-All Coding Exons)	SPIC	SPIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIB geni (Tüm Gen Dizi Analizi)	SPIB gene (Sequence Analysis-All Coding Exons)	SPIB	SPIB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SPI1 geni (Tüm Gen Dizi Analizi)	SPI1 gene (Sequence Analysis-All Coding Exons)	SPI1	SPI1, SFPI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPHKAP geni (Tüm Gen Dizi Analizi)	SPHKAP gene (Sequence Analysis-All Coding Exons)	SPHKAP	SPHKAP, SKIP, KIAA1678	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPHK2 geni (Tüm Gen Dizi Analizi)	SPHK2 gene (Sequence Analysis-All Coding Exons)	SPHK2	SPHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPHK1 geni (Tüm Gen Dizi Analizi)	SPHK1 gene (Sequence Analysis-All Coding Exons)	SPHK1	SPHK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPESP1 geni (Tüm Gen Dizi Analizi)	SPESP1 gene (Sequence Analysis-All Coding Exons)	SPESP1	SPESP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPEN geni (Tüm Gen Dizi Analizi)	SPEN gene (Sequence Analysis-All Coding Exons)	SPEN	SPEN, MINT, SHARP, HIAA0929	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPEM1 geni (Tüm Gen Dizi Analizi)	SPEM1 gene (Sequence Analysis-All Coding Exons)	SPEM1	SPEM1, C17orf83	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPEF2 geni (Tüm Gen Dizi Analizi)	SPEF2 gene (Sequence Analysis-All Coding Exons)	SPEF2	SPEF2, KPL2, KIAA1770	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TIFA geni (Tüm Gen Dizi Analizi)	TIFA gene (Sequence Analysis-All Coding Exons)	TIFA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPEF1 geni (Tüm Gen Dizi Analizi)	SPEF1 gene (Sequence Analysis-All Coding Exons)	SPEF1	C20orf28, CLAMP, SPEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPECC1 geni (Tüm Gen Dizi Analizi)	SPECC1 gene (Sequence Analysis-All Coding Exons)	SPECC1	SPECC1, HCMOGT1, NSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPDYC geni (Tüm Gen Dizi Analizi)	SPDYC gene (Sequence Analysis-All Coding Exons)	SPDYC	SPDYC, RINGOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPDYA geni (Tüm Gen Dizi Analizi)	SPDYA gene (Sequence Analysis-All Coding Exons)	SPDYA	SPDYA, SPY1, RINGOA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPDL1 geni (Tüm Gen Dizi Analizi)	SPDL1 gene (Sequence Analysis-All Coding Exons)	SPDL1	SPDL1, CCDC99, SPINDLY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPDEF geni (Tüm Gen Dizi Analizi)	SPDEF gene (Sequence Analysis-All Coding Exons)	SPDEF	SPDEF, PDEF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPCS1 geni (Tüm Gen Dizi Analizi)	SPCS1 gene (Sequence Analysis-All Coding Exons)	SPCS1	SPCS1, SPC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPC25 geni (Tüm Gen Dizi Analizi)	SPC25 gene (Sequence Analysis-All Coding Exons)	SPC25	SPC25, SPBC25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPC24 geni (Tüm Gen Dizi Analizi)	SPC24 gene (Sequence Analysis-All Coding Exons)	SPC24	SPC24, SPBC24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATS2L geni (Tüm Gen Dizi Analizi)	SPATS2L gene (Sequence Analysis-All Coding Exons)	SPATS2L	SPATS2L, SGNP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATS2 geni (Tüm Gen Dizi Analizi)	SPATS2 gene (Sequence Analysis-All Coding Exons)	SPATS2	SPATS2, SPATA10, P59SCR, SCR59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATC1L geni (Tüm Gen Dizi Analizi)	SPATC1L gene (Sequence Analysis-All Coding Exons)	SPATC1L	C21orf56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATC1 geni (Tüm Gen Dizi Analizi)	SPATC1 gene (Sequence Analysis-All Coding Exons)	SPATC1	SPATC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA8 geni (Tüm Gen Dizi Analizi)	SPATA8 gene (Sequence Analysis-All Coding Exons)	SPATA8	SPATA8, SRG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA6 geni (Tüm Gen Dizi Analizi)	SPATA6 gene (Sequence Analysis-All Coding Exons)	SPATA6	SPATA6, SRF1, HASH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA46 geni (Tüm Gen Dizi Analizi)	SPATA46 gene (Sequence Analysis-All Coding Exons)	SPATA46	SPATA46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA4 geni (Tüm Gen Dizi Analizi)	SPATA4 gene (Sequence Analysis-All Coding Exons)	SPATA4	SPATA4, TSARG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA33 geni (Tüm Gen Dizi Analizi)	SPATA33 gene (Sequence Analysis-All Coding Exons)	SPATA33	SPATA33, C16orf55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA31A7 geni (Tüm Gen Dizi Analizi)	SPATA31A7 gene (Sequence Analysis-All Coding Exons)	SPATA31A7	SPATA31A7, AEP1, FAM75A7, C9orf36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA20 geni (Tüm Gen Dizi Analizi)	SPATA20 gene (Sequence Analysis-All Coding Exons)	SPATA20	SPATA20, SSP411	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA2 geni (Tüm Gen Dizi Analizi)	SPATA2 gene (Sequence Analysis-All Coding Exons)	SPATA2	SPATA2, KIAA0757	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA19 geni (Tüm Gen Dizi Analizi)	SPATA19 gene (Sequence Analysis-All Coding Exons)	SPATA19	SPATA19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA18 geni (Tüm Gen Dizi Analizi)	SPATA18 gene (Sequence Analysis-All Coding Exons)	SPATA18	SPATA18, SPETEX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA17 geni (Tüm Gen Dizi Analizi)	SPATA17 gene (Sequence Analysis-All Coding Exons)	SPATA17	SPATA17, MSRG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA13 geni (Tüm Gen Dizi Analizi)	SPATA13 gene (Sequence Analysis-All Coding Exons)	SPATA13	SPATA13, ASEF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA12 geni (Tüm Gen Dizi Analizi)	SPATA12 gene (Sequence Analysis-All Coding Exons)	SPATA12	SPATA12, SRG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXN5 geni (Tüm Gen Dizi Analizi)	SPANXN5 gene (Sequence Analysis-All Coding Exons)	SPANXN5	SPANXN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXN4 geni (Tüm Gen Dizi Analizi)	SPANXN4 gene (Sequence Analysis-All Coding Exons)	SPANXN4	SPANXN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXN3 geni (Tüm Gen Dizi Analizi)	SPANXN3 gene (Sequence Analysis-All Coding Exons)	SPANXN3	SPANXN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXN2 geni (Tüm Gen Dizi Analizi)	SPANXN2 gene (Sequence Analysis-All Coding Exons)	SPANXN2	SPANXN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXN1 geni (Tüm Gen Dizi Analizi)	SPANXN1 gene (Sequence Analysis-All Coding Exons)	SPANXN1	SPANXN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXD geni (Tüm Gen Dizi Analizi)	SPANXD gene (Sequence Analysis-All Coding Exons)	SPANXD	SPANXD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXD geni (Tüm Gen Dizi Analizi)	SPANXD gene (Sequence Analysis-All Coding Exons)	SPANXD	SPANXE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXC geni (Tüm Gen Dizi Analizi)	SPANXC gene (Sequence Analysis-All Coding Exons)	SPANXC	SPANXC, CTP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXB1 geni (Tüm Gen Dizi Analizi)	SPANXB1 gene (Sequence Analysis-All Coding Exons)	SPANXB1	SPANXB1, SPANXB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXA2 geni (Tüm Gen Dizi Analizi)	SPANXA2 gene (Sequence Analysis-All Coding Exons)	SPANXA2	SPANXA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPANXA1 geni (Tüm Gen Dizi Analizi)	SPANXA1 gene (Sequence Analysis-All Coding Exons)	SPANXA1	SPANXA1, SPANX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAM1 geni (Tüm Gen Dizi Analizi)	SPAM1 gene (Sequence Analysis-All Coding Exons)	SPAM1	SPAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THBS4 geni (Tüm Gen Dizi Analizi)	THBS4 gene (Sequence Analysis-All Coding Exons)	THBS4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG9 geni (Tüm Gen Dizi Analizi)	SPAG9 gene (Sequence Analysis-All Coding Exons)	SPAG9	SPAG9, SYD1, KIAA0516	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG8 geni (Tüm Gen Dizi Analizi)	SPAG8 gene (Sequence Analysis-All Coding Exons)	SPAG8	SPAG8, SMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG7 geni (Tüm Gen Dizi Analizi)	SPAG7 gene (Sequence Analysis-All Coding Exons)	SPAG7	SPAG7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG6 geni (Tüm Gen Dizi Analizi)	SPAG6 gene (Sequence Analysis-All Coding Exons)	SPAG6	SPAG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG5 geni (Tüm Gen Dizi Analizi)	SPAG5 gene (Sequence Analysis-All Coding Exons)	SPAG5	SPAG5, MAP126, ASTRIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG4 geni (Tüm Gen Dizi Analizi)	SPAG4 gene (Sequence Analysis-All Coding Exons)	SPAG4	SPAG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG17 geni (Tüm Gen Dizi Analizi)	SPAG17 gene (Sequence Analysis-All Coding Exons)	SPAG17	SPAG17, PF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
THAP7 geni (Tüm Gen Dizi Analizi)	THAP7 gene (Sequence Analysis-All Coding Exons)	THAP7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG16 geni (Tüm Gen Dizi Analizi)	SPAG16 gene (Sequence Analysis-All Coding Exons)	SPAG16	SPAG16, PF20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPAG11B geni (Tüm Gen Dizi Analizi)	SPAG11B gene (Sequence Analysis-All Coding Exons)	SPAG11B	SPAG11, HE2, EP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPACA5 geni (Tüm Gen Dizi Analizi)	SPACA5 gene (Sequence Analysis-All Coding Exons)	SPACA5	SPACA5, LOC389852	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPACA4 geni (Tüm Gen Dizi Analizi)	SPACA4 gene (Sequence Analysis-All Coding Exons)	SPACA4	SPACA4, SAMP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPACA3 geni (Tüm Gen Dizi Analizi)	SPACA3 gene (Sequence Analysis-All Coding Exons)	SPACA3	SPACA3, LYZL3, SLLP1, LYC3, ALLP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPACA1 geni (Tüm Gen Dizi Analizi)	SPACA1 gene (Sequence Analysis-All Coding Exons)	SPACA1	SPACA1, SAMP32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP8 geni (Tüm Gen Dizi Analizi)	SP8 gene (Sequence Analysis-All Coding Exons)	SP8	SP8, BTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP5 geni (Tüm Gen Dizi Analizi)	SP5 gene (Sequence Analysis-All Coding Exons)	SP5	SP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP4 geni (Tüm Gen Dizi Analizi)	SP4 gene (Sequence Analysis-All Coding Exons)	SP4	SP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP3 geni (Tüm Gen Dizi Analizi)	SP3 gene (Sequence Analysis-All Coding Exons)	SP3	SP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP100 geni (Tüm Gen Dizi Analizi)	SP100 gene (Sequence Analysis-All Coding Exons)	SP100	SP100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP1 geni (Tüm Gen Dizi Analizi)	SP1 gene (Sequence Analysis-All Coding Exons)	SP1	SP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX8 geni (Tüm Gen Dizi Analizi)	SOX8 gene (Sequence Analysis-All Coding Exons)	SOX8	SOX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX7 geni (Tüm Gen Dizi Analizi)	SOX7 gene (Sequence Analysis-All Coding Exons)	SOX7	SOX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX6 geni (Tüm Gen Dizi Analizi)	SOX6 gene (Sequence Analysis-All Coding Exons)	SOX6	SOX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX4 geni (Tüm Gen Dizi Analizi)	SOX4 gene (Sequence Analysis-All Coding Exons)	SOX4	SOX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX21 geni (Tüm Gen Dizi Analizi)	SOX21 gene (Sequence Analysis-All Coding Exons)	SOX21	SOX21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX2-OT geni (Tüm Gen Dizi Analizi)	SOX2-OT gene (Sequence Analysis-All Coding Exons)	SOX2-OT	SOX2OT, NCRNA00043	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX15 geni (Tüm Gen Dizi Analizi)	SOX15 gene (Sequence Analysis-All Coding Exons)	SOX15	SOX15, SOX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX14 geni (Tüm Gen Dizi Analizi)	SOX14 gene (Sequence Analysis-All Coding Exons)	SOX14	SOX14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX13 geni (Tüm Gen Dizi Analizi)	SOX13 gene (Sequence Analysis-All Coding Exons)	SOX13	SOX13, ICA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX12 geni (Tüm Gen Dizi Analizi)	SOX12 gene (Sequence Analysis-All Coding Exons)	SOX12	SOX12, SOX22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX1 geni (Tüm Gen Dizi Analizi)	SOX1 gene (Sequence Analysis-All Coding Exons)	SOX1	SOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOSTDC1 geni (Tüm Gen Dizi Analizi)	SOSTDC1 gene (Sequence Analysis-All Coding Exons)	SOSTDC1	SOSTDC1, ECTODIN, USAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORL1 geni (Tüm Gen Dizi Analizi)	SORL1 gene (Sequence Analysis-All Coding Exons)	SORL1	SORL1, LR11, SORLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORCS3 geni (Tüm Gen Dizi Analizi)	SORCS3 gene (Sequence Analysis-All Coding Exons)	SORCS3	SORCS3, KIAA1059	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORCS2 geni (Tüm Gen Dizi Analizi)	SORCS2 gene (Sequence Analysis-All Coding Exons)	SORCS2	SORCS2, KIAA1329	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORCS1 geni (Tüm Gen Dizi Analizi)	SORCS1 gene (Sequence Analysis-All Coding Exons)	SORCS1	SORCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORBS3 geni (Tüm Gen Dizi Analizi)	SORBS3 gene (Sequence Analysis-All Coding Exons)	SORBS3	SORBS3, SCAM1, SH3D4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORBS2 geni (Tüm Gen Dizi Analizi)	SORBS2 gene (Sequence Analysis-All Coding Exons)	SORBS2	SORBS2, ARGBP2, KIAA0777	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SORBS1 geni (Tüm Gen Dizi Analizi)	SORBS1 gene (Sequence Analysis-All Coding Exons)	SORBS1	SORBS1, SH3D5, SH3P12, KIAA1296	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SON geni (Tüm Gen Dizi Analizi)	SON gene (Sequence Analysis-All Coding Exons)	SON	SON, TOKIMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOHLH2 geni (Tüm Gen Dizi Analizi)	SOHLH2 gene (Sequence Analysis-All Coding Exons)	SOHLH2	SOHLH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS6 geni (Tüm Gen Dizi Analizi)	SOCS6 gene (Sequence Analysis-All Coding Exons)	SOCS6	SOCS6, SSI4, STAT4, CIS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS5 geni (Tüm Gen Dizi Analizi)	SOCS5 gene (Sequence Analysis-All Coding Exons)	SOCS5	SOCS5, CIS6, KIAA0671	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS4 geni (Tüm Gen Dizi Analizi)	SOCS4 gene (Sequence Analysis-All Coding Exons)	SOCS4	SOCS4, SOCS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS3 geni (Tüm Gen Dizi Analizi)	SOCS3 gene (Sequence Analysis-All Coding Exons)	SOCS3	SOCS3, SSI3, CIS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SOCS2-AS1 geni (Tüm Gen Dizi Analizi)	SOCS2-AS1 gene (Sequence Analysis-All Coding Exons)	SOCS2-AS1	SOCS2AS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS1 geni (Tüm Gen Dizi Analizi)	SOCS1 gene (Sequence Analysis-All Coding Exons)	SOCS1	SOCS1, SSI1, CIS1, CISH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOAT2 geni (Tüm Gen Dizi Analizi)	SOAT2 gene (Sequence Analysis-All Coding Exons)	SOAT2	SOAT2, ACACT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOAT1 geni (Tüm Gen Dizi Analizi)	SOAT1 gene (Sequence Analysis-All Coding Exons)	SOAT1	SOAT1, STAT, ACAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX9 geni (Tüm Gen Dizi Analizi)	SNX9 gene (Sequence Analysis-All Coding Exons)	SNX9	SNX9, SH3PX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX8 geni (Tüm Gen Dizi Analizi)	SNX8 gene (Sequence Analysis-All Coding Exons)	SNX8	SNX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX7 geni (Tüm Gen Dizi Analizi)	SNX7 gene (Sequence Analysis-All Coding Exons)	SNX7	SNX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX6 geni (Tüm Gen Dizi Analizi)	SNX6 gene (Sequence Analysis-All Coding Exons)	SNX6	SNX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX5 geni (Tüm Gen Dizi Analizi)	SNX5 gene (Sequence Analysis-All Coding Exons)	SNX5	SNX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX4 geni (Tüm Gen Dizi Analizi)	SNX4 gene (Sequence Analysis-All Coding Exons)	SNX4	SNX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX3 geni (Tüm Gen Dizi Analizi)	SNX3 gene (Sequence Analysis-All Coding Exons)	SNX3	SNX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX27 geni (Tüm Gen Dizi Analizi)	SNX27 gene (Sequence Analysis-All Coding Exons)	SNX27	SNX27, MRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX20 geni (Tüm Gen Dizi Analizi)	SNX20 gene (Sequence Analysis-All Coding Exons)	SNX20	SNX20, SLIC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TFPT geni (Tüm Gen Dizi Analizi)	TFPT gene (Sequence Analysis-All Coding Exons)	TFPT		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX2 geni (Tüm Gen Dizi Analizi)	SNX2 gene (Sequence Analysis-All Coding Exons)	SNX2	SNX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX17 geni (Tüm Gen Dizi Analizi)	SNX17 gene (Sequence Analysis-All Coding Exons)	SNX17	SNX17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX16 geni (Tüm Gen Dizi Analizi)	SNX16 gene (Sequence Analysis-All Coding Exons)	SNX16	SNX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX15 geni (Tüm Gen Dizi Analizi)	SNX15 gene (Sequence Analysis-All Coding Exons)	SNX15	SNX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX13 geni (Tüm Gen Dizi Analizi)	SNX13 gene (Sequence Analysis-All Coding Exons)	SNX13	SNX13, KIAA0713	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX12 geni (Tüm Gen Dizi Analizi)	SNX12 gene (Sequence Analysis-All Coding Exons)	SNX12	SNX12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX11 geni (Tüm Gen Dizi Analizi)	SNX11 gene (Sequence Analysis-All Coding Exons)	SNX11	SNX11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNX1 geni (Tüm Gen Dizi Analizi)	SNX1 gene (Sequence Analysis-All Coding Exons)	SNX1	SNX1, SNX1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNW1 geni (Tüm Gen Dizi Analizi)	SNW1 gene (Sequence Analysis-All Coding Exons)	SNW1	SKIIP, SKIP, SNW1, Bx42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNUI13 geni (Tüm Gen Dizi Analizi)	SNUI13 gene (Sequence Analysis-All Coding Exons)	SNUI13	NHP2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNTG1 geni (Tüm Gen Dizi Analizi)	SNTG1 gene (Sequence Analysis-All Coding Exons)	SNTG1	SNTG1, SYN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNTB2 geni (Tüm Gen Dizi Analizi)	SNTB2 gene (Sequence Analysis-All Coding Exons)	SNTB2	SNTB2, SNT2B2, SNTL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNTB1 geni (Tüm Gen Dizi Analizi)	SNTB1 gene (Sequence Analysis-All Coding Exons)	SNTB1	SNT2B1, A1B, SNTB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPG geni (Tüm Gen Dizi Analizi)	SNRPG gene (Sequence Analysis-All Coding Exons)	SNRPG	SNRPG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPF geni (Tüm Gen Dizi Analizi)	SNRPF gene (Sequence Analysis-All Coding Exons)	SNRPF	SNRPF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPD2 geni (Tüm Gen Dizi Analizi)	SNRPD2 gene (Sequence Analysis-All Coding Exons)	SNRPD2	SNRPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPD1 geni (Tüm Gen Dizi Analizi)	SNRPD1 gene (Sequence Analysis-All Coding Exons)	SNRPD1	SNRPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPC geni (Tüm Gen Dizi Analizi)	SNRPC gene (Sequence Analysis-All Coding Exons)	SNRPC	SNRPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPB2 geni (Tüm Gen Dizi Analizi)	SNRPB2 gene (Sequence Analysis-All Coding Exons)	SNRPB2	SNRPB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPA1 geni (Tüm Gen Dizi Analizi)	SNRPA1 gene (Sequence Analysis-All Coding Exons)	SNRPA1	SNRPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPA geni (Tüm Gen Dizi Analizi)	SNRPA gene (Sequence Analysis-All Coding Exons)	SNRPA	SNRPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRNP70 geni (Tüm Gen Dizi Analizi)	SNRNP70 gene (Sequence Analysis-All Coding Exons)	SNRNP70	SNRP70, U1RNP, RNPU1Z, RPU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRNP40 geni (Tüm Gen Dizi Analizi)	SNRNP40 gene (Sequence Analysis-All Coding Exons)	SNRNP40	SNRNP40, SPF38, PRPF8BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRK geni (Tüm Gen Dizi Analizi)	SNRK gene (Sequence Analysis-All Coding Exons)	SNRK	SNRK, KIAA0096	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNPH geni (Tüm Gen Dizi Analizi)	SNPH gene (Sequence Analysis-All Coding Exons)	SNPH	SNPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD83B geni (Tüm Gen Dizi Analizi)	SNORD83B gene (Sequence Analysis-All Coding Exons)	SNORD83B	SNORD83B, RNU83B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD83A geni (Tüm Gen Dizi Analizi)	SNORD83A gene (Sequence Analysis-All Coding Exons)	SNORD83A	SNORD83A, RNU83A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD82 geni (Tüm Gen Dizi Analizi)	SNORD82 gene (Sequence Analysis-All Coding Exons)	SNORD82	SNORD82, RNU82	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD73A geni (Tüm Gen Dizi Analizi)	SNORD73A gene (Sequence Analysis-All Coding Exons)	SNORD73A	SNORD73A, RNU73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD43 geni (Tüm Gen Dizi Analizi)	SNORD43 gene (Sequence Analysis-All Coding Exons)	SNORD43	SNORD43, RNU43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD3A geni (Tüm Gen Dizi Analizi)	SNORD3A gene (Sequence Analysis-All Coding Exons)	SNORD3A	RNU3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD30 geni (Tüm Gen Dizi Analizi)	SNORD30 gene (Sequence Analysis-All Coding Exons)	SNORD30	RNU30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD28 geni (Tüm Gen Dizi Analizi)	SNORD28 gene (Sequence Analysis-All Coding Exons)	SNORD28	RNU28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD27 geni (Tüm Gen Dizi Analizi)	SNORD27 gene (Sequence Analysis-All Coding Exons)	SNORD27	RNU27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD26 geni (Tüm Gen Dizi Analizi)	SNORD26 gene (Sequence Analysis-All Coding Exons)	SNORD26	RNU26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD25 geni (Tüm Gen Dizi Analizi)	SNORD25 gene (Sequence Analysis-All Coding Exons)	SNORD25	RNU25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD22 geni (Tüm Gen Dizi Analizi)	SNORD22 gene (Sequence Analysis-All Coding Exons)	SNORD22	RNU22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD15A geni (Tüm Gen Dizi Analizi)	SNORD15A gene (Sequence Analysis-All Coding Exons)	SNORD15A	RNU15A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD13 geni (Tüm Gen Dizi Analizi)	SNORD13 gene (Sequence Analysis-All Coding Exons)	SNORD13	SNORD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD116-1 geni (Tüm Gen Dizi Analizi)	SNORD116-1 gene (Sequence Analysis-All Coding Exons)	SNORD116-1	SNORD116-1, PWCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD115-1 geni (Tüm Gen Dizi Analizi)	SNORD115-1 gene (Sequence Analysis-All Coding Exons)	SNORD115-1	SNORD115-1, RNHBII52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD114-1 geni (Tüm Gen Dizi Analizi)	SNORD114-1 gene (Sequence Analysis-All Coding Exons)	SNORD114-1	SNORD114-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD113-1 geni (Tüm Gen Dizi Analizi)	SNORD113-1 gene (Sequence Analysis-All Coding Exons)	SNORD113-1	SNORD113-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORD112 geni (Tüm Gen Dizi Analizi)	SNORD112 gene (Sequence Analysis-All Coding Exons)	SNORD112	SNORD112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA81 geni (Tüm Gen Dizi Analizi)	SNORA81 gene (Sequence Analysis-All Coding Exons)	SNORA81	SNORA81	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA74B geni (Tüm Gen Dizi Analizi)	SNORA74B gene (Sequence Analysis-All Coding Exons)	SNORA74B	SNORA74B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA5C geni (Tüm Gen Dizi Analizi)	SNORA5C gene (Sequence Analysis-All Coding Exons)	SNORA5C	SNORA5C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA3B geni (Tüm Gen Dizi Analizi)	SNORA3B gene (Sequence Analysis-All Coding Exons)	SNORA3B	SNORA3B, SNORA45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA2C geni (Tüm Gen Dizi Analizi)	SNORA2C gene (Sequence Analysis-All Coding Exons)	SNORA2C	SNORA2C, SNORA34, ACA34, MRS291	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA26 geni (Tüm Gen Dizi Analizi)	SNORA26 gene (Sequence Analysis-All Coding Exons)	SNORA26	SNORA26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA12 geni (Tüm Gen Dizi Analizi)	SNORA12 gene (Sequence Analysis-All Coding Exons)	SNORA12	SNORA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNORA11 geni (Tüm Gen Dizi Analizi)	SNORA11 gene (Sequence Analysis-All Coding Exons)	SNORA11	SNORA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNN geni (Tüm Gen Dizi Analizi)	SNN gene (Sequence Analysis-All Coding Exons)	SNN	SNN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNHG3 geni (Tüm Gen Dizi Analizi)	SNHG3 gene (Sequence Analysis-All Coding Exons)	SNHG3	RNU17D, U17HG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNF8 geni (Tüm Gen Dizi Analizi)	SNF8 gene (Sequence Analysis-All Coding Exons)	SNF8	SNF8, VPS22, EAP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNED1 geni (Tüm Gen Dizi Analizi)	SNED1 gene (Sequence Analysis-All Coding Exons)	SNED1	SNED1, SNEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SND1 geni (Tüm Gen Dizi Analizi)	SND1 gene (Sequence Analysis-All Coding Exons)	SND1	P100, SND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNCG geni (Tüm Gen Dizi Analizi)	SNCG gene (Sequence Analysis-All Coding Exons)	SNCG	SNCG, BCSG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNCAIP geni (Tüm Gen Dizi Analizi)	SNCAIP gene (Sequence Analysis-All Coding Exons)	SNCAIP	SNCAIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPIN geni (Tüm Gen Dizi Analizi)	SNAPIN gene (Sequence Analysis-All Coding Exons)	SNAPIN	SNAPIN, SNAPAP, BLOC1S7, BLOS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPC5 geni (Tüm Gen Dizi Analizi)	SNAPC5 gene (Sequence Analysis-All Coding Exons)	SNAPC5	SNAPC5, SNAP19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPC4 geni (Tüm Gen Dizi Analizi)	SNAPC4 gene (Sequence Analysis-All Coding Exons)	SNAPC4	SNAPC4, SNAP190	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPC3 geni (Tüm Gen Dizi Analizi)	SNAPC3 gene (Sequence Analysis-All Coding Exons)	SNAPC3	SNAPC3, SNAP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPC2 geni (Tüm Gen Dizi Analizi)	SNAPC2 gene (Sequence Analysis-All Coding Exons)	SNAPC2	SNAPC2, SNAP45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAPC1 geni (Tüm Gen Dizi Analizi)	SNAPC1 gene (Sequence Analysis-All Coding Exons)	SNAPC1	SNAPC1, SNAP43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAP91 geni (Tüm Gen Dizi Analizi)	SNAP91 gene (Sequence Analysis-All Coding Exons)	SNAP91	SNAP91, AP180, KIAA0656	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAP23 geni (Tüm Gen Dizi Analizi)	SNAP23 gene (Sequence Analysis-All Coding Exons)	SNAP23	SNAP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TEKT2 geni (Tüm Gen Dizi Analizi)	TEKT2 gene (Sequence Analysis-All Coding Exons)	TEKT2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAI3 geni (Tüm Gen Dizi Analizi)	SNAI3 gene (Sequence Analysis-All Coding Exons)	SNAI3	SNAI3, SNAIL3, SMUC, ZFP293	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNAI1 geni (Tüm Gen Dizi Analizi)	SNAI1 gene (Sequence Analysis-All Coding Exons)	SNAI1	SNAI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMYD3 geni (Tüm Gen Dizi Analizi)	SMYD3 gene (Sequence Analysis-All Coding Exons)	SMYD3	SMYD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMYD2 geni (Tüm Gen Dizi Analizi)	SMYD2 gene (Sequence Analysis-All Coding Exons)	SMYD2	SMYD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMURF2 geni (Tüm Gen Dizi Analizi)	SMURF2 gene (Sequence Analysis-All Coding Exons)	SMURF2	SMURF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TECTB geni (Tüm Gen Dizi Analizi)	TECTB gene (Sequence Analysis-All Coding Exons)	TECTB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMURF1 geni (Tüm Gen Dizi Analizi)	SMURF1 gene (Sequence Analysis-All Coding Exons)	SMURF1	SMURF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMUG1 geni (Tüm Gen Dizi Analizi)	SMUG1 gene (Sequence Analysis-All Coding Exons)	SMUG1	SMUG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMTNL1 geni (Tüm Gen Dizi Analizi)	SMTNL1 gene (Sequence Analysis-All Coding Exons)	SMTNL1	SMTNL1, CHASM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMTN geni (Tüm Gen Dizi Analizi)	SMTN gene (Sequence Analysis-All Coding Exons)	SMTN	SMTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMR3B geni (Tüm Gen Dizi Analizi)	SMR3B gene (Sequence Analysis-All Coding Exons)	SMR3B	SMR3B, SMR1B, PRL3, PBII	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SMPDL3A geni (Tüm Gen Dizi Analizi)	SMPDL3A gene (Sequence Analysis-All Coding Exons)	SMPDL3A	SMPDL3A, ASML3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMPD4 geni (Tüm Gen Dizi Analizi)	SMPD4 gene (Sequence Analysis-All Coding Exons)	SMPD4	SMPD4, NSMASE3, KIAA1418	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMOX geni (Tüm Gen Dizi Analizi)	SMOX gene (Sequence Analysis-All Coding Exons)	SMOX	SMOX, SMO, PAO1, C20orf16, PAOH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMNDC1 geni (Tüm Gen Dizi Analizi)	SMNDC1 gene (Sequence Analysis-All Coding Exons)	SMNDC1	SMNR, SPF30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMIM3 geni (Tüm Gen Dizi Analizi)	SMIM3 gene (Sequence Analysis-All Coding Exons)	SMIM3	SMIM3, NID67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMG8 geni (Tüm Gen Dizi Analizi)	SMG8 gene (Sequence Analysis-All Coding Exons)	SMG8	C17orf71, SMG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TDRKH geni (Tüm Gen Dizi Analizi)	TDRKH gene (Sequence Analysis-All Coding Exons)	TDRKH		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMG7 geni (Tüm Gen Dizi Analizi)	SMG7 gene (Sequence Analysis-All Coding Exons)	SMG7	SMG7, EST1C, KIAA0250	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMG6 geni (Tüm Gen Dizi Analizi)	SMG6 gene (Sequence Analysis-All Coding Exons)	SMG6	SMG6, EST1A, KIAA0732	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMG5 geni (Tüm Gen Dizi Analizi)	SMG5 gene (Sequence Analysis-All Coding Exons)	SMG5	SMG5, EST1B, KIAA1089	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMG1 geni (Tüm Gen Dizi Analizi)	SMG1 gene (Sequence Analysis-All Coding Exons)	SMG1	SMG1, LIP, KIAA0421	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMDT1 geni (Tüm Gen Dizi Analizi)	SMDT1 gene (Sequence Analysis-All Coding Exons)	SMDT1	SMDT1, EMRE, C22orf32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMCR8 geni (Tüm Gen Dizi Analizi)	SMCR8 gene (Sequence Analysis-All Coding Exons)	SMCR8	SMCR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMCP geni (Tüm Gen Dizi Analizi)	SMCP gene (Sequence Analysis-All Coding Exons)	SMCP	SMCP, MCSP, MCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMCO4 geni (Tüm Gen Dizi Analizi)	SMCO4 gene (Sequence Analysis-All Coding Exons)	SMCO4	C11orf75, FN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMC6 geni (Tüm Gen Dizi Analizi)	SMC6 gene (Sequence Analysis-All Coding Exons)	SMC6	SMC6L1, SMC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMC5 geni (Tüm Gen Dizi Analizi)	SMC5 gene (Sequence Analysis-All Coding Exons)	SMC5	SMC5L1, SMC5, KIAA0594	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMC4 geni (Tüm Gen Dizi Analizi)	SMC4 gene (Sequence Analysis-All Coding Exons)	SMC4	CAPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMC2 geni (Tüm Gen Dizi Analizi)	SMC2 gene (Sequence Analysis-All Coding Exons)	SMC2	CAPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCD3 geni (Tüm Gen Dizi Analizi)	SMARCD3 gene (Sequence Analysis-All Coding Exons)	SMARCD3	SMARCD3, BAF60C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCD2 geni (Tüm Gen Dizi Analizi)	SMARCD2 gene (Sequence Analysis-All Coding Exons)	SMARCD2	SMARCD2, BAF60B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCD1 geni (Tüm Gen Dizi Analizi)	SMARCD1 gene (Sequence Analysis-All Coding Exons)	SMARCD1	SMARCD1, BAF60A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCC2 geni (Tüm Gen Dizi Analizi)	SMARCC2 gene (Sequence Analysis-All Coding Exons)	SMARCC2	SMARCC2, BAF170	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCC1 geni (Tüm Gen Dizi Analizi)	SMARCC1 gene (Sequence Analysis-All Coding Exons)	SMARCC1	SMARCC1, BAF155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCA5 geni (Tüm Gen Dizi Analizi)	SMARCA5 gene (Sequence Analysis-All Coding Exons)	SMARCA5	SMARCA5, SNF2H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMARCA1 geni (Tüm Gen Dizi Analizi)	SMARCA1 gene (Sequence Analysis-All Coding Exons)	SMARCA1	SMARCA1, SNF2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMAP2 geni (Tüm Gen Dizi Analizi)	SMAP2 gene (Sequence Analysis-All Coding Exons)	SMAP2	SMAP2, SMAP1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMAP1 geni (Tüm Gen Dizi Analizi)	SMAP1 gene (Sequence Analysis-All Coding Exons)	SMAP1	SMAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMAD5 geni (Tüm Gen Dizi Analizi)	SMAD5 gene (Sequence Analysis-All Coding Exons)	SMAD5	MADH5, SMAD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMAD2 geni (Tüm Gen Dizi Analizi)	SMAD2 gene (Sequence Analysis-All Coding Exons)	SMAD2	MADH2, JV18, SMAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMAD1 geni (Tüm Gen Dizi Analizi)	SMAD1 gene (Sequence Analysis-All Coding Exons)	SMAD1	SMAD1, MADH1, MADR1, BSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLX4IP geni (Tüm Gen Dizi Analizi)	SLX4IP gene (Sequence Analysis-All Coding Exons)	SLX4IP	SLX4IP, C20orf94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLX1B geni (Tüm Gen Dizi Analizi)	SLX1B gene (Sequence Analysis-All Coding Exons)	SLX1B	SLX1B, GIYD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLX1A geni (Tüm Gen Dizi Analizi)	SLX1A gene (Sequence Analysis-All Coding Exons)	SLX1A	SLX1A, GIYD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLPI geni (Tüm Gen Dizi Analizi)	SLPI gene (Sequence Analysis-All Coding Exons)	SLPI	SLPI, HUSI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLN geni (Tüm Gen Dizi Analizi)	SLN gene (Sequence Analysis-All Coding Exons)	SLN	SLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLMAP geni (Tüm Gen Dizi Analizi)	SLMAP gene (Sequence Analysis-All Coding Exons)	SLMAP	SLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLK geni (Tüm Gen Dizi Analizi)	SLK gene (Sequence Analysis-All Coding Exons)	SLK	SLK, LOSK, KIAA0204	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLITRK5 geni (Tüm Gen Dizi Analizi)	SLITRK5 gene (Sequence Analysis-All Coding Exons)	SLITRK5	SLITRK5, KIAA0918	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLITRK4 geni (Tüm Gen Dizi Analizi)	SLITRK4 gene (Sequence Analysis-All Coding Exons)	SLITRK4	SLITRK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLITRK3 geni (Tüm Gen Dizi Analizi)	SLITRK3 gene (Sequence Analysis-All Coding Exons)	SLITRK3	SLITRK3, KIAA0848	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCFL5 geni (Tüm Gen Dizi Analizi)	TCFL5 gene (Sequence Analysis-All Coding Exons)	TCFL5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLITRK2 geni (Tüm Gen Dizi Analizi)	SLITRK2 gene (Sequence Analysis-All Coding Exons)	SLITRK2	SLITRK2, KIAA1854	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLIT3 geni (Tüm Gen Dizi Analizi)	SLIT3 gene (Sequence Analysis-All Coding Exons)	SLIT3	SLIT3, SLIL2, MEGF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLIT2 geni (Tüm Gen Dizi Analizi)	SLIT2 gene (Sequence Analysis-All Coding Exons)	SLIT2	SLIT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLIT1 geni (Tüm Gen Dizi Analizi)	SLIT1 gene (Sequence Analysis-All Coding Exons)	SLIT1	SLIT1, SLIL1, MEGF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLIRP geni (Tüm Gen Dizi Analizi)	SLIRP gene (Sequence Analysis-All Coding Exons)	SLIRP	SLIRP, C14orf156	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLFN5 geni (Tüm Gen Dizi Analizi)	SLFN5 gene (Sequence Analysis-All Coding Exons)	SLFN5	SLFN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLFN13 geni (Tüm Gen Dizi Analizi)	SLFN13 gene (Sequence Analysis-All Coding Exons)	SLFN13	SLFN13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLFN12L geni (Tüm Gen Dizi Analizi)	SLFN12L gene (Sequence Analysis-All Coding Exons)	SLFN12L	SLFN12L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLFN12 geni (Tüm Gen Dizi Analizi)	SLFN12 gene (Sequence Analysis-All Coding Exons)	SLFN12	SLFN12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLFN11 geni (Tüm Gen Dizi Analizi)	SLFN11 gene (Sequence Analysis-All Coding Exons)	SLFN11	SLFN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLF2 geni (Tüm Gen Dizi Analizi)	SLF2 gene (Sequence Analysis-All Coding Exons)	SLF2	FAM178A, C10orf6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO6A1 geni (Tüm Gen Dizi Analizi)	SLCO6A1 gene (Sequence Analysis-All Coding Exons)	SLCO6A1	SLCO6A1, GST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO5A1 geni (Tüm Gen Dizi Analizi)	SLCO5A1 gene (Sequence Analysis-All Coding Exons)	SLCO5A1	SLCO5A1, OATRPI4, SLC21A15, OATPJ, OATP5A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO4C1 geni (Tüm Gen Dizi Analizi)	SLCO4C1 gene (Sequence Analysis-All Coding Exons)	SLCO4C1	SLCO4C1, OATP4C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO4A1 geni (Tüm Gen Dizi Analizi)	SLCO4A1 gene (Sequence Analysis-All Coding Exons)	SLCO4A1	SLCO4A1, OATPE, OATP4A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO3A1 geni (Tüm Gen Dizi Analizi)	SLCO3A1 gene (Sequence Analysis-All Coding Exons)	SLCO3A1	SLCO3A1, OATPD, OATP3A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO2B1 geni (Tüm Gen Dizi Analizi)	SLCO2B1 gene (Sequence Analysis-All Coding Exons)	SLCO2B1	SLC21A9, OATPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TCEA2 geni (Tüm Gen Dizi Analizi)	TCEA2 gene (Sequence Analysis-All Coding Exons)	TCEA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO1C1 geni (Tüm Gen Dizi Analizi)	SLCO1C1 gene (Sequence Analysis-All Coding Exons)	SLCO1C1	SLCO1C1, OATPF, OATP14, SLC21A14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLCO1A2 geni (Tüm Gen Dizi Analizi)	SLCO1A2 gene (Sequence Analysis-All Coding Exons)	SLCO1A2	SLC21A3, OATP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9C1 geni (Tüm Gen Dizi Analizi)	SLC9C1 gene (Sequence Analysis-All Coding Exons)	SLC9C1	SLC9A10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9B2 geni (Tüm Gen Dizi Analizi)	SLC9B2 gene (Sequence Analysis-All Coding Exons)	SLC9B2	NHEDC2, NHA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9B1 geni (Tüm Gen Dizi Analizi)	SLC9B1 gene (Sequence Analysis-All Coding Exons)	SLC9B1	NHEDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A8 geni (Tüm Gen Dizi Analizi)	SLC9A8 gene (Sequence Analysis-All Coding Exons)	SLC9A8	SLC9A8, NHE8, KIAA0939	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A7 geni (Tüm Gen Dizi Analizi)	SLC9A7 gene (Sequence Analysis-All Coding Exons)	SLC9A7	SLC9A7, NHE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A5 geni (Tüm Gen Dizi Analizi)	SLC9A5 gene (Sequence Analysis-All Coding Exons)	SLC9A5	SLC9A5, NHE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A4 geni (Tüm Gen Dizi Analizi)	SLC9A4 gene (Sequence Analysis-All Coding Exons)	SLC9A4	SLC9A4, NHE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A3R2 geni (Tüm Gen Dizi Analizi)	SLC9A3R2 gene (Sequence Analysis-All Coding Exons)	SLC9A3R2	SLC9A3R2, SIP1, NHERF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC9A2 geni (Tüm Gen Dizi Analizi)	SLC9A2 gene (Sequence Analysis-All Coding Exons)	SLC9A2	SLC9A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC8B1 geni (Tüm Gen Dizi Analizi)	SLC8B1 gene (Sequence Analysis-All Coding Exons)	SLC8B1	SLC24A6, NCKX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC8A2 geni (Tüm Gen Dizi Analizi)	SLC8A2 gene (Sequence Analysis-All Coding Exons)	SLC8A2	SLC8A2, NCX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC8A1 geni (Tüm Gen Dizi Analizi)	SLC8A1 gene (Sequence Analysis-All Coding Exons)	SLC8A1	SLC8A1, NCX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A8 geni (Tüm Gen Dizi Analizi)	SLC7A8 gene (Sequence Analysis-All Coding Exons)	SLC7A8	SLC7A8, LAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A6 geni (Tüm Gen Dizi Analizi)	SLC7A6 gene (Sequence Analysis-All Coding Exons)	SLC7A6	SLC7A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A5 geni (Tüm Gen Dizi Analizi)	SLC7A5 gene (Sequence Analysis-All Coding Exons)	SLC7A5	SLC7A5, D16S469E, MPE16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A4 geni (Tüm Gen Dizi Analizi)	SLC7A4 gene (Sequence Analysis-All Coding Exons)	SLC7A4	SLC7A4, CAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A3 geni (Tüm Gen Dizi Analizi)	SLC7A3 gene (Sequence Analysis-All Coding Exons)	SLC7A3	SLC7A3, CAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A2 geni (Tüm Gen Dizi Analizi)	SLC7A2 gene (Sequence Analysis-All Coding Exons)	SLC7A2	SLC7A2, ATRC2, HCAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A13 geni (Tüm Gen Dizi Analizi)	SLC7A13 gene (Sequence Analysis-All Coding Exons)	SLC7A13	SLC7A13, AGT1, XAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A11 geni (Tüm Gen Dizi Analizi)	SLC7A11 gene (Sequence Analysis-All Coding Exons)	SLC7A11	SLC7A11, XCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A10 geni (Tüm Gen Dizi Analizi)	SLC7A10 gene (Sequence Analysis-All Coding Exons)	SLC7A10	SLC7A10, ASC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC7A1 geni (Tüm Gen Dizi Analizi)	SLC7A1 gene (Sequence Analysis-All Coding Exons)	SLC7A1	SLC7A1, ATRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A7 geni (Tüm Gen Dizi Analizi)	SLC6A7 gene (Sequence Analysis-All Coding Exons)	SLC6A7	SLC6A7, PROT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A6 geni (Tüm Gen Dizi Analizi)	SLC6A6 gene (Sequence Analysis-All Coding Exons)	SLC6A6	SLC6A6, TAUT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A18 geni (Tüm Gen Dizi Analizi)	SLC6A18 gene (Sequence Analysis-All Coding Exons)	SLC6A18	SLC6A18, XTRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A16 geni (Tüm Gen Dizi Analizi)	SLC6A16 gene (Sequence Analysis-All Coding Exons)	SLC6A16	SLC6A16, NTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBPL2 geni (Tüm Gen Dizi Analizi)	TBPL2 gene (Sequence Analysis-All Coding Exons)	TBPL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TBPL1 geni (Tüm Gen Dizi Analizi)	TBPL1 gene (Sequence Analysis-All Coding Exons)	TBPL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A15 geni (Tüm Gen Dizi Analizi)	SLC6A15 gene (Sequence Analysis-All Coding Exons)	SLC6A15	SLC6A15, SBAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A13 geni (Tüm Gen Dizi Analizi)	SLC6A13 gene (Sequence Analysis-All Coding Exons)	SLC6A13	SLC6A13, GAT2, GAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SLC6A12 geni (Tüm Gen Dizi Analizi)	SLC6A12 gene (Sequence Analysis-All Coding Exons)	SLC6A12	SLC6A12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC6A11 geni (Tüm Gen Dizi Analizi)	SLC6A11 gene (Sequence Analysis-All Coding Exons)	SLC6A11	SLC6A11, GAT3, GAT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC5A8 geni (Tüm Gen Dizi Analizi)	SLC5A8 gene (Sequence Analysis-All Coding Exons)	SLC5A8	SLC5A8, AIT, SMCT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC5A6 geni (Tüm Gen Dizi Analizi)	SLC5A6 gene (Sequence Analysis-All Coding Exons)	SLC5A6	SLC5A6, SMVT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC5A3 geni (Tüm Gen Dizi Analizi)	SLC5A3 gene (Sequence Analysis-All Coding Exons)	SLC5A3	SLC5A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC5A12 geni (Tüm Gen Dizi Analizi)	SLC5A12 gene (Sequence Analysis-All Coding Exons)	SLC5A12	SLC5A12, SMCT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC5A11 geni (Tüm Gen Dizi Analizi)	SLC5A11 gene (Sequence Analysis-All Coding Exons)	SLC5A11	SLC5A11, KST1, SQLT6, SMIT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC51B geni (Tüm Gen Dizi Analizi)	SLC51B gene (Sequence Analysis-All Coding Exons)	SLC51B	OSTB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC51A geni (Tüm Gen Dizi Analizi)	SLC51A gene (Sequence Analysis-All Coding Exons)	SLC51A	OSTA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC50A1 geni (Tüm Gen Dizi Analizi)	SLC50A1 gene (Sequence Analysis-All Coding Exons)	SLC50A1	SLC50A1, RAG1AP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A8 geni (Tüm Gen Dizi Analizi)	SLC4A8 gene (Sequence Analysis-All Coding Exons)	SLC4A8	SLC4A8, NBC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A7 geni (Tüm Gen Dizi Analizi)	SLC4A7 gene (Sequence Analysis-All Coding Exons)	SLC4A7	SLC4A7, NBC2, NBC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A5 geni (Tüm Gen Dizi Analizi)	SLC4A5 gene (Sequence Analysis-All Coding Exons)	SLC4A5	SLC4A5, NBC4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A3 geni (Tüm Gen Dizi Analizi)	SLC4A3 gene (Sequence Analysis-All Coding Exons)	SLC4A3	SLC2C, AE3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A2 geni (Tüm Gen Dizi Analizi)	SLC4A2 gene (Sequence Analysis-All Coding Exons)	SLC4A2	SLC4A2, AE2, EPB3L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A1AP geni (Tüm Gen Dizi Analizi)	SLC4A1AP gene (Sequence Analysis-All Coding Exons)	SLC4A1AP	SLC4A1AP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A10 geni (Tüm Gen Dizi Analizi)	SLC4A10 gene (Sequence Analysis-All Coding Exons)	SLC4A10	SLC4A10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4B1 geni (Tüm Gen Dizi Analizi)	SLC4B1 gene (Sequence Analysis-All Coding Exons)	SLC4B1	HRG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC47A2 geni (Tüm Gen Dizi Analizi)	SLC47A2 gene (Sequence Analysis-All Coding Exons)	SLC47A2	SLC47A2, MATE2, FLJ31196, MATE2K	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC47A1 geni (Tüm Gen Dizi Analizi)	SLC47A1 gene (Sequence Analysis-All Coding Exons)	SLC47A1	SLC47A1, MATE1, FLJ10847	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC46A3 geni (Tüm Gen Dizi Analizi)	SLC46A3 gene (Sequence Analysis-All Coding Exons)	SLC46A3	SLC46A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC46A2 geni (Tüm Gen Dizi Analizi)	SLC46A2 gene (Sequence Analysis-All Coding Exons)	SLC46A2	SLC46A2, TSCOT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC45A3 geni (Tüm Gen Dizi Analizi)	SLC45A3 gene (Sequence Analysis-All Coding Exons)	SLC45A3	SLC45A3, PRST, PCANAP6, dPOA6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC45A1 geni (Tüm Gen Dizi Analizi)	SLC45A1 gene (Sequence Analysis-All Coding Exons)	SLC45A1	SLC45A1, DNB5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC44A4 geni (Tüm Gen Dizi Analizi)	SLC44A4 gene (Sequence Analysis-All Coding Exons)	SLC44A4	SLC44A4, CTL4, C6orf29	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC44A2 geni (Tüm Gen Dizi Analizi)	SLC44A2 gene (Sequence Analysis-All Coding Exons)	SLC44A2	SLC44A2, CTL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC44A1 geni (Tüm Gen Dizi Analizi)	SLC44A1 gene (Sequence Analysis-All Coding Exons)	SLC44A1	SLC44A1, CTL1, CDW92	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC43A2 geni (Tüm Gen Dizi Analizi)	SLC43A2 gene (Sequence Analysis-All Coding Exons)	SLC43A2	SLC43A2, LAT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC43A1 geni (Tüm Gen Dizi Analizi)	SLC43A1 gene (Sequence Analysis-All Coding Exons)	SLC43A1	SLC43A1, LAT3, POV1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC41A3 geni (Tüm Gen Dizi Analizi)	SLC41A3 gene (Sequence Analysis-All Coding Exons)	SLC41A3	SLC41A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC41A2 geni (Tüm Gen Dizi Analizi)	SLC41A2 gene (Sequence Analysis-All Coding Exons)	SLC41A2	SLC41A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC41A1 geni (Tüm Gen Dizi Analizi)	SLC41A1 gene (Sequence Analysis-All Coding Exons)	SLC41A1	SLC41A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC3A2 geni (Tüm Gen Dizi Analizi)	SLC3A2 gene (Sequence Analysis-All Coding Exons)	SLC3A2	SLC3A2, MDU1, NACAE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A7 geni (Tüm Gen Dizi Analizi)	SLC39A7 gene (Sequence Analysis-All Coding Exons)	SLC39A7	SLC39A7, D6S2244E, HKE4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A3 geni (Tüm Gen Dizi Analizi)	SLC39A3 gene (Sequence Analysis-All Coding Exons)	SLC39A3	SLC39A3, ZIP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A2 geni (Tüm Gen Dizi Analizi)	SLC39A2 gene (Sequence Analysis-All Coding Exons)	SLC39A2	SLC39A2, ZIP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A11 geni (Tüm Gen Dizi Analizi)	SLC39A11 gene (Sequence Analysis-All Coding Exons)	SLC39A11	SLC39A11, ZIP11, C17orf26	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A1 geni (Tüm Gen Dizi Analizi)	SLC39A1 gene (Sequence Analysis-All Coding Exons)	SLC39A1	SLC39A1, ZIRTL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A9 geni (Tüm Gen Dizi Analizi)	SLC38A9 gene (Sequence Analysis-All Coding Exons)	SLC38A9	SLC38A9, URLC11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A7 geni (Tüm Gen Dizi Analizi)	SLC38A7 gene (Sequence Analysis-All Coding Exons)	SLC38A7	SLC38A7, SNA7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A6 geni (Tüm Gen Dizi Analizi)	SLC38A6 gene (Sequence Analysis-All Coding Exons)	SLC38A6	SLC38A6, SNA6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A5 geni (Tüm Gen Dizi Analizi)	SLC38A5 gene (Sequence Analysis-All Coding Exons)	SLC38A5	SLC38A5, SN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A4 geni (Tüm Gen Dizi Analizi)	SLC38A4 gene (Sequence Analysis-All Coding Exons)	SLC38A4	SLC38A4, NAT3, ATA3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A3 geni (Tüm Gen Dizi Analizi)	SLC38A3 gene (Sequence Analysis-All Coding Exons)	SLC38A3	SLC38A3, SN1, G17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A2 geni (Tüm Gen Dizi Analizi)	SLC38A2 gene (Sequence Analysis-All Coding Exons)	SLC38A2	SLC38A2, ATA2, KIAA1382	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A11 geni (Tüm Gen Dizi Analizi)	SLC38A11 gene (Sequence Analysis-All Coding Exons)	SLC38A11	SLC38A11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A10 geni (Tüm Gen Dizi Analizi)	SLC38A10 gene (Sequence Analysis-All Coding Exons)	SLC38A10	SLC38A10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC38A1 geni (Tüm Gen Dizi Analizi)	SLC38A1 gene (Sequence Analysis-All Coding Exons)	SLC38A1	SLC38A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC37A1 geni (Tüm Gen Dizi Analizi)	SLC37A1 gene (Sequence Analysis-All Coding Exons)	SLC37A1	SLC37A1, G3PP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC36A4 geni (Tüm Gen Dizi Analizi)	SLC36A4 gene (Sequence Analysis-All Coding Exons)	SLC36A4	SLC36A4, PAT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC36A1 geni (Tüm Gen Dizi Analizi)	SLC36A1 gene (Sequence Analysis-All Coding Exons)	SLC36A1	SLC36A1, LYAAT1, PAT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35G5 geni (Tüm Gen Dizi Analizi)	SLC35G5 gene (Sequence Analysis-All Coding Exons)	SLC35G5	SLC35G5, AMAC, AMAC1L2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35G1 geni (Tüm Gen Dizi Analizi)	SLC35G1 gene (Sequence Analysis-All Coding Exons)	SLC35G1	SLC35G1, POST, TMEM20, C10orf60	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35D3 geni (Tüm Gen Dizi Analizi)	SLC35D3 gene (Sequence Analysis-All Coding Exons)	SLC35D3	SLC35D3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35D2 geni (Tüm Gen Dizi Analizi)	SLC35D2 gene (Sequence Analysis-All Coding Exons)	SLC35D2	SLC35D2, HFRC1, SQV7L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35B4 geni (Tüm Gen Dizi Analizi)	SLC35B4 gene (Sequence Analysis-All Coding Exons)	SLC35B4	SLC35B4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35B3 geni (Tüm Gen Dizi Analizi)	SLC35B3 gene (Sequence Analysis-All Coding Exons)	SLC35B3	SLC35B3, PAPST2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35B2 geni (Tüm Gen Dizi Analizi)	SLC35B2 gene (Sequence Analysis-All Coding Exons)	SLC35B2	SLC35B2, PAPST1, SLL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC35B1 geni (Tüm Gen Dizi Analizi)	SLC35B1 gene (Sequence Analysis-All Coding Exons)	SLC35B1	SLC35B1, UGTREL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC32A1 geni (Tüm Gen Dizi Analizi)	SLC32A1 gene (Sequence Analysis-All Coding Exons)	SLC32A1	SLC32A1, VGAT, VIAAT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC31A2 geni (Tüm Gen Dizi Analizi)	SLC31A2 gene (Sequence Analysis-All Coding Exons)	SLC31A2	SLC13A2, COPT2, CTR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC31A1 geni (Tüm Gen Dizi Analizi)	SLC31A1 gene (Sequence Analysis-All Coding Exons)	SLC31A1	SLC31A1, COPT1, CTR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A9 geni (Tüm Gen Dizi Analizi)	SLC30A9 gene (Sequence Analysis-All Coding Exons)	SLC30A9	SLC30A9, C4orf1, HUEL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A7 geni (Tüm Gen Dizi Analizi)	SLC30A7 gene (Sequence Analysis-All Coding Exons)	SLC30A7	SLC30A7, ZNT7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A6 geni (Tüm Gen Dizi Analizi)	SLC30A6 gene (Sequence Analysis-All Coding Exons)	SLC30A6	SLC30A6, ZNT6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A5 geni (Tüm Gen Dizi Analizi)	SLC30A5 gene (Sequence Analysis-All Coding Exons)	SLC30A5	SLC30A5, ZNT5, ZTL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A4 geni (Tüm Gen Dizi Analizi)	SLC30A4 gene (Sequence Analysis-All Coding Exons)	SLC30A4	SLC30A4, ZNT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A3 geni (Tüm Gen Dizi Analizi)	SLC30A3 gene (Sequence Analysis-All Coding Exons)	SLC30A3	SLC30A3, ZNT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC30A1 geni (Tüm Gen Dizi Analizi)	SLC30A1 gene (Sequence Analysis-All Coding Exons)	SLC30A1	SLC30A1, ZNT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A8 geni (Tüm Gen Dizi Analizi)	SLC2A8 gene (Sequence Analysis-All Coding Exons)	SLC2A8	SLC2A8, GLUT8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A7 geni (Tüm Gen Dizi Analizi)	SLC2A7 gene (Sequence Analysis-All Coding Exons)	SLC2A7	SLC2A7, GLUT7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A6 geni (Tüm Gen Dizi Analizi)	SLC2A6 gene (Sequence Analysis-All Coding Exons)	SLC2A6	SLC2A6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A5 geni (Tüm Gen Dizi Analizi)	SLC2A5 gene (Sequence Analysis-All Coding Exons)	SLC2A5	SLC2A5, GLUT5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A4 geni (Tüm Gen Dizi Analizi)	SLC2A4 gene (Sequence Analysis-All Coding Exons)	SLC2A4	SLC2A4, GLUT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A3 geni (Tüm Gen Dizi Analizi)	SLC2A3 gene (Sequence Analysis-All Coding Exons)	SLC2A3	SLC2A3, GLUT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A14 geni (Tüm Gen Dizi Analizi)	SLC2A14 gene (Sequence Analysis-All Coding Exons)	SLC2A14	SLC2A14, GLUT14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A13 geni (Tüm Gen Dizi Analizi)	SLC2A13 gene (Sequence Analysis-All Coding Exons)	SLC2A13	SLC2A13, HMIT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A12 geni (Tüm Gen Dizi Analizi)	SLC2A12 gene (Sequence Analysis-All Coding Exons)	SLC2A12	SLC2A12, GLUT12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A11 geni (Tüm Gen Dizi Analizi)	SLC2A11 gene (Sequence Analysis-All Coding Exons)	SLC2A11	SLC2A11, GLUT11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC2A1 geni (Tüm Gen Dizi Analizi)	SLC2A1 gene (Sequence Analysis-All Coding Exons)	SLC2A1	HTLVR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269	PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
SLC29A4 geni (Tüm Gen Dizi Analizi)	SLC29A4 gene (Sequence Analysis-All Coding Exons)	SLC29A4	SLC29A4, PMAT, ENT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC29A2 geni (Tüm Gen Dizi Analizi)	SLC29A2 gene (Sequence Analysis-All Coding Exons)	SLC29A2	SLC29A2, HNP36, DER12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC29A1 geni (Tüm Gen Dizi Analizi)	SLC29A1 gene (Sequence Analysis-All Coding Exons)	SLC29A1	SLC29A1, ENT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC28A3 geni (Tüm Gen Dizi Analizi)	SLC28A3 gene (Sequence Analysis-All Coding Exons)	SLC28A3	SLC28A3, CNT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC28A2 geni (Tüm Gen Dizi Analizi)	SLC28A2 gene (Sequence Analysis-All Coding Exons)	SLC28A2	SLC28A2, CNT2, SPNT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC28A1 geni (Tüm Gen Dizi Analizi)	SLC28A1 gene (Sequence Analysis-All Coding Exons)	SLC28A1	SLC28A1, CNT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC27A5 geni (Tüm Gen Dizi Analizi)	SLC27A5 gene (Sequence Analysis-All Coding Exons)	SLC27A5	SLC27A5, FATP5, VLACSR, VLCSH2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC27A3 geni (Tüm Gen Dizi Analizi)	SLC27A3 gene (Sequence Analysis-All Coding Exons)	SLC27A3	SLC27A3, FATP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC27A2 geni (Tüm Gen Dizi Analizi)	SLC27A2 gene (Sequence Analysis-All Coding Exons)	SLC27A2	SLC27A2, FACVL1, VLACS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SLC27A1 geni (Tüm Gen Dizi Analizi)	SLC27A1 gene (Sequence Analysis-All Coding Exons)	SLC27A1	SLC27A1, FATP, FATP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC26A9 geni (Tüm Gen Dizi Analizi)	SLC26A9 gene (Sequence Analysis-All Coding Exons)	SLC26A9	SLC26A9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC26A6 geni (Tüm Gen Dizi Analizi)	SLC26A6 gene (Sequence Analysis-All Coding Exons)	SLC26A6	SLC26A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC26A11 geni (Tüm Gen Dizi Analizi)	SLC26A11 gene (Sequence Analysis-All Coding Exons)	SLC26A11	SLC26A11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A6 geni (Tüm Gen Dizi Analizi)	SLC25A6 gene (Sequence Analysis-All Coding Exons)	SLC25A6	SLC25A6, ANT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A6 geni (Tüm Gen Dizi Analizi)	SLC25A6 gene (Sequence Analysis-All Coding Exons)	SLC25A6	ANT3Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A53 geni (Tüm Gen Dizi Analizi)	SLC25A53 gene (Sequence Analysis-All Coding Exons)	SLC25A53	SLC25A53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A52 geni (Tüm Gen Dizi Analizi)	SLC25A52 gene (Sequence Analysis-All Coding Exons)	SLC25A52	SLC25A52, MCART2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A5 geni (Tüm Gen Dizi Analizi)	SLC25A5 gene (Sequence Analysis-All Coding Exons)	SLC25A5	SLC25A5, ANT2, T3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A48 geni (Tüm Gen Dizi Analizi)	SLC25A48 gene (Sequence Analysis-All Coding Exons)	SLC25A48	SLC25A48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A47 geni (Tüm Gen Dizi Analizi)	SLC25A47 gene (Sequence Analysis-All Coding Exons)	SLC25A47	HDMCP, C14orf68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A45 geni (Tüm Gen Dizi Analizi)	SLC25A45 gene (Sequence Analysis-All Coding Exons)	SLC25A45	SLC25A45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A44 geni (Tüm Gen Dizi Analizi)	SLC25A44 gene (Sequence Analysis-All Coding Exons)	SLC25A44	SLC25A44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A43 geni (Tüm Gen Dizi Analizi)	SLC25A43 gene (Sequence Analysis-All Coding Exons)	SLC25A43	SLC25A43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A42 geni (Tüm Gen Dizi Analizi)	SLC25A42 gene (Sequence Analysis-All Coding Exons)	SLC25A42	SLC25A42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A41 geni (Tüm Gen Dizi Analizi)	SLC25A41 gene (Sequence Analysis-All Coding Exons)	SLC25A41	SLC25A41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A40 geni (Tüm Gen Dizi Analizi)	SLC25A40 gene (Sequence Analysis-All Coding Exons)	SLC25A40	SLC25A40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A39 geni (Tüm Gen Dizi Analizi)	SLC25A39 gene (Sequence Analysis-All Coding Exons)	SLC25A39	SLC25A39, CGI69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A37 geni (Tüm Gen Dizi Analizi)	SLC25A37 gene (Sequence Analysis-All Coding Exons)	SLC25A37	SLC25A37, MFRN, MFRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A36 geni (Tüm Gen Dizi Analizi)	SLC25A36 gene (Sequence Analysis-All Coding Exons)	SLC25A36	SLC25A36, PNC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A35 geni (Tüm Gen Dizi Analizi)	SLC25A35 gene (Sequence Analysis-All Coding Exons)	SLC25A35	SLC25A35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A34 geni (Tüm Gen Dizi Analizi)	SLC25A34 gene (Sequence Analysis-All Coding Exons)	SLC25A34	SLC25A34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A33 geni (Tüm Gen Dizi Analizi)	SLC25A33 gene (Sequence Analysis-All Coding Exons)	SLC25A33	SLC25A33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A31 geni (Tüm Gen Dizi Analizi)	SLC25A31 gene (Sequence Analysis-All Coding Exons)	SLC25A31	SLC25A31, ANT4, AAC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF8 geni (Tüm Gen Dizi Analizi)	TAF8 gene (Sequence Analysis-All Coding Exons)	TAF8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A30 geni (Tüm Gen Dizi Analizi)	SLC25A30 gene (Sequence Analysis-All Coding Exons)	SLC25A30	SLC25A30, KMCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A29 geni (Tüm Gen Dizi Analizi)	SLC25A29 gene (Sequence Analysis-All Coding Exons)	SLC25A29	SLC25A29, CACL, ORNT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF6L geni (Tüm Gen Dizi Analizi)	TAF6L gene (Sequence Analysis-All Coding Exons)	TAF6L		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A28 geni (Tüm Gen Dizi Analizi)	SLC25A28 gene (Sequence Analysis-All Coding Exons)	SLC25A28	SLC25A28, MRS4L, NPD016	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A27 geni (Tüm Gen Dizi Analizi)	SLC25A27 gene (Sequence Analysis-All Coding Exons)	SLC25A27	SLC25A27, UCP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A21 geni (Tüm Gen Dizi Analizi)	SLC25A21 gene (Sequence Analysis-All Coding Exons)	SLC25A21	SLC25A21, ODC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A2 geni (Tüm Gen Dizi Analizi)	SLC25A2 gene (Sequence Analysis-All Coding Exons)	SLC25A2	SLC25A2, ORNT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A18 geni (Tüm Gen Dizi Analizi)	SLC25A18 gene (Sequence Analysis-All Coding Exons)	SLC25A18	SLC25A18, GC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A17 geni (Tüm Gen Dizi Analizi)	SLC25A17 gene (Sequence Analysis-All Coding Exons)	SLC25A17	SLC25A17, PMP34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF1L geni (Tüm Gen Dizi Analizi)	TAF1L gene (Sequence Analysis-All Coding Exons)	TAF1L		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A16 geni (Tüm Gen Dizi Analizi)	SLC25A16 gene (Sequence Analysis-All Coding Exons)	SLC25A16	SLC25A16, D10S105E, GDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A14 geni (Tüm Gen Dizi Analizi)	SLC25A14 gene (Sequence Analysis-All Coding Exons)	SLC25A14	SLC25A14, BMCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A11 geni (Tüm Gen Dizi Analizi)	SLC25A11 gene (Sequence Analysis-All Coding Exons)	SLC25A11	SLC25A11, SLC20A4, OGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A10 geni (Tüm Gen Dizi Analizi)	SLC25A10 gene (Sequence Analysis-All Coding Exons)	SLC25A10	SLC25A10, DIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC24A3 geni (Tüm Gen Dizi Analizi)	SLC24A3 gene (Sequence Analysis-All Coding Exons)	SLC24A3	SLC24A3, NCKX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
TAF13 geni (Tüm Gen Dizi Analizi)	TAF13 gene (Sequence Analysis-All Coding Exons)	TAF13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC24A2 geni (Tüm Gen Dizi Analizi)	SLC24A2 gene (Sequence Analysis-All Coding Exons)	SLC24A2	SLC24A2, NCKX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC23A2 geni (Tüm Gen Dizi Analizi)	SLC23A2 gene (Sequence Analysis-All Coding Exons)	SLC23A2	SLC23A2, SVCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC23A1 geni (Tüm Gen Dizi Analizi)	SLC23A1 gene (Sequence Analysis-All Coding Exons)	SLC23A1	SLC23A1, SVCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A9 geni (Tüm Gen Dizi Analizi)	SLC22A9 gene (Sequence Analysis-All Coding Exons)	SLC22A9	SLC22A9, OAT4, UST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A8 geni (Tüm Gen Dizi Analizi)	SLC22A8 gene (Sequence Analysis-All Coding Exons)	SLC22A8	SLC22A8, OAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A7 geni (Tüm Gen Dizi Analizi)	SLC22A7 gene (Sequence Analysis-All Coding Exons)	SLC22A7	SLC22A7, OAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A6 geni (Tüm Gen Dizi Analizi)	SLC22A6 gene (Sequence Analysis-All Coding Exons)	SLC22A6	SLC22A6, OAT1, PAHT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A3 geni (Tüm Gen Dizi Analizi)	SLC22A3 gene (Sequence Analysis-All Coding Exons)	SLC22A3	SLC22A3, EMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A25 geni (Tüm Gen Dizi Analizi)	SLC22A25 gene (Sequence Analysis-All Coding Exons)	SLC22A25	UST6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A24 geni (Tüm Gen Dizi Analizi)	SLC22A24 gene (Sequence Analysis-All Coding Exons)	SLC22A24	SLC22A24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A23 geni (Tüm Gen Dizi Analizi)	SLC22A23 gene (Sequence Analysis-All Coding Exons)	SLC22A23	SLC22A23, C6orf85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A2 geni (Tüm Gen Dizi Analizi)	SLC22A2 gene (Sequence Analysis-All Coding Exons)	SLC22A2	SLC22A2, OCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A18AS geni (Tüm Gen Dizi Analizi)	SLC22A18AS gene (Sequence Analysis-All Coding Exons)	SLC22A18AS	SLC22A1LS, ORCTL2S, BWSCR1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A17 geni (Tüm Gen Dizi Analizi)	SLC22A17 gene (Sequence Analysis-All Coding Exons)	SLC22A17	SLC22A17, NGALR, BOIT, BOCT, NGAL R2, NGAL R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A16 geni (Tüm Gen Dizi Analizi)	SLC22A16 gene (Sequence Analysis-All Coding Exons)	SLC22A16	SLC22A16, FLIPT2, CT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A15 geni (Tüm Gen Dizi Analizi)	SLC22A15 gene (Sequence Analysis-All Coding Exons)	SLC22A15	SLC22A15, FLIPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A11 geni (Tüm Gen Dizi Analizi)	SLC22A11 gene (Sequence Analysis-All Coding Exons)	SLC22A11	SLC22A11, OAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A10 geni (Tüm Gen Dizi Analizi)	SLC22A10 gene (Sequence Analysis-All Coding Exons)	SLC22A10	SLC22A10, OAT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A1 geni (Tüm Gen Dizi Analizi)	SLC22A1 gene (Sequence Analysis-All Coding Exons)	SLC22A1	SLC22A1, OCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC20A1 geni (Tüm Gen Dizi Analizi)	SLC20A1 gene (Sequence Analysis-All Coding Exons)	SLC20A1	SLC20A1, GLVR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC1A6 geni (Tüm Gen Dizi Analizi)	SLC1A6 gene (Sequence Analysis-All Coding Exons)	SLC1A6	SLC1A6, EAAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC1A5 geni (Tüm Gen Dizi Analizi)	SLC1A5 gene (Sequence Analysis-All Coding Exons)	SLC1A5	SLC1A5, RDRC, M7V1, M7V1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC19A1 geni (Tüm Gen Dizi Analizi)	SLC19A1 gene (Sequence Analysis-All Coding Exons)	SLC19A1	SLC19A1, FOLT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC18B1 geni (Tüm Gen Dizi Analizi)	SLC18B1 gene (Sequence Analysis-All Coding Exons)	SLC18B1	C6orf192	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC18A2 geni (Tüm Gen Dizi Analizi)	SLC18A2 gene (Sequence Analysis-All Coding Exons)	SLC18A2	SLC18A2, VAT2, SVMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC18A1 geni (Tüm Gen Dizi Analizi)	SLC18A1 gene (Sequence Analysis-All Coding Exons)	SLC18A1	SLC18A1, VMAT1, VAT1, CGAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC17A7 geni (Tüm Gen Dizi Analizi)	SLC17A7 gene (Sequence Analysis-All Coding Exons)	SLC17A7	SLC17A7, BNPI, VGLUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC17A6 geni (Tüm Gen Dizi Analizi)	SLC17A6 gene (Sequence Analysis-All Coding Exons)	SLC17A6	SLC17A6, VGLUT2, DNPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC17A4 geni (Tüm Gen Dizi Analizi)	SLC17A4 gene (Sequence Analysis-All Coding Exons)	SLC17A4	SLC17A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC17A2 geni (Tüm Gen Dizi Analizi)	SLC17A2 gene (Sequence Analysis-All Coding Exons)	SLC17A2	SLC17A2, NPT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC17A1 geni (Tüm Gen Dizi Analizi)	SLC17A1 gene (Sequence Analysis-All Coding Exons)	SLC17A1	SLC17A1, NPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A9 geni (Tüm Gen Dizi Analizi)	SLC16A9 gene (Sequence Analysis-All Coding Exons)	SLC16A9	SLC16A9, MCT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A8 geni (Tüm Gen Dizi Analizi)	SLC16A8 gene (Sequence Analysis-All Coding Exons)	SLC16A8	SLC16A8, MCT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A7 geni (Tüm Gen Dizi Analizi)	SLC16A7 gene (Sequence Analysis-All Coding Exons)	SLC16A7	SLC16A7, MCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A11 geni (Tüm Gen Dizi Analizi)	SLC16A11 gene (Sequence Analysis-All Coding Exons)	SLC16A11	SLC16A11, MCT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A10 geni (Tüm Gen Dizi Analizi)	SLC16A10 gene (Sequence Analysis-All Coding Exons)	SLC16A10	SLC16A10, TAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC15A4 geni (Tüm Gen Dizi Analizi)	SLC15A4 gene (Sequence Analysis-All Coding Exons)	SLC15A4	SLC15A4, PHT1, PTR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC15A3 geni (Tüm Gen Dizi Analizi)	SLC15A3 gene (Sequence Analysis-All Coding Exons)	SLC15A3	SLC15A3, PHT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC15A2 geni (Tüm Gen Dizi Analizi)	SLC15A2 gene (Sequence Analysis-All Coding Exons)	SLC15A2	SLC15A2, PEPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC15A1 geni (Tüm Gen Dizi Analizi)	SLC15A1 gene (Sequence Analysis-All Coding Exons)	SLC15A1	SLC15A1, HPECT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC14A2 geni (Tüm Gen Dizi Analizi)	SLC14A2 gene (Sequence Analysis-All Coding Exons)	SLC14A2	SLC14A2, UT2, UTR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC13A4 geni (Tüm Gen Dizi Analizi)	SLC13A4 gene (Sequence Analysis-All Coding Exons)	SLC13A4	SLC13A4, SUT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC13A3 geni (Tüm Gen Dizi Analizi)	SLC13A3 gene (Sequence Analysis-All Coding Exons)	SLC13A3	SLC13A3, NADC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC13A2 geni (Tüm Gen Dizi Analizi)	SLC13A2 gene (Sequence Analysis-All Coding Exons)	SLC13A2	SLC13A2, NADC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC13A1 geni (Tüm Gen Dizi Analizi)	SLC13A1 gene (Sequence Analysis-All Coding Exons)	SLC13A1	SLC13A1, NAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC12A9 geni (Tüm Gen Dizi Analizi)	SLC12A9 gene (Sequence Analysis-All Coding Exons)	SLC12A9	SLC12A9, CIP1, CCC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC12A7 geni (Tüm Gen Dizi Analizi)	SLC12A7 gene (Sequence Analysis-All Coding Exons)	SLC12A7	SLC12A7, KCC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC12A4 geni (Tüm Gen Dizi Analizi)	SLC12A4 gene (Sequence Analysis-All Coding Exons)	SLC12A4	SLC12A4, KCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC12A2 geni (Tüm Gen Dizi Analizi)	SLC12A2 gene (Sequence Analysis-All Coding Exons)	SLC12A2	SLC12A2, NKCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC10A6 geni (Tüm Gen Dizi Analizi)	SLC10A6 gene (Sequence Analysis-All Coding Exons)	SLC10A6	SLC10A6, SOAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC10A3 geni (Tüm Gen Dizi Analizi)	SLC10A3 gene (Sequence Analysis-All Coding Exons)	SLC10A3	SLC10A3, P3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SYT11 geni (Tüm Gen Dizi Analizi)	SYT11 gene (Sequence Analysis-All Coding Exons)	SYT11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SLC10A1 geni (Tüm Gen Dizi Analizi)	SLC10A1 gene (Sequence Analysis-All Coding Exons)	SLC10A1	SLC10A1, NTCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLBP geni (Tüm Gen Dizi Analizi)	SLBP gene (Sequence Analysis-All Coding Exons)	SLBP	SLBP, HBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAMF8 geni (Tüm Gen Dizi Analizi)	SLAMF8 gene (Sequence Analysis-All Coding Exons)	SLAMF8	SLAMF8, BLAME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAMF7 geni (Tüm Gen Dizi Analizi)	SLAMF7 gene (Sequence Analysis-All Coding Exons)	SLAMF7	SLAMF7, CRACC, CS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAMF6 geni (Tüm Gen Dizi Analizi)	SLAMF6 gene (Sequence Analysis-All Coding Exons)	SLAMF6	SLAMF6, NTBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAMF1 geni (Tüm Gen Dizi Analizi)	SLAMF1 gene (Sequence Analysis-All Coding Exons)	SLAMF1	SLAMF1, SLAM, CDW150, CD150	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAIN2 geni (Tüm Gen Dizi Analizi)	SLAIN2 gene (Sequence Analysis-All Coding Exons)	SLAIN2	SLAIN2, KIAA1458	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAIN1 geni (Tüm Gen Dizi Analizi)	SLAIN1 gene (Sequence Analysis-All Coding Exons)	SLAIN1	SLAIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLA2 geni (Tüm Gen Dizi Analizi)	SLA2 gene (Sequence Analysis-All Coding Exons)	SLA2	SLA2, SLAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLA geni (Tüm Gen Dizi Analizi)	SLA gene (Sequence Analysis-All Coding Exons)	SLA	SLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKP2 geni (Tüm Gen Dizi Analizi)	SKP2 gene (Sequence Analysis-All Coding Exons)	SKP2	SKP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKP1 geni (Tüm Gen Dizi Analizi)	SKP1 gene (Sequence Analysis-All Coding Exons)	SKP1	SKP1, SKP1A, TCEB1L, OCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKOR2 geni (Tüm Gen Dizi Analizi)	SKOR2 gene (Sequence Analysis-All Coding Exons)	SKOR2	SKOR2, FUSSEL18, CORL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKOR1 geni (Tüm Gen Dizi Analizi)	SKOR1 gene (Sequence Analysis-All Coding Exons)	SKOR1	LBXCOR1, CORL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKIL geni (Tüm Gen Dizi Analizi)	SKIL gene (Sequence Analysis-All Coding Exons)	SKIL	SKIL, SNO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKAP2 geni (Tüm Gen Dizi Analizi)	SKAP2 gene (Sequence Analysis-All Coding Exons)	SKAP2	SKAP2, SCAP2, SKAP55R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKAP1 geni (Tüm Gen Dizi Analizi)	SKAP1 gene (Sequence Analysis-All Coding Exons)	SKAP1	SKAP1, SCAP1, SKAP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKA2 geni (Tüm Gen Dizi Analizi)	SKA2 gene (Sequence Analysis-All Coding Exons)	SKA2	SKA2, FAM33A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SKA1 geni (Tüm Gen Dizi Analizi)	SKA1 gene (Sequence Analysis-All Coding Exons)	SKA1	SKA1, C18orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIX4 geni (Tüm Gen Dizi Analizi)	SIX4 gene (Sequence Analysis-All Coding Exons)	SIX4	SIX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIX2 geni (Tüm Gen Dizi Analizi)	SIX2 gene (Sequence Analysis-All Coding Exons)	SIX2	SIX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIT1 geni (Tüm Gen Dizi Analizi)	SIT1 gene (Sequence Analysis-All Coding Exons)	SIT1	SIT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT7 geni (Tüm Gen Dizi Analizi)	SIRT7 gene (Sequence Analysis-All Coding Exons)	SIRT7	SIRT7, SIR2L7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT6 geni (Tüm Gen Dizi Analizi)	SIRT6 gene (Sequence Analysis-All Coding Exons)	SIRT6	SIRT6, SIR2L6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT5 geni (Tüm Gen Dizi Analizi)	SIRT5 gene (Sequence Analysis-All Coding Exons)	SIRT5	SIRT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT4 geni (Tüm Gen Dizi Analizi)	SIRT4 gene (Sequence Analysis-All Coding Exons)	SIRT4	SIRT4, SIR2L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT3 geni (Tüm Gen Dizi Analizi)	SIRT3 gene (Sequence Analysis-All Coding Exons)	SIRT3	SIRT3, SIR2L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT2 geni (Tüm Gen Dizi Analizi)	SIRT2 gene (Sequence Analysis-All Coding Exons)	SIRT2	SIRT2, SIR2L, SIR2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRT1 geni (Tüm Gen Dizi Analizi)	SIRT1 gene (Sequence Analysis-All Coding Exons)	SIRT1	SIRT1, SIR2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRPG geni (Tüm Gen Dizi Analizi)	SIRPG gene (Sequence Analysis-All Coding Exons)	SIRPG	SIRPG, SIRPB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRPA geni (Tüm Gen Dizi Analizi)	SIRPA gene (Sequence Analysis-All Coding Exons)	SIRPA	SHPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIPA1L2 geni (Tüm Gen Dizi Analizi)	SIPA1L2 gene (Sequence Analysis-All Coding Exons)	SIPA1L2	SIPA1L2, KIAA1389	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIPA1 geni (Tüm Gen Dizi Analizi)	SIPA1 gene (Sequence Analysis-All Coding Exons)	SIPA1	SIPA1, SPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIN3B geni (Tüm Gen Dizi Analizi)	SIN3B gene (Sequence Analysis-All Coding Exons)	SIN3B	SIN3B, KIAA0700	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIM2 geni (Tüm Gen Dizi Analizi)	SIM2 gene (Sequence Analysis-All Coding Exons)	SIM2	SIM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIKE1 geni (Tüm Gen Dizi Analizi)	SIKE1 gene (Sequence Analysis-All Coding Exons)	SIKE1	SIKE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIK3 geni (Tüm Gen Dizi Analizi)	SIK3 gene (Sequence Analysis-All Coding Exons)	SIK3	SIK3, KIAA0999	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIK2 geni (Tüm Gen Dizi Analizi)	SIK2 gene (Sequence Analysis-All Coding Exons)	SIK2	SIK2, KIAA0781	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC9 geni (Tüm Gen Dizi Analizi)	SIGLEC9 gene (Sequence Analysis-All Coding Exons)	SIGLEC9	SIGLEC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC8 geni (Tüm Gen Dizi Analizi)	SIGLEC8 gene (Sequence Analysis-All Coding Exons)	SIGLEC8	SIGLEC8, SAF2, SIGLEC8L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC7 geni (Tüm Gen Dizi Analizi)	SIGLEC7 gene (Sequence Analysis-All Coding Exons)	SIGLEC7	SIGLEC7, AIRM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC6 geni (Tüm Gen Dizi Analizi)	SIGLEC6 gene (Sequence Analysis-All Coding Exons)	SIGLEC6	SIGLEC6, CD33L, CD33L1, OBBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC5 geni (Tüm Gen Dizi Analizi)	SIGLEC5 gene (Sequence Analysis-All Coding Exons)	SIGLEC5	SIGLEC5, CD33L2, OBBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC12 geni (Tüm Gen Dizi Analizi)	SIGLEC12 gene (Sequence Analysis-All Coding Exons)	SIGLEC12	SIGLEC12, SIGLEC-L1, S2V, SLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC11 geni (Tüm Gen Dizi Analizi)	SIGLEC11 gene (Sequence Analysis-All Coding Exons)	SIGLEC11	SIGLEC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC10 geni (Tüm Gen Dizi Analizi)	SIGLEC10 gene (Sequence Analysis-All Coding Exons)	SIGLEC10	SIGLEC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGLEC1 geni (Tüm Gen Dizi Analizi)	SIGLEC1 gene (Sequence Analysis-All Coding Exons)	SIGLEC1	SIGLEC1, SN, CD169	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIGIRR geni (Tüm Gen Dizi Analizi)	SIGIRR gene (Sequence Analysis-All Coding Exons)	SIGIRR	SIGIRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIAH3 geni (Tüm Gen Dizi Analizi)	SIAH3 gene (Sequence Analysis-All Coding Exons)	SIAH3	SIAH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIAH2 geni (Tüm Gen Dizi Analizi)	SIAH2 gene (Sequence Analysis-All Coding Exons)	SIAH2	SIAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIAH1 geni (Tüm Gen Dizi Analizi)	SIAH1 gene (Sequence Analysis-All Coding Exons)	SIAH1	SIAH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHTN1 geni (Tüm Gen Dizi Analizi)	SHTN1 gene (Sequence Analysis-All Coding Exons)	SHTN1	KIAA1598, SHOOTIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHROOM3 geni (Tüm Gen Dizi Analizi)	SHROOM3 gene (Sequence Analysis-All Coding Exons)	SHROOM3	SHROOM3, SHRM, KIAA1481	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHROOM2 geni (Tüm Gen Dizi Analizi)	SHROOM2 gene (Sequence Analysis-All Coding Exons)	SHROOM2	SHROOM2, APX-L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHROOM1 geni (Tüm Gen Dizi Analizi)	SHROOM1 gene (Sequence Analysis-All Coding Exons)	SHROOM1	SHROOM1, APX-L2, KIAA1960	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHQ1 geni (Tüm Gen Dizi Analizi)	SHQ1 gene (Sequence Analysis-All Coding Exons)	SHQ1	SHQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHOX2 geni (Tüm Gen Dizi Analizi)	SHOX2 gene (Sequence Analysis-All Coding Exons)	SHOX2	SHOX2, SHOT, OG12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHMT2 geni (Tüm Gen Dizi Analizi)	SHMT2 gene (Sequence Analysis-All Coding Exons)	SHMT2	SHMT2, GLYA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHMT1 geni (Tüm Gen Dizi Analizi)	SHMT1 gene (Sequence Analysis-All Coding Exons)	SHMT1	SHMT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHKBP1 geni (Tüm Gen Dizi Analizi)	SHKBP1 gene (Sequence Analysis-All Coding Exons)	SHKBP1	SHKBP1, SB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA9 geni (Tüm Gen Dizi Analizi)	SHISA9 gene (Sequence Analysis-All Coding Exons)	SHISA9	SHISA9, CKAMP44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA8 geni (Tüm Gen Dizi Analizi)	SHISA8 gene (Sequence Analysis-All Coding Exons)	SHISA8	SHISA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA7 geni (Tüm Gen Dizi Analizi)	SHISA7 gene (Sequence Analysis-All Coding Exons)	SHISA7	SHISA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA6 geni (Tüm Gen Dizi Analizi)	SHISA6 gene (Sequence Analysis-All Coding Exons)	SHISA6	SHISA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA5 geni (Tüm Gen Dizi Analizi)	SHISA5 gene (Sequence Analysis-All Coding Exons)	SHISA5	SHISA5, SCOTIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA4 geni (Tüm Gen Dizi Analizi)	SHISA4 gene (Sequence Analysis-All Coding Exons)	SHISA4	SHISA4, C1orf40, TMEM58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA3 geni (Tüm Gen Dizi Analizi)	SHISA3 gene (Sequence Analysis-All Coding Exons)	SHISA3	SHISA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHISA2 geni (Tüm Gen Dizi Analizi)	SHISA2 gene (Sequence Analysis-All Coding Exons)	SHISA2	SHISA2, C13orf13, TMEM46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUPT4H1 geni (Tüm Gen Dizi Analizi)	SUPT4H1 gene (Sequence Analysis-All Coding Exons)	SUPT4H1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHF geni (Tüm Gen Dizi Analizi)	SHF gene (Sequence Analysis-All Coding Exons)	SHF	SHF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHE geni (Tüm Gen Dizi Analizi)	SHE gene (Sequence Analysis-All Coding Exons)	SHE	SHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHD geni (Tüm Gen Dizi Analizi)	SHD gene (Sequence Analysis-All Coding Exons)	SHD	SHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHCBP1 geni (Tüm Gen Dizi Analizi)	SHCBP1 gene (Sequence Analysis-All Coding Exons)	SHCBP1	SHCBP1, PAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHC4 geni (Tüm Gen Dizi Analizi)	SHC4 gene (Sequence Analysis-All Coding Exons)	SHC4	SHC4, RALP, SHCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHC3 geni (Tüm Gen Dizi Analizi)	SHC3 gene (Sequence Analysis-All Coding Exons)	SHC3	SHC3, NSHC, RAI, SHCC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHC2 geni (Tüm Gen Dizi Analizi)	SHC2 gene (Sequence Analysis-All Coding Exons)	SHC2	SHC2, SHCB, SCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHC1 geni (Tüm Gen Dizi Analizi)	SHC1 gene (Sequence Analysis-All Coding Exons)	SHC1	SHC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHBG geni (Tüm Gen Dizi Analizi)	SHBG gene (Sequence Analysis-All Coding Exons)	SHBG	SHBG, ABP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHB geni (Tüm Gen Dizi Analizi)	SHB gene (Sequence Analysis-All Coding Exons)	SHB	SHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUMO2 geni (Tüm Gen Dizi Analizi)	SUMO2 gene (Sequence Analysis-All Coding Exons)	SUMO2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHARPIN geni (Tüm Gen Dizi Analizi)	SHARPIN gene (Sequence Analysis-All Coding Exons)	SHARPIN	SHARPIN, SIPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHANK1 geni (Tüm Gen Dizi Analizi)	SHANK1 gene (Sequence Analysis-All Coding Exons)	SHANK1	SHANK1, SSTRIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3YL1 geni (Tüm Gen Dizi Analizi)	SH3YL1 gene (Sequence Analysis-All Coding Exons)	SH3YL1	SH3YL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3RF2 geni (Tüm Gen Dizi Analizi)	SH3RF2 gene (Sequence Analysis-All Coding Exons)	SH3RF2	SH3RF2, HEPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3KBP1 geni (Tüm Gen Dizi Analizi)	SH3KBP1 gene (Sequence Analysis-All Coding Exons)	SH3KBP1	SH3KBP1, CIN85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3GL3 geni (Tüm Gen Dizi Analizi)	SH3GL3 gene (Sequence Analysis-All Coding Exons)	SH3GL3	SH3GL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3GL2 geni (Tüm Gen Dizi Analizi)	SH3GL2 gene (Sequence Analysis-All Coding Exons)	SH3GL2	SH3GL2, SHIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3D19 geni (Tüm Gen Dizi Analizi)	SH3D19 gene (Sequence Analysis-All Coding Exons)	SH3D19	SH3D19, EBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BP5 geni (Tüm Gen Dizi Analizi)	SH3BP5 gene (Sequence Analysis-All Coding Exons)	SH3BP5	SH3BP5, SAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BP4 geni (Tüm Gen Dizi Analizi)	SH3BP4 gene (Sequence Analysis-All Coding Exons)	SH3BP4	SH3BP4, BOG25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BP1 geni (Tüm Gen Dizi Analizi)	SH3BP1 gene (Sequence Analysis-All Coding Exons)	SH3BP1	SH3BP1, ARHGAP43, BGIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BGRL3 geni (Tüm Gen Dizi Analizi)	SH3BGRL3 gene (Sequence Analysis-All Coding Exons)	SH3BGRL3	SH3BGRL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BGRL2 geni (Tüm Gen Dizi Analizi)	SH3BGRL2 gene (Sequence Analysis-All Coding Exons)	SH3BGRL2	SH3BGRL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SH3BGRL geni (Tüm Gen Dizi Analizi)	SH3BGRL gene (Sequence Analysis-All Coding Exons)	SH3BGRL	SH3BGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3BGR geni (Tüm Gen Dizi Analizi)	SH3BGR gene (Sequence Analysis-All Coding Exons)	SH3BGR	SH3GBR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2D4A geni (Tüm Gen Dizi Analizi)	SH2D4A gene (Sequence Analysis-All Coding Exons)	SH2D4A	SH2D4A, SH2A, PPP1R38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2D3C geni (Tüm Gen Dizi Analizi)	SH2D3C gene (Sequence Analysis-All Coding Exons)	SH2D3C	SH2D3C, NSP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2D2A geni (Tüm Gen Dizi Analizi)	SH2D2A gene (Sequence Analysis-All Coding Exons)	SH2D2A	SH2D2A, TSAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2D1B geni (Tüm Gen Dizi Analizi)	SH2D1B gene (Sequence Analysis-All Coding Exons)	SH2D1B	SH2D1B, EAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2B2 geni (Tüm Gen Dizi Analizi)	SH2B2 gene (Sequence Analysis-All Coding Exons)	SH2B2	APS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2B1 geni (Tüm Gen Dizi Analizi)	SH2B1 gene (Sequence Analysis-All Coding Exons)	SH2B1	SH2B1, SH2B, KIAA1299	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2B1 geni (Tüm Gen Dizi Analizi)	SH2B1 gene (Sequence Analysis-All Coding Exons)	SH2B1	SH2B1, SH2B, KIAA1299	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2B1 geni (Tüm Gen Dizi Analizi)	SH2B1 gene (Sequence Analysis-All Coding Exons)	SH2B1	SH2B1, SH2B, KIAA1299	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGTA geni (Tüm Gen Dizi Analizi)	SGTA gene (Sequence Analysis-All Coding Exons)	SGTA	SGTA, SGT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGSM3 geni (Tüm Gen Dizi Analizi)	SGSM3 gene (Sequence Analysis-All Coding Exons)	SGSM3	SGSM3, RUTBC3, MAP, RUSC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SUDS3 geni (Tüm Gen Dizi Analizi)	SUDS3 gene (Sequence Analysis-All Coding Exons)	SUDS3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGSM2 geni (Tüm Gen Dizi Analizi)	SGSM2 gene (Sequence Analysis-All Coding Exons)	SGSM2	SGSM2, KIAA0397	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGPP2 geni (Tüm Gen Dizi Analizi)	SGPP2 gene (Sequence Analysis-All Coding Exons)	SGPP2	SGPP2, SPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGPP1 geni (Tüm Gen Dizi Analizi)	SGPP1 gene (Sequence Analysis-All Coding Exons)	SGPP1	SGPP1, SPPASE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGPL1 geni (Tüm Gen Dizi Analizi)	SGPL1 gene (Sequence Analysis-All Coding Exons)	SGPL1	SGPL1, SPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGO2 geni (Tüm Gen Dizi Analizi)	SGO2 gene (Sequence Analysis-All Coding Exons)	SGO2	SGOL2, SGO2, TRIPIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGMS2 geni (Tüm Gen Dizi Analizi)	SGMS2 gene (Sequence Analysis-All Coding Exons)	SGMS2	SGMS2, SMS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGMS1 geni (Tüm Gen Dizi Analizi)	SGMS1 gene (Sequence Analysis-All Coding Exons)	SGMS1	SGMS1, SMS1, TMEM23, MCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGK3 geni (Tüm Gen Dizi Analizi)	SGK3 gene (Sequence Analysis-All Coding Exons)	SGK3	SGK3, SGKL, CISK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGK2 geni (Tüm Gen Dizi Analizi)	SGK2 gene (Sequence Analysis-All Coding Exons)	SGK2	SGK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGK1 geni (Tüm Gen Dizi Analizi)	SGK1 gene (Sequence Analysis-All Coding Exons)	SGK1	SGK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGIP1 geni (Tüm Gen Dizi Analizi)	SGIP1 gene (Sequence Analysis-All Coding Exons)	SGIP1	SGIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGF29 geni (Tüm Gen Dizi Analizi)	SGF29 gene (Sequence Analysis-All Coding Exons)	SGF29	CCDC101, STAF36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STXBP3 geni (Tüm Gen Dizi Analizi)	STXBP3 gene (Sequence Analysis-All Coding Exons)	STXBP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGCZ geni (Tüm Gen Dizi Analizi)	SGCZ gene (Sequence Analysis-All Coding Exons)	SGCZ	SGCZ, ZSG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFXN5 geni (Tüm Gen Dizi Analizi)	SFXN5 gene (Sequence Analysis-All Coding Exons)	SFXN5	SFXN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFXN3 geni (Tüm Gen Dizi Analizi)	SFXN3 gene (Sequence Analysis-All Coding Exons)	SFXN3	SFXN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFXN2 geni (Tüm Gen Dizi Analizi)	SFXN2 gene (Sequence Analysis-All Coding Exons)	SFXN2	SFXN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFXN1 geni (Tüm Gen Dizi Analizi)	SFXN1 gene (Sequence Analysis-All Coding Exons)	SFXN1	SFXN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFTPD geni (Tüm Gen Dizi Analizi)	SFTPD gene (Sequence Analysis-All Coding Exons)	SFTPD	SFTPD, SFTP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFSWAP geni (Tüm Gen Dizi Analizi)	SFSWAP gene (Sequence Analysis-All Coding Exons)	SFSWAP	SFSWAP, SFRS8, SWAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFRP5 geni (Tüm Gen Dizi Analizi)	SFRP5 gene (Sequence Analysis-All Coding Exons)	SFRP5	SFRP5, SARP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFRP2 geni (Tüm Gen Dizi Analizi)	SFRP2 gene (Sequence Analysis-All Coding Exons)	SFRP2	SFRP2, SARP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFRP1 geni (Tüm Gen Dizi Analizi)	SFRP1 gene (Sequence Analysis-All Coding Exons)	SFRP1	SFRP1, FRP, SARP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFR1 geni (Tüm Gen Dizi Analizi)	SFR1 gene (Sequence Analysis-All Coding Exons)	SFR1	SFR1, MEI5, C10orf78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX18 geni (Tüm Gen Dizi Analizi)	STX18 gene (Sequence Analysis-All Coding Exons)	STX18		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFPQ geni (Tüm Gen Dizi Analizi)	SFPQ gene (Sequence Analysis-All Coding Exons)	SFPQ	SFPQ, PSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFN geni (Tüm Gen Dizi Analizi)	SFN gene (Sequence Analysis-All Coding Exons)	SFN	SFN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STX12 geni (Tüm Gen Dizi Analizi)	STX12 gene (Sequence Analysis-All Coding Exons)	STX12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFMBT2 geni (Tüm Gen Dizi Analizi)	SFMBT2 gene (Sequence Analysis-All Coding Exons)	SFMBT2	SFMBT2, KIAA1617	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFMBT1 geni (Tüm Gen Dizi Analizi)	SFMBT1 gene (Sequence Analysis-All Coding Exons)	SFMBT1	SFMBT1, RU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SFI1 geni (Tüm Gen Dizi Analizi)	SFI1 gene (Sequence Analysis-All Coding Exons)	SFI1	SFI1, KIAA0542	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3B6 geni (Tüm Gen Dizi Analizi)	SF3B6 gene (Sequence Analysis-All Coding Exons)	SF3B6	SAP14, SF3B14, P14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3B3 geni (Tüm Gen Dizi Analizi)	SF3B3 gene (Sequence Analysis-All Coding Exons)	SF3B3	SF3B3, SF3B130, SAP130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3B2 geni (Tüm Gen Dizi Analizi)	SF3B2 gene (Sequence Analysis-All Coding Exons)	SF3B2	SF3B2, SF3B145, SAP145	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3A3 geni (Tüm Gen Dizi Analizi)	SF3A3 gene (Sequence Analysis-All Coding Exons)	SF3A3	SF3A3, SF3A60, SAP61, PRP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3A2 geni (Tüm Gen Dizi Analizi)	SF3A2 gene (Sequence Analysis-All Coding Exons)	SF3A2	SF3A2, SAP62, PRP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF3A1 geni (Tüm Gen Dizi Analizi)	SF3A1 gene (Sequence Analysis-All Coding Exons)	SF3A1	SF3A1, SF3A120, SAP114, PRP21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SF1 geni (Tüm Gen Dizi Analizi)	SF1 gene (Sequence Analysis-All Coding Exons)	SF1	SF1, ZNF162, D11S636, ZFM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEZ6L2 geni (Tüm Gen Dizi Analizi)	SEZ6L2 gene (Sequence Analysis-All Coding Exons)	SEZ6L2	SEZ6L2, BSRPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEZ6L geni (Tüm Gen Dizi Analizi)	SEZ6L gene (Sequence Analysis-All Coding Exons)	SEZ6L	SEZ6L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEZ6 geni (Tüm Gen Dizi Analizi)	SEZ6 gene (Sequence Analysis-All Coding Exons)	SEZ6	SEZ6, BSRPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETMAR geni (Tüm Gen Dizi Analizi)	SETMAR gene (Sequence Analysis-All Coding Exons)	SETMAR	SETMAR, METNASE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETDB2 geni (Tüm Gen Dizi Analizi)	SETDB2 gene (Sequence Analysis-All Coding Exons)	SETDB2	SETDB2, CLLD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETDB1 geni (Tüm Gen Dizi Analizi)	SETDB1 gene (Sequence Analysis-All Coding Exons)	SETDB1	SETDB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETD7 geni (Tüm Gen Dizi Analizi)	SETD7 gene (Sequence Analysis-All Coding Exons)	SETD7	SETD7, SET7, KIAA1717	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETD6 geni (Tüm Gen Dizi Analizi)	SETD6 gene (Sequence Analysis-All Coding Exons)	SETD6	SETD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETD3 geni (Tüm Gen Dizi Analizi)	SETD3 gene (Sequence Analysis-All Coding Exons)	SETD3	SETD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETD1B geni (Tüm Gen Dizi Analizi)	SETD1B gene (Sequence Analysis-All Coding Exons)	SETD1B	SETD1B, SET1B, KIAA1076	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SETD1A geni (Tüm Gen Dizi Analizi)	SETD1A gene (Sequence Analysis-All Coding Exons)	SETD1A	SETD1A, SET1A, SET1, KIAA0339	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SET geni (Tüm Gen Dizi Analizi)	SET gene (Sequence Analysis-All Coding Exons)	SET	SET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SESN3 geni (Tüm Gen Dizi Analizi)	SESN3 gene (Sequence Analysis-All Coding Exons)	SESN3	SEST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SESN2 geni (Tüm Gen Dizi Analizi)	SESN2 gene (Sequence Analysis-All Coding Exons)	SESN2	SEST2, HI95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SESN1 geni (Tüm Gen Dizi Analizi)	SESN1 gene (Sequence Analysis-All Coding Exons)	SESN1	SESN1, SEST1, PA26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERTAD3 geni (Tüm Gen Dizi Analizi)	SERTAD3 gene (Sequence Analysis-All Coding Exons)	SERTAD3	SERTAD3, RBT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINI2 geni (Tüm Gen Dizi Analizi)	SERPINI2 gene (Sequence Analysis-All Coding Exons)	SERPINI2	SERPINI2, PI14, MEPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINE2 geni (Tüm Gen Dizi Analizi)	SERPINE2 gene (Sequence Analysis-All Coding Exons)	SERPINE2	PI7, PN1, SERPINE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB9 geni (Tüm Gen Dizi Analizi)	SERPINB9 gene (Sequence Analysis-All Coding Exons)	SERPINB9	PI9, CAP3, SERPINB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB5 geni (Tüm Gen Dizi Analizi)	SERPINB5 gene (Sequence Analysis-All Coding Exons)	SERPINB5	PI5, SERPINB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB4 geni (Tüm Gen Dizi Analizi)	SERPINB4 gene (Sequence Analysis-All Coding Exons)	SERPINB4	SERPINB4, SCCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB3 geni (Tüm Gen Dizi Analizi)	SERPINB3 gene (Sequence Analysis-All Coding Exons)	SERPINB3	SERPINB3, SCCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB2 geni (Tüm Gen Dizi Analizi)	SERPINB2 gene (Sequence Analysis-All Coding Exons)	SERPINB2	SERPINB2, PAI2, PLANH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB13 geni (Tüm Gen Dizi Analizi)	SERPINB13 gene (Sequence Analysis-All Coding Exons)	SERPINB13	SERPINB13, PI13, HJURPIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB12 geni (Tüm Gen Dizi Analizi)	SERPINB12 gene (Sequence Analysis-All Coding Exons)	SERPINB12	SERPINB12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB11 geni (Tüm Gen Dizi Analizi)	SERPINB11 gene (Sequence Analysis-All Coding Exons)	SERPINB11	SERPINB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB10 geni (Tüm Gen Dizi Analizi)	SERPINB10 gene (Sequence Analysis-All Coding Exons)	SERPINB10	SERPINB10, PI10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINB1 geni (Tüm Gen Dizi Analizi)	SERPINB1 gene (Sequence Analysis-All Coding Exons)	SERPINB1	ELANH2, EI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINA9 geni (Tüm Gen Dizi Analizi)	SERPINA9 gene (Sequence Analysis-All Coding Exons)	SERPINA9	SERPINA9, GCET1, CENTERIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINA5 geni (Tüm Gen Dizi Analizi)	SERPINA5 gene (Sequence Analysis-All Coding Exons)	SERPINA5	SERPINA5, PCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINA4 geni (Tüm Gen Dizi Analizi)	SERPINA4 gene (Sequence Analysis-All Coding Exons)	SERPINA4	SERPINA4, KST, PI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERPINA10 geni (Tüm Gen Dizi Analizi)	SERPINA10 gene (Sequence Analysis-All Coding Exons)	SERPINA10	SERPINA10, ZPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERINC5 geni (Tüm Gen Dizi Analizi)	SERINC5 gene (Sequence Analysis-All Coding Exons)	SERINC5	SERINC5, C5orf12, TPO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERINC4 geni (Tüm Gen Dizi Analizi)	SERINC4 gene (Sequence Analysis-All Coding Exons)	SERINC4	SERINC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERINC3 geni (Tüm Gen Dizi Analizi)	SERINC3 gene (Sequence Analysis-All Coding Exons)	SERINC3	SERINC3, TDE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERINC2 geni (Tüm Gen Dizi Analizi)	SERINC2 gene (Sequence Analysis-All Coding Exons)	SERINC2	SERINC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STK16 geni (Tüm Gen Dizi Analizi)	STK16 gene (Sequence Analysis-All Coding Exons)	STK16		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERINC1 geni (Tüm Gen Dizi Analizi)	SERINC1 gene (Sequence Analysis-All Coding Exons)	SERINC1	SERINC1, TMS2, KIAA1253	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERHL geni (Tüm Gen Dizi Analizi)	SERHL gene (Sequence Analysis-All Coding Exons)	SERHL	SERHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERGEF geni (Tüm Gen Dizi Analizi)	SERGEF gene (Sequence Analysis-All Coding Exons)	SERGEF	SERGEF, DELGEF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERF2 geni (Tüm Gen Dizi Analizi)	SERF2 gene (Sequence Analysis-All Coding Exons)	SERF2	SERF2, H4F5REL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SERF1A geni (Tüm Gen Dizi Analizi)	SERF1A gene (Sequence Analysis-All Coding Exons)	SERF1A	SERF1A, H4F5, SMAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SERBP1 geni (Tüm Gen Dizi Analizi)	SERBP1 gene (Sequence Analysis-All Coding Exons)	SERBP1	SERBP1, PAIRBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEPHS2 geni (Tüm Gen Dizi Analizi)	SEPHS2 gene (Sequence Analysis-All Coding Exons)	SEPHS2	SEPHS2, SPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEPHS1 geni (Tüm Gen Dizi Analizi)	SEPHS1 gene (Sequence Analysis-All Coding Exons)	SEPHS1	SEPHS1, SPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP8 geni (Tüm Gen Dizi Analizi)	SENP8 gene (Sequence Analysis-All Coding Exons)	SENP8	SENP8, NEDP1, DEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP7 geni (Tüm Gen Dizi Analizi)	SENP7 gene (Sequence Analysis-All Coding Exons)	SENP7	SENP7, KIAA1707	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP6 geni (Tüm Gen Dizi Analizi)	SENP6 gene (Sequence Analysis-All Coding Exons)	SENP6	SENP6, SSP1, SUSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP5 geni (Tüm Gen Dizi Analizi)	SENP5 gene (Sequence Analysis-All Coding Exons)	SENP5	SENP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP3 geni (Tüm Gen Dizi Analizi)	SENP3 gene (Sequence Analysis-All Coding Exons)	SENP3	SENP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP2 geni (Tüm Gen Dizi Analizi)	SENP2 gene (Sequence Analysis-All Coding Exons)	SENP2	SENP2, SMT3IP2, AXAM2, KIAA1331	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENP1 geni (Tüm Gen Dizi Analizi)	SENP1 gene (Sequence Analysis-All Coding Exons)	SENP1	SENP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SENCR geni (Tüm Gen Dizi Analizi)	SENCR gene (Sequence Analysis-All Coding Exons)	SENCR	SENCR, lncRNA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMG2 geni (Tüm Gen Dizi Analizi)	SEMG2 gene (Sequence Analysis-All Coding Exons)	SEMG2	SEMG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMG1 geni (Tüm Gen Dizi Analizi)	SEMG1 gene (Sequence Analysis-All Coding Exons)	SEMG1	SEMG1, SEMG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA6D geni (Tüm Gen Dizi Analizi)	SEMA6D gene (Sequence Analysis-All Coding Exons)	SEMA6D	SEMA6D, KIAA1479	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA6C geni (Tüm Gen Dizi Analizi)	SEMA6C gene (Sequence Analysis-All Coding Exons)	SEMA6C	SEMA6C, KIAA1869	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA6B geni (Tüm Gen Dizi Analizi)	SEMA6B gene (Sequence Analysis-All Coding Exons)	SEMA6B	SEMA6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA6A geni (Tüm Gen Dizi Analizi)	SEMA6A gene (Sequence Analysis-All Coding Exons)	SEMA6A	SEMA6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA5A geni (Tüm Gen Dizi Analizi)	SEMA5A gene (Sequence Analysis-All Coding Exons)	SEMA5A	SEMA5A, SEMF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA4F geni (Tüm Gen Dizi Analizi)	SEMA4F gene (Sequence Analysis-All Coding Exons)	SEMA4F	SEMAW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA4C geni (Tüm Gen Dizi Analizi)	SEMA4C gene (Sequence Analysis-All Coding Exons)	SEMA4C	SEMA4C, SEMAF, KIAA1739	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA4B geni (Tüm Gen Dizi Analizi)	SEMA4B gene (Sequence Analysis-All Coding Exons)	SEMA4B	SEMA4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAT5A geni (Tüm Gen Dizi Analizi)	STAT5A gene (Sequence Analysis-All Coding Exons)	STAT5A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA3F geni (Tüm Gen Dizi Analizi)	SEMA3F gene (Sequence Analysis-All Coding Exons)	SEMA3F	SEMA3F, SEMA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA3D geni (Tüm Gen Dizi Analizi)	SEMA3D gene (Sequence Analysis-All Coding Exons)	SEMA3D	SEMA3D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA3C geni (Tüm Gen Dizi Analizi)	SEMA3C gene (Sequence Analysis-All Coding Exons)	SEMA3C	SEMA3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA3B geni (Tüm Gen Dizi Analizi)	SEMA3B gene (Sequence Analysis-All Coding Exons)	SEMA3B	SEMA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEM1 geni (Tüm Gen Dizi Analizi)	SEM1 gene (Sequence Analysis-All Coding Exons)	SEM1	DSS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELPLG geni (Tüm Gen Dizi Analizi)	SELPLG gene (Sequence Analysis-All Coding Exons)	SELPLG	SELPLG, PSGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELL geni (Tüm Gen Dizi Analizi)	SELL gene (Sequence Analysis-All Coding Exons)	SELL	SELL, LYAM1, LAM1, LNHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOW geni (Tüm Gen Dizi Analizi)	SELENOW gene (Sequence Analysis-All Coding Exons)	SELENOW	SELENOW, SEPW1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOV geni (Tüm Gen Dizi Analizi)	SELENOV gene (Sequence Analysis-All Coding Exons)	SELENOV	SELENOV, SELV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOT geni (Tüm Gen Dizi Analizi)	SELENOT gene (Sequence Analysis-All Coding Exons)	SELENOT	SELENOT, SELT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOS geni (Tüm Gen Dizi Analizi)	SELENOS gene (Sequence Analysis-All Coding Exons)	SELENOS	SELENOS, SEPS1, SELS, VIMP, ADOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOP geni (Tüm Gen Dizi Analizi)	SELENOP gene (Sequence Analysis-All Coding Exons)	SELENOP	SELENOP, SEPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOO geni (Tüm Gen Dizi Analizi)	SELENOO gene (Sequence Analysis-All Coding Exons)	SELENOO	SELENOO, SELO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOM geni (Tüm Gen Dizi Analizi)	SELENOM gene (Sequence Analysis-All Coding Exons)	SELENOM	SELENOM, SELM, SEPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOK geni (Tüm Gen Dizi Analizi)	SELENOK gene (Sequence Analysis-All Coding Exons)	SELENOK	SELENOK, SELK, HSPC030	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOI geni (Tüm Gen Dizi Analizi)	SELENOI gene (Sequence Analysis-All Coding Exons)	SELENOI	SELENOI, SELI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOH geni (Tüm Gen Dizi Analizi)	SELENOH gene (Sequence Analysis-All Coding Exons)	SELENOH	SELENOH, SELH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENOF geni (Tüm Gen Dizi Analizi)	SELENOF gene (Sequence Analysis-All Coding Exons)	SELENOF	SELENOF, SEP15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SELENBP1 geni (Tüm Gen Dizi Analizi)	SELENBP1 gene (Sequence Analysis-All Coding Exons)	SELENBP1	SELENBP1, SP56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEL1L2 geni (Tüm Gen Dizi Analizi)	SEL1L2 gene (Sequence Analysis-All Coding Exons)	SEL1L2	SEL1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEL1L geni (Tüm Gen Dizi Analizi)	SEL1L gene (Sequence Analysis-All Coding Exons)	SEL1L	SEL1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEH1L geni (Tüm Gen Dizi Analizi)	SEH1L gene (Sequence Analysis-All Coding Exons)	SEH1L	SEH1L, SEH1, SEC13L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SECTM1 geni (Tüm Gen Dizi Analizi)	SECTM1 gene (Sequence Analysis-All Coding Exons)	SECTM1	SECTM1, K12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SECISBP2L geni (Tüm Gen Dizi Analizi)	SECISBP2L gene (Sequence Analysis-All Coding Exons)	SECISBP2L	SECISBP2L, SBP2L, KIAA0754	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC62 geni (Tüm Gen Dizi Analizi)	SEC62 gene (Sequence Analysis-All Coding Exons)	SEC62	SEC62, TLOC1, HTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC61G geni (Tüm Gen Dizi Analizi)	SEC61G gene (Sequence Analysis-All Coding Exons)	SEC61G	SEC61G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAG1 geni (Tüm Gen Dizi Analizi)	STAG1 gene (Sequence Analysis-All Coding Exons)	STAG1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC61B geni (Tüm Gen Dizi Analizi)	SEC61B gene (Sequence Analysis-All Coding Exons)	SEC61B	SEC61B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC31B geni (Tüm Gen Dizi Analizi)	SEC31B gene (Sequence Analysis-All Coding Exons)	SEC31B	SEC31B, SEC31L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAB2 geni (Tüm Gen Dizi Analizi)	STAB2 gene (Sequence Analysis-All Coding Exons)	STAB2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
STAB1 geni (Tüm Gen Dizi Analizi)	STAB1 gene (Sequence Analysis-All Coding Exons)	STAB1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC31A geni (Tüm Gen Dizi Analizi)	SEC31A gene (Sequence Analysis-All Coding Exons)	SEC31A	SEC31A, SEC31L1, KIAA0905	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC24C geni (Tüm Gen Dizi Analizi)	SEC24C gene (Sequence Analysis-All Coding Exons)	SEC24C	SEC24C, KIAA0079	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC24B geni (Tüm Gen Dizi Analizi)	SEC24B gene (Sequence Analysis-All Coding Exons)	SEC24B	SEC24B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST8SIA3 geni (Tüm Gen Dizi Analizi)	ST8SIA3 gene (Sequence Analysis-All Coding Exons)	ST8SIA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC24A geni (Tüm Gen Dizi Analizi)	SEC24A gene (Sequence Analysis-All Coding Exons)	SEC24A	SEC24A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC22C geni (Tüm Gen Dizi Analizi)	SEC22C gene (Sequence Analysis-All Coding Exons)	SEC22C	SEC22C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC22B geni (Tüm Gen Dizi Analizi)	SEC22B gene (Sequence Analysis-All Coding Exons)	SEC22B	SEC22L1, SEC22B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC22A geni (Tüm Gen Dizi Analizi)	SEC22A gene (Sequence Analysis-All Coding Exons)	SEC22A	SEC22A, SEC22L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC16B geni (Tüm Gen Dizi Analizi)	SEC16B gene (Sequence Analysis-All Coding Exons)	SEC16B	SEC16B, L2TP2, RGPR, SEC16S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC16A geni (Tüm Gen Dizi Analizi)	SEC16A gene (Sequence Analysis-All Coding Exons)	SEC16A	SEC16A, SEC16L, KIAA0310, p250	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC14L4 geni (Tüm Gen Dizi Analizi)	SEC14L4 gene (Sequence Analysis-All Coding Exons)	SEC14L4	SEC14L4, TAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC14L3 geni (Tüm Gen Dizi Analizi)	SEC14L3 gene (Sequence Analysis-All Coding Exons)	SEC14L3	SEC14L3, TAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC14L2 geni (Tüm Gen Dizi Analizi)	SEC14L2 gene (Sequence Analysis-All Coding Exons)	SEC14L2	SEC14L2, TAP, KIAA1186	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC14L1 geni (Tüm Gen Dizi Analizi)	SEC14L1 gene (Sequence Analysis-All Coding Exons)	SEC14L1	SEC14L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEC13 geni (Tüm Gen Dizi Analizi)	SEC13 gene (Sequence Analysis-All Coding Exons)	SEC13	SEC13L1, DS51231E, SEC13R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEBOX geni (Tüm Gen Dizi Analizi)	SEBOX gene (Sequence Analysis-All Coding Exons)	SEBOX	SEBOX, OG9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDS geni (Tüm Gen Dizi Analizi)	SDS gene (Sequence Analysis-All Coding Exons)	SDS	SDS, SDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDR9C7 geni (Tüm Gen Dizi Analizi)	SDR9C7 gene (Sequence Analysis-All Coding Exons)	SDR9C7	SDR0	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDR42E1 geni (Tüm Gen Dizi Analizi)	SDR42E1 gene (Sequence Analysis-All Coding Exons)	SDR42E1	SDR42E1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDR39U1 geni (Tüm Gen Dizi Analizi)	SDR39U1 gene (Sequence Analysis-All Coding Exons)	SDR39U1	SDR39U1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDR16C5 geni (Tüm Gen Dizi Analizi)	SDR16C5 gene (Sequence Analysis-All Coding Exons)	SDR16C5	RDHE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST3GAL2 geni (Tüm Gen Dizi Analizi)	ST3GAL2 gene (Sequence Analysis-All Coding Exons)	ST3GAL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ST3GAL1 geni (Tüm Gen Dizi Analizi)	ST3GAL1 gene (Sequence Analysis-All Coding Exons)	ST3GAL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDPR geni (Tüm Gen Dizi Analizi)	SDPR gene (Sequence Analysis-All Coding Exons)	SDPR	SDPR, SDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDK2 geni (Tüm Gen Dizi Analizi)	SDK2 gene (Sequence Analysis-All Coding Exons)	SDK2	SDK2, KIAA1514	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDK1 geni (Tüm Gen Dizi Analizi)	SDK1 gene (Sequence Analysis-All Coding Exons)	SDK1	SDK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDHAF3 geni (Tüm Gen Dizi Analizi)	SDHAF3 gene (Sequence Analysis-All Coding Exons)	SDHAF3	ACN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDF4 geni (Tüm Gen Dizi Analizi)	SDF4 gene (Sequence Analysis-All Coding Exons)	SDF4	SDF4, CAB45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDF2L1 geni (Tüm Gen Dizi Analizi)	SDF2L1 gene (Sequence Analysis-All Coding Exons)	SDF2L1	SDF2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDF2 geni (Tüm Gen Dizi Analizi)	SDF2 gene (Sequence Analysis-All Coding Exons)	SDF2	SDF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDCBP2 geni (Tüm Gen Dizi Analizi)	SDCBP2 gene (Sequence Analysis-All Coding Exons)	SDCBP2	SDCBP2, SITAC, ST2, SITAC18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSX2IP geni (Tüm Gen Dizi Analizi)	SSX2IP gene (Sequence Analysis-All Coding Exons)	SSX2IP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDCBP geni (Tüm Gen Dizi Analizi)	SDCBP gene (Sequence Analysis-All Coding Exons)	SDCBP	SDCBP, TACIP18, MDA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDC4 geni (Tüm Gen Dizi Analizi)	SDC4 gene (Sequence Analysis-All Coding Exons)	SDC4	SDC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDC2 geni (Tüm Gen Dizi Analizi)	SDC2 gene (Sequence Analysis-All Coding Exons)	SDC2	SDC2, HSPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SDC1 geni (Tüm Gen Dizi Analizi)	SDC1 gene (Sequence Analysis-All Coding Exons)	SDC1	SDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCYL3 geni (Tüm Gen Dizi Analizi)	SCYL3 gene (Sequence Analysis-All Coding Exons)	SCYL3	SCYL3, PACE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCYL2 geni (Tüm Gen Dizi Analizi)	SCYL2 gene (Sequence Analysis-All Coding Exons)	SCYL2	SCYL2, CVAK104, KIAA1360	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCX geni (Tüm Gen Dizi Analizi)	SCX gene (Sequence Analysis-All Coding Exons)	SCX	SCXA, SCX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCUBE3 geni (Tüm Gen Dizi Analizi)	SCUBE3 gene (Sequence Analysis-All Coding Exons)	SCUBE3	SCUBE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SCUBE2 geni (Tüm Gen Dizi Analizi)	SCUBE2 gene (Sequence Analysis-All Coding Exons)	SCUBE2	SCUBE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SSSCA1 geni (Tüm Gen Dizi Analizi)	SSSCA1 gene (Sequence Analysis-All Coding Exons)	SSSCA1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCUBE1 geni (Tüm Gen Dizi Analizi)	SCUBE1 gene (Sequence Analysis-All Coding Exons)	SCUBE1	SCUBE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCTR geni (Tüm Gen Dizi Analizi)	SCTR gene (Sequence Analysis-All Coding Exons)	SCTR	SCTR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCT geni (Tüm Gen Dizi Analizi)	SCT gene (Sequence Analysis-All Coding Exons)	SCT	SCT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRT1 geni (Tüm Gen Dizi Analizi)	SCRT1 gene (Sequence Analysis-All Coding Exons)	SCRT1	SCRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRN3 geni (Tüm Gen Dizi Analizi)	SCRN3 gene (Sequence Analysis-All Coding Exons)	SCRN3	SCRN3, SES3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRN2 geni (Tüm Gen Dizi Analizi)	SCRN2 gene (Sequence Analysis-All Coding Exons)	SCRN2	SCRN2, SES2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRN1 geni (Tüm Gen Dizi Analizi)	SCRN1 gene (Sequence Analysis-All Coding Exons)	SCRN1	SCRN1, SES1, KIAA0193	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRIB geni (Tüm Gen Dizi Analizi)	SCRIB gene (Sequence Analysis-All Coding Exons)	SCRIB	SCRIB, SCRB1, KIAA0147	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCRG1 geni (Tüm Gen Dizi Analizi)	SCRG1 gene (Sequence Analysis-All Coding Exons)	SCRG1	SCRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCNN1D geni (Tüm Gen Dizi Analizi)	SCNN1D gene (Sequence Analysis-All Coding Exons)	SCNN1D	SCNN1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCNM1 geni (Tüm Gen Dizi Analizi)	SCNM1 gene (Sequence Analysis-All Coding Exons)	SCNM1	SCNM1, MGC3190	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCN7A geni (Tüm Gen Dizi Analizi)	SCN7A gene (Sequence Analysis-All Coding Exons)	SCN7A	SCN7A, SCN6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCN3A geni (Tüm Gen Dizi Analizi)	SCN3A gene (Sequence Analysis-All Coding Exons)	SCN3A	SCN3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCML2 geni (Tüm Gen Dizi Analizi)	SCML2 gene (Sequence Analysis-All Coding Exons)	SCML2	SCML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCML1 geni (Tüm Gen Dizi Analizi)	SCML1 gene (Sequence Analysis-All Coding Exons)	SCML1	SCML1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCMH1 geni (Tüm Gen Dizi Analizi)	SCMH1 gene (Sequence Analysis-All Coding Exons)	SCMH1	SCMH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCLY geni (Tüm Gen Dizi Analizi)	SCLY gene (Sequence Analysis-All Coding Exons)	SCLY	SCLY, SCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCLT1 geni (Tüm Gen Dizi Analizi)	SCLT1 gene (Sequence Analysis-All Coding Exons)	SCLT1	SCLT1, CAP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCIN geni (Tüm Gen Dizi Analizi)	SCIN gene (Sequence Analysis-All Coding Exons)	SCIN	SCIN, KIAA1905	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCIMP geni (Tüm Gen Dizi Analizi)	SCIMP gene (Sequence Analysis-All Coding Exons)	SCIMP	SCIMP, C17orf87	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCHLAP1 geni (Tüm Gen Dizi Analizi)	SCHLAP1 gene (Sequence Analysis-All Coding Exons)	SCHLAP1	SCHLAP1, PCAT114, LINC00913	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB3A1 geni (Tüm Gen Dizi Analizi)	SCGB3A1 gene (Sequence Analysis-All Coding Exons)	SCGB3A1	SCGB3A1, HIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB2B2 geni (Tüm Gen Dizi Analizi)	SCGB2B2 gene (Sequence Analysis-All Coding Exons)	SCGB2B2	SCGB2B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB2A2 geni (Tüm Gen Dizi Analizi)	SCGB2A2 gene (Sequence Analysis-All Coding Exons)	SCGB2A2	MGB1, SCGB2A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB2A1 geni (Tüm Gen Dizi Analizi)	SCGB2A1 gene (Sequence Analysis-All Coding Exons)	SCGB2A1	SCGB2A1, MGB2, LPNC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB1D4 geni (Tüm Gen Dizi Analizi)	SCGB1D4 gene (Sequence Analysis-All Coding Exons)	SCGB1D4	SCGB1D4, IIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB1D2 geni (Tüm Gen Dizi Analizi)	SCGB1D2 gene (Sequence Analysis-All Coding Exons)	SCGB1D2	SCGB1D2, LPNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB1D1 geni (Tüm Gen Dizi Analizi)	SCGB1D1 gene (Sequence Analysis-All Coding Exons)	SCGB1D1	SCGB1D1, LPNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB1C1 geni (Tüm Gen Dizi Analizi)	SCGB1C1 gene (Sequence Analysis-All Coding Exons)	SCGB1C1	SCGB1C1, RYD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCGB1A1 geni (Tüm Gen Dizi Analizi)	SCGB1A1 gene (Sequence Analysis-All Coding Exons)	SCGB1A1	SCGB1A1, UGB, CC10, CCSP, SCGB1A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCG5 geni (Tüm Gen Dizi Analizi)	SCG5 gene (Sequence Analysis-All Coding Exons)	SCG5	SCG5, SGNE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCG3 geni (Tüm Gen Dizi Analizi)	SCG3 gene (Sequence Analysis-All Coding Exons)	SCG3	SCG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCG2 geni (Tüm Gen Dizi Analizi)	SCG2 gene (Sequence Analysis-All Coding Exons)	SCG2	SCG2, CHGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCEL geni (Tüm Gen Dizi Analizi)	SCEL gene (Sequence Analysis-All Coding Exons)	SCEL	SCEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCD5 geni (Tüm Gen Dizi Analizi)	SCD5 gene (Sequence Analysis-All Coding Exons)	SCD5	SCD4, ACOD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCD geni (Tüm Gen Dizi Analizi)	SCD gene (Sequence Analysis-All Coding Exons)	SCD	SCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA8 geni (Tüm Gen Dizi Analizi)	SCARNA8 gene (Sequence Analysis-All Coding Exons)	SCARNA8	SCARNA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA7 geni (Tüm Gen Dizi Analizi)	SCARNA7 gene (Sequence Analysis-All Coding Exons)	SCARNA7	SCARNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA6 geni (Tüm Gen Dizi Analizi)	SCARNA6 gene (Sequence Analysis-All Coding Exons)	SCARNA6	SCARNA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA5 geni (Tüm Gen Dizi Analizi)	SCARNA5 gene (Sequence Analysis-All Coding Exons)	SCARNA5	SCARNA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA18 geni (Tüm Gen Dizi Analizi)	SCARNA18 gene (Sequence Analysis-All Coding Exons)	SCARNA18	SCARNA18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA17 geni (Tüm Gen Dizi Analizi)	SCARNA17 gene (Sequence Analysis-All Coding Exons)	SCARNA17	SCARNA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA15 geni (Tüm Gen Dizi Analizi)	SCARNA15 gene (Sequence Analysis-All Coding Exons)	SCARNA15	SCARNA15, ACA45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA12 geni (Tüm Gen Dizi Analizi)	SCARNA12 gene (Sequence Analysis-All Coding Exons)	SCARNA12	SCARNA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARNA10 geni (Tüm Gen Dizi Analizi)	SCARNA10 gene (Sequence Analysis-All Coding Exons)	SCARNA10	SCARNA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRPK2 geni (Tüm Gen Dizi Analizi)	SRPK2 gene (Sequence Analysis-All Coding Exons)	SRPK2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SRPK1 geni (Tüm Gen Dizi Analizi)	SRPK1 gene (Sequence Analysis-All Coding Exons)	SRPK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARF1 geni (Tüm Gen Dizi Analizi)	SCARF1 gene (Sequence Analysis-All Coding Exons)	SCARF1	SCARF1, SREC, KIAA0149	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARA5 geni (Tüm Gen Dizi Analizi)	SCARA5 gene (Sequence Analysis-All Coding Exons)	SCARA5	SCARA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCARA3 geni (Tüm Gen Dizi Analizi)	SCARA3 gene (Sequence Analysis-All Coding Exons)	SCARA3	SCARA3, MSLR1, CSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAPER geni (Tüm Gen Dizi Analizi)	SCAPER gene (Sequence Analysis-All Coding Exons)	SCAPER	SCAPER, KIAA1454	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAP geni (Tüm Gen Dizi Analizi)	SCAP gene (Sequence Analysis-All Coding Exons)	SCAP	SCAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAND1 geni (Tüm Gen Dizi Analizi)	SCAND1 gene (Sequence Analysis-All Coding Exons)	SCAND1	SCAND1, RAZ1, SDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAMP5 geni (Tüm Gen Dizi Analizi)	SCAMP5 gene (Sequence Analysis-All Coding Exons)	SCAMP5	SCAMP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAMP4 geni (Tüm Gen Dizi Analizi)	SCAMP4 gene (Sequence Analysis-All Coding Exons)	SCAMP4	SCAMP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAMP3 geni (Tüm Gen Dizi Analizi)	SCAMP3 gene (Sequence Analysis-All Coding Exons)	SCAMP3	SCAMP3, PROPIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAMP2 geni (Tüm Gen Dizi Analizi)	SCAMP2 gene (Sequence Analysis-All Coding Exons)	SCAMP2	SCAMP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAMP1 geni (Tüm Gen Dizi Analizi)	SCAMP1 gene (Sequence Analysis-All Coding Exons)	SCAMP1	SCAMP1, SCAMP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAF8 geni (Tüm Gen Dizi Analizi)	SCAF8 gene (Sequence Analysis-All Coding Exons)	SCAF8	SCAF8, KIAA1116	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAF4 geni (Tüm Gen Dizi Analizi)	SCAF4 gene (Sequence Analysis-All Coding Exons)	SCAF4	SCAF4, KIAA1172	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAF11 geni (Tüm Gen Dizi Analizi)	SCAF11 gene (Sequence Analysis-All Coding Exons)	SCAF11	SCAF11, SFRS2IP, SIP1, CASC11, SRRP129	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SCAF1 geni (Tüm Gen Dizi Analizi)	SCAF1 gene (Sequence Analysis-All Coding Exons)	SCAF1	SCAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SBSN geni (Tüm Gen Dizi Analizi)	SBSN gene (Sequence Analysis-All Coding Exons)	SBSN	SBSN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SBNO2 geni (Tüm Gen Dizi Analizi)	SBNO2 gene (Sequence Analysis-All Coding Exons)	SBNO2	SBNO2, KIAA0963	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SBNO1 geni (Tüm Gen Dizi Analizi)	SBNO1 gene (Sequence Analysis-All Coding Exons)	SBNO1	SBNO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAXO1 geni (Tüm Gen Dizi Analizi)	SAXO1 gene (Sequence Analysis-All Coding Exons)	SAXO1	SAXO1, FAM154A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAV1 geni (Tüm Gen Dizi Analizi)	SAV1 gene (Sequence Analysis-All Coding Exons)	SAV1	WW45, SAV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SATB1 geni (Tüm Gen Dizi Analizi)	SATB1 gene (Sequence Analysis-All Coding Exons)	SATB1	SATB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAT2 geni (Tüm Gen Dizi Analizi)	SAT2 gene (Sequence Analysis-All Coding Exons)	SAT2	SAT2, SSAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAT1 geni (Tüm Gen Dizi Analizi)	SAT1 gene (Sequence Analysis-All Coding Exons)	SAT1	SAT1, SSAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SASH3 geni (Tüm Gen Dizi Analizi)	SASH3 gene (Sequence Analysis-All Coding Exons)	SASH3	SASH3, HACS2, CXorf9, SLY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SASH1 geni (Tüm Gen Dizi Analizi)	SASH1 gene (Sequence Analysis-All Coding Exons)	SASH1	SASH1, KIAA0790	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SASH1 geni (Tüm Gen Dizi Analizi)	SASH1 gene (Sequence Analysis-All Coding Exons)	SASH1	SASH1, KIAA0790	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SART3 geni (Tüm Gen Dizi Analizi)	SART3 gene (Sequence Analysis-All Coding Exons)	SART3	SART3, P100, KIAA0156, TIP110	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SART1 geni (Tüm Gen Dizi Analizi)	SART1 gene (Sequence Analysis-All Coding Exons)	SART1	SART1, HOMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SARS geni (Tüm Gen Dizi Analizi)	SARS gene (Sequence Analysis-All Coding Exons)	SARS	SARS, SERS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SARNP geni (Tüm Gen Dizi Analizi)	SARNP gene (Sequence Analysis-All Coding Exons)	SARNP	SARNP, CIP29, HCC1, HSPC316	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SARM1 geni (Tüm Gen Dizi Analizi)	SARM1 gene (Sequence Analysis-All Coding Exons)	SARM1	SARM, KIAA0524	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SARAF geni (Tüm Gen Dizi Analizi)	SARAF gene (Sequence Analysis-All Coding Exons)	SARAF	TMEM66, SARAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAR1A geni (Tüm Gen Dizi Analizi)	SAR1A gene (Sequence Analysis-All Coding Exons)	SAR1A	SAR1A1, SAR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAPCD2 geni (Tüm Gen Dizi Analizi)	SAPCD2 gene (Sequence Analysis-All Coding Exons)	SAPCD2	C9orf140, P42.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAP30L geni (Tüm Gen Dizi Analizi)	SAP30L gene (Sequence Analysis-All Coding Exons)	SAP30L	SAP30L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAP30BP geni (Tüm Gen Dizi Analizi)	SAP30BP gene (Sequence Analysis-All Coding Exons)	SAP30BP	SAP30BP, HTRP, HCNGP, HTRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAP30 geni (Tüm Gen Dizi Analizi)	SAP30 gene (Sequence Analysis-All Coding Exons)	SAP30	SAP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAP18 geni (Tüm Gen Dizi Analizi)	SAP18 gene (Sequence Analysis-All Coding Exons)	SAP18	SAP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAP130 geni (Tüm Gen Dizi Analizi)	SAP130 gene (Sequence Analysis-All Coding Exons)	SAP130	SAP130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAMSN1 geni (Tüm Gen Dizi Analizi)	SAMSN1 gene (Sequence Analysis-All Coding Exons)	SAMSN1	SAMSN1, HACS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAMMSON geni (Tüm Gen Dizi Analizi)	SAMMSON gene (Sequence Analysis-All Coding Exons)	SAMMSON	SAMMSON, LINC01212	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAMM50 geni (Tüm Gen Dizi Analizi)	SAMM50 gene (Sequence Analysis-All Coding Exons)	SAMM50	SAMM50, SAM50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAMD8 geni (Tüm Gen Dizi Analizi)	SAMD8 gene (Sequence Analysis-All Coding Exons)	SAMD8	SAMD8, SMSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAMD4A geni (Tüm Gen Dizi Analizi)	SAMD4A gene (Sequence Analysis-All Coding Exons)	SAMD4A	SAMD4A, SMAUG1, KIAA1053	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SAMD11 geni (Tüm Gen Dizi Analizi)	SAMD11 gene (Sequence Analysis-All Coding Exons)	SAMD11	SAMD11, MRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SALL3 geni (Tüm Gen Dizi Analizi)	SALL3 gene (Sequence Analysis-All Coding Exons)	SALL3	SALL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAGE1 geni (Tüm Gen Dizi Analizi)	SAGE1 gene (Sequence Analysis-All Coding Exons)	SAGE1	SAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAFB2 geni (Tüm Gen Dizi Analizi)	SAFB2 gene (Sequence Analysis-All Coding Exons)	SAFB2	SAFB2, KIAA0138	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAFB geni (Tüm Gen Dizi Analizi)	SAFB gene (Sequence Analysis-All Coding Exons)	SAFB	SAFB, HET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAE1 geni (Tüm Gen Dizi Analizi)	SAE1 gene (Sequence Analysis-All Coding Exons)	SAE1	SAE1, SUA1, AOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SACM1L geni (Tüm Gen Dizi Analizi)	SACM1L gene (Sequence Analysis-All Coding Exons)	SACM1L	SACM1L, SAC1, KIAA0851	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAA4 geni (Tüm Gen Dizi Analizi)	SAA4 gene (Sequence Analysis-All Coding Exons)	SAA4	SAA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAA2 geni (Tüm Gen Dizi Analizi)	SAA2 gene (Sequence Analysis-All Coding Exons)	SAA2	SAA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SAA1 geni (Tüm Gen Dizi Analizi)	SAA1 gene (Sequence Analysis-All Coding Exons)	SAA1	SAA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S1PR5 geni (Tüm Gen Dizi Analizi)	S1PR5 gene (Sequence Analysis-All Coding Exons)	S1PR5	S1PR5, EDG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S1PR4 geni (Tüm Gen Dizi Analizi)	S1PR4 gene (Sequence Analysis-All Coding Exons)	S1PR4	S1PR4, EDG6, S1P4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S1PR3 geni (Tüm Gen Dizi Analizi)	S1PR3 gene (Sequence Analysis-All Coding Exons)	S1PR3	S1PR3, EDG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S1PR1 geni (Tüm Gen Dizi Analizi)	S1PR1 gene (Sequence Analysis-All Coding Exons)	S1PR1	S1PR1, EDG1, S1P1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100Z geni (Tüm Gen Dizi Analizi)	S100Z gene (Sequence Analysis-All Coding Exons)	S100Z	S100Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100PBP geni (Tüm Gen Dizi Analizi)	S100PBP gene (Sequence Analysis-All Coding Exons)	S100PBP	S100PBP, S100PBPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100P geni (Tüm Gen Dizi Analizi)	S100P gene (Sequence Analysis-All Coding Exons)	S100P	S100P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100G geni (Tüm Gen Dizi Analizi)	S100G gene (Sequence Analysis-All Coding Exons)	S100G	CALB3, CABP9K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100B geni (Tüm Gen Dizi Analizi)	S100B gene (Sequence Analysis-All Coding Exons)	S100B	S100B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A9 geni (Tüm Gen Dizi Analizi)	S100A9 gene (Sequence Analysis-All Coding Exons)	S100A9	S100A9, CAGB, CFAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A8 geni (Tüm Gen Dizi Analizi)	S100A8 gene (Sequence Analysis-All Coding Exons)	S100A8	S100A8, CAGA, CFAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A7 geni (Tüm Gen Dizi Analizi)	S100A7 gene (Sequence Analysis-All Coding Exons)	S100A7	S100A7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A6 geni (Tüm Gen Dizi Analizi)	S100A6 gene (Sequence Analysis-All Coding Exons)	S100A6	S100A6, CACY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A5 geni (Tüm Gen Dizi Analizi)	S100A5 gene (Sequence Analysis-All Coding Exons)	S100A5	S100A5, S100D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A4 geni (Tüm Gen Dizi Analizi)	S100A4 gene (Sequence Analysis-All Coding Exons)	S100A4	S100A4, CAPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A3 geni (Tüm Gen Dizi Analizi)	S100A3 gene (Sequence Analysis-All Coding Exons)	S100A3	S100A3, S100E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A2 geni (Tüm Gen Dizi Analizi)	S100A2 gene (Sequence Analysis-All Coding Exons)	S100A2	S100A2, S100L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A14 geni (Tüm Gen Dizi Analizi)	S100A14 gene (Sequence Analysis-All Coding Exons)	S100A14	S100A14, BCMP84	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A13 geni (Tüm Gen Dizi Analizi)	S100A13 gene (Sequence Analysis-All Coding Exons)	S100A13	S100A13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A12 geni (Tüm Gen Dizi Analizi)	S100A12 gene (Sequence Analysis-All Coding Exons)	S100A12	S100A12, CAAF1, CGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A11 geni (Tüm Gen Dizi Analizi)	S100A11 gene (Sequence Analysis-All Coding Exons)	S100A11	S100A11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A10 geni (Tüm Gen Dizi Analizi)	S100A10 gene (Sequence Analysis-All Coding Exons)	S100A10	S100A10, CAL1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
S100A1 geni (Tüm Gen Dizi Analizi)	S100A1 gene (Sequence Analysis-All Coding Exons)	S100A1	S100A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RYR3 geni (Tüm Gen Dizi Analizi)	RYR3 gene (Sequence Analysis-All Coding Exons)	RYR3	RYR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RYK geni (Tüm Gen Dizi Analizi)	RYK gene (Sequence Analysis-All Coding Exons)	RYK	RYK, RYK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RYBP geni (Tüm Gen Dizi Analizi)	RYBP gene (Sequence Analysis-All Coding Exons)	RYBP	RYBP, YEAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXRG geni (Tüm Gen Dizi Analizi)	RXRG gene (Sequence Analysis-All Coding Exons)	RXRG	RXRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXRB geni (Tüm Gen Dizi Analizi)	RXRB gene (Sequence Analysis-All Coding Exons)	RXRB	RXRB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXRA geni (Tüm Gen Dizi Analizi)	RXRA gene (Sequence Analysis-All Coding Exons)	RXRA	RXRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXFP3 geni (Tüm Gen Dizi Analizi)	RXFP3 gene (Sequence Analysis-All Coding Exons)	RXFP3	RLNSR1, SALPR, GPCR135	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXFP2 geni (Tüm Gen Dizi Analizi)	RXFP2 gene (Sequence Analysis-All Coding Exons)	RXFP2	LGR8, GREAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXFP1 geni (Tüm Gen Dizi Analizi)	RXFP1 gene (Sequence Analysis-All Coding Exons)	RXFP1	RXFP1, LGR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RWDD3 geni (Tüm Gen Dizi Analizi)	RWDD3 gene (Sequence Analysis-All Coding Exons)	RWDD3	RWDD3, RSUME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUVBL2 geni (Tüm Gen Dizi Analizi)	RUVBL2 gene (Sequence Analysis-All Coding Exons)	RUVBL2	RUVBL2, TIP48, ECP51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUVBL1 geni (Tüm Gen Dizi Analizi)	RUVBL1 gene (Sequence Analysis-All Coding Exons)	RUVBL1	RUVBL1, NMP238, TIP49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIRE2 geni (Tüm Gen Dizi Analizi)	SPIRE2 gene (Sequence Analysis-All Coding Exons)	SPIRE2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPIRE1 geni (Tüm Gen Dizi Analizi)	SPIRE1 gene (Sequence Analysis-All Coding Exons)	SPIRE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUSC2 geni (Tüm Gen Dizi Analizi)	RUSC2 gene (Sequence Analysis-All Coding Exons)	RUSC2	RUSC2, KIAA0375, IPORIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUSC1 geni (Tüm Gen Dizi Analizi)	RUSC1 gene (Sequence Analysis-All Coding Exons)	RUSC1	RUSC1, NESCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPINT1 geni (Tüm Gen Dizi Analizi)	SPINT1 gene (Sequence Analysis-All Coding Exons)	SPINT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUNX3 geni (Tüm Gen Dizi Analizi)	RUNX3 gene (Sequence Analysis-All Coding Exons)	RUNX3	RUNX3, CBFA3, PEBP2A3, AML-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUNX1T1 geni (Tüm Gen Dizi Analizi)	RUNX1T1 gene (Sequence Analysis-All Coding Exons)	RUNX1T1	RUNX1T1, CBFA2T1, MM_ETO, ETO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUNDC3B geni (Tüm Gen Dizi Analizi)	RUNDC3B gene (Sequence Analysis-All Coding Exons)	RUNDC3B	RUNDC3B, RPIP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUNDC3A geni (Tüm Gen Dizi Analizi)	RUNDC3A gene (Sequence Analysis-All Coding Exons)	RUNDC3A	RUNDC3A, RPIP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUFY3 geni (Tüm Gen Dizi Analizi)	RUFY3 gene (Sequence Analysis-All Coding Exons)	RUFY3	RUFY3, SINGAR1, RIPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUFY2 geni (Tüm Gen Dizi Analizi)	RUFY2 gene (Sequence Analysis-All Coding Exons)	RUFY2	RUFY2, KIAA1537	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RUFY1 geni (Tüm Gen Dizi Analizi)	RUFY1 gene (Sequence Analysis-All Coding Exons)	RUFY1	RUFY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTP4 geni (Tüm Gen Dizi Analizi)	RTP4 gene (Sequence Analysis-All Coding Exons)	RTP4	RTP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTP3 geni (Tüm Gen Dizi Analizi)	RTP3 gene (Sequence Analysis-All Coding Exons)	RTP3	RTP3, TMEM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTP2 geni (Tüm Gen Dizi Analizi)	RTP2 gene (Sequence Analysis-All Coding Exons)	RTP2	RTP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTP1 geni (Tüm Gen Dizi Analizi)	RTP1 gene (Sequence Analysis-All Coding Exons)	RTP1	RTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTN4RL2 geni (Tüm Gen Dizi Analizi)	RTN4RL2 gene (Sequence Analysis-All Coding Exons)	RTN4RL2	RTN4RL2, NGRH1, NGR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTN4RL1 geni (Tüm Gen Dizi Analizi)	RTN4RL1 gene (Sequence Analysis-All Coding Exons)	RTN4RL1	RTN4RL1, NGRH2, NGR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTN4 geni (Tüm Gen Dizi Analizi)	RTN4 gene (Sequence Analysis-All Coding Exons)	RTN4	RTN4, NOGO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTN3 geni (Tüm Gen Dizi Analizi)	RTN3 gene (Sequence Analysis-All Coding Exons)	RTN3	RTN3, NSPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTN1 geni (Tüm Gen Dizi Analizi)	RTN1 gene (Sequence Analysis-All Coding Exons)	RTN1	RTN1, NSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTL1 geni (Tüm Gen Dizi Analizi)	RTL1 gene (Sequence Analysis-All Coding Exons)	RTL1	RTL1, PEG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTL1 geni (Tüm Gen Dizi Analizi)	RTL1 gene (Sequence Analysis-All Coding Exons)	RTL1	RTL1, PEG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTL1 geni (Tüm Gen Dizi Analizi)	RTL1 gene (Sequence Analysis-All Coding Exons)	RTL1	RTL1, PEG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTL1 geni (Tüm Gen Dizi Analizi)	RTL1 gene (Sequence Analysis-All Coding Exons)	RTL1	RTL1, PEG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTL1 geni (Tüm Gen Dizi Analizi)	RTL1 gene (Sequence Analysis-All Coding Exons)	RTL1	RTL1, PEG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTKN geni (Tüm Gen Dizi Analizi)	RTKN gene (Sequence Analysis-All Coding Exons)	RTKN	RTKN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTF1 geni (Tüm Gen Dizi Analizi)	RTF1 gene (Sequence Analysis-All Coding Exons)	RTF1	RTF1, KIAA0252	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTCB geni (Tüm Gen Dizi Analizi)	RTCB gene (Sequence Analysis-All Coding Exons)	RTCB	RTCB, C22orf28, HSPC117	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTCA geni (Tüm Gen Dizi Analizi)	RTCA gene (Sequence Analysis-All Coding Exons)	RTCA	RTCD1, RPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RTBDN geni (Tüm Gen Dizi Analizi)	RTBDN gene (Sequence Analysis-All Coding Exons)	RTBDN	RTBDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSU1 geni (Tüm Gen Dizi Analizi)	RSU1 gene (Sequence Analysis-All Coding Exons)	RSU1	RSU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSRC1 geni (Tüm Gen Dizi Analizi)	RSRC1 gene (Sequence Analysis-All Coding Exons)	RSRC1	RSRC1, SRRP53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSPO3 geni (Tüm Gen Dizi Analizi)	RSPO3 gene (Sequence Analysis-All Coding Exons)	RSPO3	RSPO3, PWTSR, CRISTIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSPO2 geni (Tüm Gen Dizi Analizi)	RSPO2 gene (Sequence Analysis-All Coding Exons)	RSPO2	RSPO2, CRISTIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSPH6A geni (Tüm Gen Dizi Analizi)	RSPH6A gene (Sequence Analysis-All Coding Exons)	RSPH6A	RSHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSPH14 geni (Tüm Gen Dizi Analizi)	RSPH14 gene (Sequence Analysis-All Coding Exons)	RSPH14	RSPH14, RTDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSL24D1 geni (Tüm Gen Dizi Analizi)	RSL24D1 gene (Sequence Analysis-All Coding Exons)	RSL24D1	RSL24D1, RLP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSL1D1 geni (Tüm Gen Dizi Analizi)	RSL1D1 gene (Sequence Analysis-All Coding Exons)	RSL1D1	RSL1D1, CSIG, PBK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSF1 geni (Tüm Gen Dizi Analizi)	RSF1 gene (Sequence Analysis-All Coding Exons)	RSF1	HBXAP, RSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSC1A1 geni (Tüm Gen Dizi Analizi)	RSC1A1 gene (Sequence Analysis-All Coding Exons)	RSC1A1	RSC1A1, RS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSBN1 geni (Tüm Gen Dizi Analizi)	RSBN1 gene (Sequence Analysis-All Coding Exons)	RSBN1	RSBN1, ROSBIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RSAD2 geni (Tüm Gen Dizi Analizi)	RSAD2 gene (Sequence Analysis-All Coding Exons)	RSAD2	RSAD2, VIPERIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRP8 geni (Tüm Gen Dizi Analizi)	RRP8 gene (Sequence Analysis-All Coding Exons)	RRP8	RRP8, NML, KIAA0409	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRP36 geni (Tüm Gen Dizi Analizi)	RRP36 gene (Sequence Analysis-All Coding Exons)	RRP36	RRP36, C9orf153	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRP1B geni (Tüm Gen Dizi Analizi)	RRP1B gene (Sequence Analysis-All Coding Exons)	RRP1B	RRP1B, KIAA0179, NNP1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRP15 geni (Tüm Gen Dizi Analizi)	RRP15 gene (Sequence Analysis-All Coding Exons)	RRP15	RRP15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

RRN3 geni (Tüm Gen Dizi Analizi)	RRN3 gene (Sequence Analysis-All Coding Exons)	RRN3	RRN3, TIF1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRM2 geni (Tüm Gen Dizi Analizi)	RRM2 gene (Sequence Analysis-All Coding Exons)	RRM2	RRM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRM1 geni (Tüm Gen Dizi Analizi)	RRM1 gene (Sequence Analysis-All Coding Exons)	RRM1	RRM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRH geni (Tüm Gen Dizi Analizi)	RRH gene (Sequence Analysis-All Coding Exons)	RRH	RRH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RREB1 geni (Tüm Gen Dizi Analizi)	RREB1 gene (Sequence Analysis-All Coding Exons)	RREB1	RREB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPATA9 geni (Tüm Gen Dizi Analizi)	SPATA9 gene (Sequence Analysis-All Coding Exons)	SPATA9		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRBP1 geni (Tüm Gen Dizi Analizi)	RRBP1 gene (Sequence Analysis-All Coding Exons)	RRBP1	RRBP1, ES130	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAS geni (Tüm Gen Dizi Analizi)	RRAS gene (Sequence Analysis-All Coding Exons)	RRAS	RRAS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAGD geni (Tüm Gen Dizi Analizi)	RRAGD gene (Sequence Analysis-All Coding Exons)	RRAGD	RRAGD, RAGD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAGC geni (Tüm Gen Dizi Analizi)	RRAGC gene (Sequence Analysis-All Coding Exons)	RRAGC	RRAGC, RAGC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAGB geni (Tüm Gen Dizi Analizi)	RRAGB gene (Sequence Analysis-All Coding Exons)	RRAGB	RRAGB, RAGB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAGA geni (Tüm Gen Dizi Analizi)	RRAGA gene (Sequence Analysis-All Coding Exons)	RRAGA	RRAGA, RAGA, FIP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRAD geni (Tüm Gen Dizi Analizi)	RRAD gene (Sequence Analysis-All Coding Exons)	RRAD	RRAD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPTOR geni (Tüm Gen Dizi Analizi)	RPTOR gene (Sequence Analysis-All Coding Exons)	RPTOR	RPTOR, RAPTOR, KIAA1303	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPTN geni (Tüm Gen Dizi Analizi)	RPTN gene (Sequence Analysis-All Coding Exons)	RPTN	RPTN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS9 geni (Tüm Gen Dizi Analizi)	RPS9 gene (Sequence Analysis-All Coding Exons)	RPS9	RPS9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS8 geni (Tüm Gen Dizi Analizi)	RPS8 gene (Sequence Analysis-All Coding Exons)	RPS8	RPS8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KB1 geni (Tüm Gen Dizi Analizi)	RPS6KB1 gene (Sequence Analysis-All Coding Exons)	RPS6KB1	RPS6KB1, S6K1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KA6 geni (Tüm Gen Dizi Analizi)	RPS6KA6 gene (Sequence Analysis-All Coding Exons)	RPS6KA6	RPS6KA6, RSK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KA5 geni (Tüm Gen Dizi Analizi)	RPS6KA5 gene (Sequence Analysis-All Coding Exons)	RPS6KA5	RPS6KA5, MSK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KA4 geni (Tüm Gen Dizi Analizi)	RPS6KA4 gene (Sequence Analysis-All Coding Exons)	RPS6KA4	RPS6KA4, RSK8, MSK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KA2 geni (Tüm Gen Dizi Analizi)	RPS6KA2 gene (Sequence Analysis-All Coding Exons)	RPS6KA2	RPS6KA2, RSK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KA1 geni (Tüm Gen Dizi Analizi)	RPS6KA1 gene (Sequence Analysis-All Coding Exons)	RPS6KA1	RPS6KA1, RSK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6 geni (Tüm Gen Dizi Analizi)	RPS6 gene (Sequence Analysis-All Coding Exons)	RPS6	RPS6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPARCL1 geni (Tüm Gen Dizi Analizi)	SPARCL1 gene (Sequence Analysis-All Coding Exons)	SPARCL1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS5 geni (Tüm Gen Dizi Analizi)	RPS5 gene (Sequence Analysis-All Coding Exons)	RPS5	RPS5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS4Y2 geni (Tüm Gen Dizi Analizi)	RPS4Y2 gene (Sequence Analysis-All Coding Exons)	RPS4Y2	RPS4Y2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS4Y1 geni (Tüm Gen Dizi Analizi)	RPS4Y1 gene (Sequence Analysis-All Coding Exons)	RPS4Y1	RPS4Y1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS4X geni (Tüm Gen Dizi Analizi)	RPS4X gene (Sequence Analysis-All Coding Exons)	RPS4X	RPS4X, CCG2, SCAR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS3A geni (Tüm Gen Dizi Analizi)	RPS3A gene (Sequence Analysis-All Coding Exons)	RPS3A	RPS3A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS3 geni (Tüm Gen Dizi Analizi)	RPS3 gene (Sequence Analysis-All Coding Exons)	RPS3	RPS3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS27L geni (Tüm Gen Dizi Analizi)	RPS27L gene (Sequence Analysis-All Coding Exons)	RPS27L	RPS27L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS27A geni (Tüm Gen Dizi Analizi)	RPS27A gene (Sequence Analysis-All Coding Exons)	RPS27A	RPS27A, UBA80, HUBCEP80, CEP80, UBCEP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS27 geni (Tüm Gen Dizi Analizi)	RPS27 gene (Sequence Analysis-All Coding Exons)	RPS27	RPS27, MPS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS25 geni (Tüm Gen Dizi Analizi)	RPS25 gene (Sequence Analysis-All Coding Exons)	RPS25	RPS25	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS23 geni (Tüm Gen Dizi Analizi)	RPS23 gene (Sequence Analysis-All Coding Exons)	RPS23	RPS23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS21 geni (Tüm Gen Dizi Analizi)	RPS21 gene (Sequence Analysis-All Coding Exons)	RPS21	RPS21	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS20 geni (Tüm Gen Dizi Analizi)	RPS20 gene (Sequence Analysis-All Coding Exons)	RPS20	RPS20	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS2 geni (Tüm Gen Dizi Analizi)	RPS2 gene (Sequence Analysis-All Coding Exons)	RPS2	RPS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS19BP1 geni (Tüm Gen Dizi Analizi)	RPS19BP1 gene (Sequence Analysis-All Coding Exons)	RPS19BP1	RPS19BP1, S19BP, MGC52010	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS18 geni (Tüm Gen Dizi Analizi)	RPS18 gene (Sequence Analysis-All Coding Exons)	RPS18	RPS18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS16 geni (Tüm Gen Dizi Analizi)	RPS16 gene (Sequence Analysis-All Coding Exons)	RPS16	RPS16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS15A geni (Tüm Gen Dizi Analizi)	RPS15A gene (Sequence Analysis-All Coding Exons)	RPS15A	RPS15A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS15 geni (Tüm Gen Dizi Analizi)	RPS15 gene (Sequence Analysis-All Coding Exons)	RPS15	RPS15, RIG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS13 geni (Tüm Gen Dizi Analizi)	RPS13 gene (Sequence Analysis-All Coding Exons)	RPS13	RPS13	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS12 geni (Tüm Gen Dizi Analizi)	RPS12 gene (Sequence Analysis-All Coding Exons)	RPS12	RPS12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS11 geni (Tüm Gen Dizi Analizi)	RPS11 gene (Sequence Analysis-All Coding Exons)	RPS11	RPS11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPRM geni (Tüm Gen Dizi Analizi)	RPRM gene (Sequence Analysis-All Coding Exons)	RPRM	RPRM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPRD2 geni (Tüm Gen Dizi Analizi)	RPRD2 gene (Sequence Analysis-All Coding Exons)	RPRD2	RPRD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPRD1B geni (Tüm Gen Dizi Analizi)	RPRD1B gene (Sequence Analysis-All Coding Exons)	RPRD1B	RPRD1B, CREPT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPRD1A geni (Tüm Gen Dizi Analizi)	RPRD1A gene (Sequence Analysis-All Coding Exons)	RPRD1A	RPRD1A, P15RS, FLJ10656	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPP40 geni (Tüm Gen Dizi Analizi)	RPP40 gene (Sequence Analysis-All Coding Exons)	RPP40	RPP40	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SPA17 geni (Tüm Gen Dizi Analizi)	SPA17 gene (Sequence Analysis-All Coding Exons)	SPA17		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPP38 geni (Tüm Gen Dizi Analizi)	RPP38 gene (Sequence Analysis-All Coding Exons)	RPP38	RPP38	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPP30 geni (Tüm Gen Dizi Analizi)	RPP30 gene (Sequence Analysis-All Coding Exons)	RPP30	RPP30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP6 geni (Tüm Gen Dizi Analizi)	SP6 gene (Sequence Analysis-All Coding Exons)	SP6		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPP21 geni (Tüm Gen Dizi Analizi)	RPP21 gene (Sequence Analysis-All Coding Exons)	RPP21	RPP21	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPP14 geni (Tüm Gen Dizi Analizi)	RPP14 gene (Sequence Analysis-All Coding Exons)	RPP14	RPP14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPN2 geni (Tüm Gen Dizi Analizi)	RPN2 gene (Sequence Analysis-All Coding Exons)	RPN2	RPN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP2 geni (Tüm Gen Dizi Analizi)	SP2 gene (Sequence Analysis-All Coding Exons)	SP2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SP140 geni (Tüm Gen Dizi Analizi)	SP140 gene (Sequence Analysis-All Coding Exons)	SP140		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPN1 geni (Tüm Gen Dizi Analizi)	RPN1 gene (Sequence Analysis-All Coding Exons)	RPN1	RPN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPLP2 geni (Tüm Gen Dizi Analizi)	RPLP2 gene (Sequence Analysis-All Coding Exons)	RPLP2	RPLP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPLP1 geni (Tüm Gen Dizi Analizi)	RPLP1 gene (Sequence Analysis-All Coding Exons)	RPLP1	RPLP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPLP0 geni (Tüm Gen Dizi Analizi)	RPLP0 gene (Sequence Analysis-All Coding Exons)	RPLP0	RPLP0	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL9 geni (Tüm Gen Dizi Analizi)	RPL9 gene (Sequence Analysis-All Coding Exons)	RPL9	RPL9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL8 geni (Tüm Gen Dizi Analizi)	RPL8 gene (Sequence Analysis-All Coding Exons)	RPL8	RPL8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL7A geni (Tüm Gen Dizi Analizi)	RPL7A gene (Sequence Analysis-All Coding Exons)	RPL7A	RPL7A, SURF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL7 geni (Tüm Gen Dizi Analizi)	RPL7 gene (Sequence Analysis-All Coding Exons)	RPL7	RPL7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL6 geni (Tüm Gen Dizi Analizi)	RPL6 gene (Sequence Analysis-All Coding Exons)	RPL6	RPL6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL41 geni (Tüm Gen Dizi Analizi)	RPL41 gene (Sequence Analysis-All Coding Exons)	RPL41	RPL41, HG12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL4 geni (Tüm Gen Dizi Analizi)	RPL4 gene (Sequence Analysis-All Coding Exons)	RPL4	RPL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL39L geni (Tüm Gen Dizi Analizi)	RPL39L gene (Sequence Analysis-All Coding Exons)	RPL39L	RPL39L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOX30 geni (Tüm Gen Dizi Analizi)	SOX30 gene (Sequence Analysis-All Coding Exons)	SOX30		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL39 geni (Tüm Gen Dizi Analizi)	RPL39 gene (Sequence Analysis-All Coding Exons)	RPL39	RPL39	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL38 geni (Tüm Gen Dizi Analizi)	RPL38 gene (Sequence Analysis-All Coding Exons)	RPL38	RPL38	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL37A geni (Tüm Gen Dizi Analizi)	RPL37A gene (Sequence Analysis-All Coding Exons)	RPL37A	RPL37A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL37 geni (Tüm Gen Dizi Analizi)	RPL37 gene (Sequence Analysis-All Coding Exons)	RPL37	RPL37	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL36AL geni (Tüm Gen Dizi Analizi)	RPL36AL gene (Sequence Analysis-All Coding Exons)	RPL36AL	RPL36AL, RPL36A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL36A geni (Tüm Gen Dizi Analizi)	RPL36A gene (Sequence Analysis-All Coding Exons)	RPL36A	RPL36A, RPL44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL34 geni (Tüm Gen Dizi Analizi)	RPL34 gene (Sequence Analysis-All Coding Exons)	RPL34	RPL34	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL30 geni (Tüm Gen Dizi Analizi)	RPL30 gene (Sequence Analysis-All Coding Exons)	RPL30	RPL30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL3 geni (Tüm Gen Dizi Analizi)	RPL3 gene (Sequence Analysis-All Coding Exons)	RPL3	RPL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL29 geni (Tüm Gen Dizi Analizi)	RPL29 gene (Sequence Analysis-All Coding Exons)	RPL29	RPL29, HIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL28 geni (Tüm Gen Dizi Analizi)	RPL28 gene (Sequence Analysis-All Coding Exons)	RPL28	RPL28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL27A geni (Tüm Gen Dizi Analizi)	RPL27A gene (Sequence Analysis-All Coding Exons)	RPL27A	RPL27A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL27 geni (Tüm Gen Dizi Analizi)	RPL27 gene (Sequence Analysis-All Coding Exons)	RPL27	RPL27	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL24 geni (Tüm Gen Dizi Analizi)	RPL24 gene (Sequence Analysis-All Coding Exons)	RPL24	RPL24	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL23A geni (Tüm Gen Dizi Analizi)	RPL23A gene (Sequence Analysis-All Coding Exons)	RPL23A	RPL23A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL23 geni (Tüm Gen Dizi Analizi)	RPL23 gene (Sequence Analysis-All Coding Exons)	RPL23	RPL23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL22 geni (Tüm Gen Dizi Analizi)	RPL22 gene (Sequence Analysis-All Coding Exons)	RPL22	RPL22, EAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL19 geni (Tüm Gen Dizi Analizi)	RPL19 gene (Sequence Analysis-All Coding Exons)	RPL19	RPL19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

RPL18A geni (Tüm Gen Dizi Analizi)	RPL18A gene (Sequence Analysis-All Coding Exons)	RPL18A	RPL18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL18 geni (Tüm Gen Dizi Analizi)	RPL18 gene (Sequence Analysis-All Coding Exons)	RPL18	RPL18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL17 geni (Tüm Gen Dizi Analizi)	RPL17 gene (Sequence Analysis-All Coding Exons)	RPL17	RPL17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL13 geni (Tüm Gen Dizi Analizi)	RPL13 gene (Sequence Analysis-All Coding Exons)	RPL13	RPL13, BBC1, D16S44E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL12 geni (Tüm Gen Dizi Analizi)	RPL12 gene (Sequence Analysis-All Coding Exons)	RPL12	RPL12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPL10A geni (Tüm Gen Dizi Analizi)	RPL10A gene (Sequence Analysis-All Coding Exons)	RPL10A	RPL10A, CSA19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPH3AL geni (Tüm Gen Dizi Analizi)	RPH3AL gene (Sequence Analysis-All Coding Exons)	RPH3AL	RPH3AL, NOC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPH3A geni (Tüm Gen Dizi Analizi)	RPH3A gene (Sequence Analysis-All Coding Exons)	RPH3A	RPH3A, KIAA0985	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPE geni (Tüm Gen Dizi Analizi)	RPE gene (Sequence Analysis-All Coding Exons)	RPE	RPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPAP3 geni (Tüm Gen Dizi Analizi)	RPAP3 gene (Sequence Analysis-All Coding Exons)	RPAP3	RPAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPAP2 geni (Tüm Gen Dizi Analizi)	RPAP2 gene (Sequence Analysis-All Coding Exons)	RPAP2	RPAP2, C1orf82	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPAP1 geni (Tüm Gen Dizi Analizi)	RPAP1 gene (Sequence Analysis-All Coding Exons)	RPAP1	RPAP1, KIAA1403	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPAIN geni (Tüm Gen Dizi Analizi)	RPAIN gene (Sequence Analysis-All Coding Exons)	RPAIN	RPAIN, RIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPA4 geni (Tüm Gen Dizi Analizi)	RPA4 gene (Sequence Analysis-All Coding Exons)	RPA4	RPA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPA3 geni (Tüm Gen Dizi Analizi)	RPA3 gene (Sequence Analysis-All Coding Exons)	RPA3	RPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPA2 geni (Tüm Gen Dizi Analizi)	RPA2 gene (Sequence Analysis-All Coding Exons)	RPA2	RPA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPA1 geni (Tüm Gen Dizi Analizi)	RPA1 gene (Sequence Analysis-All Coding Exons)	RPA1	RPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROS1 geni (Tüm Gen Dizi Analizi)	ROS1 gene (Sequence Analysis-All Coding Exons)	ROS1	ROS1, MCF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RORB geni (Tüm Gen Dizi Analizi)	RORB gene (Sequence Analysis-All Coding Exons)	RORB	RORB, RZRB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOHLH1 geni (Tüm Gen Dizi Analizi)	SOHLH1 gene (Sequence Analysis-All Coding Exons)	SOHLH1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RORA geni (Tüm Gen Dizi Analizi)	RORA gene (Sequence Analysis-All Coding Exons)	RORA	RORA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROR1 geni (Tüm Gen Dizi Analizi)	ROR1 gene (Sequence Analysis-All Coding Exons)	ROR1	NTRKR1, ROR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROPN1L geni (Tüm Gen Dizi Analizi)	ROPN1L gene (Sequence Analysis-All Coding Exons)	ROPN1L	ROPN1L, ASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS7 geni (Tüm Gen Dizi Analizi)	SOCS7 gene (Sequence Analysis-All Coding Exons)	SOCS7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROPN1 geni (Tüm Gen Dizi Analizi)	ROPN1 gene (Sequence Analysis-All Coding Exons)	ROPN1	ROPN1, ODF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROCK2 geni (Tüm Gen Dizi Analizi)	ROCK2 gene (Sequence Analysis-All Coding Exons)	ROCK2	ROCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROCK1 geni (Tüm Gen Dizi Analizi)	ROCK1 gene (Sequence Analysis-All Coding Exons)	ROCK1	ROCK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROBO4 geni (Tüm Gen Dizi Analizi)	ROBO4 gene (Sequence Analysis-All Coding Exons)	ROBO4	ROBO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ROBO1 geni (Tüm Gen Dizi Analizi)	ROBO1 gene (Sequence Analysis-All Coding Exons)	ROBO1	ROBO1, DUTT1, SAX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SOCS2 geni (Tüm Gen Dizi Analizi)	SOCS2 gene (Sequence Analysis-All Coding Exons)	SOCS2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNY4 geni (Tüm Gen Dizi Analizi)	RNY4 gene (Sequence Analysis-All Coding Exons)	RNY4	RNY4, Y4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNY3 geni (Tüm Gen Dizi Analizi)	RNY3 gene (Sequence Analysis-All Coding Exons)	RNY3	RNY3, Y3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNY1 geni (Tüm Gen Dizi Analizi)	RNY1 gene (Sequence Analysis-All Coding Exons)	RNY1	RNY1, Y1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNU6-1 geni (Tüm Gen Dizi Analizi)	RNU6-1 gene (Sequence Analysis-All Coding Exons)	RNU6-1	RNU6-1, RNU6A, RNU6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNU5A-1 geni (Tüm Gen Dizi Analizi)	RNU5A-1 gene (Sequence Analysis-All Coding Exons)	RNU5A-1	RNU5A, RNU5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNU2-1 geni (Tüm Gen Dizi Analizi)	RNU2-1 gene (Sequence Analysis-All Coding Exons)	RNU2-1	RNU2-1, RNU2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNU1-4 geni (Tüm Gen Dizi Analizi)	RNU1-4 gene (Sequence Analysis-All Coding Exons)	RNU1-4	RNU1A, RNU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNPEP geni (Tüm Gen Dizi Analizi)	RNPEP gene (Sequence Analysis-All Coding Exons)	RNPEP	RNPEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNMT geni (Tüm Gen Dizi Analizi)	RNMT gene (Sequence Analysis-All Coding Exons)	RNMT	RNMT, MET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNLS geni (Tüm Gen Dizi Analizi)	RNLS gene (Sequence Analysis-All Coding Exons)	RNLS	RNLS, C10orf59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNH1 geni (Tüm Gen Dizi Analizi)	RNH1 gene (Sequence Analysis-All Coding Exons)	RNH1	RNH1, RNH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNGTT geni (Tüm Gen Dizi Analizi)	RNGTT gene (Sequence Analysis-All Coding Exons)	RNGTT	RNGTT, HCE1, CAP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNFT1 geni (Tüm Gen Dizi Analizi)	RNFT1 gene (Sequence Analysis-All Coding Exons)	RNFT1	RNFT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF8 geni (Tüm Gen Dizi Analizi)	RNF8 gene (Sequence Analysis-All Coding Exons)	RNF8	RNF8, KIAA0646	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF7 geni (Tüm Gen Dizi Analizi)	RNF7 gene (Sequence Analysis-All Coding Exons)	RNF7	RNF7, ROC2, SAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF5 geni (Tüm Gen Dizi Analizi)	RNF5 gene (Sequence Analysis-All Coding Exons)	RNF5	RNF5, RING5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF40 geni (Tüm Gen Dizi Analizi)	RNF40 gene (Sequence Analysis-All Coding Exons)	RNF40	RNF40, BRE1B, RBP95, KIAA0661	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF4 geni (Tüm Gen Dizi Analizi)	RNF4 gene (Sequence Analysis-All Coding Exons)	RNF4	RNF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF39 geni (Tüm Gen Dizi Analizi)	RNF39 gene (Sequence Analysis-All Coding Exons)	RNF39	RNF39, HZFW, HZF, LIRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF38 geni (Tüm Gen Dizi Analizi)	RNF38 gene (Sequence Analysis-All Coding Exons)	RNF38	RNF38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF34 geni (Tüm Gen Dizi Analizi)	RNF34 gene (Sequence Analysis-All Coding Exons)	RNF34	RNF34, RFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF32 geni (Tüm Gen Dizi Analizi)	RNF32 gene (Sequence Analysis-All Coding Exons)	RNF32	RNF32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF31 geni (Tüm Gen Dizi Analizi)	RNF31 gene (Sequence Analysis-All Coding Exons)	RNF31	RNF31, ZIBRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF26 geni (Tüm Gen Dizi Analizi)	RNF26 gene (Sequence Analysis-All Coding Exons)	RNF26	RNF26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNUPN geni (Tüm Gen Dizi Analizi)	SNUPN gene (Sequence Analysis-All Coding Exons)	SNUPN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF25 geni (Tüm Gen Dizi Analizi)	RNF25 gene (Sequence Analysis-All Coding Exons)	RNF25	RNF25, AO7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNTG2 geni (Tüm Gen Dizi Analizi)	SNTG2 gene (Sequence Analysis-All Coding Exons)	SNTG2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF24 geni (Tüm Gen Dizi Analizi)	RNF24 gene (Sequence Analysis-All Coding Exons)	RNF24	RNF24, G1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF220 geni (Tüm Gen Dizi Analizi)	RNF220 gene (Sequence Analysis-All Coding Exons)	RNF220	RNF220	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF219 geni (Tüm Gen Dizi Analizi)	RNF219 gene (Sequence Analysis-All Coding Exons)	RNF219	RNF219, C13orf7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF207 geni (Tüm Gen Dizi Analizi)	RNF207 gene (Sequence Analysis-All Coding Exons)	RNF207	RNF207, C1orf188	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF20 geni (Tüm Gen Dizi Analizi)	RNF20 gene (Sequence Analysis-All Coding Exons)	RNF20	RNF20, BRE1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF2 geni (Tüm Gen Dizi Analizi)	RNF2 gene (Sequence Analysis-All Coding Exons)	RNF2	RNF2, RING2, RING1B, HIP1I, BING, BAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF19B geni (Tüm Gen Dizi Analizi)	RNF19B gene (Sequence Analysis-All Coding Exons)	RNF19B	IBRDC3, NKLAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF187 geni (Tüm Gen Dizi Analizi)	RNF187 gene (Sequence Analysis-All Coding Exons)	RNF187	RNF186, RACO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SNRPD3 geni (Tüm Gen Dizi Analizi)	SNRPD3 gene (Sequence Analysis-All Coding Exons)	SNRPD3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF186 geni (Tüm Gen Dizi Analizi)	RNF186 gene (Sequence Analysis-All Coding Exons)	RNF186	RNF186	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF181 geni (Tüm Gen Dizi Analizi)	RNF181 gene (Sequence Analysis-All Coding Exons)	RNF181	RNF181	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF180 geni (Tüm Gen Dizi Analizi)	RNF180 gene (Sequence Analysis-All Coding Exons)	RNF180	RNF180, RINES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF17 geni (Tüm Gen Dizi Analizi)	RNF17 gene (Sequence Analysis-All Coding Exons)	RNF17	RNF17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF167 geni (Tüm Gen Dizi Analizi)	RNF167 gene (Sequence Analysis-All Coding Exons)	RNF167	RNF167, RING105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF166 geni (Tüm Gen Dizi Analizi)	RNF166 gene (Sequence Analysis-All Coding Exons)	RNF166	RNF166	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF152 geni (Tüm Gen Dizi Analizi)	RNF152 gene (Sequence Analysis-All Coding Exons)	RNF152	RNF152	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF146 geni (Tüm Gen Dizi Analizi)	RNF146 gene (Sequence Analysis-All Coding Exons)	RNF146	RNF146	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF141 geni (Tüm Gen Dizi Analizi)	RNF141 gene (Sequence Analysis-All Coding Exons)	RNF141	RNF141, ZNF230	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF14 geni (Tüm Gen Dizi Analizi)	RNF14 gene (Sequence Analysis-All Coding Exons)	RNF14	RNF14, ARA54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF138 geni (Tüm Gen Dizi Analizi)	RNF138 gene (Sequence Analysis-All Coding Exons)	RNF138	RNF138, NARF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF128 geni (Tüm Gen Dizi Analizi)	RNF128 gene (Sequence Analysis-All Coding Exons)	RNF128	RNF128, GRAIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF126 geni (Tüm Gen Dizi Analizi)	RNF126 gene (Sequence Analysis-All Coding Exons)	RNF126	RNF126	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF125 geni (Tüm Gen Dizi Analizi)	RNF125 gene (Sequence Analysis-All Coding Exons)	RNF125	RNF125, TRAC1, TNORS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF123 geni (Tüm Gen Dizi Analizi)	RNF123 gene (Sequence Analysis-All Coding Exons)	RNF123	RNF123, KPC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF114 geni (Tüm Gen Dizi Analizi)	RNF114 gene (Sequence Analysis-All Coding Exons)	RNF114	RNF114, ZNF313	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF112 geni (Tüm Gen Dizi Analizi)	RNF112 gene (Sequence Analysis-All Coding Exons)	RNF112	ZNF179, BFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF111 geni (Tüm Gen Dizi Analizi)	RNF111 gene (Sequence Analysis-All Coding Exons)	RNF111	RNF111	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF11 geni (Tüm Gen Dizi Analizi)	RNF11 gene (Sequence Analysis-All Coding Exons)	RNF11	RNF11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF103 geni (Tüm Gen Dizi Analizi)	RNF103 gene (Sequence Analysis-All Coding Exons)	RNF103	ZFP103, KF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF10 geni (Tüm Gen Dizi Analizi)	RNF10 gene (Sequence Analysis-All Coding Exons)	RNF10	RNF10, KIAA0262	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RND3 geni (Tüm Gen Dizi Analizi)	RND3 gene (Sequence Analysis-All Coding Exons)	RND3	RND3, RHOE, ARHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RND2 geni (Tüm Gen Dizi Analizi)	RND2 gene (Sequence Analysis-All Coding Exons)	RND2	RND2, ARHN, RHO7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RND1 geni (Tüm Gen Dizi Analizi)	RND1 gene (Sequence Analysis-All Coding Exons)	RND1	RND1, RHO6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASEK geni (Tüm Gen Dizi Analizi)	RNASEK gene (Sequence Analysis-All Coding Exons)	RNASEK	RNASEK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE9 geni (Tüm Gen Dizi Analizi)	RNASE9 gene (Sequence Analysis-All Coding Exons)	RNASE9	RNAE9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE8 geni (Tüm Gen Dizi Analizi)	RNASE8 gene (Sequence Analysis-All Coding Exons)	RNASE8	RNASE8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

RNASE7 geni (Tüm Gen Dizi Analizi)	RNASE7 gene (Sequence Analysis-All Coding Exons)	RNASE7	RNASE7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE6 geni (Tüm Gen Dizi Analizi)	RNASE6 gene (Sequence Analysis-All Coding Exons)	RNASE6	RNASE6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE4 geni (Tüm Gen Dizi Analizi)	RNASE4 gene (Sequence Analysis-All Coding Exons)	RNASE4	RNASE4, RNS4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE3 geni (Tüm Gen Dizi Analizi)	RNASE3 gene (Sequence Analysis-All Coding Exons)	RNASE3	RNASE3, RNS3, ECP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE2 geni (Tüm Gen Dizi Analizi)	RNASE2 gene (Sequence Analysis-All Coding Exons)	RNASE2	RNASE2, RNS2, EDN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNASE1 geni (Tüm Gen Dizi Analizi)	RNASE1 gene (Sequence Analysis-All Coding Exons)	RNASE1	RNASE1, RNS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RN7SL2 geni (Tüm Gen Dizi Analizi)	RN7SL2 gene (Sequence Analysis-All Coding Exons)	RN7SL2	RN7SL2, 7L1C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RN7SL1 geni (Tüm Gen Dizi Analizi)	RN7SL1 gene (Sequence Analysis-All Coding Exons)	RN7SL1	RN7SL1, 7SL, 7L1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RN7SK geni (Tüm Gen Dizi Analizi)	RN7SK gene (Sequence Analysis-All Coding Exons)	RN7SK	RN7SK, 7SK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMST geni (Tüm Gen Dizi Analizi)	RMST gene (Sequence Analysis-All Coding Exons)	RMST	RMST, NCRMS, NCRNA00054	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMI2 geni (Tüm Gen Dizi Analizi)	RMI2 gene (Sequence Analysis-All Coding Exons)	RMI2	C16orf75, RMI2, BLAP18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMI1 geni (Tüm Gen Dizi Analizi)	RMI1 gene (Sequence Analysis-All Coding Exons)	RMI1	RMI2, BLAP75, C9orf76	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMDN3 geni (Tüm Gen Dizi Analizi)	RMDN3 gene (Sequence Analysis-All Coding Exons)	RMDN3	FAM82A2, FAM82C, RMD3, PTPIP51	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMDN2 geni (Tüm Gen Dizi Analizi)	RMDN2 gene (Sequence Analysis-All Coding Exons)	RMDN2	FAM82A1, FAM82A, RMD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RMDN1 geni (Tüm Gen Dizi Analizi)	RMDN1 gene (Sequence Analysis-All Coding Exons)	RMDN1	FAM82B, RMD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RLN3 geni (Tüm Gen Dizi Analizi)	RLN3 gene (Sequence Analysis-All Coding Exons)	RLN3	RLN3, H3, RXN3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RLN2 geni (Tüm Gen Dizi Analizi)	RLN2 gene (Sequence Analysis-All Coding Exons)	RLN2	RLN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RLN1 geni (Tüm Gen Dizi Analizi)	RLN1 gene (Sequence Analysis-All Coding Exons)	RLN1	RLN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RLF geni (Tüm Gen Dizi Analizi)	RLF gene (Sequence Analysis-All Coding Exons)	RLF	RLF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIT2 geni (Tüm Gen Dizi Analizi)	RIT2 gene (Sequence Analysis-All Coding Exons)	RIT2	RIT2, RIN, ROC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIPPLY3 geni (Tüm Gen Dizi Analizi)	RIPPLY3 gene (Sequence Analysis-All Coding Exons)	RIPPLY3	DSCR6, RIPPLY3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIPPLY1 geni (Tüm Gen Dizi Analizi)	RIPPLY1 gene (Sequence Analysis-All Coding Exons)	RIPPLY1	RIPPLY1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIPK3 geni (Tüm Gen Dizi Analizi)	RIPK3 gene (Sequence Analysis-All Coding Exons)	RIPK3	RIPK3, RIP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIPK2 geni (Tüm Gen Dizi Analizi)	RIPK2 gene (Sequence Analysis-All Coding Exons)	RIPK2	RIPK2, CARDIAK, RIP2, RICK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIPK1 geni (Tüm Gen Dizi Analizi)	RIPK1 gene (Sequence Analysis-All Coding Exons)	RIPK1	RIPK1, RIP1, RIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIOX2 geni (Tüm Gen Dizi Analizi)	RIOX2 gene (Sequence Analysis-All Coding Exons)	RIOX2	MINA, MINA53, MDIG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIOX1 geni (Tüm Gen Dizi Analizi)	RIOX1 gene (Sequence Analysis-All Coding Exons)	RIOX1	C14orf169, NO66, MAPJD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIOK3 geni (Tüm Gen Dizi Analizi)	RIOK3 gene (Sequence Analysis-All Coding Exons)	RIOK3	SUDD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RINT1 geni (Tüm Gen Dizi Analizi)	RINT1 gene (Sequence Analysis-All Coding Exons)	RINT1	RINT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RING1 geni (Tüm Gen Dizi Analizi)	RING1 gene (Sequence Analysis-All Coding Exons)	RING1	RING1, RNF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMS4 geni (Tüm Gen Dizi Analizi)	RIMS4 gene (Sequence Analysis-All Coding Exons)	RIMS4	RIMS4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMS3 geni (Tüm Gen Dizi Analizi)	RIMS3 gene (Sequence Analysis-All Coding Exons)	RIMS3	RIMS3, NIM3, KIAA0137	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMS2 geni (Tüm Gen Dizi Analizi)	RIMS2 gene (Sequence Analysis-All Coding Exons)	RIMS2	RIMS2, KIAA0751, RIM2, OBOE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMKLB geni (Tüm Gen Dizi Analizi)	RIMKLB gene (Sequence Analysis-All Coding Exons)	RIMKLB	RIMKLB, NAAGS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMBP3C geni (Tüm Gen Dizi Analizi)	RIMBP3C gene (Sequence Analysis-All Coding Exons)	RIMBP3C	RIMBP3C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMBP3B geni (Tüm Gen Dizi Analizi)	RIMBP3B gene (Sequence Analysis-All Coding Exons)	RIMBP3B	RIMBP3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMBP3 geni (Tüm Gen Dizi Analizi)	RIMBP3 gene (Sequence Analysis-All Coding Exons)	RIMBP3	RIMBP3, RIMBP3A, KIAA1668	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIMBP2 geni (Tüm Gen Dizi Analizi)	RIMBP2 gene (Sequence Analysis-All Coding Exons)	RIMBP2	RIMBP2, KIAA0318	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RILPL2 geni (Tüm Gen Dizi Analizi)	RILPL2 gene (Sequence Analysis-All Coding Exons)	RILPL2	RILPL2, RLP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RILPL1 geni (Tüm Gen Dizi Analizi)	RILPL1 gene (Sequence Analysis-All Coding Exons)	RILPL1	RILPL1, RLP1, GOSPEL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RILP geni (Tüm Gen Dizi Analizi)	RILP gene (Sequence Analysis-All Coding Exons)	RILP	RILP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMYD1 geni (Tüm Gen Dizi Analizi)	SMYD1 gene (Sequence Analysis-All Coding Exons)	SMYD1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIDA geni (Tüm Gen Dizi Analizi)	RIDA gene (Sequence Analysis-All Coding Exons)	RIDA	HRSP12, PSP, UK114	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIC8B geni (Tüm Gen Dizi Analizi)	RIC8B gene (Sequence Analysis-All Coding Exons)	RIC8B	RIC8B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIC8A geni (Tüm Gen Dizi Analizi)	RIC8A gene (Sequence Analysis-All Coding Exons)	RIC8A	RIC8A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIC3 geni (Tüm Gen Dizi Analizi)	RIC3 gene (Sequence Analysis-All Coding Exons)	RIC3	RIC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIC1 geni (Tüm Gen Dizi Analizi)	RIC1 gene (Sequence Analysis-All Coding Exons)	RIC1	CIP150, KIAA1432	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHPN1 geni (Tüm Gen Dizi Analizi)	RHPN1 gene (Sequence Analysis-All Coding Exons)	RHPN1	RHPN1, ODF5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOXF2 geni (Tüm Gen Dizi Analizi)	RHOXF2 gene (Sequence Analysis-All Coding Exons)	RHOXF2	RHOXF2, PEPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOXF1P1 geni (Tüm Gen Dizi Analizi)	RHOXF1P1 gene (Sequence Analysis-All Coding Exons)	RHOXF1P1	RHOXF1P1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOXF1 geni (Tüm Gen Dizi Analizi)	RHOXF1 gene (Sequence Analysis-All Coding Exons)	RHOXF1	RHOXF1, OTEX, PEPP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOU geni (Tüm Gen Dizi Analizi)	RHOU gene (Sequence Analysis-All Coding Exons)	RHOU	RHOU, WRCH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMPD3 geni (Tüm Gen Dizi Analizi)	SMPD3 gene (Sequence Analysis-All Coding Exons)	SMPD3		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMPD2 geni (Tüm Gen Dizi Analizi)	SMPD2 gene (Sequence Analysis-All Coding Exons)	SMPD2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOT2 geni (Tüm Gen Dizi Analizi)	RHOT2 gene (Sequence Analysis-All Coding Exons)	RHOT2	RHOT2, MIRO2, ARHT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOT1 geni (Tüm Gen Dizi Analizi)	RHOT1 gene (Sequence Analysis-All Coding Exons)	RHOT1	RHOT1, MIRO1, ARHT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOQ geni (Tüm Gen Dizi Analizi)	RHOQ gene (Sequence Analysis-All Coding Exons)	RHOQ	RHOQ, ARHQ, TC10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOJ geni (Tüm Gen Dizi Analizi)	RHOJ gene (Sequence Analysis-All Coding Exons)	RHOJ	RHOJ, ARHJ, TCL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOH geni (Tüm Gen Dizi Analizi)	RHOH gene (Sequence Analysis-All Coding Exons)	RHOH	ARHH, TTF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOG geni (Tüm Gen Dizi Analizi)	RHOG gene (Sequence Analysis-All Coding Exons)	RHOG	ARHG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOD geni (Tüm Gen Dizi Analizi)	RHOD gene (Sequence Analysis-All Coding Exons)	RHOD	RHOD, ARHD, RHOHP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOC geni (Tüm Gen Dizi Analizi)	RHOC gene (Sequence Analysis-All Coding Exons)	RHOC	RHOC, ARHC, ARH9, RHOH9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOBTB3 geni (Tüm Gen Dizi Analizi)	RHOBTB3 gene (Sequence Analysis-All Coding Exons)	RHOBTB3	RHOBTB3, KIAA0878	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOBTB2 geni (Tüm Gen Dizi Analizi)	RHOBTB2 gene (Sequence Analysis-All Coding Exons)	RHOBTB2	RHOBTB2, DBC2, KIAA0717	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOBTB1 geni (Tüm Gen Dizi Analizi)	RHOBTB1 gene (Sequence Analysis-All Coding Exons)	RHOBTB1	RHOBTB1, KIAA0740	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOB geni (Tüm Gen Dizi Analizi)	RHOB gene (Sequence Analysis-All Coding Exons)	RHOB	RHOB, ARHB, ARH6, RHOH6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHOA geni (Tüm Gen Dizi Analizi)	RHOA gene (Sequence Analysis-All Coding Exons)	RHOA	RHOA, ARHA, ARH12, RHOH12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHNO1 geni (Tüm Gen Dizi Analizi)	RHNO1 gene (Sequence Analysis-All Coding Exons)	RHNO1	RHNO1, RHINO, C12orf32	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHEB geni (Tüm Gen Dizi Analizi)	RHEB gene (Sequence Analysis-All Coding Exons)	RHEB	RHEB, RHEB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHCG geni (Tüm Gen Dizi Analizi)	RHCG gene (Sequence Analysis-All Coding Exons)	RHCG	RHCG, RHGK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHBG geni (Tüm Gen Dizi Analizi)	RHBG gene (Sequence Analysis-All Coding Exons)	RHBG	RHBG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHBDL1 geni (Tüm Gen Dizi Analizi)	RHBDL1 gene (Sequence Analysis-All Coding Exons)	RHBDL1	RHBDL, RRP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHBDF1 geni (Tüm Gen Dizi Analizi)	RHBDF1 gene (Sequence Analysis-All Coding Exons)	RHBDF1	RHBDF1, IRHOM1, DIST1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHBDD2 geni (Tüm Gen Dizi Analizi)	RHBDD2 gene (Sequence Analysis-All Coding Exons)	RHBDD2	RHBDD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGSL1 geni (Tüm Gen Dizi Analizi)	RGSL1 gene (Sequence Analysis-All Coding Exons)	RGSL1	RGSL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS7BP geni (Tüm Gen Dizi Analizi)	RGS7BP gene (Sequence Analysis-All Coding Exons)	RGS7BP	RGS7BP, R7BP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS7 geni (Tüm Gen Dizi Analizi)	RGS7 gene (Sequence Analysis-All Coding Exons)	RGS7	RGS7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS6 geni (Tüm Gen Dizi Analizi)	RGS6 gene (Sequence Analysis-All Coding Exons)	RGS6	RGS6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS4 geni (Tüm Gen Dizi Analizi)	RGS4 gene (Sequence Analysis-All Coding Exons)	RGS4	RGS4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS3 geni (Tüm Gen Dizi Analizi)	RGS3 gene (Sequence Analysis-All Coding Exons)	RGS3	RGS3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS22 geni (Tüm Gen Dizi Analizi)	RGS22 gene (Sequence Analysis-All Coding Exons)	RGS22	RGS22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS21 geni (Tüm Gen Dizi Analizi)	RGS21 gene (Sequence Analysis-All Coding Exons)	RGS21	RGS21	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS2 geni (Tüm Gen Dizi Analizi)	RGS2 gene (Sequence Analysis-All Coding Exons)	RGS2	RGS2, G0S8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS19 geni (Tüm Gen Dizi Analizi)	RGS19 gene (Sequence Analysis-All Coding Exons)	RGS19	RGS19, GAIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS16 geni (Tüm Gen Dizi Analizi)	RGS16 gene (Sequence Analysis-All Coding Exons)	RGS16	RGS16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS14 geni (Tüm Gen Dizi Analizi)	RGS14 gene (Sequence Analysis-All Coding Exons)	RGS14	RGS14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SMC1B geni (Tüm Gen Dizi Analizi)	SMC1B gene (Sequence Analysis-All Coding Exons)	SMC1B		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS12 geni (Tüm Gen Dizi Analizi)	RGS12 gene (Sequence Analysis-All Coding Exons)	RGS12	RGS12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS11 geni (Tüm Gen Dizi Analizi)	RGS11 gene (Sequence Analysis-All Coding Exons)	RGS11	RS11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS1 geni (Tüm Gen Dizi Analizi)	RGS1 gene (Sequence Analysis-All Coding Exons)	RGS1	RGS1, IER1, IR20	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD8 geni (Tüm Gen Dizi Analizi)	RGPD8 gene (Sequence Analysis-All Coding Exons)	RGPD8	RGPD8, RGP8, RANBP2L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD6 geni (Tüm Gen Dizi Analizi)	RGPD6 gene (Sequence Analysis-All Coding Exons)	RGPD6	RGPD6, RGP6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD5 geni (Tüm Gen Dizi Analizi)	RGPD5 gene (Sequence Analysis-All Coding Exons)	RGPD5	RGPD5, RGP5, BS63	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

RGPD4 geni (Tüm Gen Dizi Analizi)	RGPD4 gene (Sequence Analysis-All Coding Exons)	RGPD4	RGPD4, RGP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD3 geni (Tüm Gen Dizi Analizi)	RGPD3 gene (Sequence Analysis-All Coding Exons)	RGPD3	RGPD3, RGP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD2 geni (Tüm Gen Dizi Analizi)	RGPD2 gene (Sequence Analysis-All Coding Exons)	RGPD2	RGPD2, RGP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGPD1 geni (Tüm Gen Dizi Analizi)	RGPD1 gene (Sequence Analysis-All Coding Exons)	RGPD1	RGPD1, RGP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGP1 geni (Tüm Gen Dizi Analizi)	RGP1 gene (Sequence Analysis-All Coding Exons)	RGP1	RGP1, KIAA0258	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGN geni (Tüm Gen Dizi Analizi)	RGN gene (Sequence Analysis-All Coding Exons)	RGN	RGN, SMP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGMB geni (Tüm Gen Dizi Analizi)	RGMB gene (Sequence Analysis-All Coding Exons)	RGMB	RGMB, DRAGON	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGMA geni (Tüm Gen Dizi Analizi)	RGMA gene (Sequence Analysis-All Coding Exons)	RGMA	RGMA, RGMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGL4 geni (Tüm Gen Dizi Analizi)	RGL4 gene (Sequence Analysis-All Coding Exons)	RGL4	RGL4, RGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGL3 geni (Tüm Gen Dizi Analizi)	RGL3 gene (Sequence Analysis-All Coding Exons)	RGL3	RGL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGL2 geni (Tüm Gen Dizi Analizi)	RGL2 gene (Sequence Analysis-All Coding Exons)	RGL2	RAB2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGL1 geni (Tüm Gen Dizi Analizi)	RGL1 gene (Sequence Analysis-All Coding Exons)	RGL1	RGL1, RGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGCC geni (Tüm Gen Dizi Analizi)	RGCC gene (Sequence Analysis-All Coding Exons)	RGCC	RGC32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGAG1 geni (Tüm Gen Dizi Analizi)	RGAG1 gene (Sequence Analysis-All Coding Exons)	RGAG1	RGAG1, MAR9, KIAA1318	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFX7 geni (Tüm Gen Dizi Analizi)	RFX7 gene (Sequence Analysis-All Coding Exons)	RFX7	RFX7, RFXDC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFX3 geni (Tüm Gen Dizi Analizi)	RFX3 gene (Sequence Analysis-All Coding Exons)	RFX3	RFX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFX2 geni (Tüm Gen Dizi Analizi)	RFX2 gene (Sequence Analysis-All Coding Exons)	RFX2	RFX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFX1 geni (Tüm Gen Dizi Analizi)	RFX1 gene (Sequence Analysis-All Coding Exons)	RFX1	RFX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFWD3 geni (Tüm Gen Dizi Analizi)	RFWD3 gene (Sequence Analysis-All Coding Exons)	RFWD3	RFWD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFWD2 geni (Tüm Gen Dizi Analizi)	RFWD2 gene (Sequence Analysis-All Coding Exons)	RFWD2	RFWD2, COP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFPL4A geni (Tüm Gen Dizi Analizi)	RFPL4A gene (Sequence Analysis-All Coding Exons)	RFPL4A	RFPL4A, RNF210	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFPL3 geni (Tüm Gen Dizi Analizi)	RFPL3 gene (Sequence Analysis-All Coding Exons)	RFPL3	RFPL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFPL2 geni (Tüm Gen Dizi Analizi)	RFPL2 gene (Sequence Analysis-All Coding Exons)	RFPL2	RFPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFPL1 geni (Tüm Gen Dizi Analizi)	RFPL1 gene (Sequence Analysis-All Coding Exons)	RFPL1	RFPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFNG geni (Tüm Gen Dizi Analizi)	RFNG gene (Sequence Analysis-All Coding Exons)	RFNG	RFNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFLNB geni (Tüm Gen Dizi Analizi)	RFLNB gene (Sequence Analysis-All Coding Exons)	RFLNB	FAM101B, CFM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFLNA geni (Tüm Gen Dizi Analizi)	RFLNA gene (Sequence Analysis-All Coding Exons)	RFLNA	FAM101A, CFM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFK geni (Tüm Gen Dizi Analizi)	RFK gene (Sequence Analysis-All Coding Exons)	RFK	RFK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFFL geni (Tüm Gen Dizi Analizi)	RFFL gene (Sequence Analysis-All Coding Exons)	RFFL	RFFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFC5 geni (Tüm Gen Dizi Analizi)	RFC5 gene (Sequence Analysis-All Coding Exons)	RFC5	RFC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFC4 geni (Tüm Gen Dizi Analizi)	RFC4 gene (Sequence Analysis-All Coding Exons)	RFC4	RFC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFC3 geni (Tüm Gen Dizi Analizi)	RFC3 gene (Sequence Analysis-All Coding Exons)	RFC3	RFC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLU7 geni (Tüm Gen Dizi Analizi)	SLU7 gene (Sequence Analysis-All Coding Exons)	SLU7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFC2 geni (Tüm Gen Dizi Analizi)	RFC2 gene (Sequence Analysis-All Coding Exons)	RFC2	RFC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFC1 geni (Tüm Gen Dizi Analizi)	RFC1 gene (Sequence Analysis-All Coding Exons)	RFC1	RFC1, RECC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REXO4 geni (Tüm Gen Dizi Analizi)	REXO4 gene (Sequence Analysis-All Coding Exons)	REXO4	REXO4, XPMC2H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REXO2 geni (Tüm Gen Dizi Analizi)	REXO2 gene (Sequence Analysis-All Coding Exons)	REXO2	REXO2, RFN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REXO1 geni (Tüm Gen Dizi Analizi)	REXO1 gene (Sequence Analysis-All Coding Exons)	REXO1	REXO1, REX1, ELOABP1, TCERBBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REV3L geni (Tüm Gen Dizi Analizi)	REV3L gene (Sequence Analysis-All Coding Exons)	REV3L	REV3L, POLZ, REV3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REV1 geni (Tüm Gen Dizi Analizi)	REV1 gene (Sequence Analysis-All Coding Exons)	REV1	REV1L, REV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RETREG3 geni (Tüm Gen Dizi Analizi)	RETREG3 gene (Sequence Analysis-All Coding Exons)	RETREG3	FAM134C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RETNLB geni (Tüm Gen Dizi Analizi)	RETNLB gene (Sequence Analysis-All Coding Exons)	RETNLB	RETNLB, RELMB, FIZZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RESP18 geni (Tüm Gen Dizi Analizi)	RESP18 gene (Sequence Analysis-All Coding Exons)	RESP18	RESP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RERG geni (Tüm Gen Dizi Analizi)	RERG gene (Sequence Analysis-All Coding Exons)	RERG	RERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REPS2 geni (Tüm Gen Dizi Analizi)	REPS2 gene (Sequence Analysis-All Coding Exons)	REPS2	REPS2, POB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REPS1 geni (Tüm Gen Dizi Analizi)	REPS1 gene (Sequence Analysis-All Coding Exons)	REPS1	REPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REP15 geni (Tüm Gen Dizi Analizi)	REP15 gene (Sequence Analysis-All Coding Exons)	REP15	REP15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RENBP geni (Tüm Gen Dizi Analizi)	RENBP gene (Sequence Analysis-All Coding Exons)	RENBP	RENBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REM2 geni (Tüm Gen Dizi Analizi)	REM2 gene (Sequence Analysis-All Coding Exons)	REM2	REM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REM1 geni (Tüm Gen Dizi Analizi)	REM1 gene (Sequence Analysis-All Coding Exons)	REM1	REM1, GES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RELT geni (Tüm Gen Dizi Analizi)	RELT gene (Sequence Analysis-All Coding Exons)	RELT	RELT, TNFRSF19L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RELL2 geni (Tüm Gen Dizi Analizi)	RELL2 gene (Sequence Analysis-All Coding Exons)	RELL2	RELL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RELL1 geni (Tüm Gen Dizi Analizi)	RELL1 gene (Sequence Analysis-All Coding Exons)	RELL1	RELL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RELB geni (Tüm Gen Dizi Analizi)	RELB gene (Sequence Analysis-All Coding Exons)	RELB	RELB, IREL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RELA geni (Tüm Gen Dizi Analizi)	RELA gene (Sequence Analysis-All Coding Exons)	RELA	RELA, NFKB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REL geni (Tüm Gen Dizi Analizi)	REL gene (Sequence Analysis-All Coding Exons)	REL	REL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REG4 geni (Tüm Gen Dizi Analizi)	REG4 gene (Sequence Analysis-All Coding Exons)	REG4	REG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REG3G geni (Tüm Gen Dizi Analizi)	REG3G gene (Sequence Analysis-All Coding Exons)	REG3G	REG3G, PAP1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REG3A geni (Tüm Gen Dizi Analizi)	REG3A gene (Sequence Analysis-All Coding Exons)	REG3A	PAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REG1B geni (Tüm Gen Dizi Analizi)	REG1B gene (Sequence Analysis-All Coding Exons)	REG1B	REGL, PSPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REG1A geni (Tüm Gen Dizi Analizi)	REG1A gene (Sequence Analysis-All Coding Exons)	REG1A	REG1A, PSPS1, REG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REEP5 geni (Tüm Gen Dizi Analizi)	REEP5 gene (Sequence Analysis-All Coding Exons)	REEP5	REEP5, DS3346, DP1, C5orf18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REEP4 geni (Tüm Gen Dizi Analizi)	REEP4 gene (Sequence Analysis-All Coding Exons)	REEP4	REEP4, C8orf20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REEP3 geni (Tüm Gen Dizi Analizi)	REEP3 gene (Sequence Analysis-All Coding Exons)	REEP3	REEP3, C10orf74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RECQL5 geni (Tüm Gen Dizi Analizi)	RECQL5 gene (Sequence Analysis-All Coding Exons)	RECQL5	RECQL5, RECQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RECQL geni (Tüm Gen Dizi Analizi)	RECQL gene (Sequence Analysis-All Coding Exons)	RECQL	RECQL, RECQL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RECK geni (Tüm Gen Dizi Analizi)	RECK gene (Sequence Analysis-All Coding Exons)	RECK	RECK, ST15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
REC8 geni (Tüm Gen Dizi Analizi)	REC8 gene (Sequence Analysis-All Coding Exons)	REC8	REC8L1, REC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RDM1 geni (Tüm Gen Dizi Analizi)	RDM1 gene (Sequence Analysis-All Coding Exons)	RDM1	RDM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RDH8 geni (Tüm Gen Dizi Analizi)	RDH8 gene (Sequence Analysis-All Coding Exons)	RDH8	RDH8, PRRDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RDH14 geni (Tüm Gen Dizi Analizi)	RDH14 gene (Sequence Analysis-All Coding Exons)	RDH14	RDH14, SDR7C4, PAN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RDH10 geni (Tüm Gen Dizi Analizi)	RDH10 gene (Sequence Analysis-All Coding Exons)	RDH10	RDH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCVRN geni (Tüm Gen Dizi Analizi)	RCVRN gene (Sequence Analysis-All Coding Exons)	RCVRN	RCV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCSD1 geni (Tüm Gen Dizi Analizi)	RCSD1 gene (Sequence Analysis-All Coding Exons)	RCSD1	RCSD1, CAPZIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCOR2 geni (Tüm Gen Dizi Analizi)	RCOR2 gene (Sequence Analysis-All Coding Exons)	RCOR2	RCOR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCOR1 geni (Tüm Gen Dizi Analizi)	RCOR1 gene (Sequence Analysis-All Coding Exons)	RCOR1	RCOR, COREST, KIAA0071	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCN2 geni (Tüm Gen Dizi Analizi)	RCN2 gene (Sequence Analysis-All Coding Exons)	RCN2	RCN2, ERC55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCN1 geni (Tüm Gen Dizi Analizi)	RCN1 gene (Sequence Analysis-All Coding Exons)	RCN1	RCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCL1 geni (Tüm Gen Dizi Analizi)	RCL1 gene (Sequence Analysis-All Coding Exons)	RCL1	RCL1, RPCL1, RNAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCHY1 geni (Tüm Gen Dizi Analizi)	RCHY1 gene (Sequence Analysis-All Coding Exons)	RCHY1	RCHY1, ZNF363, PIRH2, ARNIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCE1 geni (Tüm Gen Dizi Analizi)	RCE1 gene (Sequence Analysis-All Coding Exons)	RCE1	RCE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC8A3 geni (Tüm Gen Dizi Analizi)	SLC8A3 gene (Sequence Analysis-All Coding Exons)	SLC8A3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCC1 geni (Tüm Gen Dizi Analizi)	RCC1 gene (Sequence Analysis-All Coding Exons)	RCC1	CHC1, RCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCBTB2 geni (Tüm Gen Dizi Analizi)	RCBTB2 gene (Sequence Analysis-All Coding Exons)	RCBTB2	RCBTB2, CHC1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCAN3 geni (Tüm Gen Dizi Analizi)	RCAN3 gene (Sequence Analysis-All Coding Exons)	RCAN3	RCAN3, DSCR1L2, MCIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCAN2 geni (Tüm Gen Dizi Analizi)	RCAN2 gene (Sequence Analysis-All Coding Exons)	RCAN2	RCAN2, ZAK14, DSCR1L1, MCIP2, CSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCAN1 geni (Tüm Gen Dizi Analizi)	RCAN1 gene (Sequence Analysis-All Coding Exons)	RCAN1	RCAN1, DSCR1, MCIP1, CSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RC3H2 geni (Tüm Gen Dizi Analizi)	RC3H2 gene (Sequence Analysis-All Coding Exons)	RC3H2	RC3H2, MNAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RC3H1 geni (Tüm Gen Dizi Analizi)	RC3H1 gene (Sequence Analysis-All Coding Exons)	RC3H1	RC3H1, KIAA2025	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBX1 geni (Tüm Gen Dizi Analizi)	RBX1 gene (Sequence Analysis-All Coding Exons)	RBX1	RBX1, ROC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBSN geni (Tüm Gen Dizi Analizi)	RBSN gene (Sequence Analysis-All Coding Exons)	RBSN	RBSN, ZFYVE20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBPMS geni (Tüm Gen Dizi Analizi)	RBPMS gene (Sequence Analysis-All Coding Exons)	RBPMS	RBPMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBPJL geni (Tüm Gen Dizi Analizi)	RBPJL gene (Sequence Analysis-All Coding Exons)	RBPJL	RBPJL, RBPL, SUHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

RBP5 geni (Tüm Gen Dizi Analizi)	RBP5 gene (Sequence Analysis-All Coding Exons)	RBP5	RBP5, CRBP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBP2 geni (Tüm Gen Dizi Analizi)	RBP2 gene (Sequence Analysis-All Coding Exons)	RBP2	RBP2, CRBP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBP1 geni (Tüm Gen Dizi Analizi)	RBP1 gene (Sequence Analysis-All Coding Exons)	RBP1	RBP1, CRBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBMY1A1 geni (Tüm Gen Dizi Analizi)	RBMY1A1 gene (Sequence Analysis-All Coding Exons)	RBMY1A1	RBMY1A1, RBM1, YRRM1, RBM7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBMXL2 geni (Tüm Gen Dizi Analizi)	RBMXL2 gene (Sequence Analysis-All Coding Exons)	RBMXL2	RBMXL2, HNRNPGT, HNRPGT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBMS3 geni (Tüm Gen Dizi Analizi)	RBMS3 gene (Sequence Analysis-All Coding Exons)	RBMS3	RBMS3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBMS2 geni (Tüm Gen Dizi Analizi)	RBMS2 gene (Sequence Analysis-All Coding Exons)	RBMS2	RBMS2, SCR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBMS1 geni (Tüm Gen Dizi Analizi)	RBMS1 gene (Sequence Analysis-All Coding Exons)	RBMS1	RBMS1, MSSP, SCR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM7 geni (Tüm Gen Dizi Analizi)	RBM7 gene (Sequence Analysis-All Coding Exons)	RBM7	RBM7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM6 geni (Tüm Gen Dizi Analizi)	RBM6 gene (Sequence Analysis-All Coding Exons)	RBM6	RBM6, DEF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM5 geni (Tüm Gen Dizi Analizi)	RBM5 gene (Sequence Analysis-All Coding Exons)	RBM5	RMB5, LUCA15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM45 geni (Tüm Gen Dizi Analizi)	RBM45 gene (Sequence Analysis-All Coding Exons)	RBM45	RBM45	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM42 geni (Tüm Gen Dizi Analizi)	RBM42 gene (Sequence Analysis-All Coding Exons)	RBM42	RBM42	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM4 geni (Tüm Gen Dizi Analizi)	RBM4 gene (Sequence Analysis-All Coding Exons)	RBM4	RBM4, LARK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM39 geni (Tüm Gen Dizi Analizi)	RBM39 gene (Sequence Analysis-All Coding Exons)	RBM39	RBM39, CAPER, RNPC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM38 geni (Tüm Gen Dizi Analizi)	RBM38 gene (Sequence Analysis-All Coding Exons)	RBM38	RBM38, RNPC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM3 geni (Tüm Gen Dizi Analizi)	RBM3 gene (Sequence Analysis-All Coding Exons)	RBM3	RBM3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM25 geni (Tüm Gen Dizi Analizi)	RBM25 gene (Sequence Analysis-All Coding Exons)	RBM25	RBM25, RED120	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM22 geni (Tüm Gen Dizi Analizi)	RBM22 gene (Sequence Analysis-All Coding Exons)	RBM22	RBM22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM19 geni (Tüm Gen Dizi Analizi)	RBM19 gene (Sequence Analysis-All Coding Exons)	RBM19	RBM19, KIAA0682	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM17 geni (Tüm Gen Dizi Analizi)	RBM17 gene (Sequence Analysis-All Coding Exons)	RBM17	RBM17, SPF45	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM15B geni (Tüm Gen Dizi Analizi)	RBM15B gene (Sequence Analysis-All Coding Exons)	RBM15B	RBM15B, OTT3, HUMAGCGB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM14 geni (Tüm Gen Dizi Analizi)	RBM14 gene (Sequence Analysis-All Coding Exons)	RBM14	RBM14, PSP2, COAA, SIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBL2 geni (Tüm Gen Dizi Analizi)	RBL2 gene (Sequence Analysis-All Coding Exons)	RBL2	RBL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBL1 geni (Tüm Gen Dizi Analizi)	RBL1 gene (Sequence Analysis-All Coding Exons)	RBL1	RBL1, CP107	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBKS geni (Tüm Gen Dizi Analizi)	RBKS gene (Sequence Analysis-All Coding Exons)	RBKS	RBKS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBFOX3 geni (Tüm Gen Dizi Analizi)	RBFOX3 gene (Sequence Analysis-All Coding Exons)	RBFOX3	RBFOX3, FOX3, NEUN, HRNBP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBFOX2 geni (Tüm Gen Dizi Analizi)	RBFOX2 gene (Sequence Analysis-All Coding Exons)	RBFOX2	RBFOX2, RBM9, FOX2, RTA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBFOX1 geni (Tüm Gen Dizi Analizi)	RBFOX1 gene (Sequence Analysis-All Coding Exons)	RBFOX1	RBFOX1, 2BP1, FOX1, HRNBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBBP9 geni (Tüm Gen Dizi Analizi)	RBBP9 gene (Sequence Analysis-All Coding Exons)	RBBP9	RBBP9, BOG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBBP7 geni (Tüm Gen Dizi Analizi)	RBBP7 gene (Sequence Analysis-All Coding Exons)	RBBP7	RBBP7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBBP6 geni (Tüm Gen Dizi Analizi)	RBBP6 gene (Sequence Analysis-All Coding Exons)	RBBP6	RBBP6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBBP5 geni (Tüm Gen Dizi Analizi)	RBBP5 gene (Sequence Analysis-All Coding Exons)	RBBP5	RBBP5, RBQ3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBAK geni (Tüm Gen Dizi Analizi)	RBAK gene (Sequence Analysis-All Coding Exons)	RBAK	RBAK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAVER2 geni (Tüm Gen Dizi Analizi)	RAVER2 gene (Sequence Analysis-All Coding Exons)	RAVER2	RAVER2, KIAA1579	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAVER1 geni (Tüm Gen Dizi Analizi)	RAVER1 gene (Sequence Analysis-All Coding Exons)	RAVER1	RAVER1, KIAA1978	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF9 geni (Tüm Gen Dizi Analizi)	RASSF9 gene (Sequence Analysis-All Coding Exons)	RASSF9	RASSF9, PAMCI, PCIP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF8 geni (Tüm Gen Dizi Analizi)	RASSF8 gene (Sequence Analysis-All Coding Exons)	RASSF8	RASSF8, C12orf2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF7 geni (Tüm Gen Dizi Analizi)	RASSF7 gene (Sequence Analysis-All Coding Exons)	RASSF7	RASSF7, C11orf13, HIC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF6 geni (Tüm Gen Dizi Analizi)	RASSF6 gene (Sequence Analysis-All Coding Exons)	RASSF6	RASSF6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF5 geni (Tüm Gen Dizi Analizi)	RASSF5 gene (Sequence Analysis-All Coding Exons)	RASSF5	RASSF5, NORE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF4 geni (Tüm Gen Dizi Analizi)	RASSF4 gene (Sequence Analysis-All Coding Exons)	RASSF4	RASSF4, AD037	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF3 geni (Tüm Gen Dizi Analizi)	RASSF3 gene (Sequence Analysis-All Coding Exons)	RASSF3	RASSF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF10 geni (Tüm Gen Dizi Analizi)	RASSF10 gene (Sequence Analysis-All Coding Exons)	RASSF10	RASSF10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASL11B geni (Tüm Gen Dizi Analizi)	RASL11B gene (Sequence Analysis-All Coding Exons)	RASL11B	RASL11B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC4A9 geni (Tüm Gen Dizi Analizi)	SLC4A9 gene (Sequence Analysis-All Coding Exons)	SLC4A9		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASL11A geni (Tüm Gen Dizi Analizi)	RASL11A gene (Sequence Analysis-All Coding Exons)	RASL11A	RASL11A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASL10B geni (Tüm Gen Dizi Analizi)	RASL10B gene (Sequence Analysis-All Coding Exons)	RASL10B	RASL10B, RRP17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASL10A geni (Tüm Gen Dizi Analizi)	RASL10A gene (Sequence Analysis-All Coding Exons)	RASL10A	RASL10A, RRP22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASIP1 geni (Tüm Gen Dizi Analizi)	RASIP1 gene (Sequence Analysis-All Coding Exons)	RASIP1	RASIP1, RAIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGRP4 geni (Tüm Gen Dizi Analizi)	RASGRP4 gene (Sequence Analysis-All Coding Exons)	RASGRP4	RASGRP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGRP1 geni (Tüm Gen Dizi Analizi)	RASGRP1 gene (Sequence Analysis-All Coding Exons)	RASGRP1	RASGRP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGRF2 geni (Tüm Gen Dizi Analizi)	RASGRF2 gene (Sequence Analysis-All Coding Exons)	RASGRF2	RASGRF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGEF1B geni (Tüm Gen Dizi Analizi)	RASGEF1B gene (Sequence Analysis-All Coding Exons)	RASGEF1B	RASGEF1B, GPIG4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGEF1A geni (Tüm Gen Dizi Analizi)	RASGEF1A gene (Sequence Analysis-All Coding Exons)	RASGEF1A	RASFEF1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASEF geni (Tüm Gen Dizi Analizi)	RASEF gene (Sequence Analysis-All Coding Exons)	RASEF	RASEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASD2 geni (Tüm Gen Dizi Analizi)	RASD2 gene (Sequence Analysis-All Coding Exons)	RASD2	RASD2, RHES, TEM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASD1 geni (Tüm Gen Dizi Analizi)	RASD1 gene (Sequence Analysis-All Coding Exons)	RASD1	RASD1, DEXRAS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASAL3 geni (Tüm Gen Dizi Analizi)	RASAL3 gene (Sequence Analysis-All Coding Exons)	RASAL3	RASAL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASAL2 geni (Tüm Gen Dizi Analizi)	RASAL2 gene (Sequence Analysis-All Coding Exons)	RASAL2	RASAL2, NGAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASAL1 geni (Tüm Gen Dizi Analizi)	RASAL1 gene (Sequence Analysis-All Coding Exons)	RASAL1	RASAL1, RASAL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASA4 geni (Tüm Gen Dizi Analizi)	RASA4 gene (Sequence Analysis-All Coding Exons)	RASA4	RASA4, CAPRI, GAPL, KIAA0538	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASA3 geni (Tüm Gen Dizi Analizi)	RASA3 gene (Sequence Analysis-All Coding Exons)	RASA3	RASA3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASA2 geni (Tüm Gen Dizi Analizi)	RASA2 gene (Sequence Analysis-All Coding Exons)	RASA2	RASA2, GAP1M	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RARRES3 geni (Tüm Gen Dizi Analizi)	RARRES3 gene (Sequence Analysis-All Coding Exons)	RARRES3	RARRES3, TIG3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RARRES2 geni (Tüm Gen Dizi Analizi)	RARRES2 gene (Sequence Analysis-All Coding Exons)	RARRES2	RARRES2, TIG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RARRES1 geni (Tüm Gen Dizi Analizi)	RARRES1 gene (Sequence Analysis-All Coding Exons)	RARRES1	RARRES1, TIG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RARG geni (Tüm Gen Dizi Analizi)	RARG gene (Sequence Analysis-All Coding Exons)	RARG	RARG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF6 geni (Tüm Gen Dizi Analizi)	RAPGEF6 gene (Sequence Analysis-All Coding Exons)	RAPGEF6	RAPGEF6, RAGEF2, PDZGEF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF3 geni (Tüm Gen Dizi Analizi)	RAPGEF3 gene (Sequence Analysis-All Coding Exons)	RAPGEF3	EPAC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF1 geni (Tüm Gen Dizi Analizi)	RAPGEF1 gene (Sequence Analysis-All Coding Exons)	RAPGEF1	RAPGEF1, GRF2, C3G	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAP2B geni (Tüm Gen Dizi Analizi)	RAP2B gene (Sequence Analysis-All Coding Exons)	RAP2B	RAP2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAP2A geni (Tüm Gen Dizi Analizi)	RAP2A gene (Sequence Analysis-All Coding Exons)	RAP2A	RAP2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAP1GAP geni (Tüm Gen Dizi Analizi)	RAP1GAP gene (Sequence Analysis-All Coding Exons)	RAP1GAP	RAP1GA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAP1B geni (Tüm Gen Dizi Analizi)	RAP1B gene (Sequence Analysis-All Coding Exons)	RAP1B	RAP1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAP1A geni (Tüm Gen Dizi Analizi)	RAP1A gene (Sequence Analysis-All Coding Exons)	RAP1A	RAP1A, KREV1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANGRF geni (Tüm Gen Dizi Analizi)	RANGRF gene (Sequence Analysis-All Coding Exons)	RANGRF	RANGRF, MOG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP9 geni (Tüm Gen Dizi Analizi)	RANBP9 gene (Sequence Analysis-All Coding Exons)	RANBP9	RANBP9, RANBPM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP3L geni (Tüm Gen Dizi Analizi)	RANBP3L gene (Sequence Analysis-All Coding Exons)	RANBP3L	RANBP3L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP3 geni (Tüm Gen Dizi Analizi)	RANBP3 gene (Sequence Analysis-All Coding Exons)	RANBP3	RANBP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP17 geni (Tüm Gen Dizi Analizi)	RANBP17 gene (Sequence Analysis-All Coding Exons)	RANBP17	RANBP17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP10 geni (Tüm Gen Dizi Analizi)	RANBP10 gene (Sequence Analysis-All Coding Exons)	RANBP10	RANBP10, KIAA1464	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANBP1 geni (Tüm Gen Dizi Analizi)	RANBP1 gene (Sequence Analysis-All Coding Exons)	RANBP1	RANBP1, HTF9A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAN geni (Tüm Gen Dizi Analizi)	RAN gene (Sequence Analysis-All Coding Exons)	RAN	RAN, ARA24	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAMP3 geni (Tüm Gen Dizi Analizi)	RAMP3 gene (Sequence Analysis-All Coding Exons)	RAMP3	RAMP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAMP2 geni (Tüm Gen Dizi Analizi)	RAMP2 gene (Sequence Analysis-All Coding Exons)	RAMP2	RAMP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAMP1 geni (Tüm Gen Dizi Analizi)	RAMP1 gene (Sequence Analysis-All Coding Exons)	RAMP1	RAMP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALYL geni (Tüm Gen Dizi Analizi)	RALYL gene (Sequence Analysis-All Coding Exons)	RALYL	RALYL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALGPS1 geni (Tüm Gen Dizi Analizi)	RALGPS1 gene (Sequence Analysis-All Coding Exons)	RALGPS1	RALGPS1, RALGEF2, KIAA0351	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALGDS geni (Tüm Gen Dizi Analizi)	RALGDS gene (Sequence Analysis-All Coding Exons)	RALGDS	RALGDS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALGAPA1 geni (Tüm Gen Dizi Analizi)	RALGAPA1 gene (Sequence Analysis-All Coding Exons)	RALGAPA1	RALGAPA1, GARNL1, TULIP1. GRIPE. KIAA0884	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALBP1 geni (Tüm Gen Dizi Analizi)	RALBP1 gene (Sequence Analysis-All Coding Exons)	RALBP1	RALBP1, RLIP76	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALB geni (Tüm Gen Dizi Analizi)	RALB gene (Sequence Analysis-All Coding Exons)	RALB	RALB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SLC39A6 geni (Tüm Gen Dizi Analizi)	SLC39A6 gene (Sequence Analysis-All Coding Exons)	SLC39A6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALA geni (Tüm Gen Dizi Analizi)	RALA gene (Sequence Analysis-All Coding Exons)	RALA	RALA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAI2 geni (Tüm Gen Dizi Analizi)	RAI2 gene (Sequence Analysis-All Coding Exons)	RAI2	RAI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAI14 geni (Tüm Gen Dizi Analizi)	RAI14 gene (Sequence Analysis-All Coding Exons)	RAI14	RAI14, KIAA1334	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAET1L geni (Tüm Gen Dizi Analizi)	RAET1L gene (Sequence Analysis-All Coding Exons)	RAET1L	RAET1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RADIL geni (Tüm Gen Dizi Analizi)	RADIL gene (Sequence Analysis-All Coding Exons)	RADIL	RADIL, KIAA1849	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD9B geni (Tüm Gen Dizi Analizi)	RAD9B gene (Sequence Analysis-All Coding Exons)	RAD9B	RAD9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A12 geni (Tüm Gen Dizi Analizi)	SLC39A12 gene (Sequence Analysis-All Coding Exons)	SLC39A12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD9A geni (Tüm Gen Dizi Analizi)	RAD9A gene (Sequence Analysis-All Coding Exons)	RAD9A	RAD9A, RAD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC39A10 geni (Tüm Gen Dizi Analizi)	SLC39A10 gene (Sequence Analysis-All Coding Exons)	SLC39A10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD52 geni (Tüm Gen Dizi Analizi)	RAD52 gene (Sequence Analysis-All Coding Exons)	RAD52	RAD52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD51B geni (Tüm Gen Dizi Analizi)	RAD51B gene (Sequence Analysis-All Coding Exons)	RAD51B	RAD51L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD51AP1 geni (Tüm Gen Dizi Analizi)	RAD51AP1 gene (Sequence Analysis-All Coding Exons)	RAD51AP1	RAD51AP1, PIR51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD23B geni (Tüm Gen Dizi Analizi)	RAD23B gene (Sequence Analysis-All Coding Exons)	RAD23B	RAD23B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD23A geni (Tüm Gen Dizi Analizi)	RAD23A gene (Sequence Analysis-All Coding Exons)	RAD23A	RAD23A, HHR23A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD18 geni (Tüm Gen Dizi Analizi)	RAD18 gene (Sequence Analysis-All Coding Exons)	RAD18	RAD18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD17 geni (Tüm Gen Dizi Analizi)	RAD17 gene (Sequence Analysis-All Coding Exons)	RAD17	RAD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAD1 geni (Tüm Gen Dizi Analizi)	RAD1 gene (Sequence Analysis-All Coding Exons)	RAD1	RAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RACK1 geni (Tüm Gen Dizi Analizi)	RACK1 gene (Sequence Analysis-All Coding Exons)	RACK1	GNB2L1, RACK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RACGAP1 geni (Tüm Gen Dizi Analizi)	RACGAP1 gene (Sequence Analysis-All Coding Exons)	RACGAP1	RCGAP1, MGC8/RACGAP, CYK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAC3 geni (Tüm Gen Dizi Analizi)	RAC3 gene (Sequence Analysis-All Coding Exons)	RAC3	RAC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABL6 geni (Tüm Gen Dizi Analizi)	RABL6 gene (Sequence Analysis-All Coding Exons)	RABL6	PARF, C9orf86	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABL2B geni (Tüm Gen Dizi Analizi)	RABL2B gene (Sequence Analysis-All Coding Exons)	RABL2B	RABL2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABL2A geni (Tüm Gen Dizi Analizi)	RABL2A gene (Sequence Analysis-All Coding Exons)	RABL2A	RABL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABIF geni (Tüm Gen Dizi Analizi)	RABIF gene (Sequence Analysis-All Coding Exons)	RABIF	RABIF, RASGFR3, MSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABGGTB geni (Tüm Gen Dizi Analizi)	RABGGTB gene (Sequence Analysis-All Coding Exons)	RABGGTB	RABGGTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC36A3 geni (Tüm Gen Dizi Analizi)	SLC36A3 gene (Sequence Analysis-All Coding Exons)	SLC36A3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABGGTA geni (Tüm Gen Dizi Analizi)	RABGGTA gene (Sequence Analysis-All Coding Exons)	RABGGTA	RABGGTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABGEF1 geni (Tüm Gen Dizi Analizi)	RABGEF1 gene (Sequence Analysis-All Coding Exons)	RABGEF1	RABGEF1, RABEX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABGAP1 geni (Tüm Gen Dizi Analizi)	RABGAP1 gene (Sequence Analysis-All Coding Exons)	RABGAP1	RABGAP1, GAPCENA, TBC1D11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABEP2 geni (Tüm Gen Dizi Analizi)	RABEP2 gene (Sequence Analysis-All Coding Exons)	RABEP2	RABEP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABEP1 geni (Tüm Gen Dizi Analizi)	RABEP1 gene (Sequence Analysis-All Coding Exons)	RABEP1	RABEP1, RABPT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABAC1 geni (Tüm Gen Dizi Analizi)	RABAC1 gene (Sequence Analysis-All Coding Exons)	RABAC1	PRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB9B geni (Tüm Gen Dizi Analizi)	RAB9B gene (Sequence Analysis-All Coding Exons)	RAB9B	RAB9B, RAB9L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB9A geni (Tüm Gen Dizi Analizi)	RAB9A gene (Sequence Analysis-All Coding Exons)	RAB9A	RAB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB8B geni (Tüm Gen Dizi Analizi)	RAB8B gene (Sequence Analysis-All Coding Exons)	RAB8B	RAB8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB8A geni (Tüm Gen Dizi Analizi)	RAB8A gene (Sequence Analysis-All Coding Exons)	RAB8A	RAB8A, MEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB6C geni (Tüm Gen Dizi Analizi)	RAB6C gene (Sequence Analysis-All Coding Exons)	RAB6C	RAB6C, WTH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB6B geni (Tüm Gen Dizi Analizi)	RAB6B gene (Sequence Analysis-All Coding Exons)	RAB6B	RAB6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB6A geni (Tüm Gen Dizi Analizi)	RAB6A gene (Sequence Analysis-All Coding Exons)	RAB6A	RAB6A, RAB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB5C geni (Tüm Gen Dizi Analizi)	RAB5C gene (Sequence Analysis-All Coding Exons)	RAB5C	RAB5C, RABL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB5B geni (Tüm Gen Dizi Analizi)	RAB5B gene (Sequence Analysis-All Coding Exons)	RAB5B	RAB5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB5A geni (Tüm Gen Dizi Analizi)	RAB5A gene (Sequence Analysis-All Coding Exons)	RAB5A	RAB5A, RAB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB4B geni (Tüm Gen Dizi Analizi)	RAB4B gene (Sequence Analysis-All Coding Exons)	RAB4B	RAB4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB4A geni (Tüm Gen Dizi Analizi)	RAB4A gene (Sequence Analysis-All Coding Exons)	RAB4A	RAB4A, RAB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB40AL geni (Tüm Gen Dizi Analizi)	RAB40AL gene (Sequence Analysis-All Coding Exons)	RAB40AL	RAB40AL, RLGP, MRXSMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB3D geni (Tüm Gen Dizi Analizi)	RAB3D gene (Sequence Analysis-All Coding Exons)	RAB3D	RAD3D, GOV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB3C geni (Tüm Gen Dizi Analizi)	RAB3C gene (Sequence Analysis-All Coding Exons)	RAB3C	RAB3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB3B geni (Tüm Gen Dizi Analizi)	RAB3B gene (Sequence Analysis-All Coding Exons)	RAB3B	RAB3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB3A geni (Tüm Gen Dizi Analizi)	RAB3A gene (Sequence Analysis-All Coding Exons)	RAB3A	RAB3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB38 geni (Tüm Gen Dizi Analizi)	RAB38 gene (Sequence Analysis-All Coding Exons)	RAB38	RAB38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB37 geni (Tüm Gen Dizi Analizi)	RAB37 gene (Sequence Analysis-All Coding Exons)	RAB37	RAB37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB36 geni (Tüm Gen Dizi Analizi)	RAB36 gene (Sequence Analysis-All Coding Exons)	RAB36	RAB36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB35 geni (Tüm Gen Dizi Analizi)	RAB35 gene (Sequence Analysis-All Coding Exons)	RAB35	RAB35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB34 geni (Tüm Gen Dizi Analizi)	RAB34 gene (Sequence Analysis-All Coding Exons)	RAB34	RAB34, RAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB33A geni (Tüm Gen Dizi Analizi)	RAB33A gene (Sequence Analysis-All Coding Exons)	RAB33A	RAB33A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB32 geni (Tüm Gen Dizi Analizi)	RAB32 gene (Sequence Analysis-All Coding Exons)	RAB32	RAB32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB31 geni (Tüm Gen Dizi Analizi)	RAB31 gene (Sequence Analysis-All Coding Exons)	RAB31	RAB31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB30 geni (Tüm Gen Dizi Analizi)	RAB30 gene (Sequence Analysis-All Coding Exons)	RAB30	RAB30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB2B geni (Tüm Gen Dizi Analizi)	RAB2B gene (Sequence Analysis-All Coding Exons)	RAB2B	RAB2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB2A geni (Tüm Gen Dizi Analizi)	RAB2A gene (Sequence Analysis-All Coding Exons)	RAB2A	RAB2, RAB2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB29 geni (Tüm Gen Dizi Analizi)	RAB29 gene (Sequence Analysis-All Coding Exons)	RAB29	RAB7L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB26 geni (Tüm Gen Dizi Analizi)	RAB26 gene (Sequence Analysis-All Coding Exons)	RAB26	RAB26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB25 geni (Tüm Gen Dizi Analizi)	RAB25 gene (Sequence Analysis-All Coding Exons)	RAB25	RAB25, RAB11C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB24 geni (Tüm Gen Dizi Analizi)	RAB24 gene (Sequence Analysis-All Coding Exons)	RAB24	RAB24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB22A geni (Tüm Gen Dizi Analizi)	RAB22A gene (Sequence Analysis-All Coding Exons)	RAB22A	RAB22A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB21 geni (Tüm Gen Dizi Analizi)	RAB21 gene (Sequence Analysis-All Coding Exons)	RAB21	RAB21, KIAA0118	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB1B geni (Tüm Gen Dizi Analizi)	RAB1B gene (Sequence Analysis-All Coding Exons)	RAB1B	RAB1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB1A geni (Tüm Gen Dizi Analizi)	RAB1A gene (Sequence Analysis-All Coding Exons)	RAB1A	RAB1, RAB1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC24ARG geni (Tüm Gen Dizi Analizi)	SLC24ARG gene (Sequence Analysis-All Coding Exons)	SLC24ARG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB17 geni (Tüm Gen Dizi Analizi)	RAB17 gene (Sequence Analysis-All Coding Exons)	RAB17	RAB17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB14 geni (Tüm Gen Dizi Analizi)	RAB14 gene (Sequence Analysis-All Coding Exons)	RAB14	RAB14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB13 geni (Tüm Gen Dizi Analizi)	RAB13 gene (Sequence Analysis-All Coding Exons)	RAB13	RAB13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB12 geni (Tüm Gen Dizi Analizi)	RAB12 gene (Sequence Analysis-All Coding Exons)	RAB12	RAB12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11FIP5 geni (Tüm Gen Dizi Analizi)	RAB11FIP5 gene (Sequence Analysis-All Coding Exons)	RAB11FIP5	RAB11RIP5, RIP11, KIAA0857	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11FIP4 geni (Tüm Gen Dizi Analizi)	RAB11FIP4 gene (Sequence Analysis-All Coding Exons)	RAB11FIP4	RAB11FIP4, KIAA1821	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11FIP3 geni (Tüm Gen Dizi Analizi)	RAB11FIP3 gene (Sequence Analysis-All Coding Exons)	RAB11FIP3	RAB11FIP3, EFERIN, KIAA0865	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11FIP2 geni (Tüm Gen Dizi Analizi)	RAB11FIP2 gene (Sequence Analysis-All Coding Exons)	RAB11FIP2	RAB11FIP2, KIAA0941	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11FIP1 geni (Tüm Gen Dizi Analizi)	RAB11FIP1 gene (Sequence Analysis-All Coding Exons)	RAB11FIP1	RAB11FIP1, RCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11B geni (Tüm Gen Dizi Analizi)	RAB11B gene (Sequence Analysis-All Coding Exons)	RAB11B	RAB11B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB11A geni (Tüm Gen Dizi Analizi)	RAB11A gene (Sequence Analysis-All Coding Exons)	RAB11A	RAB11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB10 geni (Tüm Gen Dizi Analizi)	RAB10 gene (Sequence Analysis-All Coding Exons)	RAB10	RAB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QTRT1 geni (Tüm Gen Dizi Analizi)	QTRT1 gene (Sequence Analysis-All Coding Exons)	QTRT1	QTRT1, TGT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QSOX2 geni (Tüm Gen Dizi Analizi)	QSOX2 gene (Sequence Analysis-All Coding Exons)	QSOX2	QSOX2, SOXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QSOX1 geni (Tüm Gen Dizi Analizi)	QSOX1 gene (Sequence Analysis-All Coding Exons)	QSOX1	QSOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QRSL1 geni (Tüm Gen Dizi Analizi)	QRSL1 gene (Sequence Analysis-All Coding Exons)	QRSL1	QRSL1, GATA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QRICH1 geni (Tüm Gen Dizi Analizi)	QRICH1 gene (Sequence Analysis-All Coding Exons)	QRICH1	QRICH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QRFP geni (Tüm Gen Dizi Analizi)	QRFP gene (Sequence Analysis-All Coding Exons)	QRFP	QRFP, P518, 26RFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QPRT geni (Tüm Gen Dizi Analizi)	QPRT gene (Sequence Analysis-All Coding Exons)	QPRT	QPRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QPCT geni (Tüm Gen Dizi Analizi)	QPCT gene (Sequence Analysis-All Coding Exons)	QPCT	QPCT, QC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QKI geni (Tüm Gen Dizi Analizi)	QKI gene (Sequence Analysis-All Coding Exons)	QKI	QKI, QK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PZP geni (Tüm Gen Dizi Analizi)	PZP gene (Sequence Analysis-All Coding Exons)	PZP	PZP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC27A6 geni (Tüm Gen Dizi Analizi)	SLC27A6 gene (Sequence Analysis-All Coding Exons)	SLC27A6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PYY2 geni (Tüm Gen Dizi Analizi)	PYY2 gene (Sequence Analysis-All Coding Exons)	PYY2	PYY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYY geni (Tüm Gen Dizi Analizi)	PYY gene (Sequence Analysis-All Coding Exons)	PYY	PYY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYHIN1 geni (Tüm Gen Dizi Analizi)	PYHIN1 gene (Sequence Analysis-All Coding Exons)	PYHIN1	PYHIN1, IFIX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYGO2 geni (Tüm Gen Dizi Analizi)	PYGO2 gene (Sequence Analysis-All Coding Exons)	PYGO2	PYGO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYGB geni (Tüm Gen Dizi Analizi)	PYGB gene (Sequence Analysis-All Coding Exons)	PYGB	PYGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYDC1 geni (Tüm Gen Dizi Analizi)	PYDC1 gene (Sequence Analysis-All Coding Exons)	PYDC1	PYDC1, POP1, PYC1, ASC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYCRL geni (Tüm Gen Dizi Analizi)	PYCRL gene (Sequence Analysis-All Coding Exons)	PYCRL	PYCRL, PYCR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC26A7 geni (Tüm Gen Dizi Analizi)	SLC26A7 gene (Sequence Analysis-All Coding Exons)	SLC26A7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYCARD geni (Tüm Gen Dizi Analizi)	PYCARD gene (Sequence Analysis-All Coding Exons)	PYCARD	PYCARD, TMS1, ASC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PXN geni (Tüm Gen Dizi Analizi)	PXN gene (Sequence Analysis-All Coding Exons)	PXN	PXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PXMP4 geni (Tüm Gen Dizi Analizi)	PXMP4 gene (Sequence Analysis-All Coding Exons)	PXMP4	PXMP4, PMP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PXK geni (Tüm Gen Dizi Analizi)	PXK gene (Sequence Analysis-All Coding Exons)	PXK	PXK, MONAKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PXDNL geni (Tüm Gen Dizi Analizi)	PXDNL gene (Sequence Analysis-All Coding Exons)	PXDNL	PXDNL, PMR1, VPO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PWRN1 geni (Tüm Gen Dizi Analizi)	PWRN1 gene (Sequence Analysis-All Coding Exons)	PWRN1	PWRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PWP2 geni (Tüm Gen Dizi Analizi)	PWP2 gene (Sequence Analysis-All Coding Exons)	PWP2	PWP2H, PWP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PWAR5 geni (Tüm Gen Dizi Analizi)	PWAR5 gene (Sequence Analysis-All Coding Exons)	PWAR5	D15S226E, PAR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PWAR1 geni (Tüm Gen Dizi Analizi)	PWAR1 gene (Sequence Analysis-All Coding Exons)	PWAR1	D15S227E, PAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PVRIG geni (Tüm Gen Dizi Analizi)	PVRIG gene (Sequence Analysis-All Coding Exons)	PVRIG	PVRIG, CD112R, C7orf15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PVR geni (Tüm Gen Dizi Analizi)	PVR gene (Sequence Analysis-All Coding Exons)	PVR	PVR, PVS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PVALB geni (Tüm Gen Dizi Analizi)	PVALB gene (Sequence Analysis-All Coding Exons)	PVALB	PVALB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUS7 geni (Tüm Gen Dizi Analizi)	PUS7 gene (Sequence Analysis-All Coding Exons)	PUS7	PUS7, KIAA1897	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUS10 geni (Tüm Gen Dizi Analizi)	PUS10 gene (Sequence Analysis-All Coding Exons)	PUS10	PUS10, DOBI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PURB geni (Tüm Gen Dizi Analizi)	PURB gene (Sequence Analysis-All Coding Exons)	PURB	PURB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUM3 geni (Tüm Gen Dizi Analizi)	PUM3 gene (Sequence Analysis-All Coding Exons)	PUM3	KIAA0020, HLA-HA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUM2 geni (Tüm Gen Dizi Analizi)	PUM2 gene (Sequence Analysis-All Coding Exons)	PUM2	PUM2, KIAA0235	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUM1 geni (Tüm Gen Dizi Analizi)	PUM1 gene (Sequence Analysis-All Coding Exons)	PUM1	PUM1, KIAA0099	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PUDP geni (Tüm Gen Dizi Analizi)	PUDP gene (Sequence Analysis-All Coding Exons)	PUDP	HDHD1A, DXF68S1E, GS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTX4 geni (Tüm Gen Dizi Analizi)	PTX4 gene (Sequence Analysis-All Coding Exons)	PTX4	PTX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTX3 geni (Tüm Gen Dizi Analizi)	PTX3 gene (Sequence Analysis-All Coding Exons)	PTX3	PTX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTTG2 geni (Tüm Gen Dizi Analizi)	PTTG2 gene (Sequence Analysis-All Coding Exons)	PTTG2	PTTG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTTG1IP geni (Tüm Gen Dizi Analizi)	PTTG1IP gene (Sequence Analysis-All Coding Exons)	PTTG1IP	PTTG1IP, C21orf1, C21orf3, PBF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTTG1 geni (Tüm Gen Dizi Analizi)	PTTG1 gene (Sequence Analysis-All Coding Exons)	PTTG1	PTTG1, EAP1, TUTR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTRHD1 geni (Tüm Gen Dizi Analizi)	PTRHD1 gene (Sequence Analysis-All Coding Exons)	PTRHD1	PRTHD1, C2orf79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRU geni (Tüm Gen Dizi Analizi)	PTPRU gene (Sequence Analysis-All Coding Exons)	PTPRU	PTPRU, GLEPP1, PTPU2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRT geni (Tüm Gen Dizi Analizi)	PTPRT gene (Sequence Analysis-All Coding Exons)	PTPRT	PTPRT, KIAA0283	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRS geni (Tüm Gen Dizi Analizi)	PTPRS gene (Sequence Analysis-All Coding Exons)	PTPRS	PTPRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRR geni (Tüm Gen Dizi Analizi)	PTPRR gene (Sequence Analysis-All Coding Exons)	PTPRR	PTPRR, PCPTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRN2 geni (Tüm Gen Dizi Analizi)	PTPRN2 gene (Sequence Analysis-All Coding Exons)	PTPRN2	PTPRN2, IAR, ICAAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRN geni (Tüm Gen Dizi Analizi)	PTPRN gene (Sequence Analysis-All Coding Exons)	PTPRN	PTPRN, IA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRM geni (Tüm Gen Dizi Analizi)	PTPRM gene (Sequence Analysis-All Coding Exons)	PTPRM	PTPRM, PTPRL1, RPTPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRK geni (Tüm Gen Dizi Analizi)	PTPRK gene (Sequence Analysis-All Coding Exons)	PTPRK	PTPRK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRH geni (Tüm Gen Dizi Analizi)	PTPRH gene (Sequence Analysis-All Coding Exons)	PTPRH	PTPRH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRG geni (Tüm Gen Dizi Analizi)	PTPRG gene (Sequence Analysis-All Coding Exons)	PTPRG	PTPRG, PTPG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRE geni (Tüm Gen Dizi Analizi)	PTPRE gene (Sequence Analysis-All Coding Exons)	PTPRE	PTPRE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRD geni (Tüm Gen Dizi Analizi)	PTPRD gene (Sequence Analysis-All Coding Exons)	PTPRD	PTPRD, PTPD, HPTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRCAP geni (Tüm Gen Dizi Analizi)	PTPRCAP gene (Sequence Analysis-All Coding Exons)	PTPRCAP	PTPRCAP, LPAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRB geni (Tüm Gen Dizi Analizi)	PTPRB gene (Sequence Analysis-All Coding Exons)	PTPRB	PTPRB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPRA geni (Tüm Gen Dizi Analizi)	PTPRA gene (Sequence Analysis-All Coding Exons)	PTPRA	PTPRA, PTPA, PTPRL2, LRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN9 geni (Tüm Gen Dizi Analizi)	PTPN9 gene (Sequence Analysis-All Coding Exons)	PTPN9	PTPN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN7 geni (Tüm Gen Dizi Analizi)	PTPN7 gene (Sequence Analysis-All Coding Exons)	PTPN7	PTPN7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN6 geni (Tüm Gen Dizi Analizi)	PTPN6 gene (Sequence Analysis-All Coding Exons)	PTPN6	PTPN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN5 geni (Tüm Gen Dizi Analizi)	PTPN5 gene (Sequence Analysis-All Coding Exons)	PTPN5	PTPN5, STEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A25 geni (Tüm Gen Dizi Analizi)	SLC25A25 gene (Sequence Analysis-All Coding Exons)	SLC25A25		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A24 geni (Tüm Gen Dizi Analizi)	SLC25A24 gene (Sequence Analysis-All Coding Exons)	SLC25A24		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC25A23 geni (Tüm Gen Dizi Analizi)	SLC25A23 gene (Sequence Analysis-All Coding Exons)	SLC25A23		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN3 geni (Tüm Gen Dizi Analizi)	PTPN3 gene (Sequence Analysis-All Coding Exons)	PTPN3	PTPN3, PTPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN23 geni (Tüm Gen Dizi Analizi)	PTPN23 gene (Sequence Analysis-All Coding Exons)	PTPN23	PTPN23, KIAA1471, HDPTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN21 geni (Tüm Gen Dizi Analizi)	PTPN21 gene (Sequence Analysis-All Coding Exons)	PTPN21	PTPN21, PTPD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN20 geni (Tüm Gen Dizi Analizi)	PTPN20 gene (Sequence Analysis-All Coding Exons)	PTPN20	PTPN20A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN20 geni (Tüm Gen Dizi Analizi)	PTPN20 gene (Sequence Analysis-All Coding Exons)	PTPN20	PTPN20B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN2 geni (Tüm Gen Dizi Analizi)	PTPN2 gene (Sequence Analysis-All Coding Exons)	PTPN2	PTPN2, PTPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN2 geni (Tüm Gen Dizi Analizi)	PTPN2 gene (Sequence Analysis-All Coding Exons)	PTPN2	PTPN2, PTPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN18 geni (Tüm Gen Dizi Analizi)	PTPN18 gene (Sequence Analysis-All Coding Exons)	PTPN18	PTPN18, BDP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN13 geni (Tüm Gen Dizi Analizi)	PTPN13 gene (Sequence Analysis-All Coding Exons)	PTPN13	PTPN13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPA geni (Tüm Gen Dizi Analizi)	PTPA gene (Sequence Analysis-All Coding Exons)	PTPA	PPP2R4, PTPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTP4A3 geni (Tüm Gen Dizi Analizi)	PTP4A3 gene (Sequence Analysis-All Coding Exons)	PTP4A3	PTP4A3, PRL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTP4A2 geni (Tüm Gen Dizi Analizi)	PTP4A2 gene (Sequence Analysis-All Coding Exons)	PTP4A2	PTP4A2, PTP4A, PRL2, HH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTP4A1 geni (Tüm Gen Dizi Analizi)	PTP4A1 gene (Sequence Analysis-All Coding Exons)	PTP4A1	PTP4A1, HH72, PRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTOV1 geni (Tüm Gen Dizi Analizi)	PTOV1 gene (Sequence Analysis-All Coding Exons)	PTOV1	PTOV1, ACID2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTN geni (Tüm Gen Dizi Analizi)	PTN gene (Sequence Analysis-All Coding Exons)	PTN	PTN, NEGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTMS geni (Tüm Gen Dizi Analizi)	PTMS gene (Sequence Analysis-All Coding Exons)	PTMS	PTMS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTMA geni (Tüm Gen Dizi Analizi)	PTMA gene (Sequence Analysis-All Coding Exons)	PTMA	PTMA, TMSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTK7 geni (Tüm Gen Dizi Analizi)	PTK7 gene (Sequence Analysis-All Coding Exons)	PTK7	PTK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTK6 geni (Tüm Gen Dizi Analizi)	PTK6 gene (Sequence Analysis-All Coding Exons)	PTK6	PTK6, BRK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTK2B geni (Tüm Gen Dizi Analizi)	PTK2B gene (Sequence Analysis-All Coding Exons)	PTK2B	PTK2B, FAK2, PYK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTK2 geni (Tüm Gen Dizi Analizi)	PTK2 gene (Sequence Analysis-All Coding Exons)	PTK2	PTK2, FADK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTH2R geni (Tüm Gen Dizi Analizi)	PTH2R gene (Sequence Analysis-All Coding Exons)	PTH2R	PTHR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTH2 geni (Tüm Gen Dizi Analizi)	PTH2 gene (Sequence Analysis-All Coding Exons)	PTH2	TIP39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGS2 geni (Tüm Gen Dizi Analizi)	PTGS2 gene (Sequence Analysis-All Coding Exons)	PTGS2	PTGS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGS1 geni (Tüm Gen Dizi Analizi)	PTGS1 gene (Sequence Analysis-All Coding Exons)	PTGS1	PTGS1, COX1, PGHS1, COX3, PCOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGR2 geni (Tüm Gen Dizi Analizi)	PTGR2 gene (Sequence Analysis-All Coding Exons)	PTGR2	ZADH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGR1 geni (Tüm Gen Dizi Analizi)	PTGR1 gene (Sequence Analysis-All Coding Exons)	PTGR1	PTGR1, LTB4DH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGIR geni (Tüm Gen Dizi Analizi)	PTGIR gene (Sequence Analysis-All Coding Exons)	PTGIR	PTGIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGFRN geni (Tüm Gen Dizi Analizi)	PTGFRN gene (Sequence Analysis-All Coding Exons)	PTGFRN	PTGFRN, FPRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGFR geni (Tüm Gen Dizi Analizi)	PTGFR gene (Sequence Analysis-All Coding Exons)	PTGFR	PTGFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGES3 geni (Tüm Gen Dizi Analizi)	PTGES3 gene (Sequence Analysis-All Coding Exons)	PTGES3	PTGES3, P23, CPGES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGES2 geni (Tüm Gen Dizi Analizi)	PTGES2 gene (Sequence Analysis-All Coding Exons)	PTGES2	PTGES2, PGES2, GBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGES geni (Tüm Gen Dizi Analizi)	PTGES gene (Sequence Analysis-All Coding Exons)	PTGES	PTGES, PGES, PIG12, MGST1 I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGER4 geni (Tüm Gen Dizi Analizi)	PTGER4 gene (Sequence Analysis-All Coding Exons)	PTGER4	PTGER4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGER3 geni (Tüm Gen Dizi Analizi)	PTGER3 gene (Sequence Analysis-All Coding Exons)	PTGER3	PTGER3, EP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGER1 geni (Tüm Gen Dizi Analizi)	PTGER1 gene (Sequence Analysis-All Coding Exons)	PTGER1	PTGER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGDS geni (Tüm Gen Dizi Analizi)	PTGDS gene (Sequence Analysis-All Coding Exons)	PTGDS	PTGDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTGDR2 geni (Tüm Gen Dizi Analizi)	PTGDR2 gene (Sequence Analysis-All Coding Exons)	PTGDR2	PTGDR2, GPR44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PTER geni (Tüm Gen Dizi Analizi)	PTER gene (Sequence Analysis-All Coding Exons)	PTER	PTER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTENP1 geni (Tüm Gen Dizi Analizi)	PTENP1 gene (Sequence Analysis-All Coding Exons)	PTENP1	PTENP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTDSS2 geni (Tüm Gen Dizi Analizi)	PTDSS2 gene (Sequence Analysis-All Coding Exons)	PTDSS2	PTDSS2, PSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCRA geni (Tüm Gen Dizi Analizi)	PTCRA gene (Sequence Analysis-All Coding Exons)	PTCRA	PTCRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCHD4 geni (Tüm Gen Dizi Analizi)	PTCHD4 gene (Sequence Analysis-All Coding Exons)	PTCHD4	PTCHD4, PTCH53, C5orf138	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCHD3 geni (Tüm Gen Dizi Analizi)	PTCHD3 gene (Sequence Analysis-All Coding Exons)	PTCHD3	PTCHD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCD3 geni (Tüm Gen Dizi Analizi)	PTCD3 gene (Sequence Analysis-All Coding Exons)	PTCD3	PTCD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A14 geni (Tüm Gen Dizi Analizi)	SLC22A14 gene (Sequence Analysis-All Coding Exons)	SLC22A14		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC22A13 geni (Tüm Gen Dizi Analizi)	SLC22A13 gene (Sequence Analysis-All Coding Exons)	SLC22A13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCD2 geni (Tüm Gen Dizi Analizi)	PTCD2 gene (Sequence Analysis-All Coding Exons)	PTCD2	PTCD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTCD1 geni (Tüm Gen Dizi Analizi)	PTCD1 gene (Sequence Analysis-All Coding Exons)	PTCD1	PTCD1, KIAA0632	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTBP3 geni (Tüm Gen Dizi Analizi)	PTBP3 gene (Sequence Analysis-All Coding Exons)	PTBP3	ROD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTBP2 geni (Tüm Gen Dizi Analizi)	PTBP2 gene (Sequence Analysis-All Coding Exons)	PTBP2	PTBP2, NPTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTBP1 geni (Tüm Gen Dizi Analizi)	PTBP1 gene (Sequence Analysis-All Coding Exons)	PTBP1	PTBP1, PTB, HNRNPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTAFR geni (Tüm Gen Dizi Analizi)	PTAFR gene (Sequence Analysis-All Coding Exons)	PTAFR	PTAFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC1A7 geni (Tüm Gen Dizi Analizi)	SLC1A7 gene (Sequence Analysis-All Coding Exons)	SLC1A7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSTPIP2 geni (Tüm Gen Dizi Analizi)	PSTPIP2 gene (Sequence Analysis-All Coding Exons)	PSTPIP2	PSTPIP2, MAYP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSTK geni (Tüm Gen Dizi Analizi)	PSTK gene (Sequence Analysis-All Coding Exons)	PSTK	PSTK, C10orf89	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSRC1 geni (Tüm Gen Dizi Analizi)	PSRC1 gene (Sequence Analysis-All Coding Exons)	PSRC1	PSRC1, DDA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSPN geni (Tüm Gen Dizi Analizi)	PSPN gene (Sequence Analysis-All Coding Exons)	PSPN	PSPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSPC1 geni (Tüm Gen Dizi Analizi)	PSPC1 gene (Sequence Analysis-All Coding Exons)	PSPC1	PSPC1, PSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSORS1C1 geni (Tüm Gen Dizi Analizi)	PSORS1C1 gene (Sequence Analysis-All Coding Exons)	PSORS1C1	SEEK1, C6orf16, PSORS1C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMG2 geni (Tüm Gen Dizi Analizi)	PSMG2 gene (Sequence Analysis-All Coding Exons)	PSMG2	PSMG2, TNFSF5IP1, HCCA3, CLAST3, PAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMG1 geni (Tüm Gen Dizi Analizi)	PSMG1 gene (Sequence Analysis-All Coding Exons)	PSMG1	PSMG1, DSCR2, C21LRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSME4 geni (Tüm Gen Dizi Analizi)	PSME4 gene (Sequence Analysis-All Coding Exons)	PSME4	PSME4, PA200, KIAA0077	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSME3 geni (Tüm Gen Dizi Analizi)	PSME3 gene (Sequence Analysis-All Coding Exons)	PSME3	PSME3, PA28G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSME2 geni (Tüm Gen Dizi Analizi)	PSME2 gene (Sequence Analysis-All Coding Exons)	PSME2	PSME2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSME1 geni (Tüm Gen Dizi Analizi)	PSME1 gene (Sequence Analysis-All Coding Exons)	PSME1	PSME1, IFI5111	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD9 geni (Tüm Gen Dizi Analizi)	PSMD9 gene (Sequence Analysis-All Coding Exons)	PSMD9	PSMD9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD7 geni (Tüm Gen Dizi Analizi)	PSMD7 gene (Sequence Analysis-All Coding Exons)	PSMD7	PSMD7, MOV34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD5 geni (Tüm Gen Dizi Analizi)	PSMD5 gene (Sequence Analysis-All Coding Exons)	PSMD5	PSMD5, SSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD4 geni (Tüm Gen Dizi Analizi)	PSMD4 gene (Sequence Analysis-All Coding Exons)	PSMD4	PSMD4, SSA, RPN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD2 geni (Tüm Gen Dizi Analizi)	PSMD2 gene (Sequence Analysis-All Coding Exons)	PSMD2	PSMD2, S2, TRAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD14 geni (Tüm Gen Dizi Analizi)	PSMD14 gene (Sequence Analysis-All Coding Exons)	PSMD14	PSMD14, POH1, PAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD13 geni (Tüm Gen Dizi Analizi)	PSMD13 gene (Sequence Analysis-All Coding Exons)	PSMD13	PSMD13, S11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD12 geni (Tüm Gen Dizi Analizi)	PSMD12 gene (Sequence Analysis-All Coding Exons)	PSMD12	PSMD12, P55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD11 geni (Tüm Gen Dizi Analizi)	PSMD11 gene (Sequence Analysis-All Coding Exons)	PSMD11	PSMD11, S9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMD10 geni (Tüm Gen Dizi Analizi)	PSMD10 gene (Sequence Analysis-All Coding Exons)	PSMD10	PSMD10, p28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC6 geni (Tüm Gen Dizi Analizi)	PSMC6 gene (Sequence Analysis-All Coding Exons)	PSMC6	PSMC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC5 geni (Tüm Gen Dizi Analizi)	PSMC5 gene (Sequence Analysis-All Coding Exons)	PSMC5	PSMC5, TRIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC4 geni (Tüm Gen Dizi Analizi)	PSMC4 gene (Sequence Analysis-All Coding Exons)	PSMC4	PSMC4, TBP7, S6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC3 geni (Tüm Gen Dizi Analizi)	PSMC3 gene (Sequence Analysis-All Coding Exons)	PSMC3	PSMC3, TBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC2 geni (Tüm Gen Dizi Analizi)	PSMC2 gene (Sequence Analysis-All Coding Exons)	PSMC2	PSMC2, MSS1, S7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMC1 geni (Tüm Gen Dizi Analizi)	PSMC1 gene (Sequence Analysis-All Coding Exons)	PSMC1	PSMC1, S4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A6 geni (Tüm Gen Dizi Analizi)	SLC16A6 gene (Sequence Analysis-All Coding Exons)	SLC16A6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A5 geni (Tüm Gen Dizi Analizi)	SLC16A5 gene (Sequence Analysis-All Coding Exons)	SLC16A5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A4 geni (Tüm Gen Dizi Analizi)	SLC16A4 gene (Sequence Analysis-All Coding Exons)	SLC16A4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC16A3 geni (Tüm Gen Dizi Analizi)	SLC16A3 gene (Sequence Analysis-All Coding Exons)	SLC16A3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB9 geni (Tüm Gen Dizi Analizi)	PSMB9 gene (Sequence Analysis-All Coding Exons)	PSMB9	PSMB9, LMP2, RING12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB7 geni (Tüm Gen Dizi Analizi)	PSMB7 gene (Sequence Analysis-All Coding Exons)	PSMB7	PSMB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB5 geni (Tüm Gen Dizi Analizi)	PSMB5 gene (Sequence Analysis-All Coding Exons)	PSMB5	PSMB5, LMPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB4 geni (Tüm Gen Dizi Analizi)	PSMB4 gene (Sequence Analysis-All Coding Exons)	PSMB4	PSMB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB3 geni (Tüm Gen Dizi Analizi)	PSMB3 gene (Sequence Analysis-All Coding Exons)	PSMB3	PSMB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB2 geni (Tüm Gen Dizi Analizi)	PSMB2 gene (Sequence Analysis-All Coding Exons)	PSMB2	PSMB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB11 geni (Tüm Gen Dizi Analizi)	PSMB11 gene (Sequence Analysis-All Coding Exons)	PSMB11	PSMB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB10 geni (Tüm Gen Dizi Analizi)	PSMB10 gene (Sequence Analysis-All Coding Exons)	PSMB10	MECL1, LMP10, PSMB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB1 geni (Tüm Gen Dizi Analizi)	PSMB1 gene (Sequence Analysis-All Coding Exons)	PSMB1	PSMB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA7 geni (Tüm Gen Dizi Analizi)	PSMA7 gene (Sequence Analysis-All Coding Exons)	PSMA7	PSMA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA5 geni (Tüm Gen Dizi Analizi)	PSMA5 gene (Sequence Analysis-All Coding Exons)	PSMA5	PSMA5, PSC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA4 geni (Tüm Gen Dizi Analizi)	PSMA4 gene (Sequence Analysis-All Coding Exons)	PSMA4	PSMA4, PSC9, HC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA3 geni (Tüm Gen Dizi Analizi)	PSMA3 gene (Sequence Analysis-All Coding Exons)	PSMA3	PSMA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA2 geni (Tüm Gen Dizi Analizi)	PSMA2 gene (Sequence Analysis-All Coding Exons)	PSMA2	PSMA2, PSC2, HC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMA1 geni (Tüm Gen Dizi Analizi)	PSMA1 gene (Sequence Analysis-All Coding Exons)	PSMA1	PSMA1, PROS30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSKH1 geni (Tüm Gen Dizi Analizi)	PSKH1 gene (Sequence Analysis-All Coding Exons)	PSKH1	PSKH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSIP1 geni (Tüm Gen Dizi Analizi)	PSIP1 gene (Sequence Analysis-All Coding Exons)	PSIP1	PSIP1, LEDGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG9 geni (Tüm Gen Dizi Analizi)	PSG9 gene (Sequence Analysis-All Coding Exons)	PSG9	PSG9, PSG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG8 geni (Tüm Gen Dizi Analizi)	PSG8 gene (Sequence Analysis-All Coding Exons)	PSG8	PSG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC12A8 geni (Tüm Gen Dizi Analizi)	SLC12A8 gene (Sequence Analysis-All Coding Exons)	SLC12A8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG7 geni (Tüm Gen Dizi Analizi)	PSG7 gene (Sequence Analysis-All Coding Exons)	PSG7	PSG7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG6 geni (Tüm Gen Dizi Analizi)	PSG6 gene (Sequence Analysis-All Coding Exons)	PSG6	PSG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG5 geni (Tüm Gen Dizi Analizi)	PSG5 gene (Sequence Analysis-All Coding Exons)	PSG5	PSG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG4 geni (Tüm Gen Dizi Analizi)	PSG4 gene (Sequence Analysis-All Coding Exons)	PSG4	PSG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG3 geni (Tüm Gen Dizi Analizi)	PSG3 gene (Sequence Analysis-All Coding Exons)	PSG3	PSG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG2 geni (Tüm Gen Dizi Analizi)	PSG2 gene (Sequence Analysis-All Coding Exons)	PSG2	PSG2, PSBG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG11 geni (Tüm Gen Dizi Analizi)	PSG11 gene (Sequence Analysis-All Coding Exons)	PSG11	PSG11, PSG13, PSG14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSG1 geni (Tüm Gen Dizi Analizi)	PSG1 gene (Sequence Analysis-All Coding Exons)	PSG1	PSG1, PSBG1, B1G1, SP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSD4 geni (Tüm Gen Dizi Analizi)	PSD4 gene (Sequence Analysis-All Coding Exons)	PSD4	PSD4, EFA6B, TIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSD3 geni (Tüm Gen Dizi Analizi)	PSD3 gene (Sequence Analysis-All Coding Exons)	PSD3	PSD3, HCA67, EFA6R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSD geni (Tüm Gen Dizi Analizi)	PSD gene (Sequence Analysis-All Coding Exons)	PSD	PSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLC10A7 geni (Tüm Gen Dizi Analizi)	SLC10A7 gene (Sequence Analysis-All Coding Exons)	SLC10A7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSCA geni (Tüm Gen Dizi Analizi)	PSCA gene (Sequence Analysis-All Coding Exons)	PSCA	PSCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRY2 geni (Tüm Gen Dizi Analizi)	PRY2 gene (Sequence Analysis-All Coding Exons)	PRY2	PRY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRY geni (Tüm Gen Dizi Analizi)	PRY gene (Sequence Analysis-All Coding Exons)	PRY	PRY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRUNE2 geni (Tüm Gen Dizi Analizi)	PRUNE2 gene (Sequence Analysis-All Coding Exons)	PRUNE2	PRUNE2, BMCC1, KIAA0367	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRTN3 geni (Tüm Gen Dizi Analizi)	PRTN3 gene (Sequence Analysis-All Coding Exons)	PRTN3	PRTN3, AGP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SLAMF9 geni (Tüm Gen Dizi Analizi)	SLAMF9 gene (Sequence Analysis-All Coding Exons)	SLAMF9		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRTG geni (Tüm Gen Dizi Analizi)	PRTG gene (Sequence Analysis-All Coding Exons)	PRTG	PRTG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRTFDC1 geni (Tüm Gen Dizi Analizi)	PRTFDC1 gene (Sequence Analysis-All Coding Exons)	PRTFDC1	PRTFDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS8 geni (Tüm Gen Dizi Analizi)	PRSS8 gene (Sequence Analysis-All Coding Exons)	PRSS8	PRSS8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS55 geni (Tüm Gen Dizi Analizi)	PRSS55 gene (Sequence Analysis-All Coding Exons)	PRSS55	PRSS55, TSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS53 geni (Tüm Gen Dizi Analizi)	PRSS53 gene (Sequence Analysis-All Coding Exons)	PRSS53	PRSS53, POL3S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS50 geni (Tüm Gen Dizi Analizi)	PRSS50 gene (Sequence Analysis-All Coding Exons)	PRSS50	PRSS50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS36 geni (Tüm Gen Dizi Analizi)	PRSS36 gene (Sequence Analysis-All Coding Exons)	PRSS36	PRSS36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PRSS33 geni (Tüm Gen Dizi Analizi)	PRSS33 gene (Sequence Analysis-All Coding Exons)	PRSS33	PRSS33, EOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS3 geni (Tüm Gen Dizi Analizi)	PRSS3 gene (Sequence Analysis-All Coding Exons)	PRSS3	PRSS3, TRY3, T9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS22 geni (Tüm Gen Dizi Analizi)	PRSS22 gene (Sequence Analysis-All Coding Exons)	PRSS22	PRSS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS21 geni (Tüm Gen Dizi Analizi)	PRSS21 gene (Sequence Analysis-All Coding Exons)	PRSS21	PRSS21, ESP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS16 geni (Tüm Gen Dizi Analizi)	PRSS16 gene (Sequence Analysis-All Coding Exons)	PRSS16	PRSS16, TSSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRX2 geni (Tüm Gen Dizi Analizi)	PRRX2 gene (Sequence Analysis-All Coding Exons)	PRRX2	PRRX2, PRX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRG4 geni (Tüm Gen Dizi Analizi)	PRRG4 gene (Sequence Analysis-All Coding Exons)	PRRG4	PRRG4, PRGP4, TMG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRG3 geni (Tüm Gen Dizi Analizi)	PRRG3 gene (Sequence Analysis-All Coding Exons)	PRRG3	PRRG3, PRGP3, TMG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRG2 geni (Tüm Gen Dizi Analizi)	PRRG2 gene (Sequence Analysis-All Coding Exons)	PRRG2	PRRG2, PRGP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRG1 geni (Tüm Gen Dizi Analizi)	PRRG1 gene (Sequence Analysis-All Coding Exons)	PRRG1	PRRG1, PRGP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRC2C geni (Tüm Gen Dizi Analizi)	PRRC2C gene (Sequence Analysis-All Coding Exons)	PRRC2C	PRRC2C, KIAA1096	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRC2A geni (Tüm Gen Dizi Analizi)	PRRC2A gene (Sequence Analysis-All Coding Exons)	PRRC2A	PRRC2A, BAT2, D6S51E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRRSL geni (Tüm Gen Dizi Analizi)	PRRSL gene (Sequence Analysis-All Coding Exons)	PRRSL	PRRSL, PROTOR2, FLJ31413	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR5 geni (Tüm Gen Dizi Analizi)	PRR5 gene (Sequence Analysis-All Coding Exons)	PRR5	PRR5, PP610	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR4 geni (Tüm Gen Dizi Analizi)	PRR4 gene (Sequence Analysis-All Coding Exons)	PRR4	PROL4, LPRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR16 geni (Tüm Gen Dizi Analizi)	PRR16 gene (Sequence Analysis-All Coding Exons)	PRR16	PRR16, LARGEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR13 geni (Tüm Gen Dizi Analizi)	PRR13 gene (Sequence Analysis-All Coding Exons)	PRR13	PRR13, TXR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR12 geni (Tüm Gen Dizi Analizi)	PRR12 gene (Sequence Analysis-All Coding Exons)	PRR12	PRR12, KIAA1205	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRR11 geni (Tüm Gen Dizi Analizi)	PRR11 gene (Sequence Analysis-All Coding Exons)	PRR11	PRR11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPSAP2 geni (Tüm Gen Dizi Analizi)	PRPSAP2 gene (Sequence Analysis-All Coding Exons)	PRPSAP2	PRPSAP2, PAP41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIVA1 geni (Tüm Gen Dizi Analizi)	SIVA1 gene (Sequence Analysis-All Coding Exons)	SIVA1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPSAP1 geni (Tüm Gen Dizi Analizi)	PRPSAP1 gene (Sequence Analysis-All Coding Exons)	PRPSAP1	PRPSAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPS2 geni (Tüm Gen Dizi Analizi)	PRPS2 gene (Sequence Analysis-All Coding Exons)	PRPS2	PRPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPS1L1 geni (Tüm Gen Dizi Analizi)	PRPS1L1 gene (Sequence Analysis-All Coding Exons)	PRPS1L1	PRPS1L1, PRPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF4B geni (Tüm Gen Dizi Analizi)	PRPF4B gene (Sequence Analysis-All Coding Exons)	PRPF4B	PRPF4B, PRP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF40A geni (Tüm Gen Dizi Analizi)	PRPF40A gene (Sequence Analysis-All Coding Exons)	PRPF40A	PRPF40A, HYPA, FNBP3, FBP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF39 geni (Tüm Gen Dizi Analizi)	PRPF39 gene (Sequence Analysis-All Coding Exons)	PRPF39	PRPF39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF38A geni (Tüm Gen Dizi Analizi)	PRPF38A gene (Sequence Analysis-All Coding Exons)	PRPF38A	PRPF38A, PRP38A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF19 geni (Tüm Gen Dizi Analizi)	PRPF19 gene (Sequence Analysis-All Coding Exons)	PRPF19	PRPF19, PRP19, PSO4, NMP200	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRPF18 geni (Tüm Gen Dizi Analizi)	PRPF18 gene (Sequence Analysis-All Coding Exons)	PRPF18	PRPF18, PRP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIRPB1 geni (Tüm Gen Dizi Analizi)	SIRPB1 gene (Sequence Analysis-All Coding Exons)	SIRPB1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROX2 geni (Tüm Gen Dizi Analizi)	PROX2 gene (Sequence Analysis-All Coding Exons)	PROX2	PROX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROX1 geni (Tüm Gen Dizi Analizi)	PROX1 gene (Sequence Analysis-All Coding Exons)	PROX1	PROX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROM2 geni (Tüm Gen Dizi Analizi)	PROM2 gene (Sequence Analysis-All Coding Exons)	PROM2	PROM2, PROML2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROKR1 geni (Tüm Gen Dizi Analizi)	PROKR1 gene (Sequence Analysis-All Coding Exons)	PROKR1	PROKR1, PKR1, GPR73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROK1 geni (Tüm Gen Dizi Analizi)	PROK1 gene (Sequence Analysis-All Coding Exons)	PROK1	PROK1, PK1, PRK1, EG:VEGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRODH2 geni (Tüm Gen Dizi Analizi)	PRODH2 gene (Sequence Analysis-All Coding Exons)	PRODH2	PRODH2, HYPDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROCR geni (Tüm Gen Dizi Analizi)	PROCR gene (Sequence Analysis-All Coding Exons)	PROCR	PROCR, EPCR, CCCA, CCD41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PROCA1 geni (Tüm Gen Dizi Analizi)	PROCA1 gene (Sequence Analysis-All Coding Exons)	PROCA1	PROCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRND geni (Tüm Gen Dizi Analizi)	PRND gene (Sequence Analysis-All Coding Exons)	PRND	PRND, DPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRNCR1 geni (Tüm Gen Dizi Analizi)	PRNCR1 gene (Sequence Analysis-All Coding Exons)	PRNCR1	PRNCR1, PCAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT9 geni (Tüm Gen Dizi Analizi)	PRMT9 gene (Sequence Analysis-All Coding Exons)	PRMT9	PRMT9, PRMT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT8 geni (Tüm Gen Dizi Analizi)	PRMT8 gene (Sequence Analysis-All Coding Exons)	PRMT8	PRMT8, HRMT1L3, HRMT1L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT6 geni (Tüm Gen Dizi Analizi)	PRMT6 gene (Sequence Analysis-All Coding Exons)	PRMT6	PRMT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT2 geni (Tüm Gen Dizi Analizi)	PRMT2 gene (Sequence Analysis-All Coding Exons)	PRMT2	HRMT1L1, PRMT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT1 geni (Tüm Gen Dizi Analizi)	PRMT1 gene (Sequence Analysis-All Coding Exons)	PRMT1	PRMT1, HRMT1L2, IR1B4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRM2 geni (Tüm Gen Dizi Analizi)	PRM2 gene (Sequence Analysis-All Coding Exons)	PRM2	PRM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRM1 geni (Tüm Gen Dizi Analizi)	PRM1 gene (Sequence Analysis-All Coding Exons)	PRM1	PRM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRLHR geni (Tüm Gen Dizi Analizi)	PRLHR gene (Sequence Analysis-All Coding Exons)	PRLHR	GPR10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRL geni (Tüm Gen Dizi Analizi)	PRL gene (Sequence Analysis-All Coding Exons)	PRL	PRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKY geni (Tüm Gen Dizi Analizi)	PRKY gene (Sequence Analysis-All Coding Exons)	PRKY	PRKY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKX geni (Tüm Gen Dizi Analizi)	PRKX gene (Sequence Analysis-All Coding Exons)	PRKX	PRKX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKG2 geni (Tüm Gen Dizi Analizi)	PRKG2 gene (Sequence Analysis-All Coding Exons)	PRKG2	PRKG2, PRKGR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKD3 geni (Tüm Gen Dizi Analizi)	PRKD3 gene (Sequence Analysis-All Coding Exons)	PRKD3	PRKCN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKD2 geni (Tüm Gen Dizi Analizi)	PRKD2 gene (Sequence Analysis-All Coding Exons)	PRKD2	PRKD2, PKD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SIDT1 geni (Tüm Gen Dizi Analizi)	SIDT1 gene (Sequence Analysis-All Coding Exons)	SIDT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKCZ geni (Tüm Gen Dizi Analizi)	PRKCZ gene (Sequence Analysis-All Coding Exons)	PRKCZ	PRKCZ, PKC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKCQ geni (Tüm Gen Dizi Analizi)	PRKCQ gene (Sequence Analysis-All Coding Exons)	PRKCQ	PRKCQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKCI geni (Tüm Gen Dizi Analizi)	PRKCI gene (Sequence Analysis-All Coding Exons)	PRKCI	PRKCI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKCE geni (Tüm Gen Dizi Analizi)	PRKCE gene (Sequence Analysis-All Coding Exons)	PRKCE	PRKCE, PKCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKCB geni (Tüm Gen Dizi Analizi)	PRKCB gene (Sequence Analysis-All Coding Exons)	PRKCB	PRKCB1, PKCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAR2B geni (Tüm Gen Dizi Analizi)	PRKAR2B gene (Sequence Analysis-All Coding Exons)	PRKAR2B	PRKAR2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAR2A geni (Tüm Gen Dizi Analizi)	PRKAR2A gene (Sequence Analysis-All Coding Exons)	PRKAR2A	PRKAR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAR1B geni (Tüm Gen Dizi Analizi)	PRKAR1B gene (Sequence Analysis-All Coding Exons)	PRKAR1B	PRKAR1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAG1 geni (Tüm Gen Dizi Analizi)	PRKAG1 gene (Sequence Analysis-All Coding Exons)	PRKAG1	PRKAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKACB geni (Tüm Gen Dizi Analizi)	PRKACB gene (Sequence Analysis-All Coding Exons)	PRKACB	PRKACB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAB2 geni (Tüm Gen Dizi Analizi)	PRKAB2 gene (Sequence Analysis-All Coding Exons)	PRKAB2	PRKAB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SHPRH geni (Tüm Gen Dizi Analizi)	SHPRH gene (Sequence Analysis-All Coding Exons)	SHPRH		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAB1 geni (Tüm Gen Dizi Analizi)	PRKAB1 gene (Sequence Analysis-All Coding Exons)	PRKAB1	PRKAB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAA2 geni (Tüm Gen Dizi Analizi)	PRKAA2 gene (Sequence Analysis-All Coding Exons)	PRKAA2	PRKAA2, AMPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRKAA1 geni (Tüm Gen Dizi Analizi)	PRKAA1 gene (Sequence Analysis-All Coding Exons)	PRKAA1	PRKAA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRIMA1 geni (Tüm Gen Dizi Analizi)	PRIMA1 gene (Sequence Analysis-All Coding Exons)	PRIMA1	PRIMA1, PRIMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRIM2 geni (Tüm Gen Dizi Analizi)	PRIM2 gene (Sequence Analysis-All Coding Exons)	PRIM2	PRIM2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRIM1 geni (Tüm Gen Dizi Analizi)	PRIM1 gene (Sequence Analysis-All Coding Exons)	PRIM1	PRIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRICKLE4 geni (Tüm Gen Dizi Analizi)	PRICKLE4 gene (Sequence Analysis-All Coding Exons)	PRICKLE4	PRICKLE4, C6orf49, OBTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRICKLE3 geni (Tüm Gen Dizi Analizi)	PRICKLE3 gene (Sequence Analysis-All Coding Exons)	PRICKLE3	PRICKLE3, LMO6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRICKLE2 geni (Tüm Gen Dizi Analizi)	PRICKLE2 gene (Sequence Analysis-All Coding Exons)	PRICKLE2	PRICKLE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRH2 geni (Tüm Gen Dizi Analizi)	PRH2 gene (Sequence Analysis-All Coding Exons)	PRH2	PRH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRH1 geni (Tüm Gen Dizi Analizi)	PRH1 gene (Sequence Analysis-All Coding Exons)	PRH1	PRH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRG3 geni (Tüm Gen Dizi Analizi)	PRG3 gene (Sequence Analysis-All Coding Exons)	PRG3	PRG3, MBPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRG2 geni (Tüm Gen Dizi Analizi)	PRG2 gene (Sequence Analysis-All Coding Exons)	PRG2	PRG2, MBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PREX2 geni (Tüm Gen Dizi Analizi)	PREX2 gene (Sequence Analysis-All Coding Exons)	PREX2	PREX2, DEPDC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PREX1 geni (Tüm Gen Dizi Analizi)	PREX1 gene (Sequence Analysis-All Coding Exons)	PREX1	PREX1, KIAA1415	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PREP geni (Tüm Gen Dizi Analizi)	PREP gene (Sequence Analysis-All Coding Exons)	PREP	PREP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRELP geni (Tüm Gen Dizi Analizi)	PRELP gene (Sequence Analysis-All Coding Exons)	PRELP	PRELP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRELID3A geni (Tüm Gen Dizi Analizi)	PRELID3A gene (Sequence Analysis-All Coding Exons)	PRELID3A	PRELID3A, SLMO1, C18orf43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRELID1 geni (Tüm Gen Dizi Analizi)	PRELID1 gene (Sequence Analysis-All Coding Exons)	PRELID1	PRELID1, PREL1, PX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PREB geni (Tüm Gen Dizi Analizi)	PREB gene (Sequence Analysis-All Coding Exons)	PREB	PREB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDX6 geni (Tüm Gen Dizi Analizi)	PRDX6 gene (Sequence Analysis-All Coding Exons)	PRDX6	PRDX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDX5 geni (Tüm Gen Dizi Analizi)	PRDX5 gene (Sequence Analysis-All Coding Exons)	PRDX5	PRDX5, AOEB166	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDX4 geni (Tüm Gen Dizi Analizi)	PRDX4 gene (Sequence Analysis-All Coding Exons)	PRDX4	PRDX4, AOE372	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDX3 geni (Tüm Gen Dizi Analizi)	PRDX3 gene (Sequence Analysis-All Coding Exons)	PRDX3	PRDX3, AOP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDX2 geni (Tüm Gen Dizi Analizi)	PRDX2 gene (Sequence Analysis-All Coding Exons)	PRDX2	PRDX2, PRX2, TDPX1, PTX1, MKEF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PRDX1 geni (Tüm Gen Dizi Analizi)	PRDX1 gene (Sequence Analysis-All Coding Exons)	PRDX1	PAGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM9 geni (Tüm Gen Dizi Analizi)	PRDM9 gene (Sequence Analysis-All Coding Exons)	PRDM9	PRDM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM4 geni (Tüm Gen Dizi Analizi)	PRDM4 gene (Sequence Analysis-All Coding Exons)	PRDM4	PRDM4, PFM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM2 geni (Tüm Gen Dizi Analizi)	PRDM2 gene (Sequence Analysis-All Coding Exons)	PRDM2	PRDM2, RIZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM14 geni (Tüm Gen Dizi Analizi)	PRDM14 gene (Sequence Analysis-All Coding Exons)	PRDM14	PRDM14, PFM11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM13 geni (Tüm Gen Dizi Analizi)	PRDM13 gene (Sequence Analysis-All Coding Exons)	PRDM13	PRDM13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM11 geni (Tüm Gen Dizi Analizi)	PRDM11 gene (Sequence Analysis-All Coding Exons)	PRDM11	PRDM11, PFM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM1 geni (Tüm Gen Dizi Analizi)	PRDM1 gene (Sequence Analysis-All Coding Exons)	PRDM1	PRDM1, BLIMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRCP geni (Tüm Gen Dizi Analizi)	PRCP gene (Sequence Analysis-All Coding Exons)	PRCP	PRCP, PCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRC1 geni (Tüm Gen Dizi Analizi)	PRC1 gene (Sequence Analysis-All Coding Exons)	PRC1	PRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRB4 geni (Tüm Gen Dizi Analizi)	PRB4 gene (Sequence Analysis-All Coding Exons)	PRB4	PRB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRB3 geni (Tüm Gen Dizi Analizi)	PRB3 gene (Sequence Analysis-All Coding Exons)	PRB3	PRB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3GLB2 geni (Tüm Gen Dizi Analizi)	SH3GLB2 gene (Sequence Analysis-All Coding Exons)	SH3GLB2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH3GLB1 geni (Tüm Gen Dizi Analizi)	SH3GLB1 gene (Sequence Analysis-All Coding Exons)	SH3GLB1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRB2 geni (Tüm Gen Dizi Analizi)	PRB2 gene (Sequence Analysis-All Coding Exons)	PRB2	PRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRB1 geni (Tüm Gen Dizi Analizi)	PRB1 gene (Sequence Analysis-All Coding Exons)	PRB1	PRB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAP1 geni (Tüm Gen Dizi Analizi)	PRAP1 gene (Sequence Analysis-All Coding Exons)	PRAP1	PRAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAME geni (Tüm Gen Dizi Analizi)	PRAME gene (Sequence Analysis-All Coding Exons)	PRAME	PRAME, MAPE, OIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAG1 geni (Tüm Gen Dizi Analizi)	PRAG1 gene (Sequence Analysis-All Coding Exons)	PRAG1	PRAG1, PRAGMIN, NACK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAF2 geni (Tüm Gen Dizi Analizi)	PRAF2 gene (Sequence Analysis-All Coding Exons)	PRAF2	PRAF2, JM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAC2 geni (Tüm Gen Dizi Analizi)	PRAC2 gene (Sequence Analysis-All Coding Exons)	PRAC2	HOXBAS5, PRAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAC1 geni (Tüm Gen Dizi Analizi)	PRAC1 gene (Sequence Analysis-All Coding Exons)	PRAC1	PRAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PQLC2 geni (Tüm Gen Dizi Analizi)	PQLC2 gene (Sequence Analysis-All Coding Exons)	PQLC2	PQLC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPY2P geni (Tüm Gen Dizi Analizi)	PPY2P gene (Sequence Analysis-All Coding Exons)	PPY2P	PPY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPY geni (Tüm Gen Dizi Analizi)	PPY gene (Sequence Analysis-All Coding Exons)	PPY	PPY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPTC7 geni (Tüm Gen Dizi Analizi)	PPTC7 gene (Sequence Analysis-All Coding Exons)	PPTC7	PPTC7, TAPP2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPT2 geni (Tüm Gen Dizi Analizi)	PPT2 gene (Sequence Analysis-All Coding Exons)	PPT2	PPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP6R3 geni (Tüm Gen Dizi Analizi)	PPP6R3 gene (Sequence Analysis-All Coding Exons)	PPP6R3	SAPS3, PP6R3, C11orf23, KIAA1458	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SH2D3A geni (Tüm Gen Dizi Analizi)	SH2D3A gene (Sequence Analysis-All Coding Exons)	SH2D3A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP6R2 geni (Tüm Gen Dizi Analizi)	PPP6R2 gene (Sequence Analysis-All Coding Exons)	PPP6R2	SAPS2, PP6R2, KIAA0685	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP6R1 geni (Tüm Gen Dizi Analizi)	PPP6R1 gene (Sequence Analysis-All Coding Exons)	PPP6R1	SAPS1, PP6R1, KIAA1115	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP6C geni (Tüm Gen Dizi Analizi)	PPP6C gene (Sequence Analysis-All Coding Exons)	PPP6C	PPP6C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP5C geni (Tüm Gen Dizi Analizi)	PPP5C gene (Sequence Analysis-All Coding Exons)	PPP5C	PPP5C, PP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4R4 geni (Tüm Gen Dizi Analizi)	PPP4R4 gene (Sequence Analysis-All Coding Exons)	PPP4R4	PPP4R4, PP4R4, KIAA1622	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4R3B geni (Tüm Gen Dizi Analizi)	PPP4R3B gene (Sequence Analysis-All Coding Exons)	PPP4R3B	PPP4R3B, PP4R3B, SMEK2, KIAA1387	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4R3A geni (Tüm Gen Dizi Analizi)	PPP4R3A gene (Sequence Analysis-All Coding Exons)	PPP4R3A	PPP4R3A, SMEK1, PP4R3A, KIAA2010	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4R2 geni (Tüm Gen Dizi Analizi)	PPP4R2 gene (Sequence Analysis-All Coding Exons)	PPP4R2	PPP4R2, PP4R2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4R1 geni (Tüm Gen Dizi Analizi)	PPP4R1 gene (Sequence Analysis-All Coding Exons)	PPP4R1	PPP4R1, PP4R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP4C geni (Tüm Gen Dizi Analizi)	PPP4C gene (Sequence Analysis-All Coding Exons)	PPP4C	PPP4C, PP4, PPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP3R2 geni (Tüm Gen Dizi Analizi)	PPP3R2 gene (Sequence Analysis-All Coding Exons)	PPP3R2	PPP3R2, PPP3RL, CBLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP3R1 geni (Tüm Gen Dizi Analizi)	PPP3R1 gene (Sequence Analysis-All Coding Exons)	PPP3R1	PPP3R1, CALNB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP3CC geni (Tüm Gen Dizi Analizi)	PPP3CC gene (Sequence Analysis-All Coding Exons)	PPP3CC	PPP3CC, CALNA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SGSM1 geni (Tüm Gen Dizi Analizi)	SGSM1 gene (Sequence Analysis-All Coding Exons)	SGSM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP3CB geni (Tüm Gen Dizi Analizi)	PPP3CB gene (Sequence Analysis-All Coding Exons)	PPP3CB	PPP3CB, CALNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP3CA geni (Tüm Gen Dizi Analizi)	PPP3CA gene (Sequence Analysis-All Coding Exons)	PPP3CA	PPP3CA, PPP2B, CALNA, CNA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R5E geni (Tüm Gen Dizi Analizi)	PPP2R5E gene (Sequence Analysis-All Coding Exons)	PPP2R5E	PPP2R5E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R5C geni (Tüm Gen Dizi Analizi)	PPP2R5C gene (Sequence Analysis-All Coding Exons)	PPP2R5C	PPP2R5C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R5B geni (Tüm Gen Dizi Analizi)	PPP2R5B gene (Sequence Analysis-All Coding Exons)	PPP2R5B	PPP2R5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R5A geni (Tüm Gen Dizi Analizi)	PPP2R5A gene (Sequence Analysis-All Coding Exons)	PPP2R5A	PPP2R5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R3C geni (Tüm Gen Dizi Analizi)	PPP2R3C gene (Sequence Analysis-All Coding Exons)	PPP2R3C	PPP2RC2, GSPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R3B geni (Tüm Gen Dizi Analizi)	PPP2R3B gene (Sequence Analysis-All Coding Exons)	PPP2R3B	PPP2R3B, PR48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R3A geni (Tüm Gen Dizi Analizi)	PPP2R3A gene (Sequence Analysis-All Coding Exons)	PPP2R3A	PPP2R3A, PR72, PR130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R2D geni (Tüm Gen Dizi Analizi)	PPP2R2D gene (Sequence Analysis-All Coding Exons)	PPP2R2D	PPP2R2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R2C geni (Tüm Gen Dizi Analizi)	PPP2R2C gene (Sequence Analysis-All Coding Exons)	PPP2R2C	PPP2R2C, PR52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2CB geni (Tüm Gen Dizi Analizi)	PPP2CB gene (Sequence Analysis-All Coding Exons)	PPP2CB	PPP2CB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2CA geni (Tüm Gen Dizi Analizi)	PPP2CA gene (Sequence Analysis-All Coding Exons)	PPP2CA	PPP2CA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R9A geni (Tüm Gen Dizi Analizi)	PPP1R9A gene (Sequence Analysis-All Coding Exons)	PPP1R9A	PPP1R9A, NRB, NRB1, KIAA1222	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R8 geni (Tüm Gen Dizi Analizi)	PPP1R8 gene (Sequence Analysis-All Coding Exons)	PPP1R8	PPP1R8, NIPP1, ARD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R7 geni (Tüm Gen Dizi Analizi)	PPP1R7 gene (Sequence Analysis-All Coding Exons)	PPP1R7	PPP1R7, SDS22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R3D geni (Tüm Gen Dizi Analizi)	PPP1R3D gene (Sequence Analysis-All Coding Exons)	PPP1R3D	PPP1R3D, PPP1R6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R3C geni (Tüm Gen Dizi Analizi)	PPP1R3C gene (Sequence Analysis-All Coding Exons)	PPP1R3C	PPP1R3C, PPP1R5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R3B geni (Tüm Gen Dizi Analizi)	PPP1R3B gene (Sequence Analysis-All Coding Exons)	PPP1R3B	PPP1R3B, GL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R26 geni (Tüm Gen Dizi Analizi)	PPP1R26 gene (Sequence Analysis-All Coding Exons)	PPP1R26	PPP1R26, KIAA0649	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R2 geni (Tüm Gen Dizi Analizi)	PPP1R2 gene (Sequence Analysis-All Coding Exons)	PPP1R2	PPP1R2, IPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R1C geni (Tüm Gen Dizi Analizi)	PPP1R1C gene (Sequence Analysis-All Coding Exons)	PPP1R1C	PPP1R1C, IPP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R1A geni (Tüm Gen Dizi Analizi)	PPP1R1A gene (Sequence Analysis-All Coding Exons)	PPP1R1A	PPP1R1A, IPP1, I1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R18 geni (Tüm Gen Dizi Analizi)	PPP1R18 gene (Sequence Analysis-All Coding Exons)	PPP1R18	KIAA1949	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R16B geni (Tüm Gen Dizi Analizi)	PPP1R16B gene (Sequence Analysis-All Coding Exons)	PPP1R16B	PPP1R16B, TIMAP, KIAA0823	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R15A geni (Tüm Gen Dizi Analizi)	PPP1R15A gene (Sequence Analysis-All Coding Exons)	PPP1R15A	PPP1R15A, GADD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R14D geni (Tüm Gen Dizi Analizi)	PPP1R14D gene (Sequence Analysis-All Coding Exons)	PPP1R14D	PPP1R14D, GBPI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R14C geni (Tüm Gen Dizi Analizi)	PPP1R14C gene (Sequence Analysis-All Coding Exons)	PPP1R14C	PPP1R14C, KEPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R14B geni (Tüm Gen Dizi Analizi)	PPP1R14B gene (Sequence Analysis-All Coding Exons)	PPP1R14B	PPP1R14B, PLCB3N, PNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R14A geni (Tüm Gen Dizi Analizi)	PPP1R14A gene (Sequence Analysis-All Coding Exons)	PPP1R14A	PPP1R14A, CPI17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R13L geni (Tüm Gen Dizi Analizi)	PPP1R13L gene (Sequence Analysis-All Coding Exons)	PPP1R13L	PPP1R13L, RAI, iASPP, NKIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R12C geni (Tüm Gen Dizi Analizi)	PPP1R12C gene (Sequence Analysis-All Coding Exons)	PPP1R12C	PPP1R12C, MBS85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R12B geni (Tüm Gen Dizi Analizi)	PPP1R12B gene (Sequence Analysis-All Coding Exons)	PPP1R12B	PPP1R12B, MYPT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R12A geni (Tüm Gen Dizi Analizi)	PPP1R12A gene (Sequence Analysis-All Coding Exons)	PPP1R12A	PPP1R12A, MYPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R11 geni (Tüm Gen Dizi Analizi)	PPP1R11 gene (Sequence Analysis-All Coding Exons)	PPP1R11	PPP1R11, TCTEX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R10 geni (Tüm Gen Dizi Analizi)	PPP1R10 gene (Sequence Analysis-All Coding Exons)	PPP1R10	PPP1R10, PNUTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1CC geni (Tüm Gen Dizi Analizi)	PPP1CC gene (Sequence Analysis-All Coding Exons)	PPP1CC	PPP1CC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1CB geni (Tüm Gen Dizi Analizi)	PPP1CB gene (Sequence Analysis-All Coding Exons)	PPP1CB	PPP1CB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1CA geni (Tüm Gen Dizi Analizi)	PPP1CA gene (Sequence Analysis-All Coding Exons)	PPP1CA	PPP1CA, PPP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPME1 geni (Tüm Gen Dizi Analizi)	PPME1 gene (Sequence Analysis-All Coding Exons)	PPME1	PPME1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1M geni (Tüm Gen Dizi Analizi)	PPM1M gene (Sequence Analysis-All Coding Exons)	PPM1M	PPM1M, PP2CE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1L geni (Tüm Gen Dizi Analizi)	PPM1L gene (Sequence Analysis-All Coding Exons)	PPM1L	PPM1L, PP2CE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1J geni (Tüm Gen Dizi Analizi)	PPM1J gene (Sequence Analysis-All Coding Exons)	PPM1J	PPM1J, PP2CZ, PPP2CZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1H geni (Tüm Gen Dizi Analizi)	PPM1H gene (Sequence Analysis-All Coding Exons)	PPM1H	PPM1H, KIAA1157	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1G geni (Tüm Gen Dizi Analizi)	PPM1G gene (Sequence Analysis-All Coding Exons)	PPM1G	PPM1G, PP2CG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1B geni (Tüm Gen Dizi Analizi)	PPM1B gene (Sequence Analysis-All Coding Exons)	PPM1B	PPM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPM1A geni (Tüm Gen Dizi Analizi)	PPM1A gene (Sequence Analysis-All Coding Exons)	PPM1A	PPM1A, PP2CA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPL geni (Tüm Gen Dizi Analizi)	PPL gene (Sequence Analysis-All Coding Exons)	PPL	PPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIP5K2 geni (Tüm Gen Dizi Analizi)	PPIP5K2 gene (Sequence Analysis-All Coding Exons)	PPIP5K2	HISPPD1, PPIP5K2, VIP2, KIAA0433	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIP5K1 geni (Tüm Gen Dizi Analizi)	PPIP5K1 gene (Sequence Analysis-All Coding Exons)	PPIP5K1	PPIP5K1, HISPPD2A, IP5I, KIAA0877	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PPIL4 geni (Tüm Gen Dizi Analizi)	PPIL4 gene (Sequence Analysis-All Coding Exons)	PPIL4	PPIL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIL3 geni (Tüm Gen Dizi Analizi)	PPIL3 gene (Sequence Analysis-All Coding Exons)	PPIL3	PPIL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIL2 geni (Tüm Gen Dizi Analizi)	PPIL2 gene (Sequence Analysis-All Coding Exons)	PPIL2	PPIL2, CYP60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIL1 geni (Tüm Gen Dizi Analizi)	PPIL1 gene (Sequence Analysis-All Coding Exons)	PPIL1	PPIL1, CYPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIH geni (Tüm Gen Dizi Analizi)	PPIH gene (Sequence Analysis-All Coding Exons)	PPIH	PPIH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIG geni (Tüm Gen Dizi Analizi)	PPIG gene (Sequence Analysis-All Coding Exons)	PPIG	PPIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIF geni (Tüm Gen Dizi Analizi)	PPIF gene (Sequence Analysis-All Coding Exons)	PPIF	PPIF, CYP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIE geni (Tüm Gen Dizi Analizi)	PPIE gene (Sequence Analysis-All Coding Exons)	PPIE	PPIE, CYP33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPID geni (Tüm Gen Dizi Analizi)	PPID gene (Sequence Analysis-All Coding Exons)	PPID	PPID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIC geni (Tüm Gen Dizi Analizi)	PPIC gene (Sequence Analysis-All Coding Exons)	PPIC	PPIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIAL4E geni (Tüm Gen Dizi Analizi)	PPIAL4E gene (Sequence Analysis-All Coding Exons)	PPIAL4E	PPIAL4A, COAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPIA geni (Tüm Gen Dizi Analizi)	PPIA gene (Sequence Analysis-All Coding Exons)	PPIA	PPIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPHLN1 geni (Tüm Gen Dizi Analizi)	PPHLN1 gene (Sequence Analysis-All Coding Exons)	PPHLN1	PPHLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIA4 geni (Tüm Gen Dizi Analizi)	PPFIA4 gene (Sequence Analysis-All Coding Exons)	PPFIA4	PPFIA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIA3 geni (Tüm Gen Dizi Analizi)	PPFIA3 gene (Sequence Analysis-All Coding Exons)	PPFIA3	PPFIA3, LPNA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIA1 geni (Tüm Gen Dizi Analizi)	PPFIA1 gene (Sequence Analysis-All Coding Exons)	PPFIA1	PPFIA1, LIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPEF2 geni (Tüm Gen Dizi Analizi)	PPEF2 gene (Sequence Analysis-All Coding Exons)	PPEF2	PPEF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPEF1 geni (Tüm Gen Dizi Analizi)	PPEF1 gene (Sequence Analysis-All Coding Exons)	PPEF1	PPEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPCS geni (Tüm Gen Dizi Analizi)	PPCS gene (Sequence Analysis-All Coding Exons)	PPCS	PPCS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPCDC geni (Tüm Gen Dizi Analizi)	PPCDC gene (Sequence Analysis-All Coding Exons)	PPCDC	PPCDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPBPP2 geni (Tüm Gen Dizi Analizi)	PPBPP2 gene (Sequence Analysis-All Coding Exons)	PPBPP2	PPBPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPBP geni (Tüm Gen Dizi Analizi)	PPBP gene (Sequence Analysis-All Coding Exons)	PPBP	PPBP, CXCL7, SCYB7, CTAP3, TGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPAT geni (Tüm Gen Dizi Analizi)	PPAT gene (Sequence Analysis-All Coding Exons)	PPAT	PPAT, GPAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPARGC1A geni (Tüm Gen Dizi Analizi)	PPARGC1A gene (Sequence Analysis-All Coding Exons)	PPARGC1A	PPARGC1A, PPARGC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPARD geni (Tüm Gen Dizi Analizi)	PPARD gene (Sequence Analysis-All Coding Exons)	PPARD	PPARD, NUC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPAN geni (Tüm Gen Dizi Analizi)	PPAN gene (Sequence Analysis-All Coding Exons)	PPAN	PPAN, SSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPA1 geni (Tüm Gen Dizi Analizi)	PPA1 gene (Sequence Analysis-All Coding Exons)	PPA1	PP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU5F1B geni (Tüm Gen Dizi Analizi)	POU5F1B gene (Sequence Analysis-All Coding Exons)	POU5F1B	POU5F1B, POU5F1P1, OCTARC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU5F1 geni (Tüm Gen Dizi Analizi)	POU5F1 gene (Sequence Analysis-All Coding Exons)	POU5F1	POU5F1, OTF3, OCT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU4F2 geni (Tüm Gen Dizi Analizi)	POU4F2 gene (Sequence Analysis-All Coding Exons)	POU4F2	BRN3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU4F1 geni (Tüm Gen Dizi Analizi)	POU4F1 gene (Sequence Analysis-All Coding Exons)	POU4F1	POU4F1, BRN3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU3F3 geni (Tüm Gen Dizi Analizi)	POU3F3 gene (Sequence Analysis-All Coding Exons)	POU3F3	POU3F3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU3F2 geni (Tüm Gen Dizi Analizi)	POU3F2 gene (Sequence Analysis-All Coding Exons)	POU3F2	POU3F2, OCT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU3F1 geni (Tüm Gen Dizi Analizi)	POU3F1 gene (Sequence Analysis-All Coding Exons)	POU3F1	POU3F1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU2F3 geni (Tüm Gen Dizi Analizi)	POU2F3 gene (Sequence Analysis-All Coding Exons)	POU2F3	POU2F3, OCT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU2F2 geni (Tüm Gen Dizi Analizi)	POU2F2 gene (Sequence Analysis-All Coding Exons)	POU2F2	POU2F2, OTF2, OCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU2F1 geni (Tüm Gen Dizi Analizi)	POU2F1 gene (Sequence Analysis-All Coding Exons)	POU2F1	OTF1, OCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POU2AF1 geni (Tüm Gen Dizi Analizi)	POU2AF1 gene (Sequence Analysis-All Coding Exons)	POU2AF1	POU2AF1, OBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTEH geni (Tüm Gen Dizi Analizi)	POTEH gene (Sequence Analysis-All Coding Exons)	POTEH	POTEH, POTE22, ACTBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTEE geni (Tüm Gen Dizi Analizi)	POTEE gene (Sequence Analysis-All Coding Exons)	POTEE	POTEE, POTE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTED geni (Tüm Gen Dizi Analizi)	POTED gene (Sequence Analysis-All Coding Exons)	POTED	POTED, ANKRD21, POTE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POSTN geni (Tüm Gen Dizi Analizi)	POSTN gene (Sequence Analysis-All Coding Exons)	POSTN	POSTN, OSF2, PN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POPDC3 geni (Tüm Gen Dizi Analizi)	POPDC3 gene (Sequence Analysis-All Coding Exons)	POPDC3	POPDC3, POP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POPDC2 geni (Tüm Gen Dizi Analizi)	POPDC2 gene (Sequence Analysis-All Coding Exons)	POPDC2	POPDC2, POP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POP7 geni (Tüm Gen Dizi Analizi)	POP7 gene (Sequence Analysis-All Coding Exons)	POP7	POP7, RPP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POP5 geni (Tüm Gen Dizi Analizi)	POP5 gene (Sequence Analysis-All Coding Exons)	POP5	POP5, HSPC004	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POP4 geni (Tüm Gen Dizi Analizi)	POP4 gene (Sequence Analysis-All Coding Exons)	POP4	RPP29, POP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POP1 geni (Tüm Gen Dizi Analizi)	POP1 gene (Sequence Analysis-All Coding Exons)	POP1	POP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PON3 geni (Tüm Gen Dizi Analizi)	PON3 gene (Sequence Analysis-All Coding Exons)	PON3	PON3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POMZP3 geni (Tüm Gen Dizi Analizi)	POMZP3 gene (Sequence Analysis-All Coding Exons)	POMZP3	POMZP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POM121C geni (Tüm Gen Dizi Analizi)	POM121C gene (Sequence Analysis-All Coding Exons)	POM121C	POM121C, POM121-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POM121 geni (Tüm Gen Dizi Analizi)	POM121 gene (Sequence Analysis-All Coding Exons)	POM121	POM121, POM121A, KIAA0618	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLRMT geni (Tüm Gen Dizi Analizi)	POLRMT gene (Sequence Analysis-All Coding Exons)	POLRMT	POLRMT, APOLMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR3D geni (Tüm Gen Dizi Analizi)	POLR3D gene (Sequence Analysis-All Coding Exons)	POLR3D	POLR3D, BN51T, TSBN51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2M geni (Tüm Gen Dizi Analizi)	POLR2M gene (Sequence Analysis-All Coding Exons)	POLR2M	POLR2M, GDOWN, GRINL1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2L geni (Tüm Gen Dizi Analizi)	POLR2L gene (Sequence Analysis-All Coding Exons)	POLR2L	POLR2L, RPB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2K geni (Tüm Gen Dizi Analizi)	POLR2K gene (Sequence Analysis-All Coding Exons)	POLR2K	POLR2K, RPB12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2J2 geni (Tüm Gen Dizi Analizi)	POLR2J2 gene (Sequence Analysis-All Coding Exons)	POLR2J2	POLR2J2, RPB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2J geni (Tüm Gen Dizi Analizi)	POLR2J gene (Sequence Analysis-All Coding Exons)	POLR2J	POLR2J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2I geni (Tüm Gen Dizi Analizi)	POLR2I gene (Sequence Analysis-All Coding Exons)	POLR2I	POLR2I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2H geni (Tüm Gen Dizi Analizi)	POLR2H gene (Sequence Analysis-All Coding Exons)	POLR2H	POLR2H, RPB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2G geni (Tüm Gen Dizi Analizi)	POLR2G gene (Sequence Analysis-All Coding Exons)	POLR2G	POLR2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2F geni (Tüm Gen Dizi Analizi)	POLR2F gene (Sequence Analysis-All Coding Exons)	POLR2F	POLR2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2E geni (Tüm Gen Dizi Analizi)	POLR2E gene (Sequence Analysis-All Coding Exons)	POLR2E	POLR2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2C geni (Tüm Gen Dizi Analizi)	POLR2C gene (Sequence Analysis-All Coding Exons)	POLR2C	POLR2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2B geni (Tüm Gen Dizi Analizi)	POLR2B gene (Sequence Analysis-All Coding Exons)	POLR2B	POLR2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2A geni (Tüm Gen Dizi Analizi)	POLR2A gene (Sequence Analysis-All Coding Exons)	POLR2A	POLR2A, RPOL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLQ geni (Tüm Gen Dizi Analizi)	POLQ gene (Sequence Analysis-All Coding Exons)	POLQ	POLQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLN geni (Tüm Gen Dizi Analizi)	POLN gene (Sequence Analysis-All Coding Exons)	POLN	POLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLM geni (Tüm Gen Dizi Analizi)	POLM gene (Sequence Analysis-All Coding Exons)	POLM	POLM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLL geni (Tüm Gen Dizi Analizi)	POLL gene (Sequence Analysis-All Coding Exons)	POLL	POLL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLK geni (Tüm Gen Dizi Analizi)	POLK gene (Sequence Analysis-All Coding Exons)	POLK	POLK, POLQ, DINB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLI geni (Tüm Gen Dizi Analizi)	POLI gene (Sequence Analysis-All Coding Exons)	POLI	POLI, RAD30B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLE4 geni (Tüm Gen Dizi Analizi)	POLE4 gene (Sequence Analysis-All Coding Exons)	POLE4	POLE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLE3 geni (Tüm Gen Dizi Analizi)	POLE3 gene (Sequence Analysis-All Coding Exons)	POLE3	POLE3, CHARAC17, YBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLE2 geni (Tüm Gen Dizi Analizi)	POLE2 gene (Sequence Analysis-All Coding Exons)	POLE2	POLE2, DPE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLDIP3 geni (Tüm Gen Dizi Analizi)	POLDIP3 gene (Sequence Analysis-All Coding Exons)	POLDIP3	POLDIP3, SKAR, PDIP46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLDIP2 geni (Tüm Gen Dizi Analizi)	POLDIP2 gene (Sequence Analysis-All Coding Exons)	POLDIP2	POLDIP2, PDIP38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLD4 geni (Tüm Gen Dizi Analizi)	POLD4 gene (Sequence Analysis-All Coding Exons)	POLD4	POLD4, POLDS, P12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLD2 geni (Tüm Gen Dizi Analizi)	POLD2 gene (Sequence Analysis-All Coding Exons)	POLD2	POLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLB geni (Tüm Gen Dizi Analizi)	POLB gene (Sequence Analysis-All Coding Exons)	POLB	POLB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POFUT2 geni (Tüm Gen Dizi Analizi)	POFUT2 gene (Sequence Analysis-All Coding Exons)	POFUT2	POFUT2, C21orf80, KIAA0958	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PODXL2 geni (Tüm Gen Dizi Analizi)	PODXL2 gene (Sequence Analysis-All Coding Exons)	PODXL2	PODXL2, EG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PODXL geni (Tüm Gen Dizi Analizi)	PODXL gene (Sequence Analysis-All Coding Exons)	PODXL	PODXL, PCLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PODN geni (Tüm Gen Dizi Analizi)	PODN gene (Sequence Analysis-All Coding Exons)	PODN	PODN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNRC2 geni (Tüm Gen Dizi Analizi)	PNRC2 gene (Sequence Analysis-All Coding Exons)	PNRC2	PNRC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNRC1 geni (Tüm Gen Dizi Analizi)	PNRC1 gene (Sequence Analysis-All Coding Exons)	PNRC1	PNRC1, PROL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNPLA7 geni (Tüm Gen Dizi Analizi)	PNPLA7 gene (Sequence Analysis-All Coding Exons)	PNPLA7	PNPLA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNPLA5 geni (Tüm Gen Dizi Analizi)	PNPLA5 gene (Sequence Analysis-All Coding Exons)	PNPLA5	PNPLA5, GS2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNPLA4 geni (Tüm Gen Dizi Analizi)	PNPLA4 gene (Sequence Analysis-All Coding Exons)	PNPLA4	DXS1283E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNPLA3 geni (Tüm Gen Dizi Analizi)	PNPLA3 gene (Sequence Analysis-All Coding Exons)	PNPLA3	PNPLA3, ADPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNOC geni (Tüm Gen Dizi Analizi)	PNOC gene (Sequence Analysis-All Coding Exons)	PNOC	PNOC, PPNOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEMA5B geni (Tüm Gen Dizi Analizi)	SEMA5B gene (Sequence Analysis-All Coding Exons)	SEMA5B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PNN geni (Tüm Gen Dizi Analizi)	PNN gene (Sequence Analysis-All Coding Exons)	PNN	PNN, DRS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMT geni (Tüm Gen Dizi Analizi)	PNMT gene (Sequence Analysis-All Coding Exons)	PNMT	PNMT, PENT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
SEM4D geni (Tüm Gen Dizi Analizi)	SEMA4D gene (Sequence Analysis-All Coding Exons)	SEMA4D		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMA6A geni (Tüm Gen Dizi Analizi)	PNMA6A gene (Sequence Analysis-All Coding Exons)	PNMA6A	PNMA6A, PNMA6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMA5 geni (Tüm Gen Dizi Analizi)	PNMA5 gene (Sequence Analysis-All Coding Exons)	PNMA5	PNMA5, KIAA1934	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMA3 geni (Tüm Gen Dizi Analizi)	PNMA3 gene (Sequence Analysis-All Coding Exons)	PNMA3	PNMA3, MA3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMA2 geni (Tüm Gen Dizi Analizi)	PNMA2 gene (Sequence Analysis-All Coding Exons)	PNMA2	PNMA2, MA2, MM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNMA1 geni (Tüm Gen Dizi Analizi)	PNMA1 gene (Sequence Analysis-All Coding Exons)	PNMA1	PNMA1, MA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNLIPRP2 geni (Tüm Gen Dizi Analizi)	PNLIPRP2 gene (Sequence Analysis-All Coding Exons)	PNLIPRP2	PNLIPRP2, PLRP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNLIPRP1 geni (Tüm Gen Dizi Analizi)	PNLIPRP1 gene (Sequence Analysis-All Coding Exons)	PNLIPRP1	PNLIPRP1, PLRP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNISR geni (Tüm Gen Dizi Analizi)	PNISR gene (Sequence Analysis-All Coding Exons)	PNISR	PNISR, SRRP130, SFRS18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PNCK geni (Tüm Gen Dizi Analizi)	PNCK gene (Sequence Analysis-All Coding Exons)	PNCK	PNCK, CAMK1B, BSTK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMS1 geni (Tüm Gen Dizi Analizi)	PMS1 gene (Sequence Analysis-All Coding Exons)	PMS1	PMS1, PMSL1, HNPCC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMPCB geni (Tüm Gen Dizi Analizi)	PMPCB gene (Sequence Analysis-All Coding Exons)	PMPCB	MPPB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMP2 geni (Tüm Gen Dizi Analizi)	PMP2 gene (Sequence Analysis-All Coding Exons)	PMP2	PMP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMM1 geni (Tüm Gen Dizi Analizi)	PMM1 gene (Sequence Analysis-All Coding Exons)	PMM1	PMM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMF1 geni (Tüm Gen Dizi Analizi)	PMF1 gene (Sequence Analysis-All Coding Exons)	PMF1	PMF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMEPA1 geni (Tüm Gen Dizi Analizi)	PMEPA1 gene (Sequence Analysis-All Coding Exons)	PMEPA1	PMEPA1, TMEPAI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMEL geni (Tüm Gen Dizi Analizi)	PMEL gene (Sequence Analysis-All Coding Exons)	PMEL	SILV, D12S53E, PMEL17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMCHL2 geni (Tüm Gen Dizi Analizi)	PMCHL2 gene (Sequence Analysis-All Coding Exons)	PMCHL2	PMCHL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMCH geni (Tüm Gen Dizi Analizi)	PMCH gene (Sequence Analysis-All Coding Exons)	PMCH	PMCH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PMAIP1 geni (Tüm Gen Dizi Analizi)	PMAIP1 gene (Sequence Analysis-All Coding Exons)	PMAIP1	PMAIP1, APR, NOXA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PM20D2 geni (Tüm Gen Dizi Analizi)	PM20D2 gene (Sequence Analysis-All Coding Exons)	PM20D2	PM20D2, ACY1L2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PM20D1 geni (Tüm Gen Dizi Analizi)	PM20D1 gene (Sequence Analysis-All Coding Exons)	PM20D1	PM20D1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXND1 geni (Tüm Gen Dizi Analizi)	PLXND1 gene (Sequence Analysis-All Coding Exons)	PLXND1	PLXND1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXND1 geni (Tüm Gen Dizi Analizi)	PLXND1 gene (Sequence Analysis-All Coding Exons)	PLXND1	PLXND1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNC1 geni (Tüm Gen Dizi Analizi)	PLXNC1 gene (Sequence Analysis-All Coding Exons)	PLXNC1	VESPR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNB3 geni (Tüm Gen Dizi Analizi)	PLXNB3 gene (Sequence Analysis-All Coding Exons)	PLXNB3	PLXNB3, PLXN5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNB2 geni (Tüm Gen Dizi Analizi)	PLXNB2 gene (Sequence Analysis-All Coding Exons)	PLXNB2	PLXNB2, MM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNB1 geni (Tüm Gen Dizi Analizi)	PLXNB1 gene (Sequence Analysis-All Coding Exons)	PLXNB1	SEP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNA4 geni (Tüm Gen Dizi Analizi)	PLXNA4 gene (Sequence Analysis-All Coding Exons)	PLXNA4	PLXNA4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNA3 geni (Tüm Gen Dizi Analizi)	PLXNA3 gene (Sequence Analysis-All Coding Exons)	PLXNA3	SEX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNA2 geni (Tüm Gen Dizi Analizi)	PLXNA2 gene (Sequence Analysis-All Coding Exons)	PLXNA2	PLXNA2, OCT, PLXN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXNA1 geni (Tüm Gen Dizi Analizi)	PLXNA1 gene (Sequence Analysis-All Coding Exons)	PLXNA1	PLXNA1, NOVP, NOV, PLXN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXDC2 geni (Tüm Gen Dizi Analizi)	PLXDC2 gene (Sequence Analysis-All Coding Exons)	PLXDC2	PLXDC2, TEM7R	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLXDC1 geni (Tüm Gen Dizi Analizi)	PLXDC1 gene (Sequence Analysis-All Coding Exons)	PLXDC1	PLXDC1, TEM7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLVAP geni (Tüm Gen Dizi Analizi)	PLVAP gene (Sequence Analysis-All Coding Exons)	PLVAP	PLVAP, PV1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLSCR4 geni (Tüm Gen Dizi Analizi)	PLSCR4 gene (Sequence Analysis-All Coding Exons)	PLSCR4	PLSCR4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLSCR3 geni (Tüm Gen Dizi Analizi)	PLSCR3 gene (Sequence Analysis-All Coding Exons)	PLSCR3	PLSCR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLSCR2 geni (Tüm Gen Dizi Analizi)	PLSCR2 gene (Sequence Analysis-All Coding Exons)	PLSCR2	PLSCR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLSCR1 geni (Tüm Gen Dizi Analizi)	PLSCR1 gene (Sequence Analysis-All Coding Exons)	PLSCR1	PLSCR1, MMTRA1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLS1 geni (Tüm Gen Dizi Analizi)	PLS1 gene (Sequence Analysis-All Coding Exons)	PLS1	PLS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLRG1 geni (Tüm Gen Dizi Analizi)	PLRG1 gene (Sequence Analysis-All Coding Exons)	PLRG1	PLRG1, PRL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPPR5 geni (Tüm Gen Dizi Analizi)	PLPPR5 gene (Sequence Analysis-All Coding Exons)	PLPPR5	PLPPR5, LPPR5, PAP2D, PAP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPPR4 geni (Tüm Gen Dizi Analizi)	PLPPR4 gene (Sequence Analysis-All Coding Exons)	PLPPR4	PLPPR4, LPPR4, PRG1, KIAA0455	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPPR3 geni (Tüm Gen Dizi Analizi)	PLPPR3 gene (Sequence Analysis-All Coding Exons)	PLPPR3	PLPPR3, LPPR3, LPR3, PRG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPP6 geni (Tüm Gen Dizi Analizi)	PLPP6 gene (Sequence Analysis-All Coding Exons)	PLPP6	PPAPDC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPP5 geni (Tüm Gen Dizi Analizi)	PLPP5 gene (Sequence Analysis-All Coding Exons)	PLPP5	PPAPDC1B, HTPAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLP2 geni (Tüm Gen Dizi Analizi)	PLP2 gene (Sequence Analysis-All Coding Exons)	PLP2	PLP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLLP geni (Tüm Gen Dizi Analizi)	PLLP gene (Sequence Analysis-All Coding Exons)	PLLP	PLLP, PMLP, TM4SF11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLK3 geni (Tüm Gen Dizi Analizi)	PLK3 gene (Sequence Analysis-All Coding Exons)	PLK3	PLK3, CNK, PRK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLK2 geni (Tüm Gen Dizi Analizi)	PLK2 gene (Sequence Analysis-All Coding Exons)	PLK2	PLK2, SNK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLIN5 geni (Tüm Gen Dizi Analizi)	PLIN5 gene (Sequence Analysis-All Coding Exons)	PLIN5	PLIN5, LSDP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLIN4 geni (Tüm Gen Dizi Analizi)	PLIN4 gene (Sequence Analysis-All Coding Exons)	PLIN4	PLIN4, KIAA1881	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLIN3 geni (Tüm Gen Dizi Analizi)	PLIN3 gene (Sequence Analysis-All Coding Exons)	PLIN3	PLIN3, M6PRBP1, TIP47	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLIN2 geni (Tüm Gen Dizi Analizi)	PLIN2 gene (Sequence Analysis-All Coding Exons)	PLIN2	PLIN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLGLB1 geni (Tüm Gen Dizi Analizi)	PLGLB1 gene (Sequence Analysis-All Coding Exons)	PLGLB1	PLGLB1, PLGL, PRGB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLET1 geni (Tüm Gen Dizi Analizi)	PLET1 gene (Sequence Analysis-All Coding Exons)	PLET1	C11orf34, PLET1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHO1 geni (Tüm Gen Dizi Analizi)	PLEKHO1 gene (Sequence Analysis-All Coding Exons)	PLEKHO1	PLEKHO1, CKIP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHM2 geni (Tüm Gen Dizi Analizi)	PLEKHM2 gene (Sequence Analysis-All Coding Exons)	PLEKHM2	PLEKHM2, SKIP, KIAA0842	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHH2 geni (Tüm Gen Dizi Analizi)	PLEKHH2 gene (Sequence Analysis-All Coding Exons)	PLEKHH2	PLEKHH2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHG6 geni (Tüm Gen Dizi Analizi)	PLEKHG6 gene (Sequence Analysis-All Coding Exons)	PLEKHG6	PLEKHG6, MYOGEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHG4 geni (Tüm Gen Dizi Analizi)	PLEKHG4 gene (Sequence Analysis-All Coding Exons)	PLEKHG4	PLEKHG4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHF2 geni (Tüm Gen Dizi Analizi)	PLEKHF2 gene (Sequence Analysis-All Coding Exons)	PLEKHF2	PLEKHF2, EAPF, PHAFIN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHF1 geni (Tüm Gen Dizi Analizi)	PLEKHF1 gene (Sequence Analysis-All Coding Exons)	PLEKHF1	PLEKHF1, LAPF, PHAFIN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHB1 geni (Tüm Gen Dizi Analizi)	PLEKHB1 gene (Sequence Analysis-All Coding Exons)	PLEKHB1	PLEKHB1, PHR1, KPL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA8 geni (Tüm Gen Dizi Analizi)	PLEKHA8 gene (Sequence Analysis-All Coding Exons)	PLEKHA8	FAPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA7 geni (Tüm Gen Dizi Analizi)	PLEKHA7 gene (Sequence Analysis-All Coding Exons)	PLEKHA7	PLEKHA7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA6 geni (Tüm Gen Dizi Analizi)	PLEKHA6 gene (Sequence Analysis-All Coding Exons)	PLEKHA6	PEPP3, KIAA0969	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA5 geni (Tüm Gen Dizi Analizi)	PLEKHA5 gene (Sequence Analysis-All Coding Exons)	PLEKHA5	PEPP2, KIAA1686	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA4 geni (Tüm Gen Dizi Analizi)	PLEKHA4 gene (Sequence Analysis-All Coding Exons)	PLEKHA4	PLEKHA4, PEPP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA3 geni (Tüm Gen Dizi Analizi)	PLEKHA3 gene (Sequence Analysis-All Coding Exons)	PLEKHA3	PLEKHA3, FAPP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA2 geni (Tüm Gen Dizi Analizi)	PLEKHA2 gene (Sequence Analysis-All Coding Exons)	PLEKHA2	PLEKHA2, TAPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEKHA1 geni (Tüm Gen Dizi Analizi)	PLEKHA1 gene (Sequence Analysis-All Coding Exons)	PLEKHA1	PLEKHA1, TAPP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEK2 geni (Tüm Gen Dizi Analizi)	PLEK2 gene (Sequence Analysis-All Coding Exons)	PLEK2	PLEK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLEK geni (Tüm Gen Dizi Analizi)	PLEK gene (Sequence Analysis-All Coding Exons)	PLEK	PLEK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLD6 geni (Tüm Gen Dizi Analizi)	PLD6 gene (Sequence Analysis-All Coding Exons)	PLD6	PLD6, ZUC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLD3 geni (Tüm Gen Dizi Analizi)	PLD3 gene (Sequence Analysis-All Coding Exons)	PLD3	PLD3, HUK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLD2 geni (Tüm Gen Dizi Analizi)	PLD2 gene (Sequence Analysis-All Coding Exons)	PLD2	PLD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLD1 geni (Tüm Gen Dizi Analizi)	PLD1 gene (Sequence Analysis-All Coding Exons)	PLD1	PLD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCXD3 geni (Tüm Gen Dizi Analizi)	PLCXD3 gene (Sequence Analysis-All Coding Exons)	PLCXD3	PLCXD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCXD2 geni (Tüm Gen Dizi Analizi)	PLCXD2 gene (Sequence Analysis-All Coding Exons)	PLCXD2	PLCXD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCXD1 geni (Tüm Gen Dizi Analizi)	PLCXD1 gene (Sequence Analysis-All Coding Exons)	PLCXD1	PLCXD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCL2 geni (Tüm Gen Dizi Analizi)	PLCL2 gene (Sequence Analysis-All Coding Exons)	PLCL2	PLCL2, KIAA1092	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCL1 geni (Tüm Gen Dizi Analizi)	PLCL1 gene (Sequence Analysis-All Coding Exons)	PLCL1	PLDL1, PLCE, PLCL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCH2 geni (Tüm Gen Dizi Analizi)	PLCH2 gene (Sequence Analysis-All Coding Exons)	PLCH2	PLCH2, PLCL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCH1 geni (Tüm Gen Dizi Analizi)	PLCH1 gene (Sequence Analysis-All Coding Exons)	PLCH1	PLCH1, PLCL3, KIAA1069	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCG1 geni (Tüm Gen Dizi Analizi)	PLCG1 gene (Sequence Analysis-All Coding Exons)	PLCG1	PLCG1, PLC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCB3 geni (Tüm Gen Dizi Analizi)	PLCB3 gene (Sequence Analysis-All Coding Exons)	PLCB3	PLCB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLB1 geni (Tüm Gen Dizi Analizi)	PLB1 gene (Sequence Analysis-All Coding Exons)	PLB1	PLB1, PLB, FLJ30866	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLAUR geni (Tüm Gen Dizi Analizi)	PLAUR gene (Sequence Analysis-All Coding Exons)	PLAUR	PLAUR, URKR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLAGL2 geni (Tüm Gen Dizi Analizi)	PLAGL2 gene (Sequence Analysis-All Coding Exons)	PLAGL2	PLAGL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLAC9 geni (Tüm Gen Dizi Analizi)	PLAC9 gene (Sequence Analysis-All Coding Exons)	PLAC9	PLAC9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PLAC8 geni (Tüm Gen Dizi Analizi)	PLAC8 gene (Sequence Analysis-All Coding Exons)	PLAC8	PLAC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLAC1 geni (Tüm Gen Dizi Analizi)	PLAC1 gene (Sequence Analysis-All Coding Exons)	PLAC1	PLAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLAA geni (Tüm Gen Dizi Analizi)	PLAA gene (Sequence Analysis-All Coding Exons)	PLAA	PLAA, PLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2R1 geni (Tüm Gen Dizi Analizi)	PLA2R1 gene (Sequence Analysis-All Coding Exons)	PLA2R1	PLA2R1, PLA2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G4D geni (Tüm Gen Dizi Analizi)	PLA2G4D gene (Sequence Analysis-All Coding Exons)	PLA2G4D	PLA2G4D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G4C geni (Tüm Gen Dizi Analizi)	PLA2G4C gene (Sequence Analysis-All Coding Exons)	PLA2G4C	PLA2G4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G4B geni (Tüm Gen Dizi Analizi)	PLA2G4B gene (Sequence Analysis-All Coding Exons)	PLA2G4B	PLA2G4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G3 geni (Tüm Gen Dizi Analizi)	PLA2G3 gene (Sequence Analysis-All Coding Exons)	PLA2G3	PLA2G3, SPLA2III	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G2F geni (Tüm Gen Dizi Analizi)	PLA2G2F gene (Sequence Analysis-All Coding Exons)	PLA2G2F	PLA2G2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G2D geni (Tüm Gen Dizi Analizi)	PLA2G2D gene (Sequence Analysis-All Coding Exons)	PLA2G2D	PLA2G2D, SPLASH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G1B geni (Tüm Gen Dizi Analizi)	PLA2G1B gene (Sequence Analysis-All Coding Exons)	PLA2G1B	PLA2G1B, PLA2A, PLA2, PPLA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G16 geni (Tüm Gen Dizi Analizi)	PLA2G16 gene (Sequence Analysis-All Coding Exons)	PLA2G16	PLA2G16, HRASLS3, HRSL3, HREV107	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G15 geni (Tüm Gen Dizi Analizi)	PLA2G15 gene (Sequence Analysis-All Coding Exons)	PLA2G15	LYPLA3, LLPL, ACS, LPLA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G12B geni (Tüm Gen Dizi Analizi)	PLA2G12B gene (Sequence Analysis-All Coding Exons)	PLA2G12B	PLA2G12B, GXIIB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G12A geni (Tüm Gen Dizi Analizi)	PLA2G12A gene (Sequence Analysis-All Coding Exons)	PLA2G12A	PLA2G12A, GXII, ROSSY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA2G10 geni (Tüm Gen Dizi Analizi)	PLA2G10 gene (Sequence Analysis-All Coding Exons)	PLA2G10	PLA2G10, SPLA2, GXSPLA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLA1A geni (Tüm Gen Dizi Analizi)	PLA1A gene (Sequence Analysis-All Coding Exons)	PLA1A	PLA1A, PSPLA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKP4 geni (Tüm Gen Dizi Analizi)	PKP4 gene (Sequence Analysis-All Coding Exons)	PKP4	PKP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKP3 geni (Tüm Gen Dizi Analizi)	PKP3 gene (Sequence Analysis-All Coding Exons)	PKP3	PKP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKNOX2 geni (Tüm Gen Dizi Analizi)	PKNOX2 gene (Sequence Analysis-All Coding Exons)	PKNOX2	PKNOX2, PREP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKNOX1 geni (Tüm Gen Dizi Analizi)	PKNOX1 gene (Sequence Analysis-All Coding Exons)	PKNOX1	PKNOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKN3 geni (Tüm Gen Dizi Analizi)	PKN3 gene (Sequence Analysis-All Coding Exons)	PKN3	PKN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKN2 geni (Tüm Gen Dizi Analizi)	PKN2 gene (Sequence Analysis-All Coding Exons)	PKN2	PKN2, PRKCL2, PRK2, PAK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKN1 geni (Tüm Gen Dizi Analizi)	PKN1 gene (Sequence Analysis-All Coding Exons)	PKN1	PKN1, PRKCL1, PRK1, PAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKMYT1 geni (Tüm Gen Dizi Analizi)	PKMYT1 gene (Sequence Analysis-All Coding Exons)	PKMYT1	PKMYT1, MYT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKM geni (Tüm Gen Dizi Analizi)	PKM gene (Sequence Analysis-All Coding Exons)	PKM	PKM, PKM2, PK3, PKM2, THBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKIG geni (Tüm Gen Dizi Analizi)	PKIG gene (Sequence Analysis-All Coding Exons)	PKIG	PKIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKIB geni (Tüm Gen Dizi Analizi)	PKIB gene (Sequence Analysis-All Coding Exons)	PKIB	PKIB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKIA geni (Tüm Gen Dizi Analizi)	PKIA gene (Sequence Analysis-All Coding Exons)	PKIA	PKIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKHD1L1 geni (Tüm Gen Dizi Analizi)	PKHD1L1 gene (Sequence Analysis-All Coding Exons)	PKHD1L1	PKHD1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKDREJ geni (Tüm Gen Dizi Analizi)	PKDREJ gene (Sequence Analysis-All Coding Exons)	PKDREJ	PKDREJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKDCC geni (Tüm Gen Dizi Analizi)	PKDCC gene (Sequence Analysis-All Coding Exons)	PKDCC	PKDCC, VLK, SGK493	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKD2L2 geni (Tüm Gen Dizi Analizi)	PKD2L2 gene (Sequence Analysis-All Coding Exons)	PKD2L2	PKD2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKD2L1 geni (Tüm Gen Dizi Analizi)	PKD2L1 gene (Sequence Analysis-All Coding Exons)	PKD2L1	PKD2L1, PKDL, PKD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKD1L3 geni (Tüm Gen Dizi Analizi)	PKD1L3 gene (Sequence Analysis-All Coding Exons)	PKD1L3	PKD1L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PKD1L2 geni (Tüm Gen Dizi Analizi)	PKD1L2 gene (Sequence Analysis-All Coding Exons)	PKD1L2	PKD1L2, PC1L2, KIAA1879	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PJA1 geni (Tüm Gen Dizi Analizi)	PJA1 gene (Sequence Analysis-All Coding Exons)	PJA1	PJA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIWIL4 geni (Tüm Gen Dizi Analizi)	PIWIL4 gene (Sequence Analysis-All Coding Exons)	PIWIL4	PIWIL4, HIWI2, MIWI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIWIL3 geni (Tüm Gen Dizi Analizi)	PIWIL3 gene (Sequence Analysis-All Coding Exons)	PIWIL3	PIWIL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIWIL2 geni (Tüm Gen Dizi Analizi)	PIWIL2 gene (Sequence Analysis-All Coding Exons)	PIWIL2	PIWIL2, MILI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIWIL1 geni (Tüm Gen Dizi Analizi)	PIWIL1 gene (Sequence Analysis-All Coding Exons)	PIWIL1	PIWIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PITPNC1 geni (Tüm Gen Dizi Analizi)	PITPNC1 gene (Sequence Analysis-All Coding Exons)	PITPNC1	PITPNC1, RDGBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PITPNB geni (Tüm Gen Dizi Analizi)	PITPNB gene (Sequence Analysis-All Coding Exons)	PITPNB	PITPNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PITPNA geni (Tüm Gen Dizi Analizi)	PITPNA gene (Sequence Analysis-All Coding Exons)	PITPNA	PITPNA, PITPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PISD geni (Tüm Gen Dizi Analizi)	PISD gene (Sequence Analysis-All Coding Exons)	PISD	PISD, PSD, PSSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIRT geni (Tüm Gen Dizi Analizi)	PIRT gene (Sequence Analysis-All Coding Exons)	PIRT	PIRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIR geni (Tüm Gen Dizi Analizi)	PIR gene (Sequence Analysis-All Coding Exons)	PIR	PIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP5KL1 geni (Tüm Gen Dizi Analizi)	PIP5KL1 gene (Sequence Analysis-All Coding Exons)	PIP5KL1	PIP5KL1, PIPKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP5K1B geni (Tüm Gen Dizi Analizi)	PIP5K1B gene (Sequence Analysis-All Coding Exons)	PIP5K1B	PIP5K1B, STM7, MSS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP5K1A geni (Tüm Gen Dizi Analizi)	PIP5K1A gene (Sequence Analysis-All Coding Exons)	PIP5K1A	PIP5K1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP4K2C geni (Tüm Gen Dizi Analizi)	PIP4K2C gene (Sequence Analysis-All Coding Exons)	PIP4K2C	PIP4K2C, PIP5K2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP4K2B geni (Tüm Gen Dizi Analizi)	PIP4K2B gene (Sequence Analysis-All Coding Exons)	PIP4K2B	PIP4K2B, PIP5P4KB, PIP5K3K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP4K2A geni (Tüm Gen Dizi Analizi)	PIP4K2A gene (Sequence Analysis-All Coding Exons)	PIP4K2A	PIP4K2A, PIP5P4KA, PIP5K2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIP geni (Tüm Gen Dizi Analizi)	PIP gene (Sequence Analysis-All Coding Exons)	PIP	PIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PINX1 geni (Tüm Gen Dizi Analizi)	PINX1 gene (Sequence Analysis-All Coding Exons)	PINX1	PINX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIN4 geni (Tüm Gen Dizi Analizi)	PIN4 gene (Sequence Analysis-All Coding Exons)	PIN4	PIN4, PAR14, EPVH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIN1P1 geni (Tüm Gen Dizi Analizi)	PIN1P1 gene (Sequence Analysis-All Coding Exons)	PIN1P1	PIN1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIN1 geni (Tüm Gen Dizi Analizi)	PIN1 gene (Sequence Analysis-All Coding Exons)	PIN1	PIN1, DOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIM3 geni (Tüm Gen Dizi Analizi)	PIM3 gene (Sequence Analysis-All Coding Exons)	PIM3	PIM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIM2 geni (Tüm Gen Dizi Analizi)	PIM2 gene (Sequence Analysis-All Coding Exons)	PIM2	PIM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIM1 geni (Tüm Gen Dizi Analizi)	PIM1 gene (Sequence Analysis-All Coding Exons)	PIM1	PIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PILRB geni (Tüm Gen Dizi Analizi)	PILRB gene (Sequence Analysis-All Coding Exons)	PILRB	PILRB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PILRA geni (Tüm Gen Dizi Analizi)	PILRA gene (Sequence Analysis-All Coding Exons)	PILRA	PILRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3R3 geni (Tüm Gen Dizi Analizi)	PIK3R3 gene (Sequence Analysis-All Coding Exons)	PIK3R3	PIK3R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3CG geni (Tüm Gen Dizi Analizi)	PIK3CG gene (Sequence Analysis-All Coding Exons)	PIK3CG	PIK3CG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3CB geni (Tüm Gen Dizi Analizi)	PIK3CB gene (Sequence Analysis-All Coding Exons)	PIK3CB	PIK3CB, P13KCB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3C2G geni (Tüm Gen Dizi Analizi)	PIK3C2G gene (Sequence Analysis-All Coding Exons)	PIK3C2G	PIK3C2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3C2B geni (Tüm Gen Dizi Analizi)	PIK3C2B gene (Sequence Analysis-All Coding Exons)	PIK3C2B	PIK3C2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3C2A geni (Tüm Gen Dizi Analizi)	PIK3C2A gene (Sequence Analysis-All Coding Exons)	PIK3C2A	PIK3C2A, CPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3AP1 geni (Tüm Gen Dizi Analizi)	PIK3AP1 gene (Sequence Analysis-All Coding Exons)	PIK3AP1	PIK3AP1, BCAP, FLJ35564	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIH1D1 geni (Tüm Gen Dizi Analizi)	PIH1D1 gene (Sequence Analysis-All Coding Exons)	PIH1D1	PIH1D1, NOP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGZ geni (Tüm Gen Dizi Analizi)	PIGZ gene (Sequence Analysis-All Coding Exons)	PIGZ	PIGZ, SMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGX geni (Tüm Gen Dizi Analizi)	PIGX gene (Sequence Analysis-All Coding Exons)	PIGX	PIGX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGU geni (Tüm Gen Dizi Analizi)	PIGU gene (Sequence Analysis-All Coding Exons)	PIGU	PIGU, CDC91L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGS geni (Tüm Gen Dizi Analizi)	PIGS gene (Sequence Analysis-All Coding Exons)	PIGS	PIGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGR geni (Tüm Gen Dizi Analizi)	PIGR gene (Sequence Analysis-All Coding Exons)	PIGR	PIGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGQ geni (Tüm Gen Dizi Analizi)	PIGQ gene (Sequence Analysis-All Coding Exons)	PIGQ	PIGQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGP geni (Tüm Gen Dizi Analizi)	PIGP gene (Sequence Analysis-All Coding Exons)	PIGP	PIGP, DSCR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGK geni (Tüm Gen Dizi Analizi)	PIGK gene (Sequence Analysis-All Coding Exons)	PIGK	PIGK, GPI8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGH geni (Tüm Gen Dizi Analizi)	PIGH gene (Sequence Analysis-All Coding Exons)	PIGH	PIGH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGF geni (Tüm Gen Dizi Analizi)	PIGF gene (Sequence Analysis-All Coding Exons)	PIGF	PIGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGC geni (Tüm Gen Dizi Analizi)	PIGC gene (Sequence Analysis-All Coding Exons)	PIGC	PIGC, GPI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIGB geni (Tüm Gen Dizi Analizi)	PIGB gene (Sequence Analysis-All Coding Exons)	PIGB	PIGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIFO geni (Tüm Gen Dizi Analizi)	PIFO gene (Sequence Analysis-All Coding Exons)	PIFO	C1orf88, PIFO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIF1 geni (Tüm Gen Dizi Analizi)	PIF1 gene (Sequence Analysis-All Coding Exons)	PIF1	PIF1, PIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIDD1 geni (Tüm Gen Dizi Analizi)	PIDD1 gene (Sequence Analysis-All Coding Exons)	PIDD1	PIDD1, LRDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PID1 geni (Tüm Gen Dizi Analizi)	PID1 gene (Sequence Analysis-All Coding Exons)	PID1	PID1, NYGGF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PICSAR geni (Tüm Gen Dizi Analizi)	PICSAR gene (Sequence Analysis-All Coding Exons)	PICSAR	PICSAR, LINC00162, NLC1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PICK1 geni (Tüm Gen Dizi Analizi)	PICK1 gene (Sequence Analysis-All Coding Exons)	PICK1	PICK1, PRKCABP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIBF1 geni (Tüm Gen Dizi Analizi)	PIBF1 gene (Sequence Analysis-All Coding Exons)	PIBF1	PIBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIAS4 geni (Tüm Gen Dizi Analizi)	PIAS4 gene (Sequence Analysis-All Coding Exons)	PIAS4	PIAS4, PIASY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIAS3 geni (Tüm Gen Dizi Analizi)	PIAS3 gene (Sequence Analysis-All Coding Exons)	PIAS3	PIAS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIAS1 geni (Tüm Gen Dizi Analizi)	PIAS1 gene (Sequence Analysis-All Coding Exons)	PIAS1	PIAS1, DDXBP1, GBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

SCGN geni (Tüm Gen Dizi Analizi)	SCGN gene (Sequence Analysis-All Coding Exons)	SCGN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIANP geni (Tüm Gen Dizi Analizi)	PIANP gene (Sequence Analysis-All Coding Exons)	PIANP	PIANP, PANP, LEDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PI4KB geni (Tüm Gen Dizi Analizi)	PI4KB gene (Sequence Analysis-All Coding Exons)	PI4KB	PIK4CB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PI4K2B geni (Tüm Gen Dizi Analizi)	PI4K2B gene (Sequence Analysis-All Coding Exons)	PI4K2B	PI4K2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PI4K2A geni (Tüm Gen Dizi Analizi)	PI4K2A gene (Sequence Analysis-All Coding Exons)	PI4K2A	PI4K2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PI3 geni (Tüm Gen Dizi Analizi)	PI3 gene (Sequence Analysis-All Coding Exons)	PI3	PI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PI15 geni (Tüm Gen Dizi Analizi)	PI15 gene (Sequence Analysis-All Coding Exons)	PI15	PI15, PI2STI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHTF2 geni (Tüm Gen Dizi Analizi)	PHTF2 gene (Sequence Analysis-All Coding Exons)	PHTF2	PHTF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHTF1 geni (Tüm Gen Dizi Analizi)	PHTF1 gene (Sequence Analysis-All Coding Exons)	PHTF1	PHTF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHRF1 geni (Tüm Gen Dizi Analizi)	PHRF1 gene (Sequence Analysis-All Coding Exons)	PHRF1	KIAA1542	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHPT1 geni (Tüm Gen Dizi Analizi)	PHPT1 gene (Sequence Analysis-All Coding Exons)	PHPT1	PHPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLPP2 geni (Tüm Gen Dizi Analizi)	PHLPP2 gene (Sequence Analysis-All Coding Exons)	PHLPP2	PHLPPL, PHLPP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLPP1 geni (Tüm Gen Dizi Analizi)	PHLPP1 gene (Sequence Analysis-All Coding Exons)	PHLPP1	PHLPP1, SCOP, KIAA0606	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLDB2 geni (Tüm Gen Dizi Analizi)	PHLDB2 gene (Sequence Analysis-All Coding Exons)	PHLDB2	PHLDB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLDB1 geni (Tüm Gen Dizi Analizi)	PHLDB1 gene (Sequence Analysis-All Coding Exons)	PHLDB1	PHLDB1, LL5A, KIAA0638	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLDA3 geni (Tüm Gen Dizi Analizi)	PHLDA3 gene (Sequence Analysis-All Coding Exons)	PHLDA3	PHLDA3, TIH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLDA2 geni (Tüm Gen Dizi Analizi)	PHLDA2 gene (Sequence Analysis-All Coding Exons)	PHLDA2	PHLDA2, TSSC3, IPL, BRW1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHLDA1 geni (Tüm Gen Dizi Analizi)	PHLDA1 gene (Sequence Analysis-All Coding Exons)	PHLDA1	PHLDA1, TDAG51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHKG1 geni (Tüm Gen Dizi Analizi)	PHKG1 gene (Sequence Analysis-All Coding Exons)	PHKG1	PHKG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHIP geni (Tüm Gen Dizi Analizi)	PHIP gene (Sequence Analysis-All Coding Exons)	PHIP	PHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF3 geni (Tüm Gen Dizi Analizi)	PHF3 gene (Sequence Analysis-All Coding Exons)	PHF3	PHF3, KIAA0244	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF23 geni (Tüm Gen Dizi Analizi)	PHF23 gene (Sequence Analysis-All Coding Exons)	PHF23	PHF23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF21B geni (Tüm Gen Dizi Analizi)	PHF21B gene (Sequence Analysis-All Coding Exons)	PHF21B	PHF21B, PHF4, BHC80L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF21A geni (Tüm Gen Dizi Analizi)	PHF21A gene (Sequence Analysis-All Coding Exons)	PHF21A	PHF21A, BHC80, KIAA1696	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF20 geni (Tüm Gen Dizi Analizi)	PHF20 gene (Sequence Analysis-All Coding Exons)	PHF20	PHF20, GLEA2, HCA58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF2 geni (Tüm Gen Dizi Analizi)	PHF2 gene (Sequence Analysis-All Coding Exons)	PHF2	PHF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF19 geni (Tüm Gen Dizi Analizi)	PHF19 gene (Sequence Analysis-All Coding Exons)	PHF19	PHF19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF10 geni (Tüm Gen Dizi Analizi)	PHF10 gene (Sequence Analysis-All Coding Exons)	PHF10	PHF10, BAF45A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHF1 geni (Tüm Gen Dizi Analizi)	PHF1 gene (Sequence Analysis-All Coding Exons)	PHF1	PHF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHB2 geni (Tüm Gen Dizi Analizi)	PHB2 gene (Sequence Analysis-All Coding Exons)	PHB2	PHB2, REA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHAX geni (Tüm Gen Dizi Analizi)	PHAX gene (Sequence Analysis-All Coding Exons)	PHAX	RNUXA, PHAX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHACTR4 geni (Tüm Gen Dizi Analizi)	PHACTR4 gene (Sequence Analysis-All Coding Exons)	PHACTR4	PHACTR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHACTR3 geni (Tüm Gen Dizi Analizi)	PHACTR3 gene (Sequence Analysis-All Coding Exons)	PHACTR3	PHACTR3, SCAPINN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHACTR2 geni (Tüm Gen Dizi Analizi)	PHACTR2 gene (Sequence Analysis-All Coding Exons)	PHACTR2	PHACTR2, KIAA0680	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHACTR1 geni (Tüm Gen Dizi Analizi)	PHACTR1 gene (Sequence Analysis-All Coding Exons)	PHACTR1	PHACTR1, KIAA1733	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGS1 geni (Tüm Gen Dizi Analizi)	PGS1 gene (Sequence Analysis-All Coding Exons)	PGS1	PGS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGRMC2 geni (Tüm Gen Dizi Analizi)	PGRMC2 gene (Sequence Analysis-All Coding Exons)	PGRMC2	PGRMC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGRMC1 geni (Tüm Gen Dizi Analizi)	PGRMC1 gene (Sequence Analysis-All Coding Exons)	PGRMC1	PGRMC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGPEP1 geni (Tüm Gen Dizi Analizi)	PGPEP1 gene (Sequence Analysis-All Coding Exons)	PGPEP1	PGPEP1, PCP, PGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGP geni (Tüm Gen Dizi Analizi)	PGP gene (Sequence Analysis-All Coding Exons)	PGP	PGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGM5 geni (Tüm Gen Dizi Analizi)	PGM5 gene (Sequence Analysis-All Coding Exons)	PGM5	PGM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGM2L1 geni (Tüm Gen Dizi Analizi)	PGM2L1 gene (Sequence Analysis-All Coding Exons)	PGM2L1	PGM2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGM2 geni (Tüm Gen Dizi Analizi)	PGM2 gene (Sequence Analysis-All Coding Exons)	PGM2	PGM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGLYRP4 geni (Tüm Gen Dizi Analizi)	PGLYRP4 gene (Sequence Analysis-All Coding Exons)	PGLYRP4	PGRPIB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGLYRP3 geni (Tüm Gen Dizi Analizi)	PGLYRP3 gene (Sequence Analysis-All Coding Exons)	PGLYRP3	PGRPIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGLYRP2 geni (Tüm Gen Dizi Analizi)	PGLYRP2 gene (Sequence Analysis-All Coding Exons)	PGLYRP2	PGRPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGLYRP1 geni (Tüm Gen Dizi Analizi)	PGLYRP1 gene (Sequence Analysis-All Coding Exons)	PGLYRP1	PGRP, PGRPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGLS geni (Tüm Gen Dizi Analizi)	PGLS gene (Sequence Analysis-All Coding Exons)	PGLS	PGLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGK2 geni (Tüm Gen Dizi Analizi)	PGK2 gene (Sequence Analysis-All Coding Exons)	PGK2	PGK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGGT1B geni (Tüm Gen Dizi Analizi)	PGGT1B gene (Sequence Analysis-All Coding Exons)	PGGT1B	PGGT1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGGHG geni (Tüm Gen Dizi Analizi)	PGGHG gene (Sequence Analysis-All Coding Exons)	PGGHG	PGGHG, ATHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGF geni (Tüm Gen Dizi Analizi)	PGF gene (Sequence Analysis-All Coding Exons)	PGF	PGF, PLGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGD geni (Tüm Gen Dizi Analizi)	PGD gene (Sequence Analysis-All Coding Exons)	PGD	PGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGC geni (Tüm Gen Dizi Analizi)	PGC gene (Sequence Analysis-All Coding Exons)	PGC	PGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGBD5 geni (Tüm Gen Dizi Analizi)	PGBD5 gene (Sequence Analysis-All Coding Exons)	PGBD5	PGBD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGAM5 geni (Tüm Gen Dizi Analizi)	PGAM5 gene (Sequence Analysis-All Coding Exons)	PGAM5	PGAM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGAM4 geni (Tüm Gen Dizi Analizi)	PGAM4 gene (Sequence Analysis-All Coding Exons)	PGAM4	PGAM4, PGAM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGAM1 geni (Tüm Gen Dizi Analizi)	PGAM1 gene (Sequence Analysis-All Coding Exons)	PGAM1	PGAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGA5 geni (Tüm Gen Dizi Analizi)	PGA5 gene (Sequence Analysis-All Coding Exons)	PGA5	PGA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PGA4 geni (Tüm Gen Dizi Analizi)	PGA4 gene (Sequence Analysis-All Coding Exons)	PGA4	PGA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFN3 geni (Tüm Gen Dizi Analizi)	PFN3 gene (Sequence Analysis-All Coding Exons)	PFN3	PFN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFN2 geni (Tüm Gen Dizi Analizi)	PFN2 gene (Sequence Analysis-All Coding Exons)	PFN2	PFN2, PFL, D3S1319E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFN1P2 geni (Tüm Gen Dizi Analizi)	PFN1P2 gene (Sequence Analysis-All Coding Exons)	PFN1P2	C1orf152, COAS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFKP geni (Tüm Gen Dizi Analizi)	PFKP gene (Sequence Analysis-All Coding Exons)	PFKP	PFKP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFKFB4 geni (Tüm Gen Dizi Analizi)	PFKFB4 gene (Sequence Analysis-All Coding Exons)	PFKFB4	PFKFB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFKFB3 geni (Tüm Gen Dizi Analizi)	PFKFB3 gene (Sequence Analysis-All Coding Exons)	PFKFB3	PFKFB3, IPFK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFKFB2 geni (Tüm Gen Dizi Analizi)	PFKFB2 gene (Sequence Analysis-All Coding Exons)	PFKFB2	PFKFB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFKFB1 geni (Tüm Gen Dizi Analizi)	PFKFB1 gene (Sequence Analysis-All Coding Exons)	PFKFB1	PFKFB1, PFRX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFDN5 geni (Tüm Gen Dizi Analizi)	PFDN5 gene (Sequence Analysis-All Coding Exons)	PFDN5	PFDN5, MM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFDN4 geni (Tüm Gen Dizi Analizi)	PFDN4 gene (Sequence Analysis-All Coding Exons)	PFDN4	PFDN4, C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFDN2 geni (Tüm Gen Dizi Analizi)	PFDN2 gene (Sequence Analysis-All Coding Exons)	PFDN2	PFDN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFDN1 geni (Tüm Gen Dizi Analizi)	PFDN1 gene (Sequence Analysis-All Coding Exons)	PFDN1	PFDN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFAS geni (Tüm Gen Dizi Analizi)	PFAS gene (Sequence Analysis-All Coding Exons)	PFAS	PFAS, FGARAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PF4V1 geni (Tüm Gen Dizi Analizi)	PF4V1 gene (Sequence Analysis-All Coding Exons)	PF4V1	PF4V1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PF4 geni (Tüm Gen Dizi Analizi)	PF4 gene (Sequence Analysis-All Coding Exons)	PF4	PF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEXSL geni (Tüm Gen Dizi Analizi)	PEXSL gene (Sequence Analysis-All Coding Exons)	PEXSL	PEXSL, PEXSR, PXR2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEX11G geni (Tüm Gen Dizi Analizi)	PEX11G gene (Sequence Analysis-All Coding Exons)	PEX11G	PEX11G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEX11A geni (Tüm Gen Dizi Analizi)	PEX11A gene (Sequence Analysis-All Coding Exons)	PEX11A	PEX11A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PET117 geni (Tüm Gen Dizi Analizi)	PET117 gene (Sequence Analysis-All Coding Exons)	PET117	PET117	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PES1 geni (Tüm Gen Dizi Analizi)	PES1 gene (Sequence Analysis-All Coding Exons)	PES1	PES1, PES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PERP geni (Tüm Gen Dizi Analizi)	PERP gene (Sequence Analysis-All Coding Exons)	PERP	PERP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PERM1 geni (Tüm Gen Dizi Analizi)	PERM1 gene (Sequence Analysis-All Coding Exons)	PERM1	PERM1, C1orf170	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PER1 geni (Tüm Gen Dizi Analizi)	PER1 gene (Sequence Analysis-All Coding Exons)	PER1	PER, RIGUI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PENK geni (Tüm Gen Dizi Analizi)	PENK gene (Sequence Analysis-All Coding Exons)	PENK	PENK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEMT geni (Tüm Gen Dizi Analizi)	PEMT gene (Sequence Analysis-All Coding Exons)	PEMT	PEMT, PEMPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PELP1 geni (Tüm Gen Dizi Analizi)	PELP1 gene (Sequence Analysis-All Coding Exons)	PELP1	PELP1, MNAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PELO geni (Tüm Gen Dizi Analizi)	PELO gene (Sequence Analysis-All Coding Exons)	PELO	PELO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PELI3 geni (Tüm Gen Dizi Analizi)	PELI3 gene (Sequence Analysis-All Coding Exons)	PELI3	PELI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PELI2 geni (Tüm Gen Dizi Analizi)	PELI2 gene (Sequence Analysis-All Coding Exons)	PELI2	PELI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PELI1 geni (Tüm Gen Dizi Analizi)	PELI1 gene (Sequence Analysis-All Coding Exons)	PELI1	PELI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEG3 geni (Tüm Gen Dizi Analizi)	PEG3 gene (Sequence Analysis-All Coding Exons)	PEG3	PEG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEG10 geni (Tüm Gen Dizi Analizi)	PEG10 gene (Sequence Analysis-All Coding Exons)	PEG10	PEG10, KIAA1051	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEF1 geni (Tüm Gen Dizi Analizi)	PEF1 gene (Sequence Analysis-All Coding Exons)	PEF1	PEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PECR geni (Tüm Gen Dizi Analizi)	PECR gene (Sequence Analysis-All Coding Exons)	PECR	PECR, TERP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PECAM1 geni (Tüm Gen Dizi Analizi)	PECAM1 gene (Sequence Analysis-All Coding Exons)	PECAM1	PECAM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEBP4 geni (Tüm Gen Dizi Analizi)	PEBP4 gene (Sequence Analysis-All Coding Exons)	PEBP4	PEBP4, CORK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEBP1 geni (Tüm Gen Dizi Analizi)	PEBP1 gene (Sequence Analysis-All Coding Exons)	PEBP1	PEBP1, PBP, RKIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEAR1 geni (Tüm Gen Dizi Analizi)	PEAR1 gene (Sequence Analysis-All Coding Exons)	PEAR1	PEAR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEAK1 geni (Tüm Gen Dizi Analizi)	PEAK1 gene (Sequence Analysis-All Coding Exons)	PEAK1	PEAK1, KIAA2002	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PEA15 geni (Tüm Gen Dizi Analizi)	PEA15 gene (Sequence Analysis-All Coding Exons)	PEA15	PEA15, HMAT1, PED	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZRN4 geni (Tüm Gen Dizi Analizi)	PDZRN4 gene (Sequence Analysis-All Coding Exons)	PDZRN4	PDZRN4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZRN3 geni (Tüm Gen Dizi Analizi)	PDZRN3 gene (Sequence Analysis-All Coding Exons)	PDZRN3	PDZRN3, KIAA1095	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZK1IP1 geni (Tüm Gen Dizi Analizi)	PDZK1IP1 gene (Sequence Analysis-All Coding Exons)	PDZK1IP1	PDZK1IP1, MAP17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZK1 geni (Tüm Gen Dizi Analizi)	PDZK1 gene (Sequence Analysis-All Coding Exons)	PDZK1	PDZK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZD8 geni (Tüm Gen Dizi Analizi)	PDZD8 gene (Sequence Analysis-All Coding Exons)	PDZD8	PDZD8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZD4 geni (Tüm Gen Dizi Analizi)	PDZD4 gene (Sequence Analysis-All Coding Exons)	PDZD4	PDZD4, PDZK4, PDZRN4L, KIAA1444, LIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZD3 geni (Tüm Gen Dizi Analizi)	PDZD3 gene (Sequence Analysis-All Coding Exons)	PDZD3	PDZD3, IKEPP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZD2 geni (Tüm Gen Dizi Analizi)	PDZD2 gene (Sequence Analysis-All Coding Exons)	PDZD2	PDZD2, PAPIN, PIN1, AIPC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDZD11 geni (Tüm Gen Dizi Analizi)	PDZD11 gene (Sequence Analysis-All Coding Exons)	PDZD11	PDZD11, AIPP1, PISP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDXK geni (Tüm Gen Dizi Analizi)	PDXK gene (Sequence Analysis-All Coding Exons)	PDXK	PDXK, PNK, PKH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDXDC1 geni (Tüm Gen Dizi Analizi)	PDXDC1 gene (Sequence Analysis-All Coding Exons)	PDXDC1	PDXDC1, KIAA0251	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDSSB geni (Tüm Gen Dizi Analizi)	PDSSB gene (Sequence Analysis-All Coding Exons)	PDSSB	AS3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDS5A geni (Tüm Gen Dizi Analizi)	PDS5A gene (Sequence Analysis-All Coding Exons)	PDS5A	PDS5A, KIAA0648, SCC112	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDRG1 geni (Tüm Gen Dizi Analizi)	PDRG1 gene (Sequence Analysis-All Coding Exons)	PDRG1	PDRG1, PDRG, C20orf126	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDPN geni (Tüm Gen Dizi Analizi)	PDPN gene (Sequence Analysis-All Coding Exons)	PDPN	PDPN, T1LA, T1A2, GP36, OTS8, aCG61.6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDPK1 geni (Tüm Gen Dizi Analizi)	PDPK1 gene (Sequence Analysis-All Coding Exons)	PDPK1	PDPK1, PDK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDP2 geni (Tüm Gen Dizi Analizi)	PDP2 gene (Sequence Analysis-All Coding Exons)	PDP2	PDP2, KIAA1348	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDLIM7 geni (Tüm Gen Dizi Analizi)	PDLIM7 gene (Sequence Analysis-All Coding Exons)	PDLIM7	PDLIM7, ENIGMA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDLIM5 geni (Tüm Gen Dizi Analizi)	PDLIM5 gene (Sequence Analysis-All Coding Exons)	PDLIM5	ENH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDLIM3 geni (Tüm Gen Dizi Analizi)	PDLIM3 gene (Sequence Analysis-All Coding Exons)	PDLIM3	ALP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDLIM2 geni (Tüm Gen Dizi Analizi)	PDLIM2 gene (Sequence Analysis-All Coding Exons)	PDLIM2	PDLIM2, MYSTIQUE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDLIM1 geni (Tüm Gen Dizi Analizi)	PDLIM1 gene (Sequence Analysis-All Coding Exons)	PDLIM1	PDLIM1, CLP36, CLIM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDK4 geni (Tüm Gen Dizi Analizi)	PDK4 gene (Sequence Analysis-All Coding Exons)	PDK4	PDK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDK2 geni (Tüm Gen Dizi Analizi)	PDK2 gene (Sequence Analysis-All Coding Exons)	PDK2	PDK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDK1 geni (Tüm Gen Dizi Analizi)	PDK1 gene (Sequence Analysis-All Coding Exons)	PDK1	PDK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDIK1L geni (Tüm Gen Dizi Analizi)	PDIK1L gene (Sequence Analysis-All Coding Exons)	PDIK1L	PDIK1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDIA6 geni (Tüm Gen Dizi Analizi)	PDIA6 gene (Sequence Analysis-All Coding Exons)	PDIA6	PDIA6, ERP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDIA5 geni (Tüm Gen Dizi Analizi)	PDIA5 gene (Sequence Analysis-All Coding Exons)	PDIA5	PDIA5, PDIR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDIA3 geni (Tüm Gen Dizi Analizi)	PDIA3 gene (Sequence Analysis-All Coding Exons)	PDIA3	GRP58	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDIA2 geni (Tüm Gen Dizi Analizi)	PDIA2 gene (Sequence Analysis-All Coding Exons)	PDIA2	PDIA2, PDIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDHA2 geni (Tüm Gen Dizi Analizi)	PDHA2 gene (Sequence Analysis-All Coding Exons)	PDHA2	PDHA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDGFD geni (Tüm Gen Dizi Analizi)	PDGFD gene (Sequence Analysis-All Coding Exons)	PDGFD	PDGFD, SCDGFB, IEGF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDGFC geni (Tüm Gen Dizi Analizi)	PDGFC gene (Sequence Analysis-All Coding Exons)	PDGFC	PDGFC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDGFA geni (Tüm Gen Dizi Analizi)	PDGFA gene (Sequence Analysis-All Coding Exons)	PDGFA	PDGFA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE9A geni (Tüm Gen Dizi Analizi)	PDE9A gene (Sequence Analysis-All Coding Exons)	PDE9A	PDE9A, HSPDE9A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE8A geni (Tüm Gen Dizi Analizi)	PDE8A gene (Sequence Analysis-All Coding Exons)	PDE8A	PDE8A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE7B geni (Tüm Gen Dizi Analizi)	PDE7B gene (Sequence Analysis-All Coding Exons)	PDE7B	PDE7B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE7A geni (Tüm Gen Dizi Analizi)	PDE7A gene (Sequence Analysis-All Coding Exons)	PDE7A	PDE7A, HCP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE5A geni (Tüm Gen Dizi Analizi)	PDE5A gene (Sequence Analysis-All Coding Exons)	PDE5A	PDE5A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE4DIP geni (Tüm Gen Dizi Analizi)	PDE4DIP gene (Sequence Analysis-All Coding Exons)	PDE4DIP	PDE4DIP, MMGL, KIAA0454, KIAA0477	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE4C geni (Tüm Gen Dizi Analizi)	PDE4C gene (Sequence Analysis-All Coding Exons)	PDE4C	PDE4C, DPDE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE4B geni (Tüm Gen Dizi Analizi)	PDE4B gene (Sequence Analysis-All Coding Exons)	PDE4B	PDE4B, DPDE4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RXFP4 geni (Tüm Gen Dizi Analizi)	RXFP4 gene (Sequence Analysis-All Coding Exons)	RXFP4		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE4A geni (Tüm Gen Dizi Analizi)	PDE4A gene (Sequence Analysis-All Coding Exons)	PDE4A	PDE4A, DPDE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE3B geni (Tüm Gen Dizi Analizi)	PDE3B gene (Sequence Analysis-All Coding Exons)	PDE3B	PDE3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE2A geni (Tüm Gen Dizi Analizi)	PDE2A gene (Sequence Analysis-All Coding Exons)	PDE2A	PDE2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE1C geni (Tüm Gen Dizi Analizi)	PDE1C gene (Sequence Analysis-All Coding Exons)	PDE1C	PDE1C, HCAM3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE1B geni (Tüm Gen Dizi Analizi)	PDE1B gene (Sequence Analysis-All Coding Exons)	PDE1B	PDE1B1, PDE51B, PDE1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE1A geni (Tüm Gen Dizi Analizi)	PDE1A gene (Sequence Analysis-All Coding Exons)	PDE1A	PDE1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDE12 geni (Tüm Gen Dizi Analizi)	PDE12 gene (Sequence Analysis-All Coding Exons)	PDE12	PDE12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCL3 geni (Tüm Gen Dizi Analizi)	PDCL3 gene (Sequence Analysis-All Coding Exons)	PDCL3	PDCL3, PHLP3, VIAF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCL2 geni (Tüm Gen Dizi Analizi)	PDCL2 gene (Sequence Analysis-All Coding Exons)	PDCL2	PDCL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCL geni (Tüm Gen Dizi Analizi)	PDCL gene (Sequence Analysis-All Coding Exons)	PDCL	PDCL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD7 geni (Tüm Gen Dizi Analizi)	PDCD7 gene (Sequence Analysis-All Coding Exons)	PDCD7	PDCD7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD6IP geni (Tüm Gen Dizi Analizi)	PDCD6IP gene (Sequence Analysis-All Coding Exons)	PDCD6IP	PDCD6IP, AIP1, ALIX, KIAA1375	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD6 geni (Tüm Gen Dizi Analizi)	PDCD6 gene (Sequence Analysis-All Coding Exons)	PDCD6	PDCD6, ALG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD5 geni (Tüm Gen Dizi Analizi)	PDCD5 gene (Sequence Analysis-All Coding Exons)	PDCD5	PDCD5, TFAR19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD4 geni (Tüm Gen Dizi Analizi)	PDCD4 gene (Sequence Analysis-All Coding Exons)	PDCD4	PDCD4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD2L geni (Tüm Gen Dizi Analizi)	PDCD2L gene (Sequence Analysis-All Coding Exons)	PDCD2L	PDCD2L, MGC13096	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD2 geni (Tüm Gen Dizi Analizi)	PDCD2 gene (Sequence Analysis-All Coding Exons)	PDCD2	PDCD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD1LG2 geni (Tüm Gen Dizi Analizi)	PDCD1LG2 gene (Sequence Analysis-All Coding Exons)	PDCD1LG2	PDCD1LG2, PDL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDCD11 geni (Tüm Gen Dizi Analizi)	PDCD11 gene (Sequence Analysis-All Coding Exons)	PDCD11	PDCD11, ALG4, NFBP, KIAA0185	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDC geni (Tüm Gen Dizi Analizi)	PDC gene (Sequence Analysis-All Coding Exons)	PDC	PDC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDAP1 geni (Tüm Gen Dizi Analizi)	PDAP1 gene (Sequence Analysis-All Coding Exons)	PDAP1	PDAP1, PAP1, PAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCYT1B geni (Tüm Gen Dizi Analizi)	PCYT1B gene (Sequence Analysis-All Coding Exons)	PCYT1B	PCYT1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCYOX1 geni (Tüm Gen Dizi Analizi)	PCYOX1 gene (Sequence Analysis-All Coding Exons)	PCYOX1	PCYOX1, PCL1, KIAA0908	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCTP geni (Tüm Gen Dizi Analizi)	PCTP gene (Sequence Analysis-All Coding Exons)	PCTP	PCTP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK7 geni (Tüm Gen Dizi Analizi)	PCSK7 gene (Sequence Analysis-All Coding Exons)	PCSK7	PCSK7, PC8, PC7, LPC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK6 geni (Tüm Gen Dizi Analizi)	PCSK6 gene (Sequence Analysis-All Coding Exons)	PCSK6	PCSK6, PACE4, SPC4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK5 geni (Tüm Gen Dizi Analizi)	PCSK5 gene (Sequence Analysis-All Coding Exons)	PCSK5	PCSK5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK4 geni (Tüm Gen Dizi Analizi)	PCSK4 gene (Sequence Analysis-All Coding Exons)	PCSK4	PCSK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK2 geni (Tüm Gen Dizi Analizi)	PCSK2 gene (Sequence Analysis-All Coding Exons)	PCSK2	PCSK2, NEC2, PC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCSK1N geni (Tüm Gen Dizi Analizi)	PCSK1N gene (Sequence Analysis-All Coding Exons)	PCSK1N	PCSK1N, PROSAAS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCP4 geni (Tüm Gen Dizi Analizi)	PCP4 gene (Sequence Analysis-All Coding Exons)	PCP4	PCP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCOTH geni (Tüm Gen Dizi Analizi)	PCOTH gene (Sequence Analysis-All Coding Exons)	PCOTH	C1QTNF9BAS1, PCOTH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCOLCE2 geni (Tüm Gen Dizi Analizi)	PCOLCE2 gene (Sequence Analysis-All Coding Exons)	PCOLCE2	PCOLCE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCOLCE geni (Tüm Gen Dizi Analizi)	PCOLCE gene (Sequence Analysis-All Coding Exons)	PCOLCE	PCOLCE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCNP geni (Tüm Gen Dizi Analizi)	PCNP gene (Sequence Analysis-All Coding Exons)	PCNP	PCNP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCMT1 geni (Tüm Gen Dizi Analizi)	PCMT1 gene (Sequence Analysis-All Coding Exons)	PCMT1	PCMT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCM1 geni (Tüm Gen Dizi Analizi)	PCM1 gene (Sequence Analysis-All Coding Exons)	PCM1	PCM1, PTC4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCLAF geni (Tüm Gen Dizi Analizi)	PCLAF gene (Sequence Analysis-All Coding Exons)	PCLAF	PAF, OEATC1, KIAA0101	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCID2 geni (Tüm Gen Dizi Analizi)	PCID2 gene (Sequence Analysis-All Coding Exons)	PCID2	PCID2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCGF6 geni (Tüm Gen Dizi Analizi)	PCGF6 gene (Sequence Analysis-All Coding Exons)	PCGF6	PCGF6, RNF134, MBLR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCGF2 geni (Tüm Gen Dizi Analizi)	PCGF2 gene (Sequence Analysis-All Coding Exons)	PCGF2	RNF110, ZNF144, MEL18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCGEM1 geni (Tüm Gen Dizi Analizi)	PCGEM1 gene (Sequence Analysis-All Coding Exons)	PCGEM1	PCGEM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCF11 geni (Tüm Gen Dizi Analizi)	PCF11 gene (Sequence Analysis-All Coding Exons)	PCF11	PCF11, KIAA0824	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PCDHGC5 geni (Tüm Gen Dizi Analizi)	PCDHGC5 gene (Sequence Analysis-All Coding Exons)	PCDHGC5	PCDHGC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGC4 geni (Tüm Gen Dizi Analizi)	PCDHGC4 gene (Sequence Analysis-All Coding Exons)	PCDHGC4	PCDHGC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGC3 geni (Tüm Gen Dizi Analizi)	PCDHGC3 gene (Sequence Analysis-All Coding Exons)	PCDHGC3	PCDHGC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB7 geni (Tüm Gen Dizi Analizi)	PCDHGB7 gene (Sequence Analysis-All Coding Exons)	PCDHGB7	PCDHGB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB6 geni (Tüm Gen Dizi Analizi)	PCDHGB6 gene (Sequence Analysis-All Coding Exons)	PCDHGB6	PCDHGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB5 geni (Tüm Gen Dizi Analizi)	PCDHGB5 gene (Sequence Analysis-All Coding Exons)	PCDHGB5	PCDHGB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB4 geni (Tüm Gen Dizi Analizi)	PCDHGB4 gene (Sequence Analysis-All Coding Exons)	PCDHGB4	PCDHGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB3 geni (Tüm Gen Dizi Analizi)	PCDHGB3 gene (Sequence Analysis-All Coding Exons)	PCDHGB3	PCDHGB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB2 geni (Tüm Gen Dizi Analizi)	PCDHGB2 gene (Sequence Analysis-All Coding Exons)	PCDHGB2	PCDHGB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGB1 geni (Tüm Gen Dizi Analizi)	PCDHGB1 gene (Sequence Analysis-All Coding Exons)	PCDHGB1	PCDHGB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA9 geni (Tüm Gen Dizi Analizi)	PCDHGA9 gene (Sequence Analysis-All Coding Exons)	PCDHGA9	PCDHGA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA8 geni (Tüm Gen Dizi Analizi)	PCDHGA8 gene (Sequence Analysis-All Coding Exons)	PCDHGA8	PCDHGA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA7 geni (Tüm Gen Dizi Analizi)	PCDHGA7 gene (Sequence Analysis-All Coding Exons)	PCDHGA7	PCDHGA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA6 geni (Tüm Gen Dizi Analizi)	PCDHGA6 gene (Sequence Analysis-All Coding Exons)	PCDHGA6	PCDHGA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA5 geni (Tüm Gen Dizi Analizi)	PCDHGA5 gene (Sequence Analysis-All Coding Exons)	PCDHGA5	PCDHGA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA4 geni (Tüm Gen Dizi Analizi)	PCDHGA4 gene (Sequence Analysis-All Coding Exons)	PCDHGA4	PCDHGA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA3 geni (Tüm Gen Dizi Analizi)	PCDHGA3 gene (Sequence Analysis-All Coding Exons)	PCDHGA3	PCDHGA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA2 geni (Tüm Gen Dizi Analizi)	PCDHGA2 gene (Sequence Analysis-All Coding Exons)	PCDHGA2	PCDHGA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA12 geni (Tüm Gen Dizi Analizi)	PCDHGA12 gene (Sequence Analysis-All Coding Exons)	PCDHGA12	PCDHGA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA11 geni (Tüm Gen Dizi Analizi)	PCDHGA11 gene (Sequence Analysis-All Coding Exons)	PCDHGA11	PCDHGA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA10 geni (Tüm Gen Dizi Analizi)	PCDHGA10 gene (Sequence Analysis-All Coding Exons)	PCDHGA10	PCDHGA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHGA1 geni (Tüm Gen Dizi Analizi)	PCDHGA1 gene (Sequence Analysis-All Coding Exons)	PCDHGA1	PCDHGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB9 geni (Tüm Gen Dizi Analizi)	PCDHB9 gene (Sequence Analysis-All Coding Exons)	PCDHB9	PCDHB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB8 geni (Tüm Gen Dizi Analizi)	PCDHB8 gene (Sequence Analysis-All Coding Exons)	PCDHB8	PCDHB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB7 geni (Tüm Gen Dizi Analizi)	PCDHB7 gene (Sequence Analysis-All Coding Exons)	PCDHB7	PCDHB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB6 geni (Tüm Gen Dizi Analizi)	PCDHB6 gene (Sequence Analysis-All Coding Exons)	PCDHB6	PCDHB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB5 geni (Tüm Gen Dizi Analizi)	PCDHB5 gene (Sequence Analysis-All Coding Exons)	PCDHB5	PCDHB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB4 geni (Tüm Gen Dizi Analizi)	PCDHB4 gene (Sequence Analysis-All Coding Exons)	PCDHB4	PCDHB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB3 geni (Tüm Gen Dizi Analizi)	PCDHB3 gene (Sequence Analysis-All Coding Exons)	PCDHB3	PCDHB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB2 geni (Tüm Gen Dizi Analizi)	PCDHB2 gene (Sequence Analysis-All Coding Exons)	PCDHB2	PCDHB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RRP1 geni (Tüm Gen Dizi Analizi)	RRP1 gene (Sequence Analysis-All Coding Exons)	RRP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB16 geni (Tüm Gen Dizi Analizi)	PCDHB16 gene (Sequence Analysis-All Coding Exons)	PCDHB16	PCDHB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB15 geni (Tüm Gen Dizi Analizi)	PCDHB15 gene (Sequence Analysis-All Coding Exons)	PCDHB15	PCDHB15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB14 geni (Tüm Gen Dizi Analizi)	PCDHB14 gene (Sequence Analysis-All Coding Exons)	PCDHB14	PCDHB14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB13 geni (Tüm Gen Dizi Analizi)	PCDHB13 gene (Sequence Analysis-All Coding Exons)	PCDHB13	PCDHB13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB12 geni (Tüm Gen Dizi Analizi)	PCDHB12 gene (Sequence Analysis-All Coding Exons)	PCDHB12	PCDHB12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB11 geni (Tüm Gen Dizi Analizi)	PCDHB11 gene (Sequence Analysis-All Coding Exons)	PCDHB11	PCDHB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB10 geni (Tüm Gen Dizi Analizi)	PCDHB10 gene (Sequence Analysis-All Coding Exons)	PCDHB10	PCHB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHB1 geni (Tüm Gen Dizi Analizi)	PCDHB1 gene (Sequence Analysis-All Coding Exons)	PCDHB1	PCDHB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHAC2 geni (Tüm Gen Dizi Analizi)	PCDHAC2 gene (Sequence Analysis-All Coding Exons)	PCDHAC2	PCDHAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHAC1 geni (Tüm Gen Dizi Analizi)	PCDHAC1 gene (Sequence Analysis-All Coding Exons)	PCDHAC1	PCDHAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA9 geni (Tüm Gen Dizi Analizi)	PCDHA9 gene (Sequence Analysis-All Coding Exons)	PCDHA9	PCDHA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA8 geni (Tüm Gen Dizi Analizi)	PCDHA8 gene (Sequence Analysis-All Coding Exons)	PCDHA8	PCDHA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA7 geni (Tüm Gen Dizi Analizi)	PCDHA7 gene (Sequence Analysis-All Coding Exons)	PCDHA7	PCDHA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA6 geni (Tüm Gen Dizi Analizi)	PCDHA6 gene (Sequence Analysis-All Coding Exons)	PCDHA6	PCDHA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA5 geni (Tüm Gen Dizi Analizi)	PCDHA5 gene (Sequence Analysis-All Coding Exons)	PCDHA5	PCDHA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA4 geni (Tüm Gen Dizi Analizi)	PCDHA4 gene (Sequence Analysis-All Coding Exons)	PCDHA4	PCDHA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA3 geni (Tüm Gen Dizi Analizi)	PCDHA3 gene (Sequence Analysis-All Coding Exons)	PCDHA3	PCDHA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA2 geni (Tüm Gen Dizi Analizi)	PCDHA2 gene (Sequence Analysis-All Coding Exons)	PCDHA2	PCDHA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA13 geni (Tüm Gen Dizi Analizi)	PCDHA13 gene (Sequence Analysis-All Coding Exons)	PCDHA13	PCDHA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA12 geni (Tüm Gen Dizi Analizi)	PCDHA12 gene (Sequence Analysis-All Coding Exons)	PCDHA12	PCDHA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA11 geni (Tüm Gen Dizi Analizi)	PCDHA11 gene (Sequence Analysis-All Coding Exons)	PCDHA11	PCDHA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA10 geni (Tüm Gen Dizi Analizi)	PCDHA10 gene (Sequence Analysis-All Coding Exons)	PCDHA10	PCDHA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RPS6KB2 geni (Tüm Gen Dizi Analizi)	RPS6KB2 gene (Sequence Analysis-All Coding Exons)	RPS6KB2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDHA1 geni (Tüm Gen Dizi Analizi)	PCDHA1 gene (Sequence Analysis-All Coding Exons)	PCDHA1	PCDHA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH9 geni (Tüm Gen Dizi Analizi)	PCDH9 gene (Sequence Analysis-All Coding Exons)	PCDH9	PCDH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH8 geni (Tüm Gen Dizi Analizi)	PCDH8 gene (Sequence Analysis-All Coding Exons)	PCDH8	PCDH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH7 geni (Tüm Gen Dizi Analizi)	PCDH7 gene (Sequence Analysis-All Coding Exons)	PCDH7	PCDH7, BHPCDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH20 geni (Tüm Gen Dizi Analizi)	PCDH20 gene (Sequence Analysis-All Coding Exons)	PCDH20	PCDH20, PCDH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH18 geni (Tüm Gen Dizi Analizi)	PCDH18 gene (Sequence Analysis-All Coding Exons)	PCDH18	PCDH18, KIAA1562	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH17 geni (Tüm Gen Dizi Analizi)	PCDH17 gene (Sequence Analysis-All Coding Exons)	PCDH17	PCDH17, PCDH68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH12 geni (Tüm Gen Dizi Analizi)	PCDH12 gene (Sequence Analysis-All Coding Exons)	PCDH12	PCDH12, VECAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH11Y geni (Tüm Gen Dizi Analizi)	PCDH11Y gene (Sequence Analysis-All Coding Exons)	PCDH11Y	PCDH11Y, PCDH22, PCDHY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH11X geni (Tüm Gen Dizi Analizi)	PCDH11X gene (Sequence Analysis-All Coding Exons)	PCDH11X	PCDH11X, PCDH11, PCDHX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH10 geni (Tüm Gen Dizi Analizi)	PCDH10 gene (Sequence Analysis-All Coding Exons)	PCDH10	PCDH10, KIAA1400	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCDH1 geni (Tüm Gen Dizi Analizi)	PCDH1 gene (Sequence Analysis-All Coding Exons)	PCDH1	PCDH1, PC42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCBP4 geni (Tüm Gen Dizi Analizi)	PCBP4 gene (Sequence Analysis-All Coding Exons)	PCBP4	PCBP4, MGC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCBP3 geni (Tüm Gen Dizi Analizi)	PCBP3 gene (Sequence Analysis-All Coding Exons)	PCBP3	PCBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCBP2 geni (Tüm Gen Dizi Analizi)	PCBP2 gene (Sequence Analysis-All Coding Exons)	PCBP2	PCBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCBP1 geni (Tüm Gen Dizi Analizi)	PCBP1 gene (Sequence Analysis-All Coding Exons)	PCBP1	PCBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCBD2 geni (Tüm Gen Dizi Analizi)	PCBD2 gene (Sequence Analysis-All Coding Exons)	PCBD2	PCBD2, DCOH2, DCOHM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCAT4 geni (Tüm Gen Dizi Analizi)	PCAT4 gene (Sequence Analysis-All Coding Exons)	PCAT4	PCAT4, GDEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCAT1 geni (Tüm Gen Dizi Analizi)	PCAT1 gene (Sequence Analysis-All Coding Exons)	PCAT1	PCAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCA3 geni (Tüm Gen Dizi Analizi)	PCA3 gene (Sequence Analysis-All Coding Exons)	PCA3	PCA3, DD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBX4 geni (Tüm Gen Dizi Analizi)	PBX4 gene (Sequence Analysis-All Coding Exons)	PBX4	PBX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBX3 geni (Tüm Gen Dizi Analizi)	PBX3 gene (Sequence Analysis-All Coding Exons)	PBX3	PBX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBX2 geni (Tüm Gen Dizi Analizi)	PBX2 gene (Sequence Analysis-All Coding Exons)	PBX2	PBX2, HOX12, G17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBRM1 geni (Tüm Gen Dizi Analizi)	PBRM1 gene (Sequence Analysis-All Coding Exons)	PBRM1	PBRM1, PB1, BAF180	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBOV1 geni (Tüm Gen Dizi Analizi)	PBOV1 gene (Sequence Analysis-All Coding Exons)	PBOV1	PBOV1, UROC28, UC28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBLD geni (Tüm Gen Dizi Analizi)	PBLD gene (Sequence Analysis-All Coding Exons)	PBLD	PBLD, MAWBP, MAWDBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PBK geni (Tüm Gen Dizi Analizi)	PBK gene (Sequence Analysis-All Coding Exons)	PBK	PBK, TOPK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAXIP1 geni (Tüm Gen Dizi Analizi)	PAXIP1 gene (Sequence Analysis-All Coding Exons)	PAXIP1	PAXIP1, PAXIP1L, PTIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAWR geni (Tüm Gen Dizi Analizi)	PAWR gene (Sequence Analysis-All Coding Exons)	PAWR	PAWR, PAR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PATZ1 geni (Tüm Gen Dizi Analizi)	PATZ1 gene (Sequence Analysis-All Coding Exons)	PATZ1	ZNF278, PATZ, MAZR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PATL2 geni (Tüm Gen Dizi Analizi)	PATL2 gene (Sequence Analysis-All Coding Exons)	PATL2	PATL2, PAT1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PATL1 geni (Tüm Gen Dizi Analizi)	PATL1 gene (Sequence Analysis-All Coding Exons)	PATL1	PATL1, PAT1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PATJ geni (Tüm Gen Dizi Analizi)	PATJ gene (Sequence Analysis-All Coding Exons)	PATJ	INADL, PATJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PATE1 geni (Tüm Gen Dizi Analizi)	PATE1 gene (Sequence Analysis-All Coding Exons)	PATE1	PATE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PASK geni (Tüm Gen Dizi Analizi)	PASK gene (Sequence Analysis-All Coding Exons)	PASK	PASK, KIAA0135	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARVG geni (Tüm Gen Dizi Analizi)	PARVG gene (Sequence Analysis-All Coding Exons)	PARVG	PARVG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARVB geni (Tüm Gen Dizi Analizi)	PARVB gene (Sequence Analysis-All Coding Exons)	PARVB	PARVB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARVA geni (Tüm Gen Dizi Analizi)	PARVA gene (Sequence Analysis-All Coding Exons)	PARVA	PARVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PART1 geni (Tüm Gen Dizi Analizi)	PART1 gene (Sequence Analysis-All Coding Exons)	PART1	PART1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

PARS2 geni (Tüm Gen Dizi Analizi)	PARS2 gene (Sequence Analysis-All Coding Exons)	PARS2	PARS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARPBP geni (Tüm Gen Dizi Analizi)	PARPBP gene (Sequence Analysis-All Coding Exons)	PARPBP	C12orf48, AROM, PARPBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP9 geni (Tüm Gen Dizi Analizi)	PARP9 gene (Sequence Analysis-All Coding Exons)	PARP9	PARP9, BAL1, BAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP4 geni (Tüm Gen Dizi Analizi)	PARP4 gene (Sequence Analysis-All Coding Exons)	PARP4	PARP4, ADPRTL1, VPARP, KIAA0177	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP3 geni (Tüm Gen Dizi Analizi)	PARP3 gene (Sequence Analysis-All Coding Exons)	PARP3	PARP3, ADPRTL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP2 geni (Tüm Gen Dizi Analizi)	PARP2 gene (Sequence Analysis-All Coding Exons)	PARP2	PARP2, ADPRTL2, ADPRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP15 geni (Tüm Gen Dizi Analizi)	PARP15 gene (Sequence Analysis-All Coding Exons)	PARP15	PARP15, BAL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP14 geni (Tüm Gen Dizi Analizi)	PARP14 gene (Sequence Analysis-All Coding Exons)	PARP14	PARP14, KIAA1268	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP12 geni (Tüm Gen Dizi Analizi)	PARP12 gene (Sequence Analysis-All Coding Exons)	PARP12	PARP12, ZC3HDC1, FLJ22693	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP11 geni (Tüm Gen Dizi Analizi)	PARP11 gene (Sequence Analysis-All Coding Exons)	PARP11	PARP11, ARTD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP1 geni (Tüm Gen Dizi Analizi)	PARP1 gene (Sequence Analysis-All Coding Exons)	PARP1	PARP1, ADPRT, PPOL, PARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARL geni (Tüm Gen Dizi Analizi)	PARL gene (Sequence Analysis-All Coding Exons)	PARL	PARL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARG geni (Tüm Gen Dizi Analizi)	PARG gene (Sequence Analysis-All Coding Exons)	PARG	PARG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARD6G geni (Tüm Gen Dizi Analizi)	PARD6G gene (Sequence Analysis-All Coding Exons)	PARD6G	PARD6G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARD6B geni (Tüm Gen Dizi Analizi)	PARD6B gene (Sequence Analysis-All Coding Exons)	PARD6B	PARD6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARD3 geni (Tüm Gen Dizi Analizi)	PARD3 gene (Sequence Analysis-All Coding Exons)	PARD3	PARD3, PAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR9 geni (Tüm Gen Dizi Analizi)	PAQR9 gene (Sequence Analysis-All Coding Exons)	PAQR9	PAQR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR8 geni (Tüm Gen Dizi Analizi)	PAQR8 gene (Sequence Analysis-All Coding Exons)	PAQR8	PAQR8, C6orf33, LMPB1, MPR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR7 geni (Tüm Gen Dizi Analizi)	PAQR7 gene (Sequence Analysis-All Coding Exons)	PAQR7	PAQR7, MPRA, PGLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR6 geni (Tüm Gen Dizi Analizi)	PAQR6 gene (Sequence Analysis-All Coding Exons)	PAQR6	PAQR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR5 geni (Tüm Gen Dizi Analizi)	PAQR5 gene (Sequence Analysis-All Coding Exons)	PAQR5	PAQR5, MPRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR4 geni (Tüm Gen Dizi Analizi)	PAQR4 gene (Sequence Analysis-All Coding Exons)	PAQR4	PAQR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAQR3 geni (Tüm Gen Dizi Analizi)	PAQR3 gene (Sequence Analysis-All Coding Exons)	PAQR3	PAQR3, RKTG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPSS1 geni (Tüm Gen Dizi Analizi)	PAPSS1 gene (Sequence Analysis-All Coding Exons)	PAPSS1	PAPSS1, ATPSK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPPA-AS1 geni (Tüm Gen Dizi Analizi)	PAPPA-AS1 gene (Sequence Analysis-All Coding Exons)	PAPPA-AS1	DIPAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPPA geni (Tüm Gen Dizi Analizi)	PAPPA gene (Sequence Analysis-All Coding Exons)	PAPPA	PAPPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPOLG geni (Tüm Gen Dizi Analizi)	PAPOLG gene (Sequence Analysis-All Coding Exons)	PAPOLG	PAPOLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPOLB geni (Tüm Gen Dizi Analizi)	PAPOLB gene (Sequence Analysis-All Coding Exons)	PAPOLB	PAPOLB, TPAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPOLA geni (Tüm Gen Dizi Analizi)	PAPOLA gene (Sequence Analysis-All Coding Exons)	PAPOLA	PAPOLA, PAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPD7 geni (Tüm Gen Dizi Analizi)	PAPD7 gene (Sequence Analysis-All Coding Exons)	PAPD7	POLS, TRF4, POLK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPD4 geni (Tüm Gen Dizi Analizi)	PAPD4 gene (Sequence Analysis-All Coding Exons)	PAPD4	PAPD4, GLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAOX geni (Tüm Gen Dizi Analizi)	PAOX gene (Sequence Analysis-All Coding Exons)	PAOX	PAOX, PAO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANX3 geni (Tüm Gen Dizi Analizi)	PANX3 gene (Sequence Analysis-All Coding Exons)	PANX3	PANX3, PX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANX2 geni (Tüm Gen Dizi Analizi)	PANX2 gene (Sequence Analysis-All Coding Exons)	PANX2	PANX2, PX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANX1 geni (Tüm Gen Dizi Analizi)	PANX1 gene (Sequence Analysis-All Coding Exons)	PANX1	PANX1, PX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANK4 geni (Tüm Gen Dizi Analizi)	PANK4 gene (Sequence Analysis-All Coding Exons)	PANK4	PANK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANK3 geni (Tüm Gen Dizi Analizi)	PANK3 gene (Sequence Analysis-All Coding Exons)	PANK3	PANK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANK1 geni (Tüm Gen Dizi Analizi)	PANK1 gene (Sequence Analysis-All Coding Exons)	PANK1	PANK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PANDAR geni (Tüm Gen Dizi Analizi)	PANDAR gene (Sequence Analysis-All Coding Exons)	PANDAR	PANDAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAM geni (Tüm Gen Dizi Analizi)	PAM gene (Sequence Analysis-All Coding Exons)	PAM	PAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PALMD geni (Tüm Gen Dizi Analizi)	PALMD gene (Sequence Analysis-All Coding Exons)	PALMD	PALMD, PALML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PALM geni (Tüm Gen Dizi Analizi)	PALM gene (Sequence Analysis-All Coding Exons)	PALM	PALM, KIAA0270	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PALD1 geni (Tüm Gen Dizi Analizi)	PALD1 gene (Sequence Analysis-All Coding Exons)	PALD1	KIAA1274, PALD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK6 geni (Tüm Gen Dizi Analizi)	PAK6 gene (Sequence Analysis-All Coding Exons)	PAK6	PAK6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK4 geni (Tüm Gen Dizi Analizi)	PAK4 gene (Sequence Analysis-All Coding Exons)	PAK4	PAK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK2 geni (Tüm Gen Dizi Analizi)	PAK2 gene (Sequence Analysis-All Coding Exons)	PAK2	PAK2, PAK65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK1IP1 geni (Tüm Gen Dizi Analizi)	PAK1IP1 gene (Sequence Analysis-All Coding Exons)	PAK1IP1	PAK1IP1, PIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK1 geni (Tüm Gen Dizi Analizi)	PAK1 gene (Sequence Analysis-All Coding Exons)	PAK1	PAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAIP2B geni (Tüm Gen Dizi Analizi)	PAIP2B gene (Sequence Analysis-All Coding Exons)	PAIP2B	PAIP2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAIP2 geni (Tüm Gen Dizi Analizi)	PAIP2 gene (Sequence Analysis-All Coding Exons)	PAIP2	PAIP2, PAIP2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAIP1 geni (Tüm Gen Dizi Analizi)	PAIP1 gene (Sequence Analysis-All Coding Exons)	PAIP1	PAIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAICS geni (Tüm Gen Dizi Analizi)	PAICS gene (Sequence Analysis-All Coding Exons)	PAICS	PAICS, AIRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAGR1 geni (Tüm Gen Dizi Analizi)	PAGR1 gene (Sequence Analysis-All Coding Exons)	PAGR1	PAGR1, C16orf53, PA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAGE4 geni (Tüm Gen Dizi Analizi)	PAGE4 gene (Sequence Analysis-All Coding Exons)	PAGE4	PAGE4, GAGEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAGE3 geni (Tüm Gen Dizi Analizi)	PAGE3 gene (Sequence Analysis-All Coding Exons)	PAGE3	PAGE3, GAGEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAGE2 geni (Tüm Gen Dizi Analizi)	PAGE2 gene (Sequence Analysis-All Coding Exons)	PAGE2	PAGE2, GAGEC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAGE1 geni (Tüm Gen Dizi Analizi)	PAGE1 gene (Sequence Analysis-All Coding Exons)	PAGE1	GAGEB1, PAGE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAG1 geni (Tüm Gen Dizi Analizi)	PAG1 gene (Sequence Analysis-All Coding Exons)	PAG1	PAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAFAH2 geni (Tüm Gen Dizi Analizi)	PAFAH2 gene (Sequence Analysis-All Coding Exons)	PAFAH2	PAFAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAFAH1B3 geni (Tüm Gen Dizi Analizi)	PAFAH1B3 gene (Sequence Analysis-All Coding Exons)	PAFAH1B3	PAFAH1B3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAFAH1B2 geni (Tüm Gen Dizi Analizi)	PAFAH1B2 gene (Sequence Analysis-All Coding Exons)	PAFAH1B2	PAFAH1B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAF1 geni (Tüm Gen Dizi Analizi)	PAF1 gene (Sequence Analysis-All Coding Exons)	PAF1	PAF1, PD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAEP geni (Tüm Gen Dizi Analizi)	PAEP gene (Sequence Analysis-All Coding Exons)	PAEP	PAEP, PP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PADI2 geni (Tüm Gen Dizi Analizi)	PADI2 gene (Sequence Analysis-All Coding Exons)	PADI2	PADI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PADI1 geni (Tüm Gen Dizi Analizi)	PADI1 gene (Sequence Analysis-All Coding Exons)	PADI1	PADI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PACSIN3 geni (Tüm Gen Dizi Analizi)	PACSIN3 gene (Sequence Analysis-All Coding Exons)	PACSIN3	PACSIN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PACSIN2 geni (Tüm Gen Dizi Analizi)	PACSIN2 gene (Sequence Analysis-All Coding Exons)	PACSIN2	PACSIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PACSIN1 geni (Tüm Gen Dizi Analizi)	PACSIN1 gene (Sequence Analysis-All Coding Exons)	PACSIN1	PACSIN1, KIAA1379	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PACS2 geni (Tüm Gen Dizi Analizi)	PACS2 gene (Sequence Analysis-All Coding Exons)	PACS2	PACS2, KIAA0602	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PACRG geni (Tüm Gen Dizi Analizi)	PACRG gene (Sequence Analysis-All Coding Exons)	PACRG	PACRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PABPC5 geni (Tüm Gen Dizi Analizi)	PABPC5 gene (Sequence Analysis-All Coding Exons)	PABPC5	PABPC5, PABP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PABPC4 geni (Tüm Gen Dizi Analizi)	PABPC4 gene (Sequence Analysis-All Coding Exons)	PABPC4	PABPC4, PABP4, IPABP, APP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PABPC1 geni (Tüm Gen Dizi Analizi)	PABPC1 gene (Sequence Analysis-All Coding Exons)	PABPC1	PABPC1, PAB1, PABP1, PABP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PA2G4 geni (Tüm Gen Dizi Analizi)	PA2G4 gene (Sequence Analysis-All Coding Exons)	PA2G4	PA2G4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P4HTM geni (Tüm Gen Dizi Analizi)	P4HTM gene (Sequence Analysis-All Coding Exons)	P4HTM	P4HTM, PH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P4HA3 geni (Tüm Gen Dizi Analizi)	P4HA3 gene (Sequence Analysis-All Coding Exons)	P4HA3	P4HA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P4HA1 geni (Tüm Gen Dizi Analizi)	P4HA1 gene (Sequence Analysis-All Coding Exons)	P4HA1	P4HA1, P4HA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P3H3 geni (Tüm Gen Dizi Analizi)	P3H3 gene (Sequence Analysis-All Coding Exons)	P3H3	P3H3, LEPREL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY8 geni (Tüm Gen Dizi Analizi)	P2RY8 gene (Sequence Analysis-All Coding Exons)	P2RY8	P2RY8, P2Y8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY6 geni (Tüm Gen Dizi Analizi)	P2RY6 gene (Sequence Analysis-All Coding Exons)	P2RY6	P2RY6, P2Y6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY4 geni (Tüm Gen Dizi Analizi)	P2RY4 gene (Sequence Analysis-All Coding Exons)	P2RY4	P2RY4, NRU, P2Y4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY2 geni (Tüm Gen Dizi Analizi)	P2RY2 gene (Sequence Analysis-All Coding Exons)	P2RY2	P2RY2, P2Y2, P2U	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY14 geni (Tüm Gen Dizi Analizi)	P2RY14 gene (Sequence Analysis-All Coding Exons)	P2RY14	P2RY14, BPR105, KIAA0001	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY13 geni (Tüm Gen Dizi Analizi)	P2RY13 gene (Sequence Analysis-All Coding Exons)	P2RY13	GPR86	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY11 geni (Tüm Gen Dizi Analizi)	P2RY11 gene (Sequence Analysis-All Coding Exons)	P2RY11	P2RY11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY10 geni (Tüm Gen Dizi Analizi)	P2RY10 gene (Sequence Analysis-All Coding Exons)	P2RY10	P2RY10, P2Y10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RY1 geni (Tüm Gen Dizi Analizi)	P2RY1 gene (Sequence Analysis-All Coding Exons)	P2RY1	P2RY1, P2Y1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RX7 geni (Tüm Gen Dizi Analizi)	P2RX7 gene (Sequence Analysis-All Coding Exons)	P2RX7	P2RX7, P2X7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RX6 geni (Tüm Gen Dizi Analizi)	P2RX6 gene (Sequence Analysis-All Coding Exons)	P2RX6	P2RXL1, P2XM, P2X6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RX4 geni (Tüm Gen Dizi Analizi)	P2RX4 gene (Sequence Analysis-All Coding Exons)	P2RX4	P2RX4, P2X4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RX3 geni (Tüm Gen Dizi Analizi)	P2RX3 gene (Sequence Analysis-All Coding Exons)	P2RX3	P2RX3, P2X3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXTR geni (Tüm Gen Dizi Analizi)	OXTR gene (Sequence Analysis-All Coding Exons)	OXTR	OXTR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXT geni (Tüm Gen Dizi Analizi)	OXT gene (Sequence Analysis-All Coding Exons)	OXT	OXT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

OXSR1 geni (Tüm Gen Dizi Analizi)	OXSR1 gene (Sequence Analysis-All Coding Exons)	OXSR1	OSR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXSM geni (Tüm Gen Dizi Analizi)	OXSM gene (Sequence Analysis-All Coding Exons)	OXSM	OXSM, KS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXR1 geni (Tüm Gen Dizi Analizi)	OXR1 gene (Sequence Analysis-All Coding Exons)	OXR1	OXR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXGR1 geni (Tüm Gen Dizi Analizi)	OXGR1 gene (Sequence Analysis-All Coding Exons)	OXGR1	GPR80	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXCT2 geni (Tüm Gen Dizi Analizi)	OXCT2 gene (Sequence Analysis-All Coding Exons)	OXCT2	OXCT2, FLJ0030	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OXA1L geni (Tüm Gen Dizi Analizi)	OXA1L gene (Sequence Analysis-All Coding Exons)	OXA1L	OXA1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OVOL3 geni (Tüm Gen Dizi Analizi)	OVOL3 gene (Sequence Analysis-All Coding Exons)	OVOL3	OVOL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OVOL1 geni (Tüm Gen Dizi Analizi)	OVOL1 gene (Sequence Analysis-All Coding Exons)	OVOL1	OVOL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OVGP1 geni (Tüm Gen Dizi Analizi)	OVGP1 gene (Sequence Analysis-All Coding Exons)	OVGP1	OVGP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OVCA2 geni (Tüm Gen Dizi Analizi)	OVCA2 gene (Sequence Analysis-All Coding Exons)	OVCA2	OVCA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTX1 geni (Tüm Gen Dizi Analizi)	OTX1 gene (Sequence Analysis-All Coding Exons)	OTX1	OTX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD7B geni (Tüm Gen Dizi Analizi)	OTUD7B gene (Sequence Analysis-All Coding Exons)	OTUD7B	OTUD7B, CEZANNE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNPS1 geni (Tüm Gen Dizi Analizi)	RNPS1 gene (Sequence Analysis-All Coding Exons)	RNPS1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNPEPL1 geni (Tüm Gen Dizi Analizi)	RNPEPL1 gene (Sequence Analysis-All Coding Exons)	RNPEPL1	RNPEPL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD7A geni (Tüm Gen Dizi Analizi)	OTUD7A gene (Sequence Analysis-All Coding Exons)	OTUD7A	OTUD7A, OTUD7, C16orf15, CEZANNE7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD6B geni (Tüm Gen Dizi Analizi)	OTUD6B gene (Sequence Analysis-All Coding Exons)	OTUD6B	OTUD6B, DUBA5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD6A geni (Tüm Gen Dizi Analizi)	OTUD6A gene (Sequence Analysis-All Coding Exons)	OTUD6A	OTUD6A, DUBA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD5 geni (Tüm Gen Dizi Analizi)	OTUD5 gene (Sequence Analysis-All Coding Exons)	OTUD5	OTUD5, DUBA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD4 geni (Tüm Gen Dizi Analizi)	OTUD4 gene (Sequence Analysis-All Coding Exons)	OTUD4	OTUD4, KIAA1046, HN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD3 geni (Tüm Gen Dizi Analizi)	OTUD3 gene (Sequence Analysis-All Coding Exons)	OTUD3	OTUD3, KIAA0459	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUD1 geni (Tüm Gen Dizi Analizi)	OTUD1 gene (Sequence Analysis-All Coding Exons)	OTUD1	OTUD1, DUBA7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUB2 geni (Tüm Gen Dizi Analizi)	OTUB2 gene (Sequence Analysis-All Coding Exons)	OTUB2	OTUB2, OTU2, OTB2, C14orf137	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTUB1 geni (Tüm Gen Dizi Analizi)	OTUB1 gene (Sequence Analysis-All Coding Exons)	OTUB1	OTUB1, OTU1, OTB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTP geni (Tüm Gen Dizi Analizi)	OTP gene (Sequence Analysis-All Coding Exons)	OTP	OTP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTOS geni (Tüm Gen Dizi Analizi)	OTOS gene (Sequence Analysis-All Coding Exons)	OTOS	OTOSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTOR geni (Tüm Gen Dizi Analizi)	OTOR gene (Sequence Analysis-All Coding Exons)	OTOR	OTOR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTOP3 geni (Tüm Gen Dizi Analizi)	OTOP3 gene (Sequence Analysis-All Coding Exons)	OTOP3	OTOP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTOP2 geni (Tüm Gen Dizi Analizi)	OTOP2 gene (Sequence Analysis-All Coding Exons)	OTOP2	OTOP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OTOP1 geni (Tüm Gen Dizi Analizi)	OTOP1 gene (Sequence Analysis-All Coding Exons)	OTOP1	OTOP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSTN geni (Tüm Gen Dizi Analizi)	OSTN gene (Sequence Analysis-All Coding Exons)	OSTN	OSTN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSTF1 geni (Tüm Gen Dizi Analizi)	OSTF1 gene (Sequence Analysis-All Coding Exons)	OSTF1	OSTF1, OSF, SH3P2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSR2 geni (Tüm Gen Dizi Analizi)	OSR2 gene (Sequence Analysis-All Coding Exons)	OSR2	OSR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSR1 geni (Tüm Gen Dizi Analizi)	OSR1 gene (Sequence Analysis-All Coding Exons)	OSR1	ODD, OSR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSM geni (Tüm Gen Dizi Analizi)	OSM gene (Sequence Analysis-All Coding Exons)	OSM	OSM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSGIN2 geni (Tüm Gen Dizi Analizi)	OSGIN2 gene (Sequence Analysis-All Coding Exons)	OSGIN2	OSGIN2, C8orf1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSGIN1 geni (Tüm Gen Dizi Analizi)	OSGIN1 gene (Sequence Analysis-All Coding Exons)	OSGIN1	OSGIN1, OKL38	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSGEP geni (Tüm Gen Dizi Analizi)	OSGEP gene (Sequence Analysis-All Coding Exons)	OSGEP	OSGEP, FLJ20411	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSCP1 geni (Tüm Gen Dizi Analizi)	OSCP1 gene (Sequence Analysis-All Coding Exons)	OSCP1	NOR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSCAR geni (Tüm Gen Dizi Analizi)	OSCAR gene (Sequence Analysis-All Coding Exons)	OSCAR	OSCAR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL9 geni (Tüm Gen Dizi Analizi)	OSBPL9 gene (Sequence Analysis-All Coding Exons)	OSBPL9	OSBPL9, ORP9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL8 geni (Tüm Gen Dizi Analizi)	OSBPL8 gene (Sequence Analysis-All Coding Exons)	OSBPL8	OSBPL8, ORP8, KIAA1451	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL7 geni (Tüm Gen Dizi Analizi)	OSBPL7 gene (Sequence Analysis-All Coding Exons)	OSBPL7	OSBPL7, ORP7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL6 geni (Tüm Gen Dizi Analizi)	OSBPL6 gene (Sequence Analysis-All Coding Exons)	OSBPL6	OSBPL6, ORP6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL5 geni (Tüm Gen Dizi Analizi)	OSBPL5 gene (Sequence Analysis-All Coding Exons)	OSBPL5	OSBPL5, ORP5, KIAA1534	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF19A geni (Tüm Gen Dizi Analizi)	RNF19A gene (Sequence Analysis-All Coding Exons)	RNF19A		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL3 geni (Tüm Gen Dizi Analizi)	OSBPL3 gene (Sequence Analysis-All Coding Exons)	OSBPL3	OSBPL3, ORP3, KIAA0704	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL1A geni (Tüm Gen Dizi Analizi)	OSBPL1A gene (Sequence Analysis-All Coding Exons)	OSBPL1A	OSBPL1A, ORP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL11 geni (Tüm Gen Dizi Analizi)	OSBPL11 gene (Sequence Analysis-All Coding Exons)	OSBPL11	OSBPL11, ORP11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBPL10 geni (Tüm Gen Dizi Analizi)	OSBPL10 gene (Sequence Analysis-All Coding Exons)	OSBPL10	OSBPL10, ORP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBP2 geni (Tüm Gen Dizi Analizi)	OSBP2 gene (Sequence Analysis-All Coding Exons)	OSBP2	OSBP2, ORP4, KIAA1664	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OSBP geni (Tüm Gen Dizi Analizi)	OSBP gene (Sequence Analysis-All Coding Exons)	OSBP	OSBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OS9 geni (Tüm Gen Dizi Analizi)	OS9 gene (Sequence Analysis-All Coding Exons)	OS9	OS9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORMDL3 geni (Tüm Gen Dizi Analizi)	ORMDL3 gene (Sequence Analysis-All Coding Exons)	ORMDL3	ORMDL3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORMDL2 geni (Tüm Gen Dizi Analizi)	ORMDL2 gene (Sequence Analysis-All Coding Exons)	ORMDL2	ORMDL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORMDL1 geni (Tüm Gen Dizi Analizi)	ORMDL1 gene (Sequence Analysis-All Coding Exons)	ORMDL1	ORMDL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORM2 geni (Tüm Gen Dizi Analizi)	ORM2 gene (Sequence Analysis-All Coding Exons)	ORM2	ORM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORM1 geni (Tüm Gen Dizi Analizi)	ORM1 gene (Sequence Analysis-All Coding Exons)	ORM1	ORM1, AGP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORC5 geni (Tüm Gen Dizi Analizi)	ORC5 gene (Sequence Analysis-All Coding Exons)	ORC5	ORC5, ORC5L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORC3 geni (Tüm Gen Dizi Analizi)	ORC3 gene (Sequence Analysis-All Coding Exons)	ORC3	ORC3, ORC3L, LAT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORC2 geni (Tüm Gen Dizi Analizi)	ORC2 gene (Sequence Analysis-All Coding Exons)	ORC2	ORC2, ORC2L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORAOV1 geni (Tüm Gen Dizi Analizi)	ORAOV1 gene (Sequence Analysis-All Coding Exons)	ORAOV1	ORAOV1, TAOS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RNF13 geni (Tüm Gen Dizi Analizi)	RNF13 gene (Sequence Analysis-All Coding Exons)	RNF13		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORAI3 geni (Tüm Gen Dizi Analizi)	ORAI3 gene (Sequence Analysis-All Coding Exons)	ORAI3	ORAI3, TMEM142C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORAI2 geni (Tüm Gen Dizi Analizi)	ORAI2 gene (Sequence Analysis-All Coding Exons)	ORAI2	ORAI2, MEM142B, C7orf19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR7D4 geni (Tüm Gen Dizi Analizi)	OR7D4 gene (Sequence Analysis-All Coding Exons)	OR7D4	OR7D4, OR19B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR6A2 geni (Tüm Gen Dizi Analizi)	OR6A2 gene (Sequence Analysis-All Coding Exons)	OR6A2	OR6A2, OR6A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ORS11 geni (Tüm Gen Dizi Analizi)	ORS11 gene (Sequence Analysis-All Coding Exons)	ORS11	ORS11, OLF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR5F1 geni (Tüm Gen Dizi Analizi)	OR5F1 gene (Sequence Analysis-All Coding Exons)	OR5F1	OR5F1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR5AN1 geni (Tüm Gen Dizi Analizi)	OR5AN1 gene (Sequence Analysis-All Coding Exons)	OR5AN1	OR5AN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR51E2 geni (Tüm Gen Dizi Analizi)	OR51E2 gene (Sequence Analysis-All Coding Exons)	OR51E2	OR51E2, PSGR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR51E1 geni (Tüm Gen Dizi Analizi)	OR51E1 gene (Sequence Analysis-All Coding Exons)	OR51E1	OR51E1, DGPCR, PSGR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR4C46 geni (Tüm Gen Dizi Analizi)	OR4C46 gene (Sequence Analysis-All Coding Exons)	OR4C46	OR4C46	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR2W3 geni (Tüm Gen Dizi Analizi)	OR2W3 gene (Sequence Analysis-All Coding Exons)	OR2W3	OR2W3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR2H2 geni (Tüm Gen Dizi Analizi)	OR2H2 gene (Sequence Analysis-All Coding Exons)	OR2H2	OR2H3, OLFR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR2F1 geni (Tüm Gen Dizi Analizi)	OR2F1 gene (Sequence Analysis-All Coding Exons)	OR2F1	OR2F1, OLF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR2D2 geni (Tüm Gen Dizi Analizi)	OR2D2 gene (Sequence Analysis-All Coding Exons)	OR2D2	OR2D2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR1F1 geni (Tüm Gen Dizi Analizi)	OR1F1 gene (Sequence Analysis-All Coding Exons)	OR1F1	OR1F1, OLFMF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR1D2 geni (Tüm Gen Dizi Analizi)	OR1D2 gene (Sequence Analysis-All Coding Exons)	OR1D2	OR1D2, OLFR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR13G1 geni (Tüm Gen Dizi Analizi)	OR13G1 gene (Sequence Analysis-All Coding Exons)	OR13G1	OR13G1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OR10A5 geni (Tüm Gen Dizi Analizi)	OR10A5 gene (Sequence Analysis-All Coding Exons)	OR10A5	OR10A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPRPN geni (Tüm Gen Dizi Analizi)	OPRPN gene (Sequence Analysis-All Coding Exons)	OPRPN	PROL1, PRL1, BPLP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPRM1 geni (Tüm Gen Dizi Analizi)	OPRM1 gene (Sequence Analysis-All Coding Exons)	OPRM1	OPRM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPRK1 geni (Tüm Gen Dizi Analizi)	OPRK1 gene (Sequence Analysis-All Coding Exons)	OPRK1	OPRK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPRD1 geni (Tüm Gen Dizi Analizi)	OPRD1 gene (Sequence Analysis-All Coding Exons)	OPRD1	OPRD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPN5 geni (Tüm Gen Dizi Analizi)	OPN5 gene (Sequence Analysis-All Coding Exons)	OPN5	OPN5, GRP136, PGR12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPN4 geni (Tüm Gen Dizi Analizi)	OPN4 gene (Sequence Analysis-All Coding Exons)	OPN4	OPN4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPN3 geni (Tüm Gen Dizi Analizi)	OPN3 gene (Sequence Analysis-All Coding Exons)	OPN3	OPN3, ECPN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPALIN geni (Tüm Gen Dizi Analizi)	OPALIN gene (Sequence Analysis-All Coding Exons)	OPALIN	OPALIN, TMEM10, TMP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OOEP geni (Tüm Gen Dizi Analizi)	OOEP gene (Sequence Analysis-All Coding Exons)	OOEP	OOEP, KHDC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ONECUT3 geni (Tüm Gen Dizi Analizi)	ONECUT3 gene (Sequence Analysis-All Coding Exons)	ONECUT3	ONECUT3, OC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ONECUT2 geni (Tüm Gen Dizi Analizi)	ONECUT2 gene (Sequence Analysis-All Coding Exons)	ONECUT2	ONECUT2, OC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ONECUT1 geni (Tüm Gen Dizi Analizi)	ONECUT1 gene (Sequence Analysis-All Coding Exons)	ONECUT1	ONECUT1, HNF6A, HNF6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OMP geni (Tüm Gen Dizi Analizi)	OMP gene (Sequence Analysis-All Coding Exons)	OMP	OMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

OMG geni (Tüm Gen Dizi Analizi)	OMG gene (Sequence Analysis-All Coding Exons)	OMG	OMG, OMGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OMA1 geni (Tüm Gen Dizi Analizi)	OMA1 gene (Sequence Analysis-All Coding Exons)	OMA1	OMA1, MPRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLIG3 geni (Tüm Gen Dizi Analizi)	OLIG3 gene (Sequence Analysis-All Coding Exons)	OLIG3	OLIG3, BHLHB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLIG2 geni (Tüm Gen Dizi Analizi)	OLIG2 gene (Sequence Analysis-All Coding Exons)	OLIG2	OLIG2, PRKCBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLIG1 geni (Tüm Gen Dizi Analizi)	OLIG1 gene (Sequence Analysis-All Coding Exons)	OLIG1	OLIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLFML3 geni (Tüm Gen Dizi Analizi)	OLFML3 gene (Sequence Analysis-All Coding Exons)	OLFML3	OLFML3, OLF44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLFML2A geni (Tüm Gen Dizi Analizi)	OLFML2A gene (Sequence Analysis-All Coding Exons)	OLFML2A	OLFML2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLFM4 geni (Tüm Gen Dizi Analizi)	OLFM4 gene (Sequence Analysis-All Coding Exons)	OLFM4	OLFM4, GC1, GW112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLFM3 geni (Tüm Gen Dizi Analizi)	OLFM3 gene (Sequence Analysis-All Coding Exons)	OLFM3	OLFM3, NOE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLFM1 geni (Tüm Gen Dizi Analizi)	OLFM1 gene (Sequence Analysis-All Coding Exons)	OLFM1	OLFM1, AMY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OLA1 geni (Tüm Gen Dizi Analizi)	OLA1 gene (Sequence Analysis-All Coding Exons)	OLA1	OLA1, GTBP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OIT3 geni (Tüm Gen Dizi Analizi)	OIT3 gene (Sequence Analysis-All Coding Exons)	OIT3	OIT3, LZP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OGT geni (Tüm Gen Dizi Analizi)	OGT gene (Sequence Analysis-All Coding Exons)	OGT	OGT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OGN geni (Tüm Gen Dizi Analizi)	OGN gene (Sequence Analysis-All Coding Exons)	OGN	OGN, OIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OGFOD1 geni (Tüm Gen Dizi Analizi)	OGFOD1 gene (Sequence Analysis-All Coding Exons)	OGFOD1	OGFOD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ODF4 geni (Tüm Gen Dizi Analizi)	ODF4 gene (Sequence Analysis-All Coding Exons)	ODF4	ODF4, OPPO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ODF3 geni (Tüm Gen Dizi Analizi)	ODF3 gene (Sequence Analysis-All Coding Exons)	ODF3	ODF3, SHIPPO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ODF2 geni (Tüm Gen Dizi Analizi)	ODF2 gene (Sequence Analysis-All Coding Exons)	ODF2	ODF2, ODF84	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ODF1 geni (Tüm Gen Dizi Analizi)	ODF1 gene (Sequence Analysis-All Coding Exons)	ODF1	ODF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ODAM geni (Tüm Gen Dizi Analizi)	ODAM gene (Sequence Analysis-All Coding Exons)	ODAM	ODAM, APIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OCM geni (Tüm Gen Dizi Analizi)	OCM gene (Sequence Analysis-All Coding Exons)	OCM	OCM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OC90 geni (Tüm Gen Dizi Analizi)	OC90 gene (Sequence Analysis-All Coding Exons)	OC90	OC90, PLA2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OBP2B geni (Tüm Gen Dizi Analizi)	OBP2B gene (Sequence Analysis-All Coding Exons)	OBP2B	OBP2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OBP2A geni (Tüm Gen Dizi Analizi)	OBP2A gene (Sequence Analysis-All Coding Exons)	OBP2A	OBP2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OAZ3 geni (Tüm Gen Dizi Analizi)	OAZ3 gene (Sequence Analysis-All Coding Exons)	OAZ3	OAZ3, AZ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OAZ2 geni (Tüm Gen Dizi Analizi)	OAZ2 gene (Sequence Analysis-All Coding Exons)	OAZ2	OAZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OAZ1 geni (Tüm Gen Dizi Analizi)	OAZ1 gene (Sequence Analysis-All Coding Exons)	OAZ1	OAZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OASL geni (Tüm Gen Dizi Analizi)	OASL gene (Sequence Analysis-All Coding Exons)	OASL	OASL, TRIP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OAS3 geni (Tüm Gen Dizi Analizi)	OAS3 gene (Sequence Analysis-All Coding Exons)	OAS3	OAS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OAS2 geni (Tüm Gen Dizi Analizi)	OAS2 gene (Sequence Analysis-All Coding Exons)	OAS2	OAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OARD1 geni (Tüm Gen Dizi Analizi)	OARD1 gene (Sequence Analysis-All Coding Exons)	OARD1	C8orf130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NYAP2 geni (Tüm Gen Dizi Analizi)	NYAP2 gene (Sequence Analysis-All Coding Exons)	NYAP2	NYPA2, KIAA1486	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NYAP1 geni (Tüm Gen Dizi Analizi)	NYAP1 gene (Sequence Analysis-All Coding Exons)	NYAP1	NYPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXT2 geni (Tüm Gen Dizi Analizi)	NXT2 gene (Sequence Analysis-All Coding Exons)	NXT2	NXT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXT1 geni (Tüm Gen Dizi Analizi)	NXT1 gene (Sequence Analysis-All Coding Exons)	NXT1	NXT1, MTR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXPH4 geni (Tüm Gen Dizi Analizi)	NXPH4 gene (Sequence Analysis-All Coding Exons)	NXPH4	NXPH4, NPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIN3 geni (Tüm Gen Dizi Analizi)	RIN3 gene (Sequence Analysis-All Coding Exons)	RIN3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXPH3 geni (Tüm Gen Dizi Analizi)	NXPH3 gene (Sequence Analysis-All Coding Exons)	NXPH3	NXPH3, NPH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIN1 geni (Tüm Gen Dizi Analizi)	RIN1 gene (Sequence Analysis-All Coding Exons)	RIN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXPH2 geni (Tüm Gen Dizi Analizi)	NXPH2 gene (Sequence Analysis-All Coding Exons)	NXPH2	NXPH2, NPH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXPH1 geni (Tüm Gen Dizi Analizi)	NXPH1 gene (Sequence Analysis-All Coding Exons)	NXPH1	NXPH1, NPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXNL2 geni (Tüm Gen Dizi Analizi)	NXNL2 gene (Sequence Analysis-All Coding Exons)	NXNL2	NXNL2, RDCVF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXNL1 geni (Tüm Gen Dizi Analizi)	NXNL1 gene (Sequence Analysis-All Coding Exons)	NXNL1	NXNL1, RDCVF, TXNL6, UNQ15561	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXN geni (Tüm Gen Dizi Analizi)	NXN gene (Sequence Analysis-All Coding Exons)	NXN	NXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXF5 geni (Tüm Gen Dizi Analizi)	NXF5 gene (Sequence Analysis-All Coding Exons)	NXF5	NXF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXF4 geni (Tüm Gen Dizi Analizi)	NXF4 gene (Sequence Analysis-All Coding Exons)	NXF4	NXF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXF3 geni (Tüm Gen Dizi Analizi)	NXF3 gene (Sequence Analysis-All Coding Exons)	NXF3	NXF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXF2 geni (Tüm Gen Dizi Analizi)	NXF2 gene (Sequence Analysis-All Coding Exons)	NXF2	NXF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NXF1 geni (Tüm Gen Dizi Analizi)	NXF1 gene (Sequence Analysis-All Coding Exons)	NXF1	NXF1, TAP, MEX67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NWD1 geni (Tüm Gen Dizi Analizi)	NWD1 gene (Sequence Analysis-All Coding Exons)	NWD1	NWD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NVL geni (Tüm Gen Dizi Analizi)	NVL gene (Sequence Analysis-All Coding Exons)	NVL	NVL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RIF1 geni (Tüm Gen Dizi Analizi)	RIF1 gene (Sequence Analysis-All Coding Exons)	RIF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUSAP1 geni (Tüm Gen Dizi Analizi)	NUSAP1 gene (Sequence Analysis-All Coding Exons)	NUSAP1	NUSAP1, ANKT, NUSAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RICTOR geni (Tüm Gen Dizi Analizi)	RICTOR gene (Sequence Analysis-All Coding Exons)	RICTOR		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUPR1 geni (Tüm Gen Dizi Analizi)	NUPR1 gene (Sequence Analysis-All Coding Exons)	NUPR1	NUPR1, COM1, p8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP98 geni (Tüm Gen Dizi Analizi)	NUP98 gene (Sequence Analysis-All Coding Exons)	NUP98	NUP98	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP88 geni (Tüm Gen Dizi Analizi)	NUP88 gene (Sequence Analysis-All Coding Exons)	NUP88	NUP88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP85 geni (Tüm Gen Dizi Analizi)	NUP85 gene (Sequence Analysis-All Coding Exons)	NUP85	NUP85, PCNT1, PCNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP58 geni (Tüm Gen Dizi Analizi)	NUP58 gene (Sequence Analysis-All Coding Exons)	NUP58	NUP58, NUPL1, KIAA0410	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP54 geni (Tüm Gen Dizi Analizi)	NUP54 gene (Sequence Analysis-All Coding Exons)	NUP54	NUP54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP50 geni (Tüm Gen Dizi Analizi)	NUP50 gene (Sequence Analysis-All Coding Exons)	NUP50	NUP50, NPAP60L, NPAP60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP43 geni (Tüm Gen Dizi Analizi)	NUP43 gene (Sequence Analysis-All Coding Exons)	NUP43	NUP43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP37 geni (Tüm Gen Dizi Analizi)	NUP37 gene (Sequence Analysis-All Coding Exons)	NUP37	NUP37, p37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP35 geni (Tüm Gen Dizi Analizi)	NUP35 gene (Sequence Analysis-All Coding Exons)	NUP35	NUP35, NP44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP210 geni (Tüm Gen Dizi Analizi)	NUP210 gene (Sequence Analysis-All Coding Exons)	NUP210	NUP210, GP210, KIAA0906	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP188 geni (Tüm Gen Dizi Analizi)	NUP188 gene (Sequence Analysis-All Coding Exons)	NUP188	NUP188, KIAA1069	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP160 geni (Tüm Gen Dizi Analizi)	NUP160 gene (Sequence Analysis-All Coding Exons)	NUP160	NUP160, KIAA0197	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP153 geni (Tüm Gen Dizi Analizi)	NUP153 gene (Sequence Analysis-All Coding Exons)	NUP153	NUP153	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUP133 geni (Tüm Gen Dizi Analizi)	NUP133 gene (Sequence Analysis-All Coding Exons)	NUP133	NUP133	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUMBL geni (Tüm Gen Dizi Analizi)	NUMBL gene (Sequence Analysis-All Coding Exons)	NUMBL	NUMBL, NUMBR, NBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUMB geni (Tüm Gen Dizi Analizi)	NUMB gene (Sequence Analysis-All Coding Exons)	NUMB	NUMB, S171	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUFIP2 geni (Tüm Gen Dizi Analizi)	NUFIP2 gene (Sequence Analysis-All Coding Exons)	NUFIP2	NUFIP2, KIAA1321, PIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUFIP1 geni (Tüm Gen Dizi Analizi)	NUFIP1 gene (Sequence Analysis-All Coding Exons)	NUFIP1	NUFIP1, NUFIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUF2 geni (Tüm Gen Dizi Analizi)	NUF2 gene (Sequence Analysis-All Coding Exons)	NUF2	NUF2, NUF2R, CDCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT21 geni (Tüm Gen Dizi Analizi)	NUDT21 gene (Sequence Analysis-All Coding Exons)	NUDT21	NUDT21, CPSF5, CFIM25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT2 geni (Tüm Gen Dizi Analizi)	NUDT2 gene (Sequence Analysis-All Coding Exons)	NUDT2	NUDT2, APAH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT18 geni (Tüm Gen Dizi Analizi)	NUDT18 gene (Sequence Analysis-All Coding Exons)	NUDT18	NUDT18, MTH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT16L1 geni (Tüm Gen Dizi Analizi)	NUDT16L1 gene (Sequence Analysis-All Coding Exons)	NUDT16L1	NUDT16L1, SOOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT16 geni (Tüm Gen Dizi Analizi)	NUDT16 gene (Sequence Analysis-All Coding Exons)	NUDT16	NUDT16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT11 geni (Tüm Gen Dizi Analizi)	NUDT11 gene (Sequence Analysis-All Coding Exons)	NUDT11	NUDT11, DIPP3B, APS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT10 geni (Tüm Gen Dizi Analizi)	NUDT10 gene (Sequence Analysis-All Coding Exons)	NUDT10	NUDT10, DIPP3A, APS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT1 geni (Tüm Gen Dizi Analizi)	NUDT1 gene (Sequence Analysis-All Coding Exons)	NUDT1	NUDT1, MTH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDCD3 geni (Tüm Gen Dizi Analizi)	NUDCD3 gene (Sequence Analysis-All Coding Exons)	NUDCD3	NUDCD3, KIAA1068, NUDCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDCD1 geni (Tüm Gen Dizi Analizi)	NUDCD1 gene (Sequence Analysis-All Coding Exons)	NUDCD1	CML66	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDC geni (Tüm Gen Dizi Analizi)	NUDC gene (Sequence Analysis-All Coding Exons)	NUDC	NUDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUCKS1 geni (Tüm Gen Dizi Analizi)	NUCKS1 gene (Sequence Analysis-All Coding Exons)	NUCKS1	NUCKS1, NUCKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RHBDL2 geni (Tüm Gen Dizi Analizi)	RHBDL2 gene (Sequence Analysis-All Coding Exons)	RHBDL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUCB2 geni (Tüm Gen Dizi Analizi)	NUCB2 gene (Sequence Analysis-All Coding Exons)	NUCB2	NUCB2, NEFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUCB1 geni (Tüm Gen Dizi Analizi)	NUCB1 gene (Sequence Analysis-All Coding Exons)	NUCB1	NUCB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUBP2 geni (Tüm Gen Dizi Analizi)	NUBP2 gene (Sequence Analysis-All Coding Exons)	NUBP2	NUBP2, CFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUBP1 geni (Tüm Gen Dizi Analizi)	NUBP1 gene (Sequence Analysis-All Coding Exons)	NUBP1	NUBP1, NBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUB1 geni (Tüm Gen Dizi Analizi)	NUB1 gene (Sequence Analysis-All Coding Exons)	NUB1	NUB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUAK2 geni (Tüm Gen Dizi Analizi)	NUAK2 gene (Sequence Analysis-All Coding Exons)	NUAK2	NUAK2, SNARK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NUAK1 geni (Tüm Gen Dizi Analizi)	NUAK1 gene (Sequence Analysis-All Coding Exons)	NUAK1	NUAK1, ARK5, KIAA0537	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTSR2 geni (Tüm Gen Dizi Analizi)	NTSR2 gene (Sequence Analysis-All Coding Exons)	NTSR2	NTSR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTSR1 geni (Tüm Gen Dizi Analizi)	NTSR1 gene (Sequence Analysis-All Coding Exons)	NTSR1	NTSR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTS geni (Tüm Gen Dizi Analizi)	NTS gene (Sequence Analysis-All Coding Exons)	NTS	NTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS8 geni (Tüm Gen Dizi Analizi)	RGS8 gene (Sequence Analysis-All Coding Exons)	RGS8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTRK3 geni (Tüm Gen Dizi Analizi)	NTRK3 gene (Sequence Analysis-All Coding Exons)	NTRK3	NTRK3, TRKC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTRK3 geni (Tüm Gen Dizi Analizi)	NTRK3 gene (Sequence Analysis-All Coding Exons)	NTRK3	NTRK3, TRKC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTRK3 geni (Tüm Gen Dizi Analizi)	NTRK3 gene (Sequence Analysis-All Coding Exons)	NTRK3	NTRK3, TRKC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTNG1 geni (Tüm Gen Dizi Analizi)	NTNG1 gene (Sequence Analysis-All Coding Exons)	NTNG1	NTNG1, LMNT1, KIAA0976	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTN4 geni (Tüm Gen Dizi Analizi)	NTN4 gene (Sequence Analysis-All Coding Exons)	NTN4	NTN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTN3 geni (Tüm Gen Dizi Analizi)	NTN3 gene (Sequence Analysis-All Coding Exons)	NTN3	NTN2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTN1 geni (Tüm Gen Dizi Analizi)	NTN1 gene (Sequence Analysis-All Coding Exons)	NTN1	NTN1, NTN1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTMT1 geni (Tüm Gen Dizi Analizi)	NTMT1 gene (Sequence Analysis-All Coding Exons)	NTMT1	NTMT1, METTL11A, NRMT, C9orf32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS20 geni (Tüm Gen Dizi Analizi)	RGS20 gene (Sequence Analysis-All Coding Exons)	RGS20		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTM geni (Tüm Gen Dizi Analizi)	NTM gene (Sequence Analysis-All Coding Exons)	NTM	NTM, HNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTF3 geni (Tüm Gen Dizi Analizi)	NTF3 gene (Sequence Analysis-All Coding Exons)	NTF3	NTF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS18 geni (Tüm Gen Dizi Analizi)	RGS18 gene (Sequence Analysis-All Coding Exons)	RGS18		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS17 geni (Tüm Gen Dizi Analizi)	RGS17 gene (Sequence Analysis-All Coding Exons)	RGS17		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NTAN1 geni (Tüm Gen Dizi Analizi)	NTAN1 gene (Sequence Analysis-All Coding Exons)	NTAN1	NTAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NT5M geni (Tüm Gen Dizi Analizi)	NT5M gene (Sequence Analysis-All Coding Exons)	NT5M	NT5M, DNT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS13 geni (Tüm Gen Dizi Analizi)	RGS13 gene (Sequence Analysis-All Coding Exons)	RGS13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NT5DC3 geni (Tüm Gen Dizi Analizi)	NT5DC3 gene (Sequence Analysis-All Coding Exons)	NT5DC3	NT5DC3, TU12B1TY, GINN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NT5C1B geni (Tüm Gen Dizi Analizi)	NT5C1B gene (Sequence Analysis-All Coding Exons)	NT5C1B	NT5C1B, CN1B, AIRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RGS10 geni (Tüm Gen Dizi Analizi)	RGS10 gene (Sequence Analysis-All Coding Exons)	RGS10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NT5C1A geni (Tüm Gen Dizi Analizi)	NT5C1A gene (Sequence Analysis-All Coding Exons)	NT5C1A	NT5C1A, CN1A, CNI, CN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NT5C geni (Tüm Gen Dizi Analizi)	NT5C gene (Sequence Analysis-All Coding Exons)	NT5C	NT5C, UMPH2, DNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSUN7 geni (Tüm Gen Dizi Analizi)	NSUN7 gene (Sequence Analysis-All Coding Exons)	NSUN7	NSUN7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSUN6 geni (Tüm Gen Dizi Analizi)	NSUN6 gene (Sequence Analysis-All Coding Exons)	NSUN6	NSUN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSUN5 geni (Tüm Gen Dizi Analizi)	NSUN5 gene (Sequence Analysis-All Coding Exons)	NSUN5	NSUN5, WBSCR20, WBSCR20A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSUN4 geni (Tüm Gen Dizi Analizi)	NSUN4 gene (Sequence Analysis-All Coding Exons)	NSUN4	NSUN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSRP1 geni (Tüm Gen Dizi Analizi)	NSRP1 gene (Sequence Analysis-All Coding Exons)	NSRP1	NSRP1, NSRP70, CCDC55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSMCE4A geni (Tüm Gen Dizi Analizi)	NSMCE4A gene (Sequence Analysis-All Coding Exons)	NSMCE4A	NSMCE4A, NSE4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSMCE1 geni (Tüm Gen Dizi Analizi)	NSMCE1 gene (Sequence Analysis-All Coding Exons)	NSMCE1	NSMCE1, NSE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSMAF geni (Tüm Gen Dizi Analizi)	NSMAF gene (Sequence Analysis-All Coding Exons)	NSMAF	NSMAF, FAN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSL1 geni (Tüm Gen Dizi Analizi)	NSL1 gene (Sequence Analysis-All Coding Exons)	NSL1	NSL1, DC8, C1orf48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSFL1C geni (Tüm Gen Dizi Analizi)	NSFL1C gene (Sequence Analysis-All Coding Exons)	NSFL1C	P47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSF geni (Tüm Gen Dizi Analizi)	NSF gene (Sequence Analysis-All Coding Exons)	NSF	NSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSD2 geni (Tüm Gen Dizi Analizi)	NSD2 gene (Sequence Analysis-All Coding Exons)	NSD2	WHSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NSA2 geni (Tüm Gen Dizi Analizi)	NSA2 gene (Sequence Analysis-All Coding Exons)	NSA2	TINP1, NSA2, HUSSY29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRXN3 geni (Tüm Gen Dizi Analizi)	NRXN3 gene (Sequence Analysis-All Coding Exons)	NRXN3	NRXN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRXN2 geni (Tüm Gen Dizi Analizi)	NRXN2 gene (Sequence Analysis-All Coding Exons)	NRXN2	NRXN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRTN geni (Tüm Gen Dizi Analizi)	NRTN gene (Sequence Analysis-All Coding Exons)	NRTN	NRTN, NTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRSN2 geni (Tüm Gen Dizi Analizi)	NRSN2 gene (Sequence Analysis-All Coding Exons)	NRSN2	NRSN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRSN1 geni (Tüm Gen Dizi Analizi)	NRSN1 gene (Sequence Analysis-All Coding Exons)	NRSN1	NRSN1, VMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRROS geni (Tüm Gen Dizi Analizi)	NRROS gene (Sequence Analysis-All Coding Exons)	NRROS	NRROS, LRRC33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRP2 geni (Tüm Gen Dizi Analizi)	NRP2 gene (Sequence Analysis-All Coding Exons)	NRP2	NRP2, VEGF165R2, NPN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRP1 geni (Tüm Gen Dizi Analizi)	NRP1 gene (Sequence Analysis-All Coding Exons)	NRP1	NRP1, NRP, VEGF165R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRN1 geni (Tüm Gen Dizi Analizi)	NRN1 gene (Sequence Analysis-All Coding Exons)	NRN1	NRN1, NRN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFX4 geni (Tüm Gen Dizi Analizi)	RFX4 gene (Sequence Analysis-All Coding Exons)	RFX4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRL geni (Tüm Gen Dizi Analizi)	NRL gene (Sequence Analysis-All Coding Exons)	NRL	NRL, D14S46E, RP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Retinal degeneration autosomal recessive clumped pigment type (RDCP) [MIM:162080]: A retinopathy characterized by night blindness since early childhood, consistent with a severe reduction in rod function. Color vision is normal although there is a relatively enhanced function of short-wavelength-sensitive cones in the macula. Signs of retinal degeneration and clusters of clumped pigment deposits in the peripheral fundus at the level of the retinal pigment epithelium are present. {ECO:0000269|PubMed:15591106}. Note=The disease is caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
NRK geni (Tüm Gen Dizi Analizi)	NRK gene (Sequence Analysis-All Coding Exons)	NRK	NRK, NESK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRIP3 geni (Tüm Gen Dizi Analizi)	NRIP3 gene (Sequence Analysis-All Coding Exons)	NRIP3	NRIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRIP1 geni (Tüm Gen Dizi Analizi)	NRIP1 gene (Sequence Analysis-All Coding Exons)	NRIP1	NRIP1, RIP140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRGN geni (Tüm Gen Dizi Analizi)	NRGN gene (Sequence Analysis-All Coding Exons)	NRGN	NRGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRG4 geni (Tüm Gen Dizi Analizi)	NRG4 gene (Sequence Analysis-All Coding Exons)	NRG4	NRG4, HRG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRG3 geni (Tüm Gen Dizi Analizi)	NRG3 gene (Sequence Analysis-All Coding Exons)	NRG3	NRG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RFPL3S geni (Tüm Gen Dizi Analizi)	RFPL3S gene (Sequence Analysis-All Coding Exons)	RFPL3S	RFPL3S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRG2 geni (Tüm Gen Dizi Analizi)	NRG2 gene (Sequence Analysis-All Coding Exons)	NRG2	NRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRF1 geni (Tüm Gen Dizi Analizi)	NRF1 gene (Sequence Analysis-All Coding Exons)	NRF1	NRF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NREP geni (Tüm Gen Dizi Analizi)	NREP gene (Sequence Analysis-All Coding Exons)	NREP	C5orf13, P311, PTZ17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRDC geni (Tüm Gen Dizi Analizi)	NRDC gene (Sequence Analysis-All Coding Exons)	NRDC	NRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRCAM geni (Tüm Gen Dizi Analizi)	NRCAM gene (Sequence Analysis-All Coding Exons)	NRCAM	NRCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRBP2 geni (Tüm Gen Dizi Analizi)	NRBP2 gene (Sequence Analysis-All Coding Exons)	NRBP2	NRBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRBP1 geni (Tüm Gen Dizi Analizi)	NRBP1 gene (Sequence Analysis-All Coding Exons)	NRBP1	NRBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRBF2 geni (Tüm Gen Dizi Analizi)	NRBF2 gene (Sequence Analysis-All Coding Exons)	NRBF2	NRBF2, COPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRAV geni (Tüm Gen Dizi Analizi)	NRAV gene (Sequence Analysis-All Coding Exons)	NRAV	NRAV, DYNLL1AS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NRAP geni (Tüm Gen Dizi Analizi)	NRAP gene (Sequence Analysis-All Coding Exons)	NRAP	NRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR6A1 geni (Tüm Gen Dizi Analizi)	NR6A1 gene (Sequence Analysis-All Coding Exons)	NR6A1	GCNF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR5A2 geni (Tüm Gen Dizi Analizi)	NR5A2 gene (Sequence Analysis-All Coding Exons)	NR5A2	NR5A2, FTF, HB1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR4A2 geni (Tüm Gen Dizi Analizi)	NR4A2 gene (Sequence Analysis-All Coding Exons)	NR4A2	NR4A2, NURR1, NOT, TINUR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR4A1 geni (Tüm Gen Dizi Analizi)	NR4A1 gene (Sequence Analysis-All Coding Exons)	NR4A1	NR4A1, HMR, NP10, GFRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR2F6 geni (Tüm Gen Dizi Analizi)	NR2F6 gene (Sequence Analysis-All Coding Exons)	NR2F6	NR2F6, ERBAL2, EAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR2E1 geni (Tüm Gen Dizi Analizi)	NR2E1 gene (Sequence Analysis-All Coding Exons)	NR2E1	NR2E1, TLX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR2C2 geni (Tüm Gen Dizi Analizi)	NR2C2 gene (Sequence Analysis-All Coding Exons)	NR2C2	NR2C2, TR4, TAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR1I3 geni (Tüm Gen Dizi Analizi)	NR1I3 gene (Sequence Analysis-All Coding Exons)	NR1I3	NR1I3, CAR, MB67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR1I2 geni (Tüm Gen Dizi Analizi)	NR1I2 gene (Sequence Analysis-All Coding Exons)	NR1I2	NR1I2, PXR, SXR, PARq	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR1H2 geni (Tüm Gen Dizi Analizi)	NR1H2 gene (Sequence Analysis-All Coding Exons)	NR1H2	NR1H2, UNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR1D2 geni (Tüm Gen Dizi Analizi)	NR1D2 gene (Sequence Analysis-All Coding Exons)	NR1D2	NR1D2, RVR, BD73	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR1D1 geni (Tüm Gen Dizi Analizi)	NR1D1 gene (Sequence Analysis-All Coding Exons)	NR1D1	NR1D1, THRAL, EAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY6R geni (Tüm Gen Dizi Analizi)	NPY6R gene (Sequence Analysis-All Coding Exons)	NPY6R	NPY6R, PP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY5R geni (Tüm Gen Dizi Analizi)	NPY5R gene (Sequence Analysis-All Coding Exons)	NPY5R	NPY5R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY4R geni (Tüm Gen Dizi Analizi)	NPY4R gene (Sequence Analysis-All Coding Exons)	NPY4R	PPYR1, NPY4R, PP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY2R geni (Tüm Gen Dizi Analizi)	NPY2R gene (Sequence Analysis-All Coding Exons)	NPY2R	NPY2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY1R geni (Tüm Gen Dizi Analizi)	NPY1R gene (Sequence Analysis-All Coding Exons)	NPY1R	NPY1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPY geni (Tüm Gen Dizi Analizi)	NPY gene (Sequence Analysis-All Coding Exons)	NPY	NPY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPW geni (Tüm Gen Dizi Analizi)	NPW gene (Sequence Analysis-All Coding Exons)	NPW	NPW, PPNPW, PPL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPVF geni (Tüm Gen Dizi Analizi)	NPVF gene (Sequence Analysis-All Coding Exons)	NPVF	NPVF, RFRP, C7orf9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPTX2 geni (Tüm Gen Dizi Analizi)	NPTX2 gene (Sequence Analysis-All Coding Exons)	NPTX2	NPTX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NPTX1 geni (Tüm Gen Dizi Analizi)	NPTX1 gene (Sequence Analysis-All Coding Exons)	NPTX1	NPTX1, NP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPTN geni (Tüm Gen Dizi Analizi)	NPTN gene (Sequence Analysis-All Coding Exons)	NPTN	NPTN, SDFR1, GPS5, NPS5, GPS6, NPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPSR1-AS1 geni (Tüm Gen Dizi Analizi)	NPSR1-AS1 gene (Sequence Analysis-All Coding Exons)	NPSR1-AS1	AAA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPS geni (Tüm Gen Dizi Analizi)	NPS gene (Sequence Analysis-All Coding Exons)	NPS	NPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPR1 geni (Tüm Gen Dizi Analizi)	NPR1 gene (Sequence Analysis-All Coding Exons)	NPR1	NPR1, ANPRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPPC geni (Tüm Gen Dizi Analizi)	NPPC gene (Sequence Analysis-All Coding Exons)	NPPC	NPPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPPB geni (Tüm Gen Dizi Analizi)	NPPB gene (Sequence Analysis-All Coding Exons)	NPPB	NPPB, BNP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPNT geni (Tüm Gen Dizi Analizi)	NPNT gene (Sequence Analysis-All Coding Exons)	NPNT	NPNT, POEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPM3 geni (Tüm Gen Dizi Analizi)	NPM3 gene (Sequence Analysis-All Coding Exons)	NPM3	NPM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPM2 geni (Tüm Gen Dizi Analizi)	NPM2 gene (Sequence Analysis-All Coding Exons)	NPM2	NPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPLOC4 geni (Tüm Gen Dizi Analizi)	NPLOC4 gene (Sequence Analysis-All Coding Exons)	NPLOC4	NPL4, KIAA1499	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPL geni (Tüm Gen Dizi Analizi)	NPL gene (Sequence Analysis-All Coding Exons)	NPL	NPL, C1orf13, C112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPIPA1 geni (Tüm Gen Dizi Analizi)	NPIPA1 gene (Sequence Analysis-All Coding Exons)	NPIPA1	NPIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPFFR2 geni (Tüm Gen Dizi Analizi)	NPFFR2 gene (Sequence Analysis-All Coding Exons)	NPFFR2	GPR74, NPFF2, NPGPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPFFR1 geni (Tüm Gen Dizi Analizi)	NPFFR1 gene (Sequence Analysis-All Coding Exons)	NPFFR1	NPFF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPEPPS geni (Tüm Gen Dizi Analizi)	NPEPPS gene (Sequence Analysis-All Coding Exons)	NPEPPS	NPEPPS, PSA, MP100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPDC1 geni (Tüm Gen Dizi Analizi)	NPDC1 gene (Sequence Analysis-All Coding Exons)	NPDC1	NPDC1, CAB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPBWR2 geni (Tüm Gen Dizi Analizi)	NPBWR2 gene (Sequence Analysis-All Coding Exons)	NPBWR2	GPR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPBWR1 geni (Tüm Gen Dizi Analizi)	NPBWR1 gene (Sequence Analysis-All Coding Exons)	NPBWR1	NPBWR1, GPR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPB geni (Tüm Gen Dizi Analizi)	NPB gene (Sequence Analysis-All Coding Exons)	NPB	NPB, PPL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAT geni (Tüm Gen Dizi Analizi)	NPAT gene (Sequence Analysis-All Coding Exons)	NPAT	NPAT, E14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAS4 geni (Tüm Gen Dizi Analizi)	NPAS4 gene (Sequence Analysis-All Coding Exons)	NPAS4	NPAS4, NXF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAS3 geni (Tüm Gen Dizi Analizi)	NPAS3 gene (Sequence Analysis-All Coding Exons)	NPAS3	NPAS3, MOP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAS2 geni (Tüm Gen Dizi Analizi)	NPAS2 gene (Sequence Analysis-All Coding Exons)	NPAS2	NPAS2, MOP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAS1 geni (Tüm Gen Dizi Analizi)	NPAS1 gene (Sequence Analysis-All Coding Exons)	NPAS1	NPAS1, MOP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPAP1 geni (Tüm Gen Dizi Analizi)	NPAP1 gene (Sequence Analysis-All Coding Exons)	NPAP1	NPAP1, C15orf2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOXO1 geni (Tüm Gen Dizi Analizi)	NOXO1 gene (Sequence Analysis-All Coding Exons)	NOXO1	NOXO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOXA1 geni (Tüm Gen Dizi Analizi)	NOXA1 gene (Sequence Analysis-All Coding Exons)	NOXA1	NOXA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOX5 geni (Tüm Gen Dizi Analizi)	NOX5 gene (Sequence Analysis-All Coding Exons)	NOX5	NOX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOX4 geni (Tüm Gen Dizi Analizi)	NOX4 gene (Sequence Analysis-All Coding Exons)	NOX4	NOX4, RENOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOX3 geni (Tüm Gen Dizi Analizi)	NOX3 gene (Sequence Analysis-All Coding Exons)	NOX3	NOX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOX1 geni (Tüm Gen Dizi Analizi)	NOX1 gene (Sequence Analysis-All Coding Exons)	NOX1	NOX1, MOX1, NOH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOVA2 geni (Tüm Gen Dizi Analizi)	NOVA2 gene (Sequence Analysis-All Coding Exons)	NOVA2	NOVA3, ANOVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOVA1 geni (Tüm Gen Dizi Analizi)	NOVA1 gene (Sequence Analysis-All Coding Exons)	NOVA1	NOVA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOV geni (Tüm Gen Dizi Analizi)	NOV gene (Sequence Analysis-All Coding Exons)	NOV	NOV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOTUM geni (Tüm Gen Dizi Analizi)	NOTUM gene (Sequence Analysis-All Coding Exons)	NOTUM	NOTUM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOTCH4 geni (Tüm Gen Dizi Analizi)	NOTCH4 gene (Sequence Analysis-All Coding Exons)	NOTCH4	NOTCH4, INT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOSTRIN geni (Tüm Gen Dizi Analizi)	NOSTRIN gene (Sequence Analysis-All Coding Exons)	NOSTRIN	NOSTRIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOSIP geni (Tüm Gen Dizi Analizi)	NOSIP gene (Sequence Analysis-All Coding Exons)	NOSIP	NOSIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOS1AP geni (Tüm Gen Dizi Analizi)	NOS1AP gene (Sequence Analysis-All Coding Exons)	NOS1AP	NOS1AP, CAPON, KIAA0464	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOS1 geni (Tüm Gen Dizi Analizi)	NOS1 gene (Sequence Analysis-All Coding Exons)	NOS1	NOS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NORAD geni (Tüm Gen Dizi Analizi)	NORAD gene (Sequence Analysis-All Coding Exons)	NORAD	NORAD, LINC00657	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOP58 geni (Tüm Gen Dizi Analizi)	NOP58 gene (Sequence Analysis-All Coding Exons)	NOP58	NOP58, NOP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOP53 geni (Tüm Gen Dizi Analizi)	NOP53 gene (Sequence Analysis-All Coding Exons)	NOP53	GLTSCR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOP2 geni (Tüm Gen Dizi Analizi)	NOP2 gene (Sequence Analysis-All Coding Exons)	NOP2	NOL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOP16 geni (Tüm Gen Dizi Analizi)	NOP16 gene (Sequence Analysis-All Coding Exons)	NOP16	NOP15, HSPC111	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOP14 geni (Tüm Gen Dizi Analizi)	NOP14 gene (Sequence Analysis-All Coding Exons)	NOP14	NOP14, NOL14, NOP14, RES425	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOMO3 geni (Tüm Gen Dizi Analizi)	NOMO3 gene (Sequence Analysis-All Coding Exons)	NOMO3	NOMO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOMO2 geni (Tüm Gen Dizi Analizi)	NOMO2 gene (Sequence Analysis-All Coding Exons)	NOMO2	NOMO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOMO1 geni (Tüm Gen Dizi Analizi)	NOMO1 gene (Sequence Analysis-All Coding Exons)	NOMO1	NOMO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOM1 geni (Tüm Gen Dizi Analizi)	NOM1 gene (Sequence Analysis-All Coding Exons)	NOM1	NOM1, C7orf3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOLC1 geni (Tüm Gen Dizi Analizi)	NOLC1 gene (Sequence Analysis-All Coding Exons)	NOLC1	NOLC1, p130, NOPP140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL8 geni (Tüm Gen Dizi Analizi)	NOL8 gene (Sequence Analysis-All Coding Exons)	NOL8	NOL8, NOP132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL7 geni (Tüm Gen Dizi Analizi)	NOL7 gene (Sequence Analysis-All Coding Exons)	NOL7	NOL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL6 geni (Tüm Gen Dizi Analizi)	NOL6 gene (Sequence Analysis-All Coding Exons)	NOL6	NOL6, NRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL4 geni (Tüm Gen Dizi Analizi)	NOL4 gene (Sequence Analysis-All Coding Exons)	NOL4	NOL4, NOLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RCC2 geni (Tüm Gen Dizi Analizi)	RCC2 gene (Sequence Analysis-All Coding Exons)	RCC2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL11 geni (Tüm Gen Dizi Analizi)	NOL11 gene (Sequence Analysis-All Coding Exons)	NOL11	NOL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOL10 geni (Tüm Gen Dizi Analizi)	NOL10 gene (Sequence Analysis-All Coding Exons)	NOL10	NOL10, PQBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOD1 geni (Tüm Gen Dizi Analizi)	NOD1 gene (Sequence Analysis-All Coding Exons)	NOD1	NOD1, CARD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOCT geni (Tüm Gen Dizi Analizi)	NOCT gene (Sequence Analysis-All Coding Exons)	NOCT	CCRN4L, CCR4L, CCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOC4L geni (Tüm Gen Dizi Analizi)	NOC4L gene (Sequence Analysis-All Coding Exons)	NOC4L	NOC4L, NOC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOC3L geni (Tüm Gen Dizi Analizi)	NOC3L gene (Sequence Analysis-All Coding Exons)	NOC3L	NOC3L, FAD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOC2L geni (Tüm Gen Dizi Analizi)	NOC2L gene (Sequence Analysis-All Coding Exons)	NOC2L	NOC2L, NIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOA1 geni (Tüm Gen Dizi Analizi)	NOA1 gene (Sequence Analysis-All Coding Exons)	NOA1	NOA1, C4orf14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NNAT geni (Tüm Gen Dizi Analizi)	NNAT gene (Sequence Analysis-All Coding Exons)	NNAT	NNAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMUR2 geni (Tüm Gen Dizi Analizi)	NMUR2 gene (Sequence Analysis-All Coding Exons)	NMUR2	NMUR2, NMU2R, FM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMUR1 geni (Tüm Gen Dizi Analizi)	NMUR1 gene (Sequence Analysis-All Coding Exons)	NMUR1	NMUR1, GPR66, FM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMT2 geni (Tüm Gen Dizi Analizi)	NMT2 gene (Sequence Analysis-All Coding Exons)	NMT2	NMT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMT1 geni (Tüm Gen Dizi Analizi)	NMT1 gene (Sequence Analysis-All Coding Exons)	NMT1	NMT1, NMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBP7 geni (Tüm Gen Dizi Analizi)	RBP7 gene (Sequence Analysis-All Coding Exons)	RBP7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMNAT3 geni (Tüm Gen Dizi Analizi)	NMNAT3 gene (Sequence Analysis-All Coding Exons)	NMNAT3	NMNAT3, PNAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMNAT2 geni (Tüm Gen Dizi Analizi)	NMNAT2 gene (Sequence Analysis-All Coding Exons)	NMNAT2	NMNAT2, PNAT2, KIAA0479	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMI geni (Tüm Gen Dizi Analizi)	NMI gene (Sequence Analysis-All Coding Exons)	NMI	NMI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME7 geni (Tüm Gen Dizi Analizi)	NME7 gene (Sequence Analysis-All Coding Exons)	NME7	NME7, MN23H7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME6 geni (Tüm Gen Dizi Analizi)	NME6 gene (Sequence Analysis-All Coding Exons)	NME6	NME6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME5 geni (Tüm Gen Dizi Analizi)	NME5 gene (Sequence Analysis-All Coding Exons)	NME5	NME5, NM23H5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME4 geni (Tüm Gen Dizi Analizi)	NME4 gene (Sequence Analysis-All Coding Exons)	NME4	NME4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME3 geni (Tüm Gen Dizi Analizi)	NME3 gene (Sequence Analysis-All Coding Exons)	NME3	NME3, NM23H3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NME2 geni (Tüm Gen Dizi Analizi)	NME2 gene (Sequence Analysis-All Coding Exons)	NME2	NME2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMD3 geni (Tüm Gen Dizi Analizi)	NMD3 gene (Sequence Analysis-All Coding Exons)	NMD3	NMD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMBR geni (Tüm Gen Dizi Analizi)	NMBR gene (Sequence Analysis-All Coding Exons)	NMBR	NMBR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMB geni (Tüm Gen Dizi Analizi)	NMB gene (Sequence Analysis-All Coding Exons)	NMB	NMB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRX1 geni (Tüm Gen Dizi Analizi)	NLRX1 gene (Sequence Analysis-All Coding Exons)	NLRX1	NLRX1, NOD9, CLR11.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP9 geni (Tüm Gen Dizi Analizi)	NLRP9 gene (Sequence Analysis-All Coding Exons)	NLRP9	NALP9, NOD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP8 geni (Tüm Gen Dizi Analizi)	NLRP8 gene (Sequence Analysis-All Coding Exons)	NLRP8	NALP8, NOD16, PAN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP6 geni (Tüm Gen Dizi Analizi)	NLRP6 gene (Sequence Analysis-All Coding Exons)	NLRP6	NALP6, PYPAF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP5 geni (Tüm Gen Dizi Analizi)	NLRP5 gene (Sequence Analysis-All Coding Exons)	NLRP5	NALP5, MATER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP4 geni (Tüm Gen Dizi Analizi)	NLRP4 gene (Sequence Analysis-All Coding Exons)	NLRP4	NALP4, PAN2, PYPAF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP2 geni (Tüm Gen Dizi Analizi)	NLRP2 gene (Sequence Analysis-All Coding Exons)	NLRP2	NALP2, PAN1, PYPAF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP14 geni (Tüm Gen Dizi Analizi)	NLRP14 gene (Sequence Analysis-All Coding Exons)	NLRP14	NLRP1, NALP14, NOD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP13 geni (Tüm Gen Dizi Analizi)	NLRP13 gene (Sequence Analysis-All Coding Exons)	NLRP13	NALP13, NOD14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRP11 geni (Tüm Gen Dizi Analizi)	NLRP11 gene (Sequence Analysis-All Coding Exons)	NLRP11	NALP11, PYPAF7, NOD17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NLRP10 geni (Tüm Gen Dizi Analizi)	NLRP10 gene (Sequence Analysis-All Coding Exons)	NLRP10	NLRP10, NALP10, PYNOD, NOS8	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRC5 geni (Tüm Gen Dizi Analizi)	NLRC5 gene (Sequence Analysis-All Coding Exons)	NLRC5	NLRC5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLRC3 geni (Tüm Gen Dizi Analizi)	NLRC3 gene (Sequence Analysis-All Coding Exons)	NLRC3	NLRC3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLN geni (Tüm Gen Dizi Analizi)	NLN gene (Sequence Analysis-All Coding Exons)	NLN	NLN, KIAA1226, AGTBP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLGN4Y geni (Tüm Gen Dizi Analizi)	NLGN4Y gene (Sequence Analysis-All Coding Exons)	NLGN4Y	NLGN4Y, KIAA0951	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLGN2 geni (Tüm Gen Dizi Analizi)	NLGN2 gene (Sequence Analysis-All Coding Exons)	NLGN2	NLGN2, KIAA1366	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLGN1 geni (Tüm Gen Dizi Analizi)	NLGN1 gene (Sequence Analysis-All Coding Exons)	NLGN1	NLGN1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX6-3 geni (Tüm Gen Dizi Analizi)	NKX6-3 gene (Sequence Analysis-All Coding Exons)	NKX6-3	NKX6-3, NKX6.3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBM12 geni (Tüm Gen Dizi Analizi)	RBM12 gene (Sequence Analysis-All Coding Exons)	RBM12		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX6-2 geni (Tüm Gen Dizi Analizi)	NKX6-2 gene (Sequence Analysis-All Coding Exons)	NKX6-2	NKX6B, NKX6.2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX6-1 geni (Tüm Gen Dizi Analizi)	NKX6-1 gene (Sequence Analysis-All Coding Exons)	NKX6-1	NKX6-1, NKX6A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX3-1 geni (Tüm Gen Dizi Analizi)	NKX3-1 gene (Sequence Analysis-All Coding Exons)	NKX3-1	NKX3-1, NKX3A, BAPX2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX2-8 geni (Tüm Gen Dizi Analizi)	NKX2-8 gene (Sequence Analysis-All Coding Exons)	NKX2-8	NKX2-8, NKX2H, NKX2.8	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX2-4 geni (Tüm Gen Dizi Analizi)	NKX2-4 gene (Sequence Analysis-All Coding Exons)	NKX2-4	NKX2-4, NKX2D	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX2-3 geni (Tüm Gen Dizi Analizi)	NKX2-3 gene (Sequence Analysis-All Coding Exons)	NKX2-3	NKX2C, NK2.3, CSX3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKX2-2 geni (Tüm Gen Dizi Analizi)	NKX2-2 gene (Sequence Analysis-All Coding Exons)	NKX2-2	NKX2-2, NKX2B	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKTR geni (Tüm Gen Dizi Analizi)	NKTR gene (Sequence Analysis-All Coding Exons)	NKTR	NKTR	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKRF geni (Tüm Gen Dizi Analizi)	NKRF gene (Sequence Analysis-All Coding Exons)	NKRF	NKRF, NRF	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKIRAS2 geni (Tüm Gen Dizi Analizi)	NKIRAS2 gene (Sequence Analysis-All Coding Exons)	NKIRAS2	NKIRAS2, KBRAS2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKD2 geni (Tüm Gen Dizi Analizi)	NKD2 gene (Sequence Analysis-All Coding Exons)	NKD2	NKD2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKD1 geni (Tüm Gen Dizi Analizi)	NKD1 gene (Sequence Analysis-All Coding Exons)	NKD1	NKD1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKAP geni (Tüm Gen Dizi Analizi)	NKAP gene (Sequence Analysis-All Coding Exons)	NKAP	NKAP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKAIN4 geni (Tüm Gen Dizi Analizi)	NKAIN4 gene (Sequence Analysis-All Coding Exons)	NKAIN4	NKAIN4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKAIN2 geni (Tüm Gen Dizi Analizi)	NKAIN2 gene (Sequence Analysis-All Coding Exons)	NKAIN2	NKAIN2, TCBA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RBBP4 geni (Tüm Gen Dizi Analizi)	RBBP4 gene (Sequence Analysis-All Coding Exons)	RBBP4		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKAIN1 geni (Tüm Gen Dizi Analizi)	NKAIN1 gene (Sequence Analysis-All Coding Exons)	NKAIN1	NKAIN1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIT2 geni (Tüm Gen Dizi Analizi)	NIT2 gene (Sequence Analysis-All Coding Exons)	NIT2	NIT2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIT1 geni (Tüm Gen Dizi Analizi)	NIT1 gene (Sequence Analysis-All Coding Exons)	NIT1	NIT1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NISCH geni (Tüm Gen Dizi Analizi)	NISCH gene (Sequence Analysis-All Coding Exons)	NISCH	NISCH, IRAS, KIAA0975	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIPSNAP3A geni (Tüm Gen Dizi Analizi)	NIPSNAP3A gene (Sequence Analysis-All Coding Exons)	NIPSNAP3A	NIPSNAP3A, TASSC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIPSNAP2 geni (Tüm Gen Dizi Analizi)	NIPSNAP2 gene (Sequence Analysis-All Coding Exons)	NIPSNAP2	GBAS, NIPSNAP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIPSNAP1 geni (Tüm Gen Dizi Analizi)	NIPSNAP1 gene (Sequence Analysis-All Coding Exons)	NIPSNAP1	NIPSNAP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIPA2 geni (Tüm Gen Dizi Analizi)	NIPA2 gene (Sequence Analysis-All Coding Exons)	NIPA2	NIPA2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NINJ2 geni (Tüm Gen Dizi Analizi)	NINJ2 gene (Sequence Analysis-All Coding Exons)	NINJ2	NINJ2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NINJ1 geni (Tüm Gen Dizi Analizi)	NINJ1 gene (Sequence Analysis-All Coding Exons)	NINJ1	NINJ1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIFK geni (Tüm Gen Dizi Analizi)	NIFK gene (Sequence Analysis-All Coding Exons)	NIFK	MKI67IP, NIFK, NOPP34	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIF3L1 geni (Tüm Gen Dizi Analizi)	NIF3L1 gene (Sequence Analysis-All Coding Exons)	NIF3L1	NIF3L1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NID2 geni (Tüm Gen Dizi Analizi)	NID2 gene (Sequence Analysis-All Coding Exons)	NID2	NID2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NID1 geni (Tüm Gen Dizi Analizi)	NID1 gene (Sequence Analysis-All Coding Exons)	NID1	NID	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NICN1 geni (Tüm Gen Dizi Analizi)	NICN1 gene (Sequence Analysis-All Coding Exons)	NICN1	NICN1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NHLH2 geni (Tüm Gen Dizi Analizi)	NHLH2 gene (Sequence Analysis-All Coding Exons)	NHLH2	NHLH2, HEN2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NHLH1 geni (Tüm Gen Dizi Analizi)	NHLH1 gene (Sequence Analysis-All Coding Exons)	NHLH1	NHLH1, HEN1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NGRN geni (Tüm Gen Dizi Analizi)	NGRN gene (Sequence Analysis-All Coding Exons)	NGRN	NGRN	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NGFR geni (Tüm Gen Dizi Analizi)	NGFR gene (Sequence Analysis-All Coding Exons)	NGFR	NGFR, TNFRSF16	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASSF2 geni (Tüm Gen Dizi Analizi)	RASSF2 gene (Sequence Analysis-All Coding Exons)	RASSF2		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NGEF geni (Tüm Gen Dizi Analizi)	NGEF gene (Sequence Analysis-All Coding Exons)	NGEF	NGEF	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NGDN geni (Tüm Gen Dizi Analizi)	NGDN gene (Sequence Analysis-All Coding Exons)	NGDN	NGDN, NGD	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NGB geni (Tüm Gen Dizi Analizi)	NGB gene (Sequence Analysis-All Coding Exons)	NGB	NGB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFYC geni (Tüm Gen Dizi Analizi)	NFYC gene (Sequence Analysis-All Coding Exons)	NFYC	NFYC, CBFC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFYB geni (Tüm Gen Dizi Analizi)	NFYB gene (Sequence Analysis-All Coding Exons)	NFYB	NFYB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFYA geni (Tüm Gen Dizi Analizi)	NFYA gene (Sequence Analysis-All Coding Exons)	NFYA	NFYA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFX1 geni (Tüm Gen Dizi Analizi)	NFX1 gene (Sequence Analysis-All Coding Exons)	NFX1	NFX1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFS1 geni (Tüm Gen Dizi Analizi)	NFS1 gene (Sequence Analysis-All Coding Exons)	NFS1	NFS1, NIFS	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGRP3 geni (Tüm Gen Dizi Analizi)	RASGRP3 gene (Sequence Analysis-All Coding Exons)	RASGRP3		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFRKB geni (Tüm Gen Dizi Analizi)	NFRKB gene (Sequence Analysis-All Coding Exons)	NFRKB	NFRKB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFKBIZ geni (Tüm Gen Dizi Analizi)	NFKBIZ gene (Sequence Analysis-All Coding Exons)	NFKBIZ	NFKBIZ, INAP, MAIL, IKBZ	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFKBIE geni (Tüm Gen Dizi Analizi)	NFKBIE gene (Sequence Analysis-All Coding Exons)	NFKBIE	NFKBIE, IKBE	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RASGRF1 geni (Tüm Gen Dizi Analizi)	RASGRF1 gene (Sequence Analysis-All Coding Exons)	RASGRF1		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFKBIB geni (Tüm Gen Dizi Analizi)	NFKBIB gene (Sequence Analysis-All Coding Exons)	NFKBIB	NFKBIB, IKBB, TRIP9	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFIL3 geni (Tüm Gen Dizi Analizi)	NFIL3 gene (Sequence Analysis-All Coding Exons)	NFIL3	NFIL3, NFIL3A, E4BP4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFIC geni (Tüm Gen Dizi Analizi)	NFIC gene (Sequence Analysis-All Coding Exons)	NFIC	NFIC	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFIB geni (Tüm Gen Dizi Analizi)	NFIB gene (Sequence Analysis-All Coding Exons)	NFIB	NFIB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFIA geni (Tüm Gen Dizi Analizi)	NFIA gene (Sequence Analysis-All Coding Exons)	NFIA	NFIA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFE2L3 geni (Tüm Gen Dizi Analizi)	NFE2L3 gene (Sequence Analysis-All Coding Exons)	NFE2L3	NFE2L3, NRF3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFE2L2 geni (Tüm Gen Dizi Analizi)	NFE2L2 gene (Sequence Analysis-All Coding Exons)	NFE2L2	NFE2L2, NRF2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFE2L1 geni (Tüm Gen Dizi Analizi)	NFE2L1 gene (Sequence Analysis-All Coding Exons)	NFE2L1	NFE2L1, NRF1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFE2 geni (Tüm Gen Dizi Analizi)	NFE2 gene (Sequence Analysis-All Coding Exons)	NFE2	NFE2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFATC4 geni (Tüm Gen Dizi Analizi)	NFATC4 gene (Sequence Analysis-All Coding Exons)	NFATC4	NFATC4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFATC3 geni (Tüm Gen Dizi Analizi)	NFATC3 gene (Sequence Analysis-All Coding Exons)	NFATC3	NFATC3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFATC2IP geni (Tüm Gen Dizi Analizi)	NFATC2IP gene (Sequence Analysis-All Coding Exons)	NFATC2IP	NFATC2IP, NIP45	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFATC2 geni (Tüm Gen Dizi Analizi)	NFATC2 gene (Sequence Analysis-All Coding Exons)	NFATC2	NFATC2, NFATP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFATC1 geni (Tüm Gen Dizi Analizi)	NFATC1 gene (Sequence Analysis-All Coding Exons)	NFATC1	NFATC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFAT5 geni (Tüm Gen Dizi Analizi)	NFAT5 gene (Sequence Analysis-All Coding Exons)	NFAT5	NFAT5, KIAA0827, NFATL1, TONEBP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFASC geni (Tüm Gen Dizi Analizi)	NFASC gene (Sequence Analysis-All Coding Exons)	NFASC	NFASC, KIAA0756	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFAM1 geni (Tüm Gen Dizi Analizi)	NFAM1 gene (Sequence Analysis-All Coding Exons)	NFAM1	NFAM1, CNAIP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEUROG2 geni (Tüm Gen Dizi Analizi)	NEUROG2 gene (Sequence Analysis-All Coding Exons)	NEUROG2	NEUROG2, NGN2, ATOH4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEUROG1 geni (Tüm Gen Dizi Analizi)	NEUROG1 gene (Sequence Analysis-All Coding Exons)	NEUROG1	NEUROG1, NEUROD3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEUROD6 geni (Tüm Gen Dizi Analizi)	NEUROD6 gene (Sequence Analysis-All Coding Exons)	NEUROD6	NEUROD6, ATOH2, MATH2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEUROD4 geni (Tüm Gen Dizi Analizi)	NEUROD4 gene (Sequence Analysis-All Coding Exons)	NEUROD4	NEUROD4, MATH3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEUROD2 geni (Tüm Gen Dizi Analizi)	NEUROD2 gene (Sequence Analysis-All Coding Exons)	NEUROD2	NEUROD2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEURL4 geni (Tüm Gen Dizi Analizi)	NEURL4 gene (Sequence Analysis-All Coding Exons)	NEURL4	NEURL4, KIAA1787	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEURL3 geni (Tüm Gen Dizi Analizi)	NEURL3 gene (Sequence Analysis-All Coding Exons)	NEURL3	NEURL3, LINCR	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPH1 geni (Tüm Gen Dizi Analizi)	RAPH1 gene (Sequence Analysis-All Coding Exons)	RAPH1		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEURL1B geni (Tüm Gen Dizi Analizi)	NEURL1B gene (Sequence Analysis-All Coding Exons)	NEURL1B	NEURL1B, NEUR2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF5 geni (Tüm Gen Dizi Analizi)	RAPGEF5 gene (Sequence Analysis-All Coding Exons)	RAPGEF5		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF4 geni (Tüm Gen Dizi Analizi)	RAPGEF4 gene (Sequence Analysis-All Coding Exons)	RAPGEF4		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEURL1 geni (Tüm Gen Dizi Analizi)	NEURL1 gene (Sequence Analysis-All Coding Exons)	NEURL1	NEURL1, NEURL, NEUR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAPGEF2 geni (Tüm Gen Dizi Analizi)	RAPGEF2 gene (Sequence Analysis-All Coding Exons)	RAPGEF2		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEU4 geni (Tüm Gen Dizi Analizi)	NEU4 gene (Sequence Analysis-All Coding Exons)	NEU4	NEU4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEU3 geni (Tüm Gen Dizi Analizi)	NEU3 gene (Sequence Analysis-All Coding Exons)	NEU3	NEU3, SIAL3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEU2 geni (Tüm Gen Dizi Analizi)	NEU2 gene (Sequence Analysis-All Coding Exons)	NEU2	NEU2, SIAL2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEOT2 geni (Tüm Gen Dizi Analizi)	NETO2 gene (Sequence Analysis-All Coding Exons)	NETO2	NEOT2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NETO1 geni (Tüm Gen Dizi Analizi)	NETO1 gene (Sequence Analysis-All Coding Exons)	NETO1	NETO1, BCTL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NES geni (Tüm Gen Dizi Analizi)	NES gene (Sequence Analysis-All Coding Exons)	NES	NES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEPRO geni (Tüm Gen Dizi Analizi)	NEPRO gene (Sequence Analysis-All Coding Exons)	NEPRO	NEPRO, C3orf17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEO1 geni (Tüm Gen Dizi Analizi)	NEO1 gene (Sequence Analysis-All Coding Exons)	NEO1	NEO1, NGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RANGAP1 geni (Tüm Gen Dizi Analizi)	RANGAP1 gene (Sequence Analysis-All Coding Exons)	RANGAP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NENF geni (Tüm Gen Dizi Analizi)	NENF gene (Sequence Analysis-All Coding Exons)	NENF	NENF, CIR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEMP2 geni (Tüm Gen Dizi Analizi)	NEMP2 gene (Sequence Analysis-All Coding Exons)	NEMP2	NEMP2, TMEM194B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEMP1 geni (Tüm Gen Dizi Analizi)	NEMP1 gene (Sequence Analysis-All Coding Exons)	NEMP1	NEMP1, TMEM194A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEMF geni (Tüm Gen Dizi Analizi)	NEMF gene (Sequence Analysis-All Coding Exons)	NEMF	NEMF, SDCCAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELL2 geni (Tüm Gen Dizi Analizi)	NELL2 gene (Sequence Analysis-All Coding Exons)	NELL2	NELL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELL1 geni (Tüm Gen Dizi Analizi)	NELL1 gene (Sequence Analysis-All Coding Exons)	NELL1	NELL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELFE geni (Tüm Gen Dizi Analizi)	NELFE gene (Sequence Analysis-All Coding Exons)	NELFE	RDBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELFCD geni (Tüm Gen Dizi Analizi)	NELFCD gene (Sequence Analysis-All Coding Exons)	NELFCD	TH1L, TH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELFB geni (Tüm Gen Dizi Analizi)	NELFB gene (Sequence Analysis-All Coding Exons)	NELFB	COBRA1, NELFB, KIAA1182	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NELFA geni (Tüm Gen Dizi Analizi)	NELFA gene (Sequence Analysis-All Coding Exons)	NELFA	WHSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK7 geni (Tüm Gen Dizi Analizi)	NEK7 gene (Sequence Analysis-All Coding Exons)	NEK7	NEK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK6 geni (Tüm Gen Dizi Analizi)	NEK6 gene (Sequence Analysis-All Coding Exons)	NEK6	NEK6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RALY geni (Tüm Gen Dizi Analizi)	RALY gene (Sequence Analysis-All Coding Exons)	RALY		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK5 geni (Tüm Gen Dizi Analizi)	NEK5 gene (Sequence Analysis-All Coding Exons)	NEK5	NEK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK4 geni (Tüm Gen Dizi Analizi)	NEK4 gene (Sequence Analysis-All Coding Exons)	NEK4	NEK4, STK2, NRK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK3 geni (Tüm Gen Dizi Analizi)	NEK3 gene (Sequence Analysis-All Coding Exons)	NEK3	NEK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEK11 geni (Tüm Gen Dizi Analizi)	NEK11 gene (Sequence Analysis-All Coding Exons)	NEK11	NEK11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEIL1 geni (Tüm Gen Dizi Analizi)	NEIL1 gene (Sequence Analysis-All Coding Exons)	NEIL1	NEIL1, NEI1, FPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEGR1 geni (Tüm Gen Dizi Analizi)	NEGR1 gene (Sequence Analysis-All Coding Exons)	NEGR1	NEGR1, KILON	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEFM geni (Tüm Gen Dizi Analizi)	NEFM gene (Sequence Analysis-All Coding Exons)	NEFM	NEFM, NEF3, NFM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEDD9 geni (Tüm Gen Dizi Analizi)	NEDD9 gene (Sequence Analysis-All Coding Exons)	NEDD9	NEDD9, HEF1, CASL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEDD8 geni (Tüm Gen Dizi Analizi)	NEDD8 gene (Sequence Analysis-All Coding Exons)	NEDD8	NEDD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEDD4 geni (Tüm Gen Dizi Analizi)	NEDD4 gene (Sequence Analysis-All Coding Exons)	NEDD4	NEDD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEDD1 geni (Tüm Gen Dizi Analizi)	NEDD1 gene (Sequence Analysis-All Coding Exons)	NEDD1	NEDD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NECTIN3 geni (Tüm Gen Dizi Analizi)	NECTIN3 gene (Sequence Analysis-All Coding Exons)	NECTIN3	PVRL3, PRR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NECTIN2 geni (Tüm Gen Dizi Analizi)	NECTIN2 gene (Sequence Analysis-All Coding Exons)	NECTIN2	PVRL2, HVEB, PVRR2, PRR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NECAP2 geni (Tüm Gen Dizi Analizi)	NECAP2 gene (Sequence Analysis-All Coding Exons)	NECAP2	NECAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NECAB3 geni (Tüm Gen Dizi Analizi)	NECAB3 gene (Sequence Analysis-All Coding Exons)	NECAB3	NECAB3, EFCBP3, NP1, APBA2BP, XB51, STIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEBL geni (Tüm Gen Dizi Analizi)	NEBL gene (Sequence Analysis-All Coding Exons)	NEBL	NEBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEAT1 geni (Tüm Gen Dizi Analizi)	NEAT1 gene (Sequence Analysis-All Coding Exons)	NEAT1	NEAT1, TNCRNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFV3 geni (Tüm Gen Dizi Analizi)	NDUFV3 gene (Sequence Analysis-All Coding Exons)	NDUFV3	NDUFV3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFS5 geni (Tüm Gen Dizi Analizi)	NDUFS5 gene (Sequence Analysis-All Coding Exons)	NDUFS5	NDUFS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAET1G geni (Tüm Gen Dizi Analizi)	RAET1G gene (Sequence Analysis-All Coding Exons)	RAET1G		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAET1E geni (Tüm Gen Dizi Analizi)	RAET1E gene (Sequence Analysis-All Coding Exons)	RAET1E		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAE1 geni (Tüm Gen Dizi Analizi)	RAE1 gene (Sequence Analysis-All Coding Exons)	RAE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFC2 geni (Tüm Gen Dizi Analizi)	NDUFC2 gene (Sequence Analysis-All Coding Exons)	NDUFC2	NDUFC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFC1 geni (Tüm Gen Dizi Analizi)	NDUFC1 gene (Sequence Analysis-All Coding Exons)	NDUFC1	NDUFC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB8 geni (Tüm Gen Dizi Analizi)	NDUFB8 gene (Sequence Analysis-All Coding Exons)	NDUFB8	NDUFB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB7 geni (Tüm Gen Dizi Analizi)	NDUFB7 gene (Sequence Analysis-All Coding Exons)	NDUFB7	NDUFB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB4 geni (Tüm Gen Dizi Analizi)	NDUFB4 gene (Sequence Analysis-All Coding Exons)	NDUFB4	NDUFB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB10 geni (Tüm Gen Dizi Analizi)	NDUFB10 gene (Sequence Analysis-All Coding Exons)	NDUFB10	NDUFB10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB1 geni (Tüm Gen Dizi Analizi)	NDUFB1 gene (Sequence Analysis-All Coding Exons)	NDUFB1	NDUFB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFAF7 geni (Tüm Gen Dizi Analizi)	NDUFAF7 gene (Sequence Analysis-All Coding Exons)	NDUFAF7	NDUFAF7, MIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFAB1 geni (Tüm Gen Dizi Analizi)	NDUFAB1 gene (Sequence Analysis-All Coding Exons)	NDUFAB1	NDUFAB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA8 geni (Tüm Gen Dizi Analizi)	NDUFA8 gene (Sequence Analysis-All Coding Exons)	NDUFA8	NDUFA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA7 geni (Tüm Gen Dizi Analizi)	NDUFA7 gene (Sequence Analysis-All Coding Exons)	NDUFA7	NDUFA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA6 geni (Tüm Gen Dizi Analizi)	NDUFA6 gene (Sequence Analysis-All Coding Exons)	NDUFA6	NDUFA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA5 geni (Tüm Gen Dizi Analizi)	NDUFA5 gene (Sequence Analysis-All Coding Exons)	NDUFA5	NDUFA5, UQOR13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA4 geni (Tüm Gen Dizi Analizi)	NDUFA4 gene (Sequence Analysis-All Coding Exons)	NDUFA4	NDUFA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFA3 geni (Tüm Gen Dizi Analizi)	NDUFA3 gene (Sequence Analysis-All Coding Exons)	NDUFA3	NDUFA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDST4 geni (Tüm Gen Dizi Analizi)	NDST4 gene (Sequence Analysis-All Coding Exons)	NDST4	NDST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDST2 geni (Tüm Gen Dizi Analizi)	NDST2 gene (Sequence Analysis-All Coding Exons)	NDST2	NDST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDRG4 geni (Tüm Gen Dizi Analizi)	NDRG4 gene (Sequence Analysis-All Coding Exons)	NDRG4	NDRG4, SMAP8, KIAA1180	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDRG3 geni (Tüm Gen Dizi Analizi)	NDRG3 gene (Sequence Analysis-All Coding Exons)	NDRG3	NDRG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDRG2 geni (Tüm Gen Dizi Analizi)	NDRG2 gene (Sequence Analysis-All Coding Exons)	NDRG2	NDRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDOR1 geni (Tüm Gen Dizi Analizi)	NDOR1 gene (Sequence Analysis-All Coding Exons)	NDOR1	NR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDNF geni (Tüm Gen Dizi Analizi)	NDNF gene (Sequence Analysis-All Coding Exons)	NDNF	NDNF, C4orf31, NORD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDFIP2 geni (Tüm Gen Dizi Analizi)	NDFIP2 gene (Sequence Analysis-All Coding Exons)	NDFIP2	NDFIP2, NW4/BP5A, KIAA1165	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDFIP1 geni (Tüm Gen Dizi Analizi)	NDFIP1 gene (Sequence Analysis-All Coding Exons)	NDFIP1	NDFIP1, N4WBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDEL1 geni (Tüm Gen Dizi Analizi)	NDEL1 gene (Sequence Analysis-All Coding Exons)	NDEL1	NUDEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDC1 geni (Tüm Gen Dizi Analizi)	NDC1 gene (Sequence Analysis-All Coding Exons)	NDC1	TMEM48, NDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCS1 geni (Tüm Gen Dizi Analizi)	NCS1 gene (Sequence Analysis-All Coding Exons)	NCS1	FREQ, NCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCR3LG1 geni (Tüm Gen Dizi Analizi)	NCR3LG1 gene (Sequence Analysis-All Coding Exons)	NCR3LG1	B7H6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAC1 geni (Tüm Gen Dizi Analizi)	RAC1 gene (Sequence Analysis-All Coding Exons)	RAC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCR2 geni (Tüm Gen Dizi Analizi)	NCR2 gene (Sequence Analysis-All Coding Exons)	NCR2	NCR2, LY95, NKP44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCR1 geni (Tüm Gen Dizi Analizi)	NCR1 gene (Sequence Analysis-All Coding Exons)	NCR1	NCR1, LY94, NKP46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOR2 geni (Tüm Gen Dizi Analizi)	NCOR2 gene (Sequence Analysis-All Coding Exons)	NCOR2	NCOR2, SMRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOR1 geni (Tüm Gen Dizi Analizi)	NCOR1 gene (Sequence Analysis-All Coding Exons)	NCOR1	NCOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA7 geni (Tüm Gen Dizi Analizi)	NCOA7 gene (Sequence Analysis-All Coding Exons)	NCOA7	NCOA7, ERAP140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA6 geni (Tüm Gen Dizi Analizi)	NCOA6 gene (Sequence Analysis-All Coding Exons)	NCOA6	NCOA6, ASC2, RAP250, NRC, PRIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA5 geni (Tüm Gen Dizi Analizi)	NCOA5 gene (Sequence Analysis-All Coding Exons)	NCOA5	NCOA5, CIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABGAP1L geni (Tüm Gen Dizi Analizi)	RABGAP1L gene (Sequence Analysis-All Coding Exons)	RABGAP1L		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA4 geni (Tüm Gen Dizi Analizi)	NCOA4 gene (Sequence Analysis-All Coding Exons)	NCOA4	NCOA4, ELE1, PTC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RABEPK geni (Tüm Gen Dizi Analizi)	RABEPK gene (Sequence Analysis-All Coding Exons)	RABEPK		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA3 geni (Tüm Gen Dizi Analizi)	NCOA3 gene (Sequence Analysis-All Coding Exons)	NCOA3	NCOA3, AIB1, TNRC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA2 geni (Tüm Gen Dizi Analizi)	NCOA2 gene (Sequence Analysis-All Coding Exons)	NCOA2	NCOA2, GRIP1, TIF2, SRC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCOA1 geni (Tüm Gen Dizi Analizi)	NCOA1 gene (Sequence Analysis-All Coding Exons)	NCOA1	NCOA1, SRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCL geni (Tüm Gen Dizi Analizi)	NCL gene (Sequence Analysis-All Coding Exons)	NCL	NCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCKIPSD geni (Tüm Gen Dizi Analizi)	NCKIPSD gene (Sequence Analysis-All Coding Exons)	NCKIPSD	NCKIPSD, AF3P21, SPIN90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCKAPSL geni (Tüm Gen Dizi Analizi)	NCKAPSL gene (Sequence Analysis-All Coding Exons)	NCKAPSL	NCKAPSL, KIAA1602	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCKAP5 geni (Tüm Gen Dizi Analizi)	NCKAP5 gene (Sequence Analysis-All Coding Exons)	NCKAP5	NCKAP5, NAP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCKAP1L geni (Tüm Gen Dizi Analizi)	NCKAP1L gene (Sequence Analysis-All Coding Exons)	NCKAP1L	HEM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCKAP1 geni (Tüm Gen Dizi Analizi)	NCKAP1 gene (Sequence Analysis-All Coding Exons)	NCKAP1	NCKAP1, NAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCK2 geni (Tüm Gen Dizi Analizi)	NCK2 gene (Sequence Analysis-All Coding Exons)	NCK2	NCK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCK1 geni (Tüm Gen Dizi Analizi)	NCK1 gene (Sequence Analysis-All Coding Exons)	NCK1	NCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCEH1 geni (Tüm Gen Dizi Analizi)	NCEH1 gene (Sequence Analysis-All Coding Exons)	NCEH1	NCEH1, NCEH, KIAA1363	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCDN geni (Tüm Gen Dizi Analizi)	NCDN gene (Sequence Analysis-All Coding Exons)	NCDN	NCDN, KIAA0607	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCCRP1 geni (Tüm Gen Dizi Analizi)	NCCRP1 gene (Sequence Analysis-All Coding Exons)	NCCRP1	NCCRP1, FBXO50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCBP3 geni (Tüm Gen Dizi Analizi)	NCBP3 gene (Sequence Analysis-All Coding Exons)	NCBP3	NCBP3, C17orf85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NCBP2 geni (Tüm Gen Dizi Analizi)	NCBP2 gene (Sequence Analysis-All Coding Exons)	NCBP2	NCBP2, CBP20, NIP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCBP1 geni (Tüm Gen Dizi Analizi)	NCBP1 gene (Sequence Analysis-All Coding Exons)	NCBP1	NCBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB3IP geni (Tüm Gen Dizi Analizi)	RAB3IP gene (Sequence Analysis-All Coding Exons)	RAB3IP		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPH2 geni (Tüm Gen Dizi Analizi)	NCAPH2 gene (Sequence Analysis-All Coding Exons)	NCAPH2	NCAPH2, CAPH2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPH geni (Tüm Gen Dizi Analizi)	NCAPH gene (Sequence Analysis-All Coding Exons)	NCAPH	BRRN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPG2 geni (Tüm Gen Dizi Analizi)	NCAPG2 gene (Sequence Analysis-All Coding Exons)	NCAPG2	NCAPG2, CAPG2, MTB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPG geni (Tüm Gen Dizi Analizi)	NCAPG gene (Sequence Analysis-All Coding Exons)	NCAPG	NCAPG, CAPG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPD3 geni (Tüm Gen Dizi Analizi)	NCAPD3 gene (Sequence Analysis-All Coding Exons)	NCAPD3	NCAPD3, CAPD3, KIAA0056	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAPD2 geni (Tüm Gen Dizi Analizi)	NCAPD2 gene (Sequence Analysis-All Coding Exons)	NCAPD2	NCAPD2, CNAP1, KIAA0159	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAN geni (Tüm Gen Dizi Analizi)	NCAN gene (Sequence Analysis-All Coding Exons)	NCAN	CSPG3, NCAN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAM2 geni (Tüm Gen Dizi Analizi)	NCAM2 gene (Sequence Analysis-All Coding Exons)	NCAM2	NCAM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCAM1 geni (Tüm Gen Dizi Analizi)	NCAM1 gene (Sequence Analysis-All Coding Exons)	NCAM1	NCAM1, MSK39	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCALD geni (Tüm Gen Dizi Analizi)	NCALD gene (Sequence Analysis-All Coding Exons)	NCALD	NCALD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBR1 geni (Tüm Gen Dizi Analizi)	NBR1 gene (Sequence Analysis-All Coding Exons)	NBR1	NBR1, M17S2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF9 geni (Tüm Gen Dizi Analizi)	NBPF9 gene (Sequence Analysis-All Coding Exons)	NBPF9	NBPF9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF8 geni (Tüm Gen Dizi Analizi)	NBPF8 gene (Sequence Analysis-All Coding Exons)	NBPF8	NBPF8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF6 geni (Tüm Gen Dizi Analizi)	NBPF6 gene (Sequence Analysis-All Coding Exons)	NBPF6	NBPF6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF4 geni (Tüm Gen Dizi Analizi)	NBPF4 gene (Sequence Analysis-All Coding Exons)	NBPF4	NBPF4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF3 geni (Tüm Gen Dizi Analizi)	NBPF3 gene (Sequence Analysis-All Coding Exons)	NBPF3	NBPF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF20 geni (Tüm Gen Dizi Analizi)	NBPF20 gene (Sequence Analysis-All Coding Exons)	NBPF20	NBPF20	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF15 geni (Tüm Gen Dizi Analizi)	NBPF15 gene (Sequence Analysis-All Coding Exons)	NBPF15	NBPF15, MGC8902	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF15 geni (Tüm Gen Dizi Analizi)	NBPF15 gene (Sequence Analysis-All Coding Exons)	NBPF15	NBPF16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF14 geni (Tüm Gen Dizi Analizi)	NBPF14 gene (Sequence Analysis-All Coding Exons)	NBPF14	NBPF14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF12 geni (Tüm Gen Dizi Analizi)	NBPF12 gene (Sequence Analysis-All Coding Exons)	NBPF12	NBPF12, COAS1, KIAA1245	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF11 geni (Tüm Gen Dizi Analizi)	NBPF11 gene (Sequence Analysis-All Coding Exons)	NBPF11	NBPF11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF10 geni (Tüm Gen Dizi Analizi)	NBPF10 gene (Sequence Analysis-All Coding Exons)	NBPF10	NBPF10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
RAB27B geni (Tüm Gen Dizi Analizi)	RAB27B gene (Sequence Analysis-All Coding Exons)	RAB27B		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBL1 geni (Tüm Gen Dizi Analizi)	NBL1 gene (Sequence Analysis-All Coding Exons)	NBL1	D1S1733E, DAN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBEAL1 geni (Tüm Gen Dizi Analizi)	NBEAL1 gene (Sequence Analysis-All Coding Exons)	NBEAL1	NBEAL1, ALS2CR17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBEA geni (Tüm Gen Dizi Analizi)	NBEA gene (Sequence Analysis-All Coding Exons)	NBEA	NBEA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAXD geni (Tüm Gen Dizi Analizi)	NAXD gene (Sequence Analysis-All Coding Exons)	NAXD	CARKD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAV3 geni (Tüm Gen Dizi Analizi)	NAV3 gene (Sequence Analysis-All Coding Exons)	NAV3	NAV3, POMFIL1, KIAA0938	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAV2 geni (Tüm Gen Dizi Analizi)	NAV2 gene (Sequence Analysis-All Coding Exons)	NAV2	NAV2, RAINB1, KIAA1419	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAV1 geni (Tüm Gen Dizi Analizi)	NAV1 gene (Sequence Analysis-All Coding Exons)	NAV1	NAV1, POMFIL3, KIAA1151	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT8B geni (Tüm Gen Dizi Analizi)	NAT8B gene (Sequence Analysis-All Coding Exons)	NAT8B	NAT8B, CML2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT8 geni (Tüm Gen Dizi Analizi)	NAT8 gene (Sequence Analysis-All Coding Exons)	NAT8	NAT8, TSCS10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT6 geni (Tüm Gen Dizi Analizi)	NAT6 gene (Sequence Analysis-All Coding Exons)	NAT6	FUS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT16 geni (Tüm Gen Dizi Analizi)	NAT16 gene (Sequence Analysis-All Coding Exons)	NAT16	NAT16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT10 geni (Tüm Gen Dizi Analizi)	NAT10 gene (Sequence Analysis-All Coding Exons)	NAT10	NAT10, ALP, KIAA1709	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAT1 geni (Tüm Gen Dizi Analizi)	NAT1 gene (Sequence Analysis-All Coding Exons)	NAT1	NAT1, AAC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NASP geni (Tüm Gen Dizi Analizi)	NASP gene (Sequence Analysis-All Coding Exons)	NASP	NASP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NARS geni (Tüm Gen Dizi Analizi)	NARS gene (Sequence Analysis-All Coding Exons)	NARS	NARS, ASNRS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NARFL geni (Tüm Gen Dizi Analizi)	NARFL gene (Sequence Analysis-All Coding Exons)	NARFL	NARFL, IOP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPSA geni (Tüm Gen Dizi Analizi)	NAPSA gene (Sequence Analysis-All Coding Exons)	NAPSA	NAPA, NAP1, SNAPA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPRT geni (Tüm Gen Dizi Analizi)	NAPRT gene (Sequence Analysis-All Coding Exons)	NAPRT	NAPRT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPG geni (Tüm Gen Dizi Analizi)	NAPG gene (Sequence Analysis-All Coding Exons)	NAPG	NAPG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPEPLD geni (Tüm Gen Dizi Analizi)	NAPEPLD gene (Sequence Analysis-All Coding Exons)	NAPEPLD	NAPEPLD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPB geni (Tüm Gen Dizi Analizi)	NAPB gene (Sequence Analysis-All Coding Exons)	NAPB	NAPB, SNAPB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAPA geni (Tüm Gen Dizi Analizi)	NAPA gene (Sequence Analysis-All Coding Exons)	NAPA	NAPA, SNAPA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAP1L5 geni (Tüm Gen Dizi Analizi)	NAP1L5 gene (Sequence Analysis-All Coding Exons)	NAP1L5	NAP1L5, DRLM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAP1L4 geni (Tüm Gen Dizi Analizi)	NAP1L4 gene (Sequence Analysis-All Coding Exons)	NAP1L4	NAP1L4, NAP2L, NAP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAP1L3 geni (Tüm Gen Dizi Analizi)	NAP1L3 gene (Sequence Analysis-All Coding Exons)	NAP1L3	NAP1L3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAP1L2 geni (Tüm Gen Dizi Analizi)	NAP1L2 gene (Sequence Analysis-All Coding Exons)	NAP1L2	NAP1L2, BPX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAP1L1 geni (Tüm Gen Dizi Analizi)	NAP1L1 gene (Sequence Analysis-All Coding Exons)	NAP1L1	NAP1L1, NAP1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
QRFPR geni (Tüm Gen Dizi Analizi)	QRFPR gene (Sequence Analysis-All Coding Exons)	QRFPR		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NANP geni (Tüm Gen Dizi Analizi)	NANP gene (Sequence Analysis-All Coding Exons)	NANP	NANP, HDHD4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NANOS3 geni (Tüm Gen Dizi Analizi)	NANOS3 gene (Sequence Analysis-All Coding Exons)	NANOS3	NANOS3, NOS3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NANOS2 geni (Tüm Gen Dizi Analizi)	NANOS2 gene (Sequence Analysis-All Coding Exons)	NANOS2	NANOS2, NOS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NANOG geni (Tüm Gen Dizi Analizi)	NANOG gene (Sequence Analysis-All Coding Exons)	NANOG	NANOG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAMPT geni (Tüm Gen Dizi Analizi)	NAMPT gene (Sequence Analysis-All Coding Exons)	NAMPT	NAMPT, PBEF1, VF, PBEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAIP geni (Tüm Gen Dizi Analizi)	NAIP gene (Sequence Analysis-All Coding Exons)	NAIP	BIRC1, NAIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAIP geni (Tüm Gen Dizi Analizi)	NAIP gene (Sequence Analysis-All Coding Exons)	NAIP	BIRC1, NAIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAIP geni (Tüm Gen Dizi Analizi)	NAIP gene (Sequence Analysis-All Coding Exons)	NAIP	BIRC1, NAIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAIF1 geni (Tüm Gen Dizi Analizi)	NAIF1 gene (Sequence Analysis-All Coding Exons)	NAIF1	C9orf90, NAIF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAGPA geni (Tüm Gen Dizi Analizi)	NAGPA gene (Sequence Analysis-All Coding Exons)	NAGPA	NAGPA, UCE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAGK geni (Tüm Gen Dizi Analizi)	NAGK gene (Sequence Analysis-All Coding Exons)	NAGK	NAGK, GNK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAE1 geni (Tüm Gen Dizi Analizi)	NAE1 gene (Sequence Analysis-All Coding Exons)	NAE1	NAE1, APPBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYGO1 geni (Tüm Gen Dizi Analizi)	PYGO1 gene (Sequence Analysis-All Coding Exons)	PYGO1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NADSYN1 geni (Tüm Gen Dizi Analizi)	NADSYN1 gene (Sequence Analysis-All Coding Exons)	NADSYN1	NADSYN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NADK geni (Tüm Gen Dizi Analizi)	NADK gene (Sequence Analysis-All Coding Exons)	NADK	NADK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NACC2 geni (Tüm Gen Dizi Analizi)	NACC2 gene (Sequence Analysis-All Coding Exons)	NACC2	NACC2, RBB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PYDC2 geni (Tüm Gen Dizi Analizi)	PYDC2 gene (Sequence Analysis-All Coding Exons)	PYDC2	PYDC2, POP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NACA geni (Tüm Gen Dizi Analizi)	NACA gene (Sequence Analysis-All Coding Exons)	NACA	NACA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NABP2 geni (Tüm Gen Dizi Analizi)	NABP2 gene (Sequence Analysis-All Coding Exons)	NABP2	OBFC2B, SSB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NABP1 geni (Tüm Gen Dizi Analizi)	NABP1 gene (Sequence Analysis-All Coding Exons)	NABP1	OBFC2A, SSB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAB2 geni (Tüm Gen Dizi Analizi)	NAB2 gene (Sequence Analysis-All Coding Exons)	NAB2	NAB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAB1 geni (Tüm Gen Dizi Analizi)	NAB1 gene (Sequence Analysis-All Coding Exons)	NAB1	NAB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAALADL2 geni (Tüm Gen Dizi Analizi)	NAALADL2 gene (Sequence Analysis-All Coding Exons)	NAALADL2	NAALADL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAALADL1 geni (Tüm Gen Dizi Analizi)	NAALADL1 gene (Sequence Analysis-All Coding Exons)	NAALADL1	NAALADL1, I100	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAALAD2 geni (Tüm Gen Dizi Analizi)	NAALAD2 gene (Sequence Analysis-All Coding Exons)	NAALAD2	NAALAD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAAA geni (Tüm Gen Dizi Analizi)	NAAA gene (Sequence Analysis-All Coding Exons)	NAAA	NAAA, ASAHL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAA60 geni (Tüm Gen Dizi Analizi)	NAA60 gene (Sequence Analysis-All Coding Exons)	NAA60	NAA60, NAT15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAA50 geni (Tüm Gen Dizi Analizi)	NAA50 gene (Sequence Analysis-All Coding Exons)	NAA50	NAT13, NAT5, SAN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAA25 geni (Tüm Gen Dizi Analizi)	NAA25 gene (Sequence Analysis-All Coding Exons)	NAA25	MDM20, C12orf30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAA20 geni (Tüm Gen Dizi Analizi)	NAA20 gene (Sequence Analysis-All Coding Exons)	NAA20	NAT5, NAT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NAA15 geni (Tüm Gen Dizi Analizi)	NAA15 gene (Sequence Analysis-All Coding Exons)	NAA15	NAA15, NARG1, NATH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
N6AMT1 geni (Tüm Gen Dizi Analizi)	N6AMT1 gene (Sequence Analysis-All Coding Exons)	N6AMT1	N6AMT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
N4BP2L2 geni (Tüm Gen Dizi Analizi)	N4BP2L2 gene (Sequence Analysis-All Coding Exons)	N4BP2L2	N4BP2L2, PFAAP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MZT2B geni (Tüm Gen Dizi Analizi)	MZT2B gene (Sequence Analysis-All Coding Exons)	MZT2B	FAM128B, MOZART2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MZT2A geni (Tüm Gen Dizi Analizi)	MZT2A gene (Sequence Analysis-All Coding Exons)	MZT2A	FAM128A, MOZART2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MZT1 geni (Tüm Gen Dizi Analizi)	MZT1 gene (Sequence Analysis-All Coding Exons)	MZT1	C13orf37, MOZART1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MZF1 geni (Tüm Gen Dizi Analizi)	MZF1 gene (Sequence Analysis-All Coding Exons)	MZF1	ZNF42, MZF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MZB1 geni (Tüm Gen Dizi Analizi)	MZB1 gene (Sequence Analysis-All Coding Exons)	MZB1	MZB1, PACAP, MGC29506	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MYZAP geni (Tüm Gen Dizi Analizi)	MYZAP gene (Sequence Analysis-All Coding Exons)	MYZAP	GUP, GCOM1, MYOZAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYT1 geni (Tüm Gen Dizi Analizi)	MYT1 gene (Sequence Analysis-All Coding Exons)	MYT1	MYT1, PLPB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYSM1 geni (Tüm Gen Dizi Analizi)	MYSM1 gene (Sequence Analysis-All Coding Exons)	MYSM1	MYSM1, 2ADUB, KIAA1915	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYRIP geni (Tüm Gen Dizi Analizi)	MYRIP gene (Sequence Analysis-All Coding Exons)	MYRIP	MYRIP, SLAC2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYRF geni (Tüm Gen Dizi Analizi)	MYRF gene (Sequence Analysis-All Coding Exons)	MYRF	C11orf9, KIAA0954	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOZ3 geni (Tüm Gen Dizi Analizi)	MYOZ3 gene (Sequence Analysis-All Coding Exons)	MYOZ3	MYOZ3, FRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOZ1 geni (Tüm Gen Dizi Analizi)	MYOZ1 gene (Sequence Analysis-All Coding Exons)	MYOZ1	MYOZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOM3 geni (Tüm Gen Dizi Analizi)	MYOM3 gene (Sequence Analysis-All Coding Exons)	MYOM3	MYOM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOM2 geni (Tüm Gen Dizi Analizi)	MYOM2 gene (Sequence Analysis-All Coding Exons)	MYOM2	MYOM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOM1 geni (Tüm Gen Dizi Analizi)	MYOM1 gene (Sequence Analysis-All Coding Exons)	MYOM1	MYOM1, SKELEMIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOG geni (Tüm Gen Dizi Analizi)	MYOG gene (Sequence Analysis-All Coding Exons)	MYOG	MYOG, MYF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOF geni (Tüm Gen Dizi Analizi)	MYOF gene (Sequence Analysis-All Coding Exons)	MYOF	MYOF, FER1L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOD1 geni (Tüm Gen Dizi Analizi)	MYOD1 gene (Sequence Analysis-All Coding Exons)	MYOD1	MYOD1, MYF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYOCD geni (Tüm Gen Dizi Analizi)	MYOCD gene (Sequence Analysis-All Coding Exons)	MYOCD	MYOCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO9A geni (Tüm Gen Dizi Analizi)	MYO9A gene (Sequence Analysis-All Coding Exons)	MYO9A	MYO9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO7B geni (Tüm Gen Dizi Analizi)	MYO7B gene (Sequence Analysis-All Coding Exons)	MYO7B	MYO7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO5C geni (Tüm Gen Dizi Analizi)	MYO5C gene (Sequence Analysis-All Coding Exons)	MYO5C	MYO5C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO3B geni (Tüm Gen Dizi Analizi)	MYO3B gene (Sequence Analysis-All Coding Exons)	MYO3B	MYO3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1H geni (Tüm Gen Dizi Analizi)	MYO1H gene (Sequence Analysis-All Coding Exons)	MYO1H	MYO1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1G geni (Tüm Gen Dizi Analizi)	MYO1G gene (Sequence Analysis-All Coding Exons)	MYO1G	MYO1G, HA2, HLA-HA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1F geni (Tüm Gen Dizi Analizi)	MYO1F gene (Sequence Analysis-All Coding Exons)	MYO1F	MYO1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1D geni (Tüm Gen Dizi Analizi)	MYO1D gene (Sequence Analysis-All Coding Exons)	MYO1D	MYO1D, KIAA0727	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1C geni (Tüm Gen Dizi Analizi)	MYO1C gene (Sequence Analysis-All Coding Exons)	MYO1C	MYO1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1B geni (Tüm Gen Dizi Analizi)	MYO1B gene (Sequence Analysis-All Coding Exons)	MYO1B	MYO1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO1A geni (Tüm Gen Dizi Analizi)	MYO1A gene (Sequence Analysis-All Coding Exons)	MYO1A	MYO1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO19 geni (Tüm Gen Dizi Analizi)	MYO19 gene (Sequence Analysis-All Coding Exons)	MYO19	MYO19, MYOHD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO18A geni (Tüm Gen Dizi Analizi)	MYO18A gene (Sequence Analysis-All Coding Exons)	MYO18A	MYO18A, SPR210	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO16 geni (Tüm Gen Dizi Analizi)	MYO16 gene (Sequence Analysis-All Coding Exons)	MYO16	MYO16, MYAP3, KIAA0865	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYO10 geni (Tüm Gen Dizi Analizi)	MYO10 gene (Sequence Analysis-All Coding Exons)	MYO10	MYO10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYNN geni (Tüm Gen Dizi Analizi)	MYNN gene (Sequence Analysis-All Coding Exons)	MYNN	MYNN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYLPF geni (Tüm Gen Dizi Analizi)	MYLPF gene (Sequence Analysis-All Coding Exons)	MYLPF	MYLPF, MLC2B, MRLC2, MYL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYLK3 geni (Tüm Gen Dizi Analizi)	MYLK3 gene (Sequence Analysis-All Coding Exons)	MYLK3	MYLK3, MLCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYLIP geni (Tüm Gen Dizi Analizi)	MYLIP gene (Sequence Analysis-All Coding Exons)	MYLIP	MYLIP, MIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL9 geni (Tüm Gen Dizi Analizi)	MYL9 gene (Sequence Analysis-All Coding Exons)	MYL9	MYL9, MLC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL7 geni (Tüm Gen Dizi Analizi)	MYL7 gene (Sequence Analysis-All Coding Exons)	MYL7	MYL7, MYL2A, MLC2A, MYLC2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL6B geni (Tüm Gen Dizi Analizi)	MYL6B gene (Sequence Analysis-All Coding Exons)	MYL6B	MYL6B, MLC1SA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL6 geni (Tüm Gen Dizi Analizi)	MYL6 gene (Sequence Analysis-All Coding Exons)	MYL6	MYL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL5 geni (Tüm Gen Dizi Analizi)	MYL5 gene (Sequence Analysis-All Coding Exons)	MYL5	MYL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL12B geni (Tüm Gen Dizi Analizi)	MYL12B gene (Sequence Analysis-All Coding Exons)	MYL12B	MYL12B, MRLC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL10 geni (Tüm Gen Dizi Analizi)	MYL10 gene (Sequence Analysis-All Coding Exons)	MYL10	MYL10, PLRLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYL1 geni (Tüm Gen Dizi Analizi)	MYL1 gene (Sequence Analysis-All Coding Exons)	MYL1	MYL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH7B geni (Tüm Gen Dizi Analizi)	MYH7B gene (Sequence Analysis-All Coding Exons)	MYH7B	MYH7B, MYH14, KIAA1512	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH4 geni (Tüm Gen Dizi Analizi)	MYH4 gene (Sequence Analysis-All Coding Exons)	MYH4	MYH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPN4 geni (Tüm Gen Dizi Analizi)	PTPN4 gene (Sequence Analysis-All Coding Exons)	PTPN4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH15 geni (Tüm Gen Dizi Analizi)	MYH15 gene (Sequence Analysis-All Coding Exons)	MYH15	MYH15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH13 geni (Tüm Gen Dizi Analizi)	MYH13 gene (Sequence Analysis-All Coding Exons)	MYH13	MYH13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH10 geni (Tüm Gen Dizi Analizi)	MYH10 gene (Sequence Analysis-All Coding Exons)	MYH10	MYH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYH1 geni (Tüm Gen Dizi Analizi)	MYH1 gene (Sequence Analysis-All Coding Exons)	MYH1	MYH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYF5 geni (Tüm Gen Dizi Analizi)	MYF5 gene (Sequence Analysis-All Coding Exons)	MYF5	MYF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYEOV geni (Tüm Gen Dizi Analizi)	MYEOV gene (Sequence Analysis-All Coding Exons)	MYEOV	MYEOV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYDGF geni (Tüm Gen Dizi Analizi)	MYDGF gene (Sequence Analysis-All Coding Exons)	MYDGF	C19orf10, SF20, IL25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCT1 geni (Tüm Gen Dizi Analizi)	MYCT1 gene (Sequence Analysis-All Coding Exons)	MYCT1	MYCT1, MTLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCNUT geni (Tüm Gen Dizi Analizi)	MYCNUT gene (Sequence Analysis-All Coding Exons)	MYCNUT	MYCNUT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCNOS geni (Tüm Gen Dizi Analizi)	MYCNOS gene (Sequence Analysis-All Coding Exons)	MYCNOS	MYCNOS, NCYM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCL geni (Tüm Gen Dizi Analizi)	MYCL gene (Sequence Analysis-All Coding Exons)	MYCL	MYCL, LMYC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCBPAP geni (Tüm Gen Dizi Analizi)	MYCBPAP gene (Sequence Analysis-All Coding Exons)	MYCBPAP	MYCBPAP, AMAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCBP2 geni (Tüm Gen Dizi Analizi)	MYCBP2 gene (Sequence Analysis-All Coding Exons)	MYCBP2	MYCBP2, PAM, KIAA0916	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYCBP geni (Tüm Gen Dizi Analizi)	MYCBP gene (Sequence Analysis-All Coding Exons)	MYCBP	MYCBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYBPH geni (Tüm Gen Dizi Analizi)	MYBPH gene (Sequence Analysis-All Coding Exons)	MYBPH	MYBPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYBPC2 geni (Tüm Gen Dizi Analizi)	MYBPC2 gene (Sequence Analysis-All Coding Exons)	MYBPC2	MYBPC2, MYBPCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYBL2 geni (Tüm Gen Dizi Analizi)	MYBL2 gene (Sequence Analysis-All Coding Exons)	MYBL2	MYBL2, BMYB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYBL1 geni (Tüm Gen Dizi Analizi)	MYBL1 gene (Sequence Analysis-All Coding Exons)	MYBL1	MYBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYBBP1A geni (Tüm Gen Dizi Analizi)	MYBBP1A gene (Sequence Analysis-All Coding Exons)	MYBBP1A	MYBBP1A, P160	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PTPMT1 geni (Tüm Gen Dizi Analizi)	PTPMT1 gene (Sequence Analysis-All Coding Exons)	PTPMT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MYADM geni (Tüm Gen Dizi Analizi)	MYADM gene (Sequence Analysis-All Coding Exons)	MYADM	MYADM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MXRA8 geni (Tüm Gen Dizi Analizi)	MXRA8 gene (Sequence Analysis-All Coding Exons)	MXRA8	MXRA8, ASP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MXRA5 geni (Tüm Gen Dizi Analizi)	MXRA5 gene (Sequence Analysis-All Coding Exons)	MXRA5	MXRA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MXD3 geni (Tüm Gen Dizi Analizi)	MXD3 gene (Sequence Analysis-All Coding Exons)	MXD3	MXD3, MAD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MXD1 geni (Tüm Gen Dizi Analizi)	MXD1 gene (Sequence Analysis-All Coding Exons)	MXD1	MAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MX2 geni (Tüm Gen Dizi Analizi)	MX2 gene (Sequence Analysis-All Coding Exons)	MX2	MX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MX1 geni (Tüm Gen Dizi Analizi)	MX1 gene (Sequence Analysis-All Coding Exons)	MX1	MX1, MX, IFI78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MVP geni (Tüm Gen Dizi Analizi)	MVP gene (Sequence Analysis-All Coding Exons)	MVP	MVP, LRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUSTN1 geni (Tüm Gen Dizi Analizi)	MUSTN1 gene (Sequence Analysis-All Coding Exons)	MUSTN1	MUSTN1, MUSTANG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUSB1 geni (Tüm Gen Dizi Analizi)	MUSB1 gene (Sequence Analysis-All Coding Exons)	MUSB1	MUSB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUL1 geni (Tüm Gen Dizi Analizi)	MUL1 gene (Sequence Analysis-All Coding Exons)	MUL1	MUL1, MULAN, C1orf166	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUCL1 geni (Tüm Gen Dizi Analizi)	MUCL1 gene (Sequence Analysis-All Coding Exons)	MUCL1	SBEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC6 geni (Tüm Gen Dizi Analizi)	MUC6 gene (Sequence Analysis-All Coding Exons)	MUC6	MUC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC5AC geni (Tüm Gen Dizi Analizi)	MUC5AC gene (Sequence Analysis-All Coding Exons)	MUC5AC	MUC5AC, MUC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC4 geni (Tüm Gen Dizi Analizi)	MUC4 gene (Sequence Analysis-All Coding Exons)	MUC4	MUC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC3A geni (Tüm Gen Dizi Analizi)	MUC3A gene (Sequence Analysis-All Coding Exons)	MUC3A	MUC3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC22 geni (Tüm Gen Dizi Analizi)	MUC22 gene (Sequence Analysis-All Coding Exons)	MUC22	MUC22, PBMUCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC21 geni (Tüm Gen Dizi Analizi)	MUC21 gene (Sequence Analysis-All Coding Exons)	MUC21	MUC21, C6orf205	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC20 geni (Tüm Gen Dizi Analizi)	MUC20 gene (Sequence Analysis-All Coding Exons)	MUC20	MUC20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC17 geni (Tüm Gen Dizi Analizi)	MUC17 gene (Sequence Analysis-All Coding Exons)	MUC17	MUC17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC16 geni (Tüm Gen Dizi Analizi)	MUC16 gene (Sequence Analysis-All Coding Exons)	MUC16	MUC16, CA125	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC13 geni (Tüm Gen Dizi Analizi)	MUC13 gene (Sequence Analysis-All Coding Exons)	MUC13	MUC13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC12 geni (Tüm Gen Dizi Analizi)	MUC12 gene (Sequence Analysis-All Coding Exons)	MUC12	MUC12, MUC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTX2 geni (Tüm Gen Dizi Analizi)	MTX2 gene (Sequence Analysis-All Coding Exons)	MTX2	MTX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTX1 geni (Tüm Gen Dizi Analizi)	MTX1 gene (Sequence Analysis-All Coding Exons)	MTX1	MTX1, MTXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTSS1L geni (Tüm Gen Dizi Analizi)	MTSS1L gene (Sequence Analysis-All Coding Exons)	MTSS1L	MTSS1L, ABBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTSS1 geni (Tüm Gen Dizi Analizi)	MTSS1 gene (Sequence Analysis-All Coding Exons)	MTSS1	MTSS1, MIM, KIAA0429	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTRNR2L1 geni (Tüm Gen Dizi Analizi)	MTRNR2L1 gene (Sequence Analysis-All Coding Exons)	MTRNR2L1	MTRNR2L1, HN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTRF1 geni (Tüm Gen Dizi Analizi)	MTRF1 gene (Sequence Analysis-All Coding Exons)	MTRF1	MTRF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MTPN geni (Tüm Gen Dizi Analizi)	MTPN gene (Sequence Analysis-All Coding Exons)	MTPN	MTPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTNR1A geni (Tüm Gen Dizi Analizi)	MTNR1A gene (Sequence Analysis-All Coding Exons)	MTNR1A	MTNR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR9 geni (Tüm Gen Dizi Analizi)	MTMR9 gene (Sequence Analysis-All Coding Exons)	MTMR9	MTMR9, MTMR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR7 geni (Tüm Gen Dizi Analizi)	MTMR7 gene (Sequence Analysis-All Coding Exons)	MTMR7	MTMR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR6 geni (Tüm Gen Dizi Analizi)	MTMR6 gene (Sequence Analysis-All Coding Exons)	MTMR6	MTMR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR4 geni (Tüm Gen Dizi Analizi)	MTMR4 gene (Sequence Analysis-All Coding Exons)	MTMR4	MTMR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR3 geni (Tüm Gen Dizi Analizi)	MTMR3 gene (Sequence Analysis-All Coding Exons)	MTMR3	MTMR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR12 geni (Tüm Gen Dizi Analizi)	MTMR12 gene (Sequence Analysis-All Coding Exons)	MTMR12	MTMR12, 3PAP, KIAA1682	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTMR1 geni (Tüm Gen Dizi Analizi)	MTMR1 gene (Sequence Analysis-All Coding Exons)	MTMR1	MTMR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTIF2 geni (Tüm Gen Dizi Analizi)	MTIF2 gene (Sequence Analysis-All Coding Exons)	MTIF2	MTIF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTHFSD geni (Tüm Gen Dizi Analizi)	MTHFSD gene (Sequence Analysis-All Coding Exons)	MTHFSD	MTHFSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTHFS geni (Tüm Gen Dizi Analizi)	MTHFS gene (Sequence Analysis-All Coding Exons)	MTHFS	MTHFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTHFD2L geni (Tüm Gen Dizi Analizi)	MTHFD2L gene (Sequence Analysis-All Coding Exons)	MTHFD2L	MTHFD2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTHFD1L geni (Tüm Gen Dizi Analizi)	MTHFD1L gene (Sequence Analysis-All Coding Exons)	MTHFD1L	MTHFD1L, MTC1THFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTG2 geni (Tüm Gen Dizi Analizi)	MTG2 gene (Sequence Analysis-All Coding Exons)	MTG2	GTPBP5, OBGH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTFP1 geni (Tüm Gen Dizi Analizi)	MTFP1 gene (Sequence Analysis-All Coding Exons)	MTFP1	MTFP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTF2 geni (Tüm Gen Dizi Analizi)	MTF2 gene (Sequence Analysis-All Coding Exons)	MTF2	MTF2, PCL2, M96	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTF1 geni (Tüm Gen Dizi Analizi)	MTF1 gene (Sequence Analysis-All Coding Exons)	MTF1	MTF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTERF4 geni (Tüm Gen Dizi Analizi)	MTERF4 gene (Sequence Analysis-All Coding Exons)	MTERF4	MTERF4, MTERFD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTERF3 geni (Tüm Gen Dizi Analizi)	MTERF3 gene (Sequence Analysis-All Coding Exons)	MTERF3	MTERF3, MTERFD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTERF2 geni (Tüm Gen Dizi Analizi)	MTERF2 gene (Sequence Analysis-All Coding Exons)	MTERF2	MTERF2, MTERFD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTERF1 geni (Tüm Gen Dizi Analizi)	MTERF1 gene (Sequence Analysis-All Coding Exons)	MTERF1	MTERF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTDH geni (Tüm Gen Dizi Analizi)	MTDH gene (Sequence Analysis-All Coding Exons)	MTDH	MTDH, AEG1, LYRIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTCP1 geni (Tüm Gen Dizi Analizi)	MTCP1 gene (Sequence Analysis-All Coding Exons)	MTCP1	MTCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTCL1 geni (Tüm Gen Dizi Analizi)	MTCL1 gene (Sequence Analysis-All Coding Exons)	MTCL1	MTCL1, KIAA0802	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTCH2 geni (Tüm Gen Dizi Analizi)	MTCH2 gene (Sequence Analysis-All Coding Exons)	MTCH2	MTCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTCH1 geni (Tüm Gen Dizi Analizi)	MTCH1 gene (Sequence Analysis-All Coding Exons)	MTCH1	MTCH1, PSAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTBP geni (Tüm Gen Dizi Analizi)	MTBP gene (Sequence Analysis-All Coding Exons)	MTBP	MTBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTA2 geni (Tüm Gen Dizi Analizi)	MTA2 gene (Sequence Analysis-All Coding Exons)	MTA2	MTA2, MTA1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTA1 geni (Tüm Gen Dizi Analizi)	MTA1 gene (Sequence Analysis-All Coding Exons)	MTA1	MTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT4 geni (Tüm Gen Dizi Analizi)	MT4 gene (Sequence Analysis-All Coding Exons)	MT4	MT4, MTIV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT3 geni (Tüm Gen Dizi Analizi)	MT3 gene (Sequence Analysis-All Coding Exons)	MT3	MT3, GIFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT2A geni (Tüm Gen Dizi Analizi)	MT2A gene (Sequence Analysis-All Coding Exons)	MT2A	MT2A, MT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1X geni (Tüm Gen Dizi Analizi)	MT1X gene (Sequence Analysis-All Coding Exons)	MT1X	MT1X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1M geni (Tüm Gen Dizi Analizi)	MT1M gene (Sequence Analysis-All Coding Exons)	MT1M	MT1K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1L geni (Tüm Gen Dizi Analizi)	MT1L gene (Sequence Analysis-All Coding Exons)	MT1L	MT1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1JP geni (Tüm Gen Dizi Analizi)	MT1JP gene (Sequence Analysis-All Coding Exons)	MT1JP	MT1J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1H geni (Tüm Gen Dizi Analizi)	MT1H gene (Sequence Analysis-All Coding Exons)	MT1H	MT1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1G geni (Tüm Gen Dizi Analizi)	MT1G gene (Sequence Analysis-All Coding Exons)	MT1G	MT1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1F geni (Tüm Gen Dizi Analizi)	MT1F gene (Sequence Analysis-All Coding Exons)	MT1F	MT1F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1E geni (Tüm Gen Dizi Analizi)	MT1E gene (Sequence Analysis-All Coding Exons)	MT1E	MT1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1B geni (Tüm Gen Dizi Analizi)	MT1B gene (Sequence Analysis-All Coding Exons)	MT1B	MT1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT1A geni (Tüm Gen Dizi Analizi)	MT1A gene (Sequence Analysis-All Coding Exons)	MT1A	MT1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MST1 geni (Tüm Gen Dizi Analizi)	MST1 gene (Sequence Analysis-All Coding Exons)	MST1	MST1, HGFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSS51 geni (Tüm Gen Dizi Analizi)	MSS51 gene (Sequence Analysis-All Coding Exons)	MSS51	MSS51, ZMYND17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSRB2 geni (Tüm Gen Dizi Analizi)	MSRB2 gene (Sequence Analysis-All Coding Exons)	MSRB2	MSRB2, CBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSRB1 geni (Tüm Gen Dizi Analizi)	MSRB1 gene (Sequence Analysis-All Coding Exons)	MSRB1	MSRB1, SEPX1, SELX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSMP geni (Tüm Gen Dizi Analizi)	MSMP gene (Sequence Analysis-All Coding Exons)	MSMP	MSMP, PSMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSLN geni (Tüm Gen Dizi Analizi)	MSLN gene (Sequence Analysis-All Coding Exons)	MSLN	MSLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSL3 geni (Tüm Gen Dizi Analizi)	MSL3 gene (Sequence Analysis-All Coding Exons)	MSL3	MSL3L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSL2 geni (Tüm Gen Dizi Analizi)	MSL2 gene (Sequence Analysis-All Coding Exons)	MSL2	MSL2, KIAA1585	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSL1 geni (Tüm Gen Dizi Analizi)	MSL1 gene (Sequence Analysis-All Coding Exons)	MSL1	MSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSORS1C2 geni (Tüm Gen Dizi Analizi)	PSORS1C2 gene (Sequence Analysis-All Coding Exons)	PSORS1C2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSI2 geni (Tüm Gen Dizi Analizi)	MSI2 gene (Sequence Analysis-All Coding Exons)	MSI2	MSI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSI1 geni (Tüm Gen Dizi Analizi)	MSI1 gene (Sequence Analysis-All Coding Exons)	MSI1	MSI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSH4 geni (Tüm Gen Dizi Analizi)	MSH4 gene (Sequence Analysis-All Coding Exons)	MSH4	MSH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSGN1 geni (Tüm Gen Dizi Analizi)	MSGN1 gene (Sequence Analysis-All Coding Exons)	MSGN1	MSGN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSC geni (Tüm Gen Dizi Analizi)	MSC gene (Sequence Analysis-All Coding Exons)	MSC	MSC, ABF1, MYOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A8 geni (Tüm Gen Dizi Analizi)	MS4A8 gene (Sequence Analysis-All Coding Exons)	MS4A8	MS4A8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A7 geni (Tüm Gen Dizi Analizi)	MS4A7 gene (Sequence Analysis-All Coding Exons)	MS4A7	MS4A7, CFFM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A6E geni (Tüm Gen Dizi Analizi)	MS4A6E gene (Sequence Analysis-All Coding Exons)	MS4A6E	MS4A6E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A6A geni (Tüm Gen Dizi Analizi)	MS4A6A gene (Sequence Analysis-All Coding Exons)	MS4A6A	MS4A6A, MS4A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A5 geni (Tüm Gen Dizi Analizi)	MS4A5 gene (Sequence Analysis-All Coding Exons)	MS4A5	MS4A5, TETM4, CD20L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A4E geni (Tüm Gen Dizi Analizi)	MS4A4E gene (Sequence Analysis-All Coding Exons)	MS4A4E	MS4A4E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A4A geni (Tüm Gen Dizi Analizi)	MS4A4A gene (Sequence Analysis-All Coding Exons)	MS4A4A	MS4A4A, MS4A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A3 geni (Tüm Gen Dizi Analizi)	MS4A3 gene (Sequence Analysis-All Coding Exons)	MS4A3	MS4A3, HTM4, CD20L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A12 geni (Tüm Gen Dizi Analizi)	MS4A12 gene (Sequence Analysis-All Coding Exons)	MS4A12	MS4A12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MS4A10 geni (Tüm Gen Dizi Analizi)	MS4A10 gene (Sequence Analysis-All Coding Exons)	MS4A10	MS4A10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRVI1 geni (Tüm Gen Dizi Analizi)	MRVI1 gene (Sequence Analysis-All Coding Exons)	MRVI1	MRVI1, IRAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRRF geni (Tüm Gen Dizi Analizi)	MRRF gene (Sequence Analysis-All Coding Exons)	MRRF	MRRF, RRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS9 geni (Tüm Gen Dizi Analizi)	MRPS9 gene (Sequence Analysis-All Coding Exons)	MRPS9	MRPS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS7 geni (Tüm Gen Dizi Analizi)	MRPS7 gene (Sequence Analysis-All Coding Exons)	MRPS7	MRPS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS6 geni (Tüm Gen Dizi Analizi)	MRPS6 gene (Sequence Analysis-All Coding Exons)	MRPS6	MRPS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS5 geni (Tüm Gen Dizi Analizi)	MRPS5 gene (Sequence Analysis-All Coding Exons)	MRPS5	MRPS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS36 geni (Tüm Gen Dizi Analizi)	MRPS36 gene (Sequence Analysis-All Coding Exons)	MRPS36	MRPS36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS35 geni (Tüm Gen Dizi Analizi)	MRPS35 gene (Sequence Analysis-All Coding Exons)	MRPS35	MRPS35, MRPS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS34 geni (Tüm Gen Dizi Analizi)	MRPS34 gene (Sequence Analysis-All Coding Exons)	MRPS34	MRPS34, MRPS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS33 geni (Tüm Gen Dizi Analizi)	MRPS33 gene (Sequence Analysis-All Coding Exons)	MRPS33	MRPS33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS31 geni (Tüm Gen Dizi Analizi)	MRPS31 gene (Sequence Analysis-All Coding Exons)	MRPS31	MRPS31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS30 geni (Tüm Gen Dizi Analizi)	MRPS30 gene (Sequence Analysis-All Coding Exons)	MRPS30	MRPS30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PSMB6 geni (Tüm Gen Dizi Analizi)	PSMB6 gene (Sequence Analysis-All Coding Exons)	PSMB6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS28 geni (Tüm Gen Dizi Analizi)	MRPS28 gene (Sequence Analysis-All Coding Exons)	MRPS28	MRPS28, MRPS35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS27 geni (Tüm Gen Dizi Analizi)	MRPS27 gene (Sequence Analysis-All Coding Exons)	MRPS27	MRPS27, KIAA0264	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS26 geni (Tüm Gen Dizi Analizi)	MRPS26 gene (Sequence Analysis-All Coding Exons)	MRPS26	MRPS26, MRPS13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS25 geni (Tüm Gen Dizi Analizi)	MRPS25 gene (Sequence Analysis-All Coding Exons)	MRPS25	MRPS25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS24 geni (Tüm Gen Dizi Analizi)	MRPS24 gene (Sequence Analysis-All Coding Exons)	MRPS24	MRPS24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS23 geni (Tüm Gen Dizi Analizi)	MRPS23 gene (Sequence Analysis-All Coding Exons)	MRPS23	MRPS23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS21 geni (Tüm Gen Dizi Analizi)	MRPS21 gene (Sequence Analysis-All Coding Exons)	MRPS21	MRPS21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS2 geni (Tüm Gen Dizi Analizi)	MRPS2 gene (Sequence Analysis-All Coding Exons)	MRPS2	MRPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS18C geni (Tüm Gen Dizi Analizi)	MRPS18C gene (Sequence Analysis-All Coding Exons)	MRPS18C	MRPS18C, MRPS18-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS18B geni (Tüm Gen Dizi Analizi)	MRPS18B gene (Sequence Analysis-All Coding Exons)	MRPS18B	MRPS18B, MRPS18-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS18A geni (Tüm Gen Dizi Analizi)	MRPS18A gene (Sequence Analysis-All Coding Exons)	MRPS18A	MRPS18A, MRPS18-3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS17 geni (Tüm Gen Dizi Analizi)	MRPS17 gene (Sequence Analysis-All Coding Exons)	MRPS17	MRPS17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MRPS15 geni (Tüm Gen Dizi Analizi)	MRPS15 gene (Sequence Analysis-All Coding Exons)	MRPS15	MRPS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS14 geni (Tüm Gen Dizi Analizi)	MRPS14 gene (Sequence Analysis-All Coding Exons)	MRPS14	MRPS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS12 geni (Tüm Gen Dizi Analizi)	MRPS12 gene (Sequence Analysis-All Coding Exons)	MRPS12	MRPS12, RPSM12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS11 geni (Tüm Gen Dizi Analizi)	MRPS11 gene (Sequence Analysis-All Coding Exons)	MRPS11	MRPS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPS10 geni (Tüm Gen Dizi Analizi)	MRPS10 gene (Sequence Analysis-All Coding Exons)	MRPS10	MRPS10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL9 geni (Tüm Gen Dizi Analizi)	MRPL9 gene (Sequence Analysis-All Coding Exons)	MRPL9	MRPL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL58 geni (Tüm Gen Dizi Analizi)	MRPL58 gene (Sequence Analysis-All Coding Exons)	MRPL58	ICT1, DS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL57 geni (Tüm Gen Dizi Analizi)	MRPL57 gene (Sequence Analysis-All Coding Exons)	MRPL57	MRP63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL55 geni (Tüm Gen Dizi Analizi)	MRPL55 gene (Sequence Analysis-All Coding Exons)	MRPL55	MRPL55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL54 geni (Tüm Gen Dizi Analizi)	MRPL54 gene (Sequence Analysis-All Coding Exons)	MRPL54	MRPL54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL53 geni (Tüm Gen Dizi Analizi)	MRPL53 gene (Sequence Analysis-All Coding Exons)	MRPL53	MRPL53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL52 geni (Tüm Gen Dizi Analizi)	MRPL52 gene (Sequence Analysis-All Coding Exons)	MRPL52	MRPL52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL51 geni (Tüm Gen Dizi Analizi)	MRPL51 gene (Sequence Analysis-All Coding Exons)	MRPL51	MRPL51, MRP64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL50 geni (Tüm Gen Dizi Analizi)	MRPL50 gene (Sequence Analysis-All Coding Exons)	MRPL50	MRPL50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL49 geni (Tüm Gen Dizi Analizi)	MRPL49 gene (Sequence Analysis-All Coding Exons)	MRPL49	MRPL49, NOF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL48 geni (Tüm Gen Dizi Analizi)	MRPL48 gene (Sequence Analysis-All Coding Exons)	MRPL48	MRPL48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL47 geni (Tüm Gen Dizi Analizi)	MRPL47 gene (Sequence Analysis-All Coding Exons)	MRPL47	MRPL47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL46 geni (Tüm Gen Dizi Analizi)	MRPL46 gene (Sequence Analysis-All Coding Exons)	MRPL46	MRPL46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL45 geni (Tüm Gen Dizi Analizi)	MRPL45 gene (Sequence Analysis-All Coding Exons)	MRPL45	MRPL45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL43 geni (Tüm Gen Dizi Analizi)	MRPL43 gene (Sequence Analysis-All Coding Exons)	MRPL43	MRPL43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL42 geni (Tüm Gen Dizi Analizi)	MRPL42 gene (Sequence Analysis-All Coding Exons)	MRPL42	MRPL42, MRPL31, MRPS32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL41 geni (Tüm Gen Dizi Analizi)	MRPL41 gene (Sequence Analysis-All Coding Exons)	MRPL41	MRPL41, MRPL27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL40 geni (Tüm Gen Dizi Analizi)	MRPL40 gene (Sequence Analysis-All Coding Exons)	MRPL40	MRPL40, NLVCF, MRPL22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL4 geni (Tüm Gen Dizi Analizi)	MRPL4 gene (Sequence Analysis-All Coding Exons)	MRPL4	MRPL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL39 geni (Tüm Gen Dizi Analizi)	MRPL39 gene (Sequence Analysis-All Coding Exons)	MRPL39	MRPL39, MRPL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL38 geni (Tüm Gen Dizi Analizi)	MRPL38 gene (Sequence Analysis-All Coding Exons)	MRPL38	MRPL38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL37 geni (Tüm Gen Dizi Analizi)	MRPL37 gene (Sequence Analysis-All Coding Exons)	MRPL37	MRPL37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL36 geni (Tüm Gen Dizi Analizi)	MRPL36 gene (Sequence Analysis-All Coding Exons)	MRPL36	MRPL36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL35 geni (Tüm Gen Dizi Analizi)	MRPL35 gene (Sequence Analysis-All Coding Exons)	MRPL35	MRPL35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL34 geni (Tüm Gen Dizi Analizi)	MRPL34 gene (Sequence Analysis-All Coding Exons)	MRPL34	MRPL34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL33 geni (Tüm Gen Dizi Analizi)	MRPL33 gene (Sequence Analysis-All Coding Exons)	MRPL33	MRPL33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL32 geni (Tüm Gen Dizi Analizi)	MRPL32 gene (Sequence Analysis-All Coding Exons)	MRPL32	MRPL32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL30 geni (Tüm Gen Dizi Analizi)	MRPL30 gene (Sequence Analysis-All Coding Exons)	MRPL30	MRPL30, MRPL28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL28 geni (Tüm Gen Dizi Analizi)	MRPL28 gene (Sequence Analysis-All Coding Exons)	MRPL28	MRPL28, MAAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL27 geni (Tüm Gen Dizi Analizi)	MRPL27 gene (Sequence Analysis-All Coding Exons)	MRPL27	MRPL27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL24 geni (Tüm Gen Dizi Analizi)	MRPL24 gene (Sequence Analysis-All Coding Exons)	MRPL24	MRPL24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL23 geni (Tüm Gen Dizi Analizi)	MRPL23 gene (Sequence Analysis-All Coding Exons)	MRPL23	RPL23L, L23MRP, MRPL23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL22 geni (Tüm Gen Dizi Analizi)	MRPL22 gene (Sequence Analysis-All Coding Exons)	MRPL22	MRPL22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL21 geni (Tüm Gen Dizi Analizi)	MRPL21 gene (Sequence Analysis-All Coding Exons)	MRPL21	MRPL21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL20 geni (Tüm Gen Dizi Analizi)	MRPL20 gene (Sequence Analysis-All Coding Exons)	MRPL20	MRPL20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL2 geni (Tüm Gen Dizi Analizi)	MRPL2 gene (Sequence Analysis-All Coding Exons)	MRPL2	MRPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL19 geni (Tüm Gen Dizi Analizi)	MRPL19 gene (Sequence Analysis-All Coding Exons)	MRPL19	MRPL19, MRPL15, KIAA0104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL18 geni (Tüm Gen Dizi Analizi)	MRPL18 gene (Sequence Analysis-All Coding Exons)	MRPL18	MRPL18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL17 geni (Tüm Gen Dizi Analizi)	MRPL17 gene (Sequence Analysis-All Coding Exons)	MRPL17	MRPL17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL16 geni (Tüm Gen Dizi Analizi)	MRPL16 gene (Sequence Analysis-All Coding Exons)	MRPL16	MRPL16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL15 geni (Tüm Gen Dizi Analizi)	MRPL15 gene (Sequence Analysis-All Coding Exons)	MRPL15	MRPL15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL14 geni (Tüm Gen Dizi Analizi)	MRPL14 gene (Sequence Analysis-All Coding Exons)	MRPL14	MRPL14, MRPL32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL13 geni (Tüm Gen Dizi Analizi)	MRPL13 gene (Sequence Analysis-All Coding Exons)	MRPL13	MRPL13, RPML13, L13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL12 geni (Tüm Gen Dizi Analizi)	MRPL12 gene (Sequence Analysis-All Coding Exons)	MRPL12	MRPL12, RPML12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL11 geni (Tüm Gen Dizi Analizi)	MRPL11 gene (Sequence Analysis-All Coding Exons)	MRPL11	MRPL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRSS27 geni (Tüm Gen Dizi Analizi)	PRSS27 gene (Sequence Analysis-All Coding Exons)	PRSS27		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL10 geni (Tüm Gen Dizi Analizi)	MRPL10 gene (Sequence Analysis-All Coding Exons)	MRPL10	MRPL10, MRPL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRPL1 geni (Tüm Gen Dizi Analizi)	MRPL1 gene (Sequence Analysis-All Coding Exons)	MRPL1	MRPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRO geni (Tüm Gen Dizi Analizi)	MRO gene (Sequence Analysis-All Coding Exons)	MRO	MRO, B29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRNIP geni (Tüm Gen Dizi Analizi)	MRNIP gene (Sequence Analysis-All Coding Exons)	MRNIP	MRNIP, C5orf45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRM3 geni (Tüm Gen Dizi Analizi)	MRM3 gene (Sequence Analysis-All Coding Exons)	MRM3	RNMTL1, HC90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRM2 geni (Tüm Gen Dizi Analizi)	MRM2 gene (Sequence Analysis-All Coding Exons)	MRM2	FTSJ2, FJH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRLN geni (Tüm Gen Dizi Analizi)	MRLN gene (Sequence Analysis-All Coding Exons)	MRLN	MRLN, LINC00948	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRI1 geni (Tüm Gen Dizi Analizi)	MRI1 gene (Sequence Analysis-All Coding Exons)	MRI1	MRI1, MRDI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRX4 geni (Tüm Gen Dizi Analizi)	MRGPRX4 gene (Sequence Analysis-All Coding Exons)	MRGPRX4	MRGPRX4, MRGX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRX3 geni (Tüm Gen Dizi Analizi)	MRGPRX3 gene (Sequence Analysis-All Coding Exons)	MRGPRX3	MRGPRX3, MRGX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRX2 geni (Tüm Gen Dizi Analizi)	MRGPRX2 gene (Sequence Analysis-All Coding Exons)	MRGPRX2	MRGPRX2, MRGX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRX1 geni (Tüm Gen Dizi Analizi)	MRGPRX1 gene (Sequence Analysis-All Coding Exons)	MRGPRX1	MRGPRX1, MRGX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRG geni (Tüm Gen Dizi Analizi)	MRGPRG gene (Sequence Analysis-All Coding Exons)	MRGPRG	MRGPRG, MRGG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRF geni (Tüm Gen Dizi Analizi)	MRGPRF gene (Sequence Analysis-All Coding Exons)	MRGPRF	MRGPRF, MRGF, RTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRE geni (Tüm Gen Dizi Analizi)	MRGPRE gene (Sequence Analysis-All Coding Exons)	MRGPRE	MRGPRE, MRGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGPRD geni (Tüm Gen Dizi Analizi)	MRGPRD gene (Sequence Analysis-All Coding Exons)	MRGPRD	MRGPRD, MRGD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRGBP geni (Tüm Gen Dizi Analizi)	MRGBP gene (Sequence Analysis-All Coding Exons)	MRGBP	MRGBP, C20orf20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRFAP1 geni (Tüm Gen Dizi Analizi)	MRFAP1 gene (Sequence Analysis-All Coding Exons)	MRFAP1	MRFAP1, PAM14, PGRI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MREG geni (Tüm Gen Dizi Analizi)	MREG gene (Sequence Analysis-All Coding Exons)	MREG	MREG, DSU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRC2 geni (Tüm Gen Dizi Analizi)	MRC2 gene (Sequence Analysis-All Coding Exons)	MRC2	MRC2, ENDO180, UPARAP, CD280	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRC1 geni (Tüm Gen Dizi Analizi)	MRC1 gene (Sequence Analysis-All Coding Exons)	MRC1	MRC1, MMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MRAS geni (Tüm Gen Dizi Analizi)	MRAS gene (Sequence Analysis-All Coding Exons)	MRAS	MRAS, RRAS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MR1 geni (Tüm Gen Dizi Analizi)	MR1 gene (Sequence Analysis-All Coding Exons)	MR1	MR1, HLALS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPZL3 geni (Tüm Gen Dizi Analizi)	MPZL3 gene (Sequence Analysis-All Coding Exons)	MPZL3	MPZL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPZL2 geni (Tüm Gen Dizi Analizi)	MPZL2 gene (Sequence Analysis-All Coding Exons)	MPZL2	MPZL2, EVA1, EVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPV17L2 geni (Tüm Gen Dizi Analizi)	MPV17L2 gene (Sequence Analysis-All Coding Exons)	MPV17L2	MPV17L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPST geni (Tüm Gen Dizi Analizi)	MPST gene (Sequence Analysis-All Coding Exons)	MPST	MPST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPRIP geni (Tüm Gen Dizi Analizi)	MPRIP gene (Sequence Analysis-All Coding Exons)	MPRIP	MPRIP, MRIP, KIAA0864, RIP3, P116RIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPPED2 geni (Tüm Gen Dizi Analizi)	MPPED2 gene (Sequence Analysis-All Coding Exons)	MPPED2	MPPED2, C11orf8, D11S302E, 239FB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPPED1 geni (Tüm Gen Dizi Analizi)	MPPED1 gene (Sequence Analysis-All Coding Exons)	MPPED1	MPPED1, C22orf1, 239AB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPPE1 geni (Tüm Gen Dizi Analizi)	MPPE1 gene (Sequence Analysis-All Coding Exons)	MPPE1	MPPE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP7 geni (Tüm Gen Dizi Analizi)	MPP7 gene (Sequence Analysis-All Coding Exons)	MPP7	MPP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP6 geni (Tüm Gen Dizi Analizi)	MPP6 gene (Sequence Analysis-All Coding Exons)	MPP6	MPP6, VAM1, PALS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP5 geni (Tüm Gen Dizi Analizi)	MPP5 gene (Sequence Analysis-All Coding Exons)	MPP5	MPP5, PALS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP4 geni (Tüm Gen Dizi Analizi)	MPP4 gene (Sequence Analysis-All Coding Exons)	MPP4	MPP4, DLG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP3 geni (Tüm Gen Dizi Analizi)	MPP3 gene (Sequence Analysis-All Coding Exons)	MPP3	MPP3, DLG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP2 geni (Tüm Gen Dizi Analizi)	MPP2 gene (Sequence Analysis-All Coding Exons)	MPP2	MPP2, DLG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPP1 geni (Tüm Gen Dizi Analizi)	MPP1 gene (Sequence Analysis-All Coding Exons)	MPP1	MPP1, PEMP, EMP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPHOSPH9 geni (Tüm Gen Dizi Analizi)	MPHOSPH9 gene (Sequence Analysis-All Coding Exons)	MPHOSPH9	MPHOSPH9, MPP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPHOSPH8 geni (Tüm Gen Dizi Analizi)	MPHOSPH8 gene (Sequence Analysis-All Coding Exons)	MPHOSPH8	MPHOSPH8, TWA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPHOSPH6 geni (Tüm Gen Dizi Analizi)	MPHOSPH6 gene (Sequence Analysis-All Coding Exons)	MPHOSPH6	MPHOSPH6, MPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPG geni (Tüm Gen Dizi Analizi)	MPG gene (Sequence Analysis-All Coding Exons)	MPG	MPG, MDG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MPEG1 geni (Tüm Gen Dizi Analizi)	MPEG1 gene (Sequence Analysis-All Coding Exons)	MPEG1	MPEG1, MPG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPC2 geni (Tüm Gen Dizi Analizi)	MPC2 gene (Sequence Analysis-All Coding Exons)	MPC2	BRP44, MPC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOXD1 geni (Tüm Gen Dizi Analizi)	MOXD1 gene (Sequence Analysis-All Coding Exons)	MOXD1	MOXD1, MOX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOV10L1 geni (Tüm Gen Dizi Analizi)	MOV10L1 gene (Sequence Analysis-All Coding Exons)	MOV10L1	MOV10L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOV10 geni (Tüm Gen Dizi Analizi)	MOV10 gene (Sequence Analysis-All Coding Exons)	MOV10	MOV10, KIAA1631	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOSPD1 geni (Tüm Gen Dizi Analizi)	MOSPD1 gene (Sequence Analysis-All Coding Exons)	MOSPD1	MOSPD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOS geni (Tüm Gen Dizi Analizi)	MOS gene (Sequence Analysis-All Coding Exons)	MOS	MOS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MORF4L2 geni (Tüm Gen Dizi Analizi)	MORF4L2 gene (Sequence Analysis-All Coding Exons)	MORF4L2	MORFL2, MRGX, KIAA0026	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MORF4L1 geni (Tüm Gen Dizi Analizi)	MORF4L1 gene (Sequence Analysis-All Coding Exons)	MORF4L1	MORF4L1, MRG15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MORC4 geni (Tüm Gen Dizi Analizi)	MORC4 gene (Sequence Analysis-All Coding Exons)	MORC4	MORC4, ZCW4, ZCWCC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MORC3 geni (Tüm Gen Dizi Analizi)	MORC3 gene (Sequence Analysis-All Coding Exons)	MORC3	MORC3, NXP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MORC1 geni (Tüm Gen Dizi Analizi)	MORC1 gene (Sequence Analysis-All Coding Exons)	MORC1	MORC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MON2 geni (Tüm Gen Dizi Analizi)	MON2 gene (Sequence Analysis-All Coding Exons)	MON2	MON2, KIAA1040	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MON1B geni (Tüm Gen Dizi Analizi)	MON1B gene (Sequence Analysis-All Coding Exons)	MON1B	MON1B, SRG1, KIAA0872	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MON1A geni (Tüm Gen Dizi Analizi)	MON1A gene (Sequence Analysis-All Coding Exons)	MON1A	MON1A, SAND1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOK geni (Tüm Gen Dizi Analizi)	MOK gene (Sequence Analysis-All Coding Exons)	MOK	RAGE, MOK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOGAT3 geni (Tüm Gen Dizi Analizi)	MOGAT3 gene (Sequence Analysis-All Coding Exons)	MOGAT3	MOGAT3, MGAT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOGAT2 geni (Tüm Gen Dizi Analizi)	MOGAT2 gene (Sequence Analysis-All Coding Exons)	MOGAT2	MOGAT2, MGAT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOGAT1 geni (Tüm Gen Dizi Analizi)	MOGAT1 gene (Sequence Analysis-All Coding Exons)	MOGAT1	MOGAT1, MGAT1, DGAT2L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOBP geni (Tüm Gen Dizi Analizi)	MOBP gene (Sequence Analysis-All Coding Exons)	MOBP	MOBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOB4 geni (Tüm Gen Dizi Analizi)	MOB4 gene (Sequence Analysis-All Coding Exons)	MOB4	MOBKL3, PREI3, MOB1, MOB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOB2 geni (Tüm Gen Dizi Analizi)	MOB2 gene (Sequence Analysis-All Coding Exons)	MOB2	HCCA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOAP1 geni (Tüm Gen Dizi Analizi)	MOAP1 gene (Sequence Analysis-All Coding Exons)	MOAP1	MOAP1, MAP1, PNMA4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MNT geni (Tüm Gen Dizi Analizi)	MNT gene (Sequence Analysis-All Coding Exons)	MNT	MNT, ROX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MNS1 geni (Tüm Gen Dizi Analizi)	MNS1 gene (Sequence Analysis-All Coding Exons)	MNS1	MNS1, FLJ11222	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MNDA geni (Tüm Gen Dizi Analizi)	MNDA gene (Sequence Analysis-All Coding Exons)	MNDA	MNDA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MND1 geni (Tüm Gen Dizi Analizi)	MND1 gene (Sequence Analysis-All Coding Exons)	MND1	MND1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MNAT1 geni (Tüm Gen Dizi Analizi)	MNAT1 gene (Sequence Analysis-All Coding Exons)	MNAT1	MNAT1, MAT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMS22L geni (Tüm Gen Dizi Analizi)	MMS22L gene (Sequence Analysis-All Coding Exons)	MMS22L	MMS22L, C6orf167	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMS19 geni (Tüm Gen Dizi Analizi)	MMS19 gene (Sequence Analysis-All Coding Exons)	MMS19	MMS19, MMS19L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP8 geni (Tüm Gen Dizi Analizi)	MMP8 gene (Sequence Analysis-All Coding Exons)	MMP8	MMP8, CLG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP7 geni (Tüm Gen Dizi Analizi)	MMP7 gene (Sequence Analysis-All Coding Exons)	MMP7	MMP7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP28 geni (Tüm Gen Dizi Analizi)	MMP28 gene (Sequence Analysis-All Coding Exons)	MMP28	MMP28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP26 geni (Tüm Gen Dizi Analizi)	MMP26 gene (Sequence Analysis-All Coding Exons)	MMP26	MMP26	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP25 geni (Tüm Gen Dizi Analizi)	MMP25 gene (Sequence Analysis-All Coding Exons)	MMP25	MMP25, MMP20A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP24 geni (Tüm Gen Dizi Analizi)	MMP24 gene (Sequence Analysis-All Coding Exons)	MMP24	MMP25	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP23B geni (Tüm Gen Dizi Analizi)	MMP23B gene (Sequence Analysis-All Coding Exons)	MMP23B	MMP23B, MMP22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP17 geni (Tüm Gen Dizi Analizi)	MMP17 gene (Sequence Analysis-All Coding Exons)	MMP17	MMP17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP16 geni (Tüm Gen Dizi Analizi)	MMP16 gene (Sequence Analysis-All Coding Exons)	MMP16	MMP16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP15 geni (Tüm Gen Dizi Analizi)	MMP15 gene (Sequence Analysis-All Coding Exons)	MMP15	MMP15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP12 geni (Tüm Gen Dizi Analizi)	MMP12 gene (Sequence Analysis-All Coding Exons)	MMP12	MMP12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP11 geni (Tüm Gen Dizi Analizi)	MMP11 gene (Sequence Analysis-All Coding Exons)	MMP11	MMP11, STMY3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMP10 geni (Tüm Gen Dizi Analizi)	MMP10 gene (Sequence Analysis-All Coding Exons)	MMP10	MMP10, STMY2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMD2 geni (Tüm Gen Dizi Analizi)	MMD2 gene (Sequence Analysis-All Coding Exons)	MMD2	MMD2, PAQR10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMD geni (Tüm Gen Dizi Analizi)	MMD gene (Sequence Analysis-All Coding Exons)	MMD	MMD, MMD1, PAQR11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT5 geni (Tüm Gen Dizi Analizi)	PRMT5 gene (Sequence Analysis-All Coding Exons)	PRMT5		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRMT3 geni (Tüm Gen Dizi Analizi)	PRMT3 gene (Sequence Analysis-All Coding Exons)	PRMT3		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLXIPL geni (Tüm Gen Dizi Analizi)	MLXIPL gene (Sequence Analysis-All Coding Exons)	MLXIPL	MLXIPL, WBSCR14, MONDOB, CHREBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLXIP geni (Tüm Gen Dizi Analizi)	MLXIP gene (Sequence Analysis-All Coding Exons)	MLXIP	MLXIP, MONDOA, KIAA0867	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLX geni (Tüm Gen Dizi Analizi)	MLX gene (Sequence Analysis-All Coding Exons)	MLX	MLX, TCFL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLST8 geni (Tüm Gen Dizi Analizi)	MLST8 gene (Sequence Analysis-All Coding Exons)	MLST8	MLST8, GBL, LST8, WAT1, POP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLNR geni (Tüm Gen Dizi Analizi)	MLNR gene (Sequence Analysis-All Coding Exons)	MLNR	MLNR, GPR38	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLN geni (Tüm Gen Dizi Analizi)	MLN gene (Sequence Analysis-All Coding Exons)	MLN	MLN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLLT6 geni (Tüm Gen Dizi Analizi)	MLLT6 gene (Sequence Analysis-All Coding Exons)	MLLT6	MLLT6, AF17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRLH geni (Tüm Gen Dizi Analizi)	PRLH gene (Sequence Analysis-All Coding Exons)	PRLH		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLLT3 geni (Tüm Gen Dizi Analizi)	MLLT3 gene (Sequence Analysis-All Coding Exons)	MLLT3	MLLT3, AF9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLLT11 geni (Tüm Gen Dizi Analizi)	MLLT11 gene (Sequence Analysis-All Coding Exons)	MLLT11	MLLT11, AF1Q	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLLT1 geni (Tüm Gen Dizi Analizi)	MLLT1 gene (Sequence Analysis-All Coding Exons)	MLLT1	MLLT1, ENL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLKL geni (Tüm Gen Dizi Analizi)	MLKL gene (Sequence Analysis-All Coding Exons)	MLKL	MLKL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLIP geni (Tüm Gen Dizi Analizi)	MLIP gene (Sequence Analysis-All Coding Exons)	MLIP	MLIP, C6orf142	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLF2 geni (Tüm Gen Dizi Analizi)	MLF2 gene (Sequence Analysis-All Coding Exons)	MLF2	MLF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLEC geni (Tüm Gen Dizi Analizi)	MLEC gene (Sequence Analysis-All Coding Exons)	MLEC	MLEC, KIAA0152	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MLANA geni (Tüm Gen Dizi Analizi)	MLANA gene (Sequence Analysis-All Coding Exons)	MLANA	MLANA, MART1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKX geni (Tüm Gen Dizi Analizi)	MKX gene (Sequence Analysis-All Coding Exons)	MKX	MKX, IFRX, IRXL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKRN2 geni (Tüm Gen Dizi Analizi)	MKRN2 gene (Sequence Analysis-All Coding Exons)	MKRN2	MKRN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKNK2 geni (Tüm Gen Dizi Analizi)	MKNK2 gene (Sequence Analysis-All Coding Exons)	MKNK2	MKNK2, MNK2, GPRK7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKNK1 geni (Tüm Gen Dizi Analizi)	MKNK1 gene (Sequence Analysis-All Coding Exons)	MKNK1	MKNK1, MNK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKLN1 geni (Tüm Gen Dizi Analizi)	MKLN1 gene (Sequence Analysis-All Coding Exons)	MKLN1	MKLN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKL2 geni (Tüm Gen Dizi Analizi)	MKL2 gene (Sequence Analysis-All Coding Exons)	MKL2	MKL2, MRTFB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKI67 geni (Tüm Gen Dizi Analizi)	MKI67 gene (Sequence Analysis-All Coding Exons)	MKI67	MKI67	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIXL1 geni (Tüm Gen Dizi Analizi)	MIXL1 gene (Sequence Analysis-All Coding Exons)	MIXL1	MIXL1, MIXL, MIX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MISP geni (Tüm Gen Dizi Analizi)	MISP gene (Sequence Analysis-All Coding Exons)	MISP	MISP, C19orf21	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIS12 geni (Tüm Gen Dizi Analizi)	MIS12 gene (Sequence Analysis-All Coding Exons)	MIS12	MIS12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7I geni (Tüm Gen Dizi Analizi)	MIRLET7I gene (Sequence Analysis-All Coding Exons)	MIRLET7I	MIRLET7I, LET7I, MIRNLET7I	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7G geni (Tüm Gen Dizi Analizi)	MIRLET7G gene (Sequence Analysis-All Coding Exons)	MIRLET7G	MIRLET7G, LET7G, MIRNLET7G	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7F2 geni (Tüm Gen Dizi Analizi)	MIRLET7F2 gene (Sequence Analysis-All Coding Exons)	MIRLET7F2	MIRLET7F2, LET7F2, MIRNLET7F2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7F1 geni (Tüm Gen Dizi Analizi)	MIRLET7F1 gene (Sequence Analysis-All Coding Exons)	MIRLET7F1	MIRLET7F1, LET7F1, MIRNLET7F1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7E geni (Tüm Gen Dizi Analizi)	MIRLET7E gene (Sequence Analysis-All Coding Exons)	MIRLET7E	MIRLET7E, LET7E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7D geni (Tüm Gen Dizi Analizi)	MIRLET7D gene (Sequence Analysis-All Coding Exons)	MIRLET7D	MIRLET7D, LET7D, MIRNLET7D	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7C geni (Tüm Gen Dizi Analizi)	MIRLET7C gene (Sequence Analysis-All Coding Exons)	MIRLET7C	MIRLET7C, MIRNLET7C, LET7C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7B geni (Tüm Gen Dizi Analizi)	MIRLET7B gene (Sequence Analysis-All Coding Exons)	MIRLET7B	MIRLET7B, LET7B, MIRNLET7B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7A3 geni (Tüm Gen Dizi Analizi)	MIRLET7A3 gene (Sequence Analysis-All Coding Exons)	MIRLET7A3	MIRLET7A3, LET7A3, MIRNLET7A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7A2 geni (Tüm Gen Dizi Analizi)	MIRLET7A2 gene (Sequence Analysis-All Coding Exons)	MIRLET7A2	MIRLET7A2, LET7A2, MIRNLET7A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIRLET7A1 geni (Tüm Gen Dizi Analizi)	MIRLET7A1 gene (Sequence Analysis-All Coding Exons)	MIRLET7A1	MIRLET7A1, LET7A1, MIRNLET7A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR99B geni (Tüm Gen Dizi Analizi)	MIR99B gene (Sequence Analysis-All Coding Exons)	MIR99B	MIR99B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR99A geni (Tüm Gen Dizi Analizi)	MIR99A gene (Sequence Analysis-All Coding Exons)	MIR99A	MIR99A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR98 geni (Tüm Gen Dizi Analizi)	MIR98 gene (Sequence Analysis-All Coding Exons)	MIR98	MIR98, MIRN98	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR95 geni (Tüm Gen Dizi Analizi)	MIR95 gene (Sequence Analysis-All Coding Exons)	MIR95	MIR95, MIRN95	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR93 geni (Tüm Gen Dizi Analizi)	MIR93 gene (Sequence Analysis-All Coding Exons)	MIR93	MIR93, MIRN9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR92A1 geni (Tüm Gen Dizi Analizi)	MIR92A1 gene (Sequence Analysis-All Coding Exons)	MIR92A1	MIR92A1, MIR92-1, MIRN92-1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR9-3 geni (Tüm Gen Dizi Analizi)	MIR9-3 gene (Sequence Analysis-All Coding Exons)	MIR9-3	MIR9-3, MIRN9-3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR9-2 geni (Tüm Gen Dizi Analizi)	MIR9-2 gene (Sequence Analysis-All Coding Exons)	MIR9-2	MIR9-2, MIRN9-2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR9-1 geni (Tüm Gen Dizi Analizi)	MIR9-1 gene (Sequence Analysis-All Coding Exons)	MIR9-1	MIR9-1, MIRN9-1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MIR877 geni (Tüm Gen Dizi Analizi)	MIR877 gene (Sequence Analysis-All Coding Exons)	MIR877	MIR877, MIRN877	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR873 geni (Tüm Gen Dizi Analizi)	MIR873 gene (Sequence Analysis-All Coding Exons)	MIR873	MIR873	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR802 geni (Tüm Gen Dizi Analizi)	MIR802 gene (Sequence Analysis-All Coding Exons)	MIR802	MIR802	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR718 geni (Tüm Gen Dizi Analizi)	MIR718 gene (Sequence Analysis-All Coding Exons)	MIR718	MIR718	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR7-1 geni (Tüm Gen Dizi Analizi)	MIR7-1 gene (Sequence Analysis-All Coding Exons)	MIR7-1	MIR7-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR675 geni (Tüm Gen Dizi Analizi)	MIR675 gene (Sequence Analysis-All Coding Exons)	MIR675	MIR675	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR671 geni (Tüm Gen Dizi Analizi)	MIR671 gene (Sequence Analysis-All Coding Exons)	MIR671	MIR671	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR661 geni (Tüm Gen Dizi Analizi)	MIR661 gene (Sequence Analysis-All Coding Exons)	MIR661	MIR661, MIRN661	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR659 geni (Tüm Gen Dizi Analizi)	MIR659 gene (Sequence Analysis-All Coding Exons)	MIR659	MIR659	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR656 geni (Tüm Gen Dizi Analizi)	MIR656 gene (Sequence Analysis-All Coding Exons)	MIR656	MIR656	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR650 geni (Tüm Gen Dizi Analizi)	MIR650 gene (Sequence Analysis-All Coding Exons)	MIR650	MIR650	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR648 geni (Tüm Gen Dizi Analizi)	MIR648 gene (Sequence Analysis-All Coding Exons)	MIR648	MIR648	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR616 geni (Tüm Gen Dizi Analizi)	MIR616 gene (Sequence Analysis-All Coding Exons)	MIR616	MIR616	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR610 geni (Tüm Gen Dizi Analizi)	MIR610 gene (Sequence Analysis-All Coding Exons)	MIR610	MIR610, MIRN610	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR590 geni (Tüm Gen Dizi Analizi)	MIR590 gene (Sequence Analysis-All Coding Exons)	MIR590	MIR590	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR574 geni (Tüm Gen Dizi Analizi)	MIR574 gene (Sequence Analysis-All Coding Exons)	MIR574	MIR574, MIR574-3p	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR570 geni (Tüm Gen Dizi Analizi)	MIR570 gene (Sequence Analysis-All Coding Exons)	MIR570	MIR570	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR558 geni (Tüm Gen Dizi Analizi)	MIR558 gene (Sequence Analysis-All Coding Exons)	MIR558	MIR558	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR551A geni (Tüm Gen Dizi Analizi)	MIR551A gene (Sequence Analysis-All Coding Exons)	MIR551A	MIR551A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR520H geni (Tüm Gen Dizi Analizi)	MIR520H gene (Sequence Analysis-All Coding Exons)	MIR520H	MIR520H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR520G geni (Tüm Gen Dizi Analizi)	MIR520G gene (Sequence Analysis-All Coding Exons)	MIR520G	MIR520G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR520C geni (Tüm Gen Dizi Analizi)	MIR520C gene (Sequence Analysis-All Coding Exons)	MIR520C	MIR520C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR519D geni (Tüm Gen Dizi Analizi)	MIR519D gene (Sequence Analysis-All Coding Exons)	MIR519D	MIR519D, MIRN519D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR510 geni (Tüm Gen Dizi Analizi)	MIR510 gene (Sequence Analysis-All Coding Exons)	MIR510	MIR510	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR509-3 geni (Tüm Gen Dizi Analizi)	MIR509-3 gene (Sequence Analysis-All Coding Exons)	MIR509-3	MIR509-3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR509-1 geni (Tüm Gen Dizi Analizi)	MIR509-1 gene (Sequence Analysis-All Coding Exons)	MIR509-1	MIR509-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR508 geni (Tüm Gen Dizi Analizi)	MIR508 gene (Sequence Analysis-All Coding Exons)	MIR508	MIR508	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR506 geni (Tüm Gen Dizi Analizi)	MIR506 gene (Sequence Analysis-All Coding Exons)	MIR506	MIR506	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR503 geni (Tüm Gen Dizi Analizi)	MIR503 gene (Sequence Analysis-All Coding Exons)	MIR503	MIR503	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR502 geni (Tüm Gen Dizi Analizi)	MIR502 gene (Sequence Analysis-All Coding Exons)	MIR502	MIR502	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR499A geni (Tüm Gen Dizi Analizi)	MIR499A gene (Sequence Analysis-All Coding Exons)	MIR499A	MIR499, MIRN499	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR497 geni (Tüm Gen Dizi Analizi)	MIR497 gene (Sequence Analysis-All Coding Exons)	MIR497	MIR497	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR495 geni (Tüm Gen Dizi Analizi)	MIR495 gene (Sequence Analysis-All Coding Exons)	MIR495	MIR551A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR494 geni (Tüm Gen Dizi Analizi)	MIR494 gene (Sequence Analysis-All Coding Exons)	MIR494	MIR494	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR490 geni (Tüm Gen Dizi Analizi)	MIR490 gene (Sequence Analysis-All Coding Exons)	MIR490	MIR490	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR489 geni (Tüm Gen Dizi Analizi)	MIR489 gene (Sequence Analysis-All Coding Exons)	MIR489	MIR489	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR487B geni (Tüm Gen Dizi Analizi)	MIR487B gene (Sequence Analysis-All Coding Exons)	MIR487B	MIR487B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR485 geni (Tüm Gen Dizi Analizi)	MIR485 gene (Sequence Analysis-All Coding Exons)	MIR485	MIR485	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRDM7 geni (Tüm Gen Dizi Analizi)	PRDM7 gene (Sequence Analysis-All Coding Exons)	PRDM7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR451A geni (Tüm Gen Dizi Analizi)	MIR451A gene (Sequence Analysis-All Coding Exons)	MIR451A	MIR451, MIRN451	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR449B geni (Tüm Gen Dizi Analizi)	MIR449B gene (Sequence Analysis-All Coding Exons)	MIR449B	MIR449B, MIRN449B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR449A geni (Tüm Gen Dizi Analizi)	MIR449A gene (Sequence Analysis-All Coding Exons)	MIR449A	MIR449A, MIRN449A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR448 geni (Tüm Gen Dizi Analizi)	MIR448 gene (Sequence Analysis-All Coding Exons)	MIR448	MIR448, MIRN448	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR4449 geni (Tüm Gen Dizi Analizi)	MIR4449 gene (Sequence Analysis-All Coding Exons)	MIR4449	MIR4449	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR433 geni (Tüm Gen Dizi Analizi)	MIR433 gene (Sequence Analysis-All Coding Exons)	MIR433	MIR433, MIRN433	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR431 geni (Tüm Gen Dizi Analizi)	MIR431 gene (Sequence Analysis-All Coding Exons)	MIR431	MIR431, MIRN431	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR429 geni (Tüm Gen Dizi Analizi)	MIR429 gene (Sequence Analysis-All Coding Exons)	MIR429	MIR429, MIRN429	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR4276 geni (Tüm Gen Dizi Analizi)	MIR4276 gene (Sequence Analysis-All Coding Exons)	MIR4276	MIR4276	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR4271 geni (Tüm Gen Dizi Analizi)	MIR4271 gene (Sequence Analysis-All Coding Exons)	MIR4271	MIR4271	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR424 geni (Tüm Gen Dizi Analizi)	MIR424 gene (Sequence Analysis-All Coding Exons)	MIR424	MIR424, MIRN424, MIR322	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR410 geni (Tüm Gen Dizi Analizi)	MIR410 gene (Sequence Analysis-All Coding Exons)	MIR410	MIR410	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR409 geni (Tüm Gen Dizi Analizi)	MIR409 gene (Sequence Analysis-All Coding Exons)	MIR409	MIR409, MIRN409	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR380 geni (Tüm Gen Dizi Analizi)	MIR380 gene (Sequence Analysis-All Coding Exons)	MIR380	MIR380, MIRN380	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR379 geni (Tüm Gen Dizi Analizi)	MIR379 gene (Sequence Analysis-All Coding Exons)	MIR379	MIR379	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR378A geni (Tüm Gen Dizi Analizi)	MIR378A gene (Sequence Analysis-All Coding Exons)	MIR378A	MIR378, MIRN378	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR376C geni (Tüm Gen Dizi Analizi)	MIR376C gene (Sequence Analysis-All Coding Exons)	MIR376C	MIR376C, MIR368	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR376B geni (Tüm Gen Dizi Analizi)	MIR376B gene (Sequence Analysis-All Coding Exons)	MIR376B	MIR376B, MIRN376B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR376A2 geni (Tüm Gen Dizi Analizi)	MIR376A2 gene (Sequence Analysis-All Coding Exons)	MIR376A2	MIR376A-2, MIRN376A-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR376A1 geni (Tüm Gen Dizi Analizi)	MIR376A1 gene (Sequence Analysis-All Coding Exons)	MIR376A1	MIR376A-1, MIRN376A-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR375 geni (Tüm Gen Dizi Analizi)	MIR375 gene (Sequence Analysis-All Coding Exons)	MIR375	MIR375, MIRN375	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PRAM1 geni (Tüm Gen Dizi Analizi)	PRAM1 gene (Sequence Analysis-All Coding Exons)	PRAM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR373 geni (Tüm Gen Dizi Analizi)	MIR373 gene (Sequence Analysis-All Coding Exons)	MIR373	MIR373, MIRN373	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR372 geni (Tüm Gen Dizi Analizi)	MIR372 gene (Sequence Analysis-All Coding Exons)	MIR372	MIR372, MIRN372	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR371A geni (Tüm Gen Dizi Analizi)	MIR371A gene (Sequence Analysis-All Coding Exons)	MIR371A	MIR371A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR370 geni (Tüm Gen Dizi Analizi)	MIR370 gene (Sequence Analysis-All Coding Exons)	MIR370	MIR370, MIRN370	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR369 geni (Tüm Gen Dizi Analizi)	MIR369 gene (Sequence Analysis-All Coding Exons)	MIR369	MIR369, MIR369-3, MIRN369-3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR367 geni (Tüm Gen Dizi Analizi)	MIR367 gene (Sequence Analysis-All Coding Exons)	MIR367	MIR367	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR365A geni (Tüm Gen Dizi Analizi)	MIR365A gene (Sequence Analysis-All Coding Exons)	MIR365A	MIR365A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR34C geni (Tüm Gen Dizi Analizi)	MIR34C gene (Sequence Analysis-All Coding Exons)	MIR34C	MIR34C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR34B geni (Tüm Gen Dizi Analizi)	MIR34B gene (Sequence Analysis-All Coding Exons)	MIR34B	MIR34B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR34A geni (Tüm Gen Dizi Analizi)	MIR34A gene (Sequence Analysis-All Coding Exons)	MIR34A	MIR34A, MIRN34A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR346 geni (Tüm Gen Dizi Analizi)	MIR346 gene (Sequence Analysis-All Coding Exons)	MIR346	MIR346	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR33B geni (Tüm Gen Dizi Analizi)	MIR33B gene (Sequence Analysis-All Coding Exons)	MIR33B	MIR33B, MIRN33B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR33A geni (Tüm Gen Dizi Analizi)	MIR33A gene (Sequence Analysis-All Coding Exons)	MIR33A	MIR33A, MIRN33A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR339 geni (Tüm Gen Dizi Analizi)	MIR339 gene (Sequence Analysis-All Coding Exons)	MIR339	MIR339	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR338 geni (Tüm Gen Dizi Analizi)	MIR338 gene (Sequence Analysis-All Coding Exons)	MIR338	MIR338, MIRN338	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR328 geni (Tüm Gen Dizi Analizi)	MIR328 gene (Sequence Analysis-All Coding Exons)	MIR328	MIR328, MIRN328	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR326 geni (Tüm Gen Dizi Analizi)	MIR326 gene (Sequence Analysis-All Coding Exons)	MIR326	MIR326, MIRN326	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR320A geni (Tüm Gen Dizi Analizi)	MIR320A gene (Sequence Analysis-All Coding Exons)	MIR320A	MIR320A, MIRN320A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR32 geni (Tüm Gen Dizi Analizi)	MIR32 gene (Sequence Analysis-All Coding Exons)	MIR32	MIR32, MIRN32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR3120 geni (Tüm Gen Dizi Analizi)	MIR3120 gene (Sequence Analysis-All Coding Exons)	MIR3120	MIR3120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR31 geni (Tüm Gen Dizi Analizi)	MIR31 gene (Sequence Analysis-All Coding Exons)	MIR31	MIR31, MIRN31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR30C1 geni (Tüm Gen Dizi Analizi)	MIR30C1 gene (Sequence Analysis-All Coding Exons)	MIR30C1	MIR30C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR30A geni (Tüm Gen Dizi Analizi)	MIR30A gene (Sequence Analysis-All Coding Exons)	MIR30A	MIR30A, MIRN30A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR302D geni (Tüm Gen Dizi Analizi)	MIR302D gene (Sequence Analysis-All Coding Exons)	MIR302D	MIR302D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR302C geni (Tüm Gen Dizi Analizi)	MIR302C gene (Sequence Analysis-All Coding Exons)	MIR302C	MIR302C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR302B geni (Tüm Gen Dizi Analizi)	MIR302B gene (Sequence Analysis-All Coding Exons)	MIR302B	MIR302B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR302A geni (Tüm Gen Dizi Analizi)	MIR302A gene (Sequence Analysis-All Coding Exons)	MIR302A	MIR302A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR301A geni (Tüm Gen Dizi Analizi)	MIR301A gene (Sequence Analysis-All Coding Exons)	MIR301A	MIR301A, MIR301	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR29C geni (Tüm Gen Dizi Analizi)	MIR29C gene (Sequence Analysis-All Coding Exons)	MIR29C	MIR29C, MIRN29C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR29B1 geni (Tüm Gen Dizi Analizi)	MIR29B1 gene (Sequence Analysis-All Coding Exons)	MIR29B1	MIR29B1, MIRN29B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR29A geni (Tüm Gen Dizi Analizi)	MIR29A gene (Sequence Analysis-All Coding Exons)	MIR29A	MIR29A, MIRN29A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR298 geni (Tüm Gen Dizi Analizi)	MIR298 gene (Sequence Analysis-All Coding Exons)	MIR298	MIR298	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MIR297 geni (Tüm Gen Dizi Analizi)	MIR297 gene (Sequence Analysis-All Coding Exons)	MIR297	MIR297	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR296 geni (Tüm Gen Dizi Analizi)	MIR296 gene (Sequence Analysis-All Coding Exons)	MIR296	MIR296, MIRN296	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR28 geni (Tüm Gen Dizi Analizi)	MIR28 gene (Sequence Analysis-All Coding Exons)	MIR28	MIR28, MIRN28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR27B geni (Tüm Gen Dizi Analizi)	MIR27B gene (Sequence Analysis-All Coding Exons)	MIR27B	MIR27B, MIRN27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR27A geni (Tüm Gen Dizi Analizi)	MIR27A gene (Sequence Analysis-All Coding Exons)	MIR27A	MIR27, MIRN27A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR26B geni (Tüm Gen Dizi Analizi)	MIR26B gene (Sequence Analysis-All Coding Exons)	MIR26B	MIR26B, MIRN26B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP2R2A geni (Tüm Gen Dizi Analizi)	PPP2R2A gene (Sequence Analysis-All Coding Exons)	PPP2R2A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR26A2 geni (Tüm Gen Dizi Analizi)	MIR26A2 gene (Sequence Analysis-All Coding Exons)	MIR26A2	MIR26A2, MIRN26A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR26A1 geni (Tüm Gen Dizi Analizi)	MIR26A1 gene (Sequence Analysis-All Coding Exons)	MIR26A1	MIR26A, MIRN26A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR25 geni (Tüm Gen Dizi Analizi)	MIR25 gene (Sequence Analysis-All Coding Exons)	MIR25	MIR25, MIRN25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR24-2 geni (Tüm Gen Dizi Analizi)	MIR24-2 gene (Sequence Analysis-All Coding Exons)	MIR24-2	MIR24-2, MIRN24-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R9B geni (Tüm Gen Dizi Analizi)	PPP1R9B gene (Sequence Analysis-All Coding Exons)	PPP1R9B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR24-1 geni (Tüm Gen Dizi Analizi)	MIR24-1 gene (Sequence Analysis-All Coding Exons)	MIR24-1	MIR189, MIRN24-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR23B geni (Tüm Gen Dizi Analizi)	MIR23B gene (Sequence Analysis-All Coding Exons)	MIR23B	MIR23B, MIRN23B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR23A geni (Tüm Gen Dizi Analizi)	MIR23A gene (Sequence Analysis-All Coding Exons)	MIR23A	MIR23A, MIRN23A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR224 geni (Tüm Gen Dizi Analizi)	MIR224 gene (Sequence Analysis-All Coding Exons)	MIR224	MIR224, MIRN224	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR223 geni (Tüm Gen Dizi Analizi)	MIR223 gene (Sequence Analysis-All Coding Exons)	MIR223	MIR223, MIRN223	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR222 geni (Tüm Gen Dizi Analizi)	MIR222 gene (Sequence Analysis-All Coding Exons)	MIR222	MIR222, MIRN222	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR221 geni (Tüm Gen Dizi Analizi)	MIR221 gene (Sequence Analysis-All Coding Exons)	MIR221	MIR221, MIRN221	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR22 geni (Tüm Gen Dizi Analizi)	MIR22 gene (Sequence Analysis-All Coding Exons)	MIR22	MIR22, MIRN22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR219A1 geni (Tüm Gen Dizi Analizi)	MIR219A1 gene (Sequence Analysis-All Coding Exons)	MIR219A1	MIR219-1, MIRN219-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR218-2 geni (Tüm Gen Dizi Analizi)	MIR218-2 gene (Sequence Analysis-All Coding Exons)	MIR218-2	MIR218-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R1B geni (Tüm Gen Dizi Analizi)	PPP1R1B gene (Sequence Analysis-All Coding Exons)	PPP1R1B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR218-1 geni (Tüm Gen Dizi Analizi)	MIR218-1 gene (Sequence Analysis-All Coding Exons)	MIR218-1	MIR218-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR217 geni (Tüm Gen Dizi Analizi)	MIR217 gene (Sequence Analysis-All Coding Exons)	MIR217	MIR217	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR216A geni (Tüm Gen Dizi Analizi)	MIR216A gene (Sequence Analysis-All Coding Exons)	MIR216A	MIR216, MIRN216	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR215 geni (Tüm Gen Dizi Analizi)	MIR215 gene (Sequence Analysis-All Coding Exons)	MIR215	MIR215, MIRN215	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R16A geni (Tüm Gen Dizi Analizi)	PPP1R16A gene (Sequence Analysis-All Coding Exons)	PPP1R16A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR214 geni (Tüm Gen Dizi Analizi)	MIR214 gene (Sequence Analysis-All Coding Exons)	MIR214	MIR214, MIRN214	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR212 geni (Tüm Gen Dizi Analizi)	MIR212 gene (Sequence Analysis-All Coding Exons)	MIR212	MIR212, MIRN212	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR211 geni (Tüm Gen Dizi Analizi)	MIR211 gene (Sequence Analysis-All Coding Exons)	MIR211	MIR211, MIRN211	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR210 geni (Tüm Gen Dizi Analizi)	MIR210 gene (Sequence Analysis-All Coding Exons)	MIR210	MIR210, MIRN210	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR21 geni (Tüm Gen Dizi Analizi)	MIR21 gene (Sequence Analysis-All Coding Exons)	MIR21	MIR21, MIRN21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR20B geni (Tüm Gen Dizi Analizi)	MIR20B gene (Sequence Analysis-All Coding Exons)	MIR20B	MIR20B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR20A geni (Tüm Gen Dizi Analizi)	MIR20A gene (Sequence Analysis-All Coding Exons)	MIR20A	MIR20A, MIRN20A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPP1R13B geni (Tüm Gen Dizi Analizi)	PPP1R13B gene (Sequence Analysis-All Coding Exons)	PPP1R13B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR208B geni (Tüm Gen Dizi Analizi)	MIR208B gene (Sequence Analysis-All Coding Exons)	MIR208B	MIR208B, MIRN208B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR208A geni (Tüm Gen Dizi Analizi)	MIR208A gene (Sequence Analysis-All Coding Exons)	MIR208A	MIR208A, MIRN208A, MIR208, MIRN208	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR206 geni (Tüm Gen Dizi Analizi)	MIR206 gene (Sequence Analysis-All Coding Exons)	MIR206	MIR206, MIRN206	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR205 geni (Tüm Gen Dizi Analizi)	MIR205 gene (Sequence Analysis-All Coding Exons)	MIR205	MIR205, MIRN205	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR203A geni (Tüm Gen Dizi Analizi)	MIR203A gene (Sequence Analysis-All Coding Exons)	MIR203A	MIR203, MIRN203	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR200C geni (Tüm Gen Dizi Analizi)	MIR200C gene (Sequence Analysis-All Coding Exons)	MIR200C	MIR200C, MIRN200C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR200B geni (Tüm Gen Dizi Analizi)	MIR200B gene (Sequence Analysis-All Coding Exons)	MIR200B	MIR200B, MIRN200B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR200A geni (Tüm Gen Dizi Analizi)	MIR200A gene (Sequence Analysis-All Coding Exons)	MIR200A	MIR200A, MIRN200A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR19B2 geni (Tüm Gen Dizi Analizi)	MIR19B2 gene (Sequence Analysis-All Coding Exons)	MIR19B2	MIR19B2, MIRN19B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR19B1 geni (Tüm Gen Dizi Analizi)	MIR19B1 gene (Sequence Analysis-All Coding Exons)	MIR19B1	MIR19B1, MIRN19B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR19A geni (Tüm Gen Dizi Analizi)	MIR19A gene (Sequence Analysis-All Coding Exons)	MIR19A	MIR19A, MIRN19A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR199B geni (Tüm Gen Dizi Analizi)	MIR199B gene (Sequence Analysis-All Coding Exons)	MIR199B	MIR199B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR199A2 geni (Tüm Gen Dizi Analizi)	MIR199A2 gene (Sequence Analysis-All Coding Exons)	MIR199A2	MIR199A2, MIRN199A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR199A1 geni (Tüm Gen Dizi Analizi)	MIR199A1 gene (Sequence Analysis-All Coding Exons)	MIR199A1	MIR199A1, MIRN199A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR197 geni (Tüm Gen Dizi Analizi)	MIR197 gene (Sequence Analysis-All Coding Exons)	MIR197	MIR197	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR196B geni (Tüm Gen Dizi Analizi)	MIR196B gene (Sequence Analysis-All Coding Exons)	MIR196B	MIR196B, MIRN196B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR196A2 geni (Tüm Gen Dizi Analizi)	MIR196A2 gene (Sequence Analysis-All Coding Exons)	MIR196A2	MIR196A2, MIRN196A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR196A1 geni (Tüm Gen Dizi Analizi)	MIR196A1 gene (Sequence Analysis-All Coding Exons)	MIR196A1	MIR196A1, MIRN196A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR195 geni (Tüm Gen Dizi Analizi)	MIR195 gene (Sequence Analysis-All Coding Exons)	MIR195	MIR195, MIRN195	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR194-2 geni (Tüm Gen Dizi Analizi)	MIR194-2 gene (Sequence Analysis-All Coding Exons)	MIR194-2	MIR194-2, MIRN194-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR194-1 geni (Tüm Gen Dizi Analizi)	MIR194-1 gene (Sequence Analysis-All Coding Exons)	MIR194-1	MIR194-1, MIRN194-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR193B geni (Tüm Gen Dizi Analizi)	MIR193B gene (Sequence Analysis-All Coding Exons)	MIR193B	MIR193B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR193A geni (Tüm Gen Dizi Analizi)	MIR193A gene (Sequence Analysis-All Coding Exons)	MIR193A	MIR193A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR192 geni (Tüm Gen Dizi Analizi)	MIR192 gene (Sequence Analysis-All Coding Exons)	MIR192	MIR192, MIRN192	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR191S geni (Tüm Gen Dizi Analizi)	MIR191S gene (Sequence Analysis-All Coding Exons)	MIR191S	MIR191S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR191 geni (Tüm Gen Dizi Analizi)	MIR191 gene (Sequence Analysis-All Coding Exons)	MIR191	MIR191	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR190A geni (Tüm Gen Dizi Analizi)	MIR190A gene (Sequence Analysis-All Coding Exons)	MIR190A	MIR190A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1909 geni (Tüm Gen Dizi Analizi)	MIR1909 gene (Sequence Analysis-All Coding Exons)	MIR1909	MIR1909	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR18A geni (Tüm Gen Dizi Analizi)	MIR18A gene (Sequence Analysis-All Coding Exons)	MIR18A	MIR18A, MIRN18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR187 geni (Tüm Gen Dizi Analizi)	MIR187 gene (Sequence Analysis-All Coding Exons)	MIR187	MIR187, MIRN187	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR185 geni (Tüm Gen Dizi Analizi)	MIR185 gene (Sequence Analysis-All Coding Exons)	MIR185	MIR185	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR183 geni (Tüm Gen Dizi Analizi)	MIR183 gene (Sequence Analysis-All Coding Exons)	MIR183	MIR183, MIRN183	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR182 geni (Tüm Gen Dizi Analizi)	MIR182 gene (Sequence Analysis-All Coding Exons)	MIR182	MIR182, MIRN182	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR181C geni (Tüm Gen Dizi Analizi)	MIR181C gene (Sequence Analysis-All Coding Exons)	MIR181C	MIR181C, MIRN181C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR181B2 geni (Tüm Gen Dizi Analizi)	MIR181B2 gene (Sequence Analysis-All Coding Exons)	MIR181B2	MIR181B2, MIRN181B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR181B1 geni (Tüm Gen Dizi Analizi)	MIR181B1 gene (Sequence Analysis-All Coding Exons)	MIR181B1	MIR181B1, MIRN181B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIBP2 geni (Tüm Gen Dizi Analizi)	PPFIBP2 gene (Sequence Analysis-All Coding Exons)	PPFIBP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIBP1 geni (Tüm Gen Dizi Analizi)	PPFIBP1 gene (Sequence Analysis-All Coding Exons)	PPFIBP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR181A2 geni (Tüm Gen Dizi Analizi)	MIR181A2 gene (Sequence Analysis-All Coding Exons)	MIR181A2	MIR181A2, MIRN181A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR181A1 geni (Tüm Gen Dizi Analizi)	MIR181A1 gene (Sequence Analysis-All Coding Exons)	MIR181A1	MIR181A1, MIR213, MIRN181A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PPFIA2 geni (Tüm Gen Dizi Analizi)	PPFIA2 gene (Sequence Analysis-All Coding Exons)	PPFIA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR17 geni (Tüm Gen Dizi Analizi)	MIR17 gene (Sequence Analysis-All Coding Exons)	MIR17	MIR17, MIR91, MIRN17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR16-1 geni (Tüm Gen Dizi Analizi)	MIR16-1 gene (Sequence Analysis-All Coding Exons)	MIR16-1	MIR16-1, MIRN16-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR15A geni (Tüm Gen Dizi Analizi)	MIR15A gene (Sequence Analysis-All Coding Exons)	MIR15A	MIR15A, MIRN15A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR155 geni (Tüm Gen Dizi Analizi)	MIR155 gene (Sequence Analysis-All Coding Exons)	MIR155	MIR155, BIC, MIRN155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR152 geni (Tüm Gen Dizi Analizi)	MIR152 gene (Sequence Analysis-All Coding Exons)	MIR152	MIR152, MIRN152	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR150 geni (Tüm Gen Dizi Analizi)	MIR150 gene (Sequence Analysis-All Coding Exons)	MIR150	MIR150, MIRN150	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR149 geni (Tüm Gen Dizi Analizi)	MIR149 gene (Sequence Analysis-All Coding Exons)	MIR149	MIR149	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR148B geni (Tüm Gen Dizi Analizi)	MIR148B gene (Sequence Analysis-All Coding Exons)	MIR148B	MIR148B, MIRN148B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR148A geni (Tüm Gen Dizi Analizi)	MIR148A gene (Sequence Analysis-All Coding Exons)	MIR148A	MIR148A, MIRN148A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR146B geni (Tüm Gen Dizi Analizi)	MIR146B gene (Sequence Analysis-All Coding Exons)	MIR146B	MIR146B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR146A geni (Tüm Gen Dizi Analizi)	MIR146A gene (Sequence Analysis-All Coding Exons)	MIR146A	MIR146A, MIRN146A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR145 geni (Tüm Gen Dizi Analizi)	MIR145 gene (Sequence Analysis-All Coding Exons)	MIR145	MIR145, MIRN145	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR144 geni (Tüm Gen Dizi Analizi)	MIR144 gene (Sequence Analysis-All Coding Exons)	MIR144	MIR144, MIRN144	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR143 geni (Tüm Gen Dizi Analizi)	MIR143 gene (Sequence Analysis-All Coding Exons)	MIR143	MIR143, MIRN143	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR142 geni (Tüm Gen Dizi Analizi)	MIR142 gene (Sequence Analysis-All Coding Exons)	MIR142	MIR142	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR141 geni (Tüm Gen Dizi Analizi)	MIR141 gene (Sequence Analysis-All Coding Exons)	MIR141	MIR141, MIRN141	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MIR140 geni (Tüm Gen Dizi Analizi)	MIR140 gene (Sequence Analysis-All Coding Exons)	MIR140	MIR140, MIRN140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR139 geni (Tüm Gen Dizi Analizi)	MIR139 gene (Sequence Analysis-All Coding Exons)	MIR139	MIR139, MIR139-3p	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR138-2 geni (Tüm Gen Dizi Analizi)	MIR138-2 gene (Sequence Analysis-All Coding Exons)	MIR138-2	MIR138-2, MIRN138-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR138-1 geni (Tüm Gen Dizi Analizi)	MIR138-1 gene (Sequence Analysis-All Coding Exons)	MIR138-1	MIR138-1, MIRN138-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR137 geni (Tüm Gen Dizi Analizi)	MIR137 gene (Sequence Analysis-All Coding Exons)	MIR137	MIR137, MIRN137	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR136 geni (Tüm Gen Dizi Analizi)	MIR136 gene (Sequence Analysis-All Coding Exons)	MIR136	MIR136, MIRN136	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR134 geni (Tüm Gen Dizi Analizi)	MIR134 gene (Sequence Analysis-All Coding Exons)	MIR134	MIR134	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR133B geni (Tüm Gen Dizi Analizi)	MIR133B gene (Sequence Analysis-All Coding Exons)	MIR133B	MIR133B, MIRN133B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR133A2 geni (Tüm Gen Dizi Analizi)	MIR133A2 gene (Sequence Analysis-All Coding Exons)	MIR133A2	MIR133A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR133A1 geni (Tüm Gen Dizi Analizi)	MIR133A1 gene (Sequence Analysis-All Coding Exons)	MIR133A1	MIR133A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR132 geni (Tüm Gen Dizi Analizi)	MIR132 gene (Sequence Analysis-All Coding Exons)	MIR132	MIR132, MIRN132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR130B geni (Tüm Gen Dizi Analizi)	MIR130B gene (Sequence Analysis-All Coding Exons)	MIR130B	MIR130B, MIRN130B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR130A geni (Tüm Gen Dizi Analizi)	MIR130A gene (Sequence Analysis-All Coding Exons)	MIR130A	MIR130A, MIRN130A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1292 geni (Tüm Gen Dizi Analizi)	MIR1292 gene (Sequence Analysis-All Coding Exons)	MIR1292	MIR1292, MIRN1292	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR128-2 geni (Tüm Gen Dizi Analizi)	MIR128-2 gene (Sequence Analysis-All Coding Exons)	MIR128-2	MIR128-2, MIR128B, MIRN128-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR128-1 geni (Tüm Gen Dizi Analizi)	MIR128-1 gene (Sequence Analysis-All Coding Exons)	MIR128-1	MIR128-1, MIRN128-1, MIR1798	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR127 geni (Tüm Gen Dizi Analizi)	MIR127 gene (Sequence Analysis-All Coding Exons)	MIR127	MIR127, MIRN127	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1260B geni (Tüm Gen Dizi Analizi)	MIR1260B gene (Sequence Analysis-All Coding Exons)	MIR1260B	MIR1260B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR126 geni (Tüm Gen Dizi Analizi)	MIR126 gene (Sequence Analysis-All Coding Exons)	MIR126	MIR126, MIRN126	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR125B2 geni (Tüm Gen Dizi Analizi)	MIR125B2 gene (Sequence Analysis-All Coding Exons)	MIR125B2	MIR125B2, MIRN125B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR125B1 geni (Tüm Gen Dizi Analizi)	MIR125B1 gene (Sequence Analysis-All Coding Exons)	MIR125B1	MIR125B1, MIRN125B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR125A geni (Tüm Gen Dizi Analizi)	MIR125A gene (Sequence Analysis-All Coding Exons)	MIR125A	MIR125A, MIRN125A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTEG geni (Tüm Gen Dizi Analizi)	POTEG gene (Sequence Analysis-All Coding Exons)	POTEG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1258 geni (Tüm Gen Dizi Analizi)	MIR1258 gene (Sequence Analysis-All Coding Exons)	MIR1258	MIR1258	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR124-1 geni (Tüm Gen Dizi Analizi)	MIR124-1 gene (Sequence Analysis-All Coding Exons)	MIR124-1	MIR124-1, MIR124A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTEB geni (Tüm Gen Dizi Analizi)	POTEB gene (Sequence Analysis-All Coding Exons)	POTEB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POTEA geni (Tüm Gen Dizi Analizi)	POTEA gene (Sequence Analysis-All Coding Exons)	POTEA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1231 geni (Tüm Gen Dizi Analizi)	MIR1231 gene (Sequence Analysis-All Coding Exons)	MIR1231	MIR1231	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1225 geni (Tüm Gen Dizi Analizi)	MIR1225 gene (Sequence Analysis-All Coding Exons)	MIR1225	MIR1225, MIRN1225	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1224 geni (Tüm Gen Dizi Analizi)	MIR1224 gene (Sequence Analysis-All Coding Exons)	MIR1224	MIR1224, MIRN1224	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR122 geni (Tüm Gen Dizi Analizi)	MIR122 gene (Sequence Analysis-All Coding Exons)	MIR122	MIR122A, MIRN122A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR10B geni (Tüm Gen Dizi Analizi)	MIR10B gene (Sequence Analysis-All Coding Exons)	MIR10B	MIR10B, MIRN10B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR10A geni (Tüm Gen Dizi Analizi)	MIR10A gene (Sequence Analysis-All Coding Exons)	MIR10A	MIR10A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR107 geni (Tüm Gen Dizi Analizi)	MIR107 gene (Sequence Analysis-All Coding Exons)	MIR107	MIR107, MIRN107	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR106B geni (Tüm Gen Dizi Analizi)	MIR106B gene (Sequence Analysis-All Coding Exons)	MIR106B	MIR106B, MIRN106B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR106A geni (Tüm Gen Dizi Analizi)	MIR106A gene (Sequence Analysis-All Coding Exons)	MIR106A	MIR106A, MIRN106A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR105-2 geni (Tüm Gen Dizi Analizi)	MIR105-2 gene (Sequence Analysis-All Coding Exons)	MIR105-2	MIR105-2, MIRN105-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR105-1 geni (Tüm Gen Dizi Analizi)	MIR105-1 gene (Sequence Analysis-All Coding Exons)	MIR105-1	MIR105-1, MIRN105-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR103A2 geni (Tüm Gen Dizi Analizi)	MIR103A2 gene (Sequence Analysis-All Coding Exons)	MIR103A2	MIR103-2, MIRN103-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR103A1 geni (Tüm Gen Dizi Analizi)	MIR103A1 gene (Sequence Analysis-All Coding Exons)	MIR103A1	MIR103-1, MIRN103-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR101-2 geni (Tüm Gen Dizi Analizi)	MIR101-2 gene (Sequence Analysis-All Coding Exons)	MIR101-2	MIR101-2, MIRN101-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR101-1 geni (Tüm Gen Dizi Analizi)	MIR101-1 gene (Sequence Analysis-All Coding Exons)	MIR101-1	MIR101-1, MIRN101-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR100HG geni (Tüm Gen Dizi Analizi)	MIR100HG gene (Sequence Analysis-All Coding Exons)	MIR100HG	MIR100HG, AGD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR100 geni (Tüm Gen Dizi Analizi)	MIR100 gene (Sequence Analysis-All Coding Exons)	MIR100	MIR100, MIRN100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1-2 geni (Tüm Gen Dizi Analizi)	MIR1-2 gene (Sequence Analysis-All Coding Exons)	MIR1-2	MIR1-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIR1-1 geni (Tüm Gen Dizi Analizi)	MIR1-1 gene (Sequence Analysis-All Coding Exons)	MIR1-1	MIR1-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPOL1 geni (Tüm Gen Dizi Analizi)	MIPOL1 gene (Sequence Analysis-All Coding Exons)	MIPOL1	MIPOL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIOS geni (Tüm Gen Dizi Analizi)	MIOS gene (Sequence Analysis-All Coding Exons)	MIOS	MIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MINOS1 geni (Tüm Gen Dizi Analizi)	MINOS1 gene (Sequence Analysis-All Coding Exons)	MINOS1	MINOS1, MIO10, MIC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MINK1 geni (Tüm Gen Dizi Analizi)	MINK1 gene (Sequence Analysis-All Coding Exons)	MINK1	MINK1, MINK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MINDY3 geni (Tüm Gen Dizi Analizi)	MINDY3 gene (Sequence Analysis-All Coding Exons)	MINDY3	C10orf97, CARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIGA1 geni (Tüm Gen Dizi Analizi)	MIGA1 gene (Sequence Analysis-All Coding Exons)	MIGA1	FAM73A, MIGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIF4GD geni (Tüm Gen Dizi Analizi)	MIF4GD gene (Sequence Analysis-All Coding Exons)	MIF4GD	MIF4GD, SLIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIER1 geni (Tüm Gen Dizi Analizi)	MIER1 gene (Sequence Analysis-All Coding Exons)	MIER1	MIER1, KIAA1610	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIEN1 geni (Tüm Gen Dizi Analizi)	MIEN1 gene (Sequence Analysis-All Coding Exons)	MIEN1	C17orf37, RDX12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIEF2 geni (Tüm Gen Dizi Analizi)	MIEF2 gene (Sequence Analysis-All Coding Exons)	MIEF2	MEIF2, SMCR7, MID49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIEF1 geni (Tüm Gen Dizi Analizi)	MIEF1 gene (Sequence Analysis-All Coding Exons)	MIEF1	MEIF1, SMCR7L, MID51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIDN geni (Tüm Gen Dizi Analizi)	MIDN gene (Sequence Analysis-All Coding Exons)	MIDN	MIDN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MID1IP1 geni (Tüm Gen Dizi Analizi)	MID1IP1 gene (Sequence Analysis-All Coding Exons)	MID1IP1	MID1IP1, MIG12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICU3 geni (Tüm Gen Dizi Analizi)	MICU3 gene (Sequence Analysis-All Coding Exons)	MICU3	MICU3, EFHA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICU2 geni (Tüm Gen Dizi Analizi)	MICU2 gene (Sequence Analysis-All Coding Exons)	MICU2	MICU2, EFHA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICB geni (Tüm Gen Dizi Analizi)	MICB gene (Sequence Analysis-All Coding Exons)	MICB	MICB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICAL2L geni (Tüm Gen Dizi Analizi)	MICAL2L gene (Sequence Analysis-All Coding Exons)	MICAL2L	MICAL2L, EBITEIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICAL3 geni (Tüm Gen Dizi Analizi)	MICAL3 gene (Sequence Analysis-All Coding Exons)	MICAL3	MICAL3, KIAA1364	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICAL2 geni (Tüm Gen Dizi Analizi)	MICAL2 gene (Sequence Analysis-All Coding Exons)	MICAL2	MICAL2, KIAA0750	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR3K geni (Tüm Gen Dizi Analizi)	POLR3K gene (Sequence Analysis-All Coding Exons)	POLR3K		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICAL1 geni (Tüm Gen Dizi Analizi)	MICAL1 gene (Sequence Analysis-All Coding Exons)	MICAL1	MICAL1, NICAL, MICAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MICA geni (Tüm Gen Dizi Analizi)	MICA gene (Sequence Analysis-All Coding Exons)	MICA	MICA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIB2 geni (Tüm Gen Dizi Analizi)	MIB2 gene (Sequence Analysis-All Coding Exons)	MIB2	MIB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIA3 geni (Tüm Gen Dizi Analizi)	MIA3 gene (Sequence Analysis-All Coding Exons)	MIA3	MIA3, TANGO1, TANGO, KIAA0768	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIA2 geni (Tüm Gen Dizi Analizi)	MIA2 gene (Sequence Analysis-All Coding Exons)	MIA2	MGEA, MEA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIA geni (Tüm Gen Dizi Analizi)	MIA gene (Sequence Analysis-All Coding Exons)	MIA	MIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGST3 geni (Tüm Gen Dizi Analizi)	MGST3 gene (Sequence Analysis-All Coding Exons)	MGST3	MGST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGST2 geni (Tüm Gen Dizi Analizi)	MGST2 gene (Sequence Analysis-All Coding Exons)	MGST2	MGST2, GST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGST1 geni (Tüm Gen Dizi Analizi)	MGST1 gene (Sequence Analysis-All Coding Exons)	MGST1	MGST1, GST12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGRN1 geni (Tüm Gen Dizi Analizi)	MGRN1 gene (Sequence Analysis-All Coding Exons)	MGRN1	MGRN1, KIAA0544	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGLL geni (Tüm Gen Dizi Analizi)	MGLL gene (Sequence Analysis-All Coding Exons)	MGLL	MGLL, MGL, HUK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGEA5 geni (Tüm Gen Dizi Analizi)	MGEA5 gene (Sequence Analysis-All Coding Exons)	MGEA5	MGEA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT5B geni (Tüm Gen Dizi Analizi)	MGAT5B gene (Sequence Analysis-All Coding Exons)	MGAT5B	MGAT5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR2D geni (Tüm Gen Dizi Analizi)	POLR2D gene (Sequence Analysis-All Coding Exons)	POLR2D		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT5 geni (Tüm Gen Dizi Analizi)	MGAT5 gene (Sequence Analysis-All Coding Exons)	MGAT5	MGAT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT4D geni (Tüm Gen Dizi Analizi)	MGAT4D gene (Sequence Analysis-All Coding Exons)	MGAT4D	LOC152586	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT4C geni (Tüm Gen Dizi Analizi)	MGAT4C gene (Sequence Analysis-All Coding Exons)	MGAT4C	GNTIVH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT4B geni (Tüm Gen Dizi Analizi)	MGAT4B gene (Sequence Analysis-All Coding Exons)	MGAT4B	MGAT4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT4A geni (Tüm Gen Dizi Analizi)	MGAT4A gene (Sequence Analysis-All Coding Exons)	MGAT4A	MGAT4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT3 geni (Tüm Gen Dizi Analizi)	MGAT3 gene (Sequence Analysis-All Coding Exons)	MGAT3	MGAT3, GNT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLR1B geni (Tüm Gen Dizi Analizi)	POLR1B gene (Sequence Analysis-All Coding Exons)	POLR1B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAT1 geni (Tüm Gen Dizi Analizi)	MGAT1 gene (Sequence Analysis-All Coding Exons)	MGAT1	MGAT1, GLYT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGAM geni (Tüm Gen Dizi Analizi)	MGAM gene (Sequence Analysis-All Coding Exons)	MGAM	MGAM, MGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MGA geni (Tüm Gen Dizi Analizi)	MGA gene (Sequence Analysis-All Coding Exons)	MGA	MGA, KIAA0518	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFSD6 geni (Tüm Gen Dizi Analizi)	MFSD6 gene (Sequence Analysis-All Coding Exons)	MFSD6	MFSD6, MMR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFSD4B geni (Tüm Gen Dizi Analizi)	MFSD4B gene (Sequence Analysis-All Coding Exons)	MFSD4B	MFSD4B, NAGLT1, Kiaa1919	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFSD10 geni (Tüm Gen Dizi Analizi)	MFSD10 gene (Sequence Analysis-All Coding Exons)	MFSD10	TETRAN, TPO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MFNG geni (Tüm Gen Dizi Analizi)	MFNG gene (Sequence Analysis-All Coding Exons)	MFNG	MFNG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFGE8 geni (Tüm Gen Dizi Analizi)	MFGE8 gene (Sequence Analysis-All Coding Exons)	MFGE8	MFGE8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFAP4 geni (Tüm Gen Dizi Analizi)	MFAP4 gene (Sequence Analysis-All Coding Exons)	MFAP4	MFAP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFAP3L geni (Tüm Gen Dizi Analizi)	MFAP3L gene (Sequence Analysis-All Coding Exons)	MFAP3L	MFAP3L, KIAA0626	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFAP3 geni (Tüm Gen Dizi Analizi)	MFAP3 gene (Sequence Analysis-All Coding Exons)	MFAP3	MFAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFAP2 geni (Tüm Gen Dizi Analizi)	MFAP2 gene (Sequence Analysis-All Coding Exons)	MFAP2	MFAP2, MAGP, MAGP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFAP1 geni (Tüm Gen Dizi Analizi)	MFAP1 gene (Sequence Analysis-All Coding Exons)	MFAP1	MFAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEX3D geni (Tüm Gen Dizi Analizi)	MEX3D gene (Sequence Analysis-All Coding Exons)	MEX3D	MEX3D, RKHD1, TINO, KIAA2031	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEX3C geni (Tüm Gen Dizi Analizi)	MEX3C gene (Sequence Analysis-All Coding Exons)	MEX3C	MEX3C, RKHD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEX3B geni (Tüm Gen Dizi Analizi)	MEX3B gene (Sequence Analysis-All Coding Exons)	MEX3B	MEX3B, RKHD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEX3A geni (Tüm Gen Dizi Analizi)	MEX3A gene (Sequence Analysis-All Coding Exons)	MEX3A	MEX3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL9 geni (Tüm Gen Dizi Analizi)	METTL9 gene (Sequence Analysis-All Coding Exons)	METTL9	METTL9, DREV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL3 geni (Tüm Gen Dizi Analizi)	METTL3 gene (Sequence Analysis-All Coding Exons)	METTL3	METTL3, IME4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL22 geni (Tüm Gen Dizi Analizi)	METTL22 gene (Sequence Analysis-All Coding Exons)	METTL22	METTL22, C16orf68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL21C geni (Tüm Gen Dizi Analizi)	METTL21C gene (Sequence Analysis-All Coding Exons)	METTL21C	METTL21C, C13orf39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL21A geni (Tüm Gen Dizi Analizi)	METTL21A gene (Sequence Analysis-All Coding Exons)	METTL21A	METTL21A, FAM119A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
POLD3 geni (Tüm Gen Dizi Analizi)	POLD3 gene (Sequence Analysis-All Coding Exons)	POLD3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL18 geni (Tüm Gen Dizi Analizi)	METTL18 gene (Sequence Analysis-All Coding Exons)	METTL18	METTL18, C1orf156	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL17 geni (Tüm Gen Dizi Analizi)	METTL17 gene (Sequence Analysis-All Coding Exons)	METTL17	METTL17, METT11D1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL14 geni (Tüm Gen Dizi Analizi)	METTL14 gene (Sequence Analysis-All Coding Exons)	METTL14	METTL14, KIAA1627	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL1 geni (Tüm Gen Dizi Analizi)	METTL1 gene (Sequence Analysis-All Coding Exons)	METTL1	METTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METRNL geni (Tüm Gen Dizi Analizi)	METRNL gene (Sequence Analysis-All Coding Exons)	METRNL	METRNL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METRN geni (Tüm Gen Dizi Analizi)	METRN gene (Sequence Analysis-All Coding Exons)	METRN	METRN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METAP2 geni (Tüm Gen Dizi Analizi)	METAP2 gene (Sequence Analysis-All Coding Exons)	METAP2	METAP2, p67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METAP1D geni (Tüm Gen Dizi Analizi)	METAP1D gene (Sequence Analysis-All Coding Exons)	METAP1D	METAP1D, MAP1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METAP1 geni (Tüm Gen Dizi Analizi)	METAP1 gene (Sequence Analysis-All Coding Exons)	METAP1	METAP1, KIAA0094	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MESP1 geni (Tüm Gen Dizi Analizi)	MESP1 gene (Sequence Analysis-All Coding Exons)	MESP1	MESP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MESDC2 geni (Tüm Gen Dizi Analizi)	MESDC2 gene (Sequence Analysis-All Coding Exons)	MESDC2	MESDC2, MES5, BOCA, KIAA0081	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MESDC1 geni (Tüm Gen Dizi Analizi)	MESDC1 gene (Sequence Analysis-All Coding Exons)	MESDC1	MESDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEPE geni (Tüm Gen Dizi Analizi)	MEPE gene (Sequence Analysis-All Coding Exons)	MEPE	MEPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEPCE geni (Tüm Gen Dizi Analizi)	MEPCE gene (Sequence Analysis-All Coding Exons)	MEPCE	MEPCE, BCDIN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEP1B geni (Tüm Gen Dizi Analizi)	MEP1B gene (Sequence Analysis-All Coding Exons)	MEP1B	MEP1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEP1A geni (Tüm Gen Dizi Analizi)	MEP1A gene (Sequence Analysis-All Coding Exons)	MEP1A	MEP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEOX2 geni (Tüm Gen Dizi Analizi)	MEOX2 gene (Sequence Analysis-All Coding Exons)	MEOX2	MEOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEMO1 geni (Tüm Gen Dizi Analizi)	MEMO1 gene (Sequence Analysis-All Coding Exons)	MEMO1	MEMO1, MEMO, C2orf4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MELTF geni (Tüm Gen Dizi Analizi)	MELTF gene (Sequence Analysis-All Coding Exons)	MELTF	MFI2, MAP97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MELK geni (Tüm Gen Dizi Analizi)	MELK gene (Sequence Analysis-All Coding Exons)	MELK	MELK, KIAA0175	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEIS2 geni (Tüm Gen Dizi Analizi)	MEIS2 gene (Sequence Analysis-All Coding Exons)	MEIS2	MEIS2, MRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEIS1 geni (Tüm Gen Dizi Analizi)	MEIS1 gene (Sequence Analysis-All Coding Exons)	MEIS1	MEIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEIOC geni (Tüm Gen Dizi Analizi)	MEIOC gene (Sequence Analysis-All Coding Exons)	MEIOC	MEIOC, C17orf104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEIKIN geni (Tüm Gen Dizi Analizi)	MEIKIN gene (Sequence Analysis-All Coding Exons)	MEIKIN	MEIKIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEIG1 geni (Tüm Gen Dizi Analizi)	MEIG1 gene (Sequence Analysis-All Coding Exons)	MEIG1	MEIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEGF9 geni (Tüm Gen Dizi Analizi)	MEGF9 gene (Sequence Analysis-All Coding Exons)	MEGF9	MEGF9, EGFL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEGF6 geni (Tüm Gen Dizi Analizi)	MEGF6 gene (Sequence Analysis-All Coding Exons)	MEGF6	MEGF6, EGFL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEGF11 geni (Tüm Gen Dizi Analizi)	MEGF11 gene (Sequence Analysis-All Coding Exons)	MEGF11	MEGF11, KIAA1781	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG8 geni (Tüm Gen Dizi Analizi)	MEG8 gene (Sequence Analysis-All Coding Exons)	MEG8	MEG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG3 geni (Tüm Gen Dizi Analizi)	MEG3 gene (Sequence Analysis-All Coding Exons)	MEG3	MEG3, GTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG3 geni (Tüm Gen Dizi Analizi)	MEG3 gene (Sequence Analysis-All Coding Exons)	MEG3	MEG3, GTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG3 geni (Tüm Gen Dizi Analizi)	MEG3 gene (Sequence Analysis-All Coding Exons)	MEG3	MEG3, GTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG3 geni (Tüm Gen Dizi Analizi)	MEG3 gene (Sequence Analysis-All Coding Exons)	MEG3	MEG3, GTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEG3 geni (Tüm Gen Dizi Analizi)	MEG3 gene (Sequence Analysis-All Coding Exons)	MEG3	MEG3, GTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEF2D geni (Tüm Gen Dizi Analizi)	MEF2D gene (Sequence Analysis-All Coding Exons)	MEF2D	MEF2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEF2B geni (Tüm Gen Dizi Analizi)	MEF2B gene (Sequence Analysis-All Coding Exons)	MEF2B	MEF2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED9 geni (Tüm Gen Dizi Analizi)	MED9 gene (Sequence Analysis-All Coding Exons)	MED9	MED9, MED25, FLJ10193	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED8 geni (Tüm Gen Dizi Analizi)	MED8 gene (Sequence Analysis-All Coding Exons)	MED8	MED8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED7 geni (Tüm Gen Dizi Analizi)	MED7 gene (Sequence Analysis-All Coding Exons)	MED7	MED7, CRSP9, CRSP33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED4 geni (Tüm Gen Dizi Analizi)	MED4 gene (Sequence Analysis-All Coding Exons)	MED4	MED4, DRIP36, HSPC126	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED30 geni (Tüm Gen Dizi Analizi)	MED30 gene (Sequence Analysis-All Coding Exons)	MED30	MED30, TRAP25, THRAP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED29 geni (Tüm Gen Dizi Analizi)	MED29 gene (Sequence Analysis-All Coding Exons)	MED29	MED29, IXL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED28 geni (Tüm Gen Dizi Analizi)	MED28 gene (Sequence Analysis-All Coding Exons)	MED28	MED28, EG1, MAGICIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED27 geni (Tüm Gen Dizi Analizi)	MED27 gene (Sequence Analysis-All Coding Exons)	MED27	MED27, CRSP8, CRAP34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED26 geni (Tüm Gen Dizi Analizi)	MED26 gene (Sequence Analysis-All Coding Exons)	MED26	MED26, CRSP7, CRSP70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED24 geni (Tüm Gen Dizi Analizi)	MED24 gene (Sequence Analysis-All Coding Exons)	MED24	MED24, TRAP100, KIAA0130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED22 geni (Tüm Gen Dizi Analizi)	MED22 gene (Sequence Analysis-All Coding Exons)	MED22	SURF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED21 geni (Tüm Gen Dizi Analizi)	MED21 gene (Sequence Analysis-All Coding Exons)	MED21	MED21, SURB7, SRB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED20 geni (Tüm Gen Dizi Analizi)	MED20 gene (Sequence Analysis-All Coding Exons)	MED20	MED20, TRFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED19 geni (Tüm Gen Dizi Analizi)	MED19 gene (Sequence Analysis-All Coding Exons)	MED19	MED19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED18 geni (Tüm Gen Dizi Analizi)	MED18 gene (Sequence Analysis-All Coding Exons)	MED18	MED18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED15 geni (Tüm Gen Dizi Analizi)	MED15 gene (Sequence Analysis-All Coding Exons)	MED15	MED15, PCQAP, TIG1, ARC105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED14 geni (Tüm Gen Dizi Analizi)	MED14 gene (Sequence Analysis-All Coding Exons)	MED14	MED14, CRSP2, CXorf4, TRAP170, EXLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED13 geni (Tüm Gen Dizi Analizi)	MED13 gene (Sequence Analysis-All Coding Exons)	MED13	MED13, THRAP1, TRAP240	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED12L geni (Tüm Gen Dizi Analizi)	MED12L gene (Sequence Analysis-All Coding Exons)	MED12L	MED12L, NOPAR, KIAA1635	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED11 geni (Tüm Gen Dizi Analizi)	MED11 gene (Sequence Analysis-All Coding Exons)	MED11	MED11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED10 geni (Tüm Gen Dizi Analizi)	MED10 gene (Sequence Analysis-All Coding Exons)	MED10	MED10, NUT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED1 geni (Tüm Gen Dizi Analizi)	MED1 gene (Sequence Analysis-All Coding Exons)	MED1	MED1, PPARBP, PBP, TRAP220	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEAF6 geni (Tüm Gen Dizi Analizi)	MEAF6 gene (Sequence Analysis-All Coding Exons)	MEAF6	MEAF6, C1orf149, EAF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MEA1 geni (Tüm Gen Dizi Analizi)	MEA1 gene (Sequence Analysis-All Coding Exons)	MEA1	MEA1, MEA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ME3 geni (Tüm Gen Dizi Analizi)	ME3 gene (Sequence Analysis-All Coding Exons)	ME3	ME3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ME2 geni (Tüm Gen Dizi Analizi)	ME2 gene (Sequence Analysis-All Coding Exons)	ME2	ME2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ME1 geni (Tüm Gen Dizi Analizi)	ME1 gene (Sequence Analysis-All Coding Exons)	ME1	ME1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDM4 geni (Tüm Gen Dizi Analizi)	MDM4 gene (Sequence Analysis-All Coding Exons)	MDM4	MDM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDM1 geni (Tüm Gen Dizi Analizi)	MDM1 gene (Sequence Analysis-All Coding Exons)	MDM1	MDM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDK geni (Tüm Gen Dizi Analizi)	MDK gene (Sequence Analysis-All Coding Exons)	MDK	MDK, NEGF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDH1 geni (Tüm Gen Dizi Analizi)	MDH1 gene (Sequence Analysis-All Coding Exons)	MDH1	MDH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDGA2 geni (Tüm Gen Dizi Analizi)	MDGA2 gene (Sequence Analysis-All Coding Exons)	MDGA2	MDGA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDGA1 geni (Tüm Gen Dizi Analizi)	MDGA1 gene (Sequence Analysis-All Coding Exons)	MDGA1	MDGA1, GPIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDFIC geni (Tüm Gen Dizi Analizi)	MDFIC gene (Sequence Analysis-All Coding Exons)	MDFIC	MDFIC, HIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDFI geni (Tüm Gen Dizi Analizi)	MDFI gene (Sequence Analysis-All Coding Exons)	MDFI	MDFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDC1 geni (Tüm Gen Dizi Analizi)	MDC1 gene (Sequence Analysis-All Coding Exons)	MDC1	MDC1, NFBD1, KIAA0170	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCUR1 geni (Tüm Gen Dizi Analizi)	MCUR1 gene (Sequence Analysis-All Coding Exons)	MCUR1	MCUR1, CCDC90A, FMP32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCU geni (Tüm Gen Dizi Analizi)	MCU gene (Sequence Analysis-All Coding Exons)	MCU	MCU, CCDC109A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCTS1 geni (Tüm Gen Dizi Analizi)	MCTS1 gene (Sequence Analysis-All Coding Exons)	MCTS1	MCTS1, MCT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MCTP2 geni (Tüm Gen Dizi Analizi)	MCTP2 gene (Sequence Analysis-All Coding Exons)	MCTP2	MCTP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCTP1 geni (Tüm Gen Dizi Analizi)	MCTP1 gene (Sequence Analysis-All Coding Exons)	MCTP1	MCTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCRS1 geni (Tüm Gen Dizi Analizi)	MCRS1 gene (Sequence Analysis-All Coding Exons)	MCRS1	MCRS1, MSP58, MCRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCRIP1 geni (Tüm Gen Dizi Analizi)	MCRIP1 gene (Sequence Analysis-All Coding Exons)	MCRIP1	FAM195B, MCRIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCOLN3 geni (Tüm Gen Dizi Analizi)	MCOLN3 gene (Sequence Analysis-All Coding Exons)	MCOLN3	MCOLN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCOLN2 geni (Tüm Gen Dizi Analizi)	MCOLN2 gene (Sequence Analysis-All Coding Exons)	MCOLN2	MCOLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCMBP geni (Tüm Gen Dizi Analizi)	MCMBP gene (Sequence Analysis-All Coding Exons)	MCMBP	MCMBP, C10orf119	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCM7 geni (Tüm Gen Dizi Analizi)	MCM7 gene (Sequence Analysis-All Coding Exons)	MCM7	MCM7, MCM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCM5 geni (Tüm Gen Dizi Analizi)	MCM5 gene (Sequence Analysis-All Coding Exons)	MCM5	MCM5, CDC46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCM3AP geni (Tüm Gen Dizi Analizi)	MCM3AP gene (Sequence Analysis-All Coding Exons)	MCM3AP	MCM3AP, MAP80, GANP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCM3 geni (Tüm Gen Dizi Analizi)	MCM3 gene (Sequence Analysis-All Coding Exons)	MCM3	MCM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCM10 geni (Tüm Gen Dizi Analizi)	MCM10 gene (Sequence Analysis-All Coding Exons)	MCM10	MCM10, CNA43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCL1 geni (Tüm Gen Dizi Analizi)	MCL1 gene (Sequence Analysis-All Coding Exons)	MCL1	MCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCIDAS geni (Tüm Gen Dizi Analizi)	MCIDAS gene (Sequence Analysis-All Coding Exons)	MCIDAS	IDA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCHR2 geni (Tüm Gen Dizi Analizi)	MCHR2 gene (Sequence Analysis-All Coding Exons)	MCHR2	MCHR2, MCH2R, SLT, MCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCHR1 geni (Tüm Gen Dizi Analizi)	MCHR1 gene (Sequence Analysis-All Coding Exons)	MCHR1	MCHR1, GPR24, SLC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCF2L geni (Tüm Gen Dizi Analizi)	MCF2L gene (Sequence Analysis-All Coding Exons)	MCF2L	MCF2L, OST, KIAA0362	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCF2 geni (Tüm Gen Dizi Analizi)	MCF2 gene (Sequence Analysis-All Coding Exons)	MCF2	MCF2, DBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCEMP1 geni (Tüm Gen Dizi Analizi)	MCEMP1 gene (Sequence Analysis-All Coding Exons)	MCEMP1	MCEMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCCD1 geni (Tüm Gen Dizi Analizi)	MCCD1 gene (Sequence Analysis-All Coding Exons)	MCCD1	MCCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCAT geni (Tüm Gen Dizi Analizi)	MCAT gene (Sequence Analysis-All Coding Exons)	MCAT	MCAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPP3 geni (Tüm Gen Dizi Analizi)	PLPP3 gene (Sequence Analysis-All Coding Exons)	PLPP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPP2 geni (Tüm Gen Dizi Analizi)	PLPP2 gene (Sequence Analysis-All Coding Exons)	PLPP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLPP1 geni (Tüm Gen Dizi Analizi)	PLPP1 gene (Sequence Analysis-All Coding Exons)	PLPP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MCAM geni (Tüm Gen Dizi Analizi)	MCAM gene (Sequence Analysis-All Coding Exons)	MCAM	MCAM, MUC18, CD146	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MC5R geni (Tüm Gen Dizi Analizi)	MC5R gene (Sequence Analysis-All Coding Exons)	MC5R	MC5R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBTPS1 geni (Tüm Gen Dizi Analizi)	MBTPS1 gene (Sequence Analysis-All Coding Exons)	MBTPS1	MBTPS1, S1P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBP geni (Tüm Gen Dizi Analizi)	MBP gene (Sequence Analysis-All Coding Exons)	MBP	MBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBOAT4 geni (Tüm Gen Dizi Analizi)	MBOAT4 gene (Sequence Analysis-All Coding Exons)	MBOAT4	MBOAT4, GOAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBOAT2 geni (Tüm Gen Dizi Analizi)	MBOAT2 gene (Sequence Analysis-All Coding Exons)	MBOAT2	MBOAT2, LPCAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBOAT1 geni (Tüm Gen Dizi Analizi)	MBOAT1 gene (Sequence Analysis-All Coding Exons)	MBOAT1	MBOAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBNL3 geni (Tüm Gen Dizi Analizi)	MBNL3 gene (Sequence Analysis-All Coding Exons)	MBNL3	MBNL3, MBXL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBNL1 geni (Tüm Gen Dizi Analizi)	MBNL1 gene (Sequence Analysis-All Coding Exons)	MBNL1	MBNL1, KIAA0428, EXP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBIP geni (Tüm Gen Dizi Analizi)	MBIP gene (Sequence Analysis-All Coding Exons)	MBIP	MBIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD4 geni (Tüm Gen Dizi Analizi)	MBD4 gene (Sequence Analysis-All Coding Exons)	MBD4	MBD4, MED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD3L2 geni (Tüm Gen Dizi Analizi)	MBD3L2 gene (Sequence Analysis-All Coding Exons)	MBD3L2	MBD3L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD3L1 geni (Tüm Gen Dizi Analizi)	MBD3L1 gene (Sequence Analysis-All Coding Exons)	MBD3L1	MBD3L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLK1 geni (Tüm Gen Dizi Analizi)	PLK1 gene (Sequence Analysis-All Coding Exons)	PLK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD3 geni (Tüm Gen Dizi Analizi)	MBD3 gene (Sequence Analysis-All Coding Exons)	MBD3	MBD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD2 geni (Tüm Gen Dizi Analizi)	MBD2 gene (Sequence Analysis-All Coding Exons)	MBD2	MBD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBD1 geni (Tüm Gen Dizi Analizi)	MBD1 gene (Sequence Analysis-All Coding Exons)	MBD1	MBD1, PCM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MB21D1 geni (Tüm Gen Dizi Analizi)	MB21D1 gene (Sequence Analysis-All Coding Exons)	MB21D1	MB21D1, C6orf150	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MB geni (Tüm Gen Dizi Analizi)	MB gene (Sequence Analysis-All Coding Exons)	MB	MB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAZ geni (Tüm Gen Dizi Analizi)	MAZ gene (Sequence Analysis-All Coding Exons)	MAZ	MAZ, ZF87, PUR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAVS geni (Tüm Gen Dizi Analizi)	MAVS gene (Sequence Analysis-All Coding Exons)	MAVS	MAVS, VISA, IPS1, MAVS, CARDIF, KIAA1271	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAU2 geni (Tüm Gen Dizi Analizi)	MAU2 gene (Sequence Analysis-All Coding Exons)	MAU2	MAU2, SCC4, KIAA0892	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MATN4 geni (Tüm Gen Dizi Analizi)	MATN4 gene (Sequence Analysis-All Coding Exons)	MATN4	MATN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MATN2 geni (Tüm Gen Dizi Analizi)	MATN2 gene (Sequence Analysis-All Coding Exons)	MATN2	MATN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MATN1 geni (Tüm Gen Dizi Analizi)	MATN1 gene (Sequence Analysis-All Coding Exons)	MATN1	MATN1, CRTM, CMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MATK geni (Tüm Gen Dizi Analizi)	MATK gene (Sequence Analysis-All Coding Exons)	MATK	MATK, HYL, CTK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAT2A geni (Tüm Gen Dizi Analizi)	MAT2A gene (Sequence Analysis-All Coding Exons)	MAT2A	MAT2A, MATA2, SAMS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MASTL geni (Tüm Gen Dizi Analizi)	MASTL gene (Sequence Analysis-All Coding Exons)	MASTL	MASTL, FLJ14813, GWL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAST3 geni (Tüm Gen Dizi Analizi)	MAST3 gene (Sequence Analysis-All Coding Exons)	MAST3	MAST3, KIAA0561	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAST2 geni (Tüm Gen Dizi Analizi)	MAST2 gene (Sequence Analysis-All Coding Exons)	MAST2	MAST2, MAST205, KIAA0807	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAST1 geni (Tüm Gen Dizi Analizi)	MAST1 gene (Sequence Analysis-All Coding Exons)	MAST1	MAST1, SAST, KIAA0973	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAS1L geni (Tüm Gen Dizi Analizi)	MAS1L gene (Sequence Analysis-All Coding Exons)	MAS1L	MAS1L, MRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAS1 geni (Tüm Gen Dizi Analizi)	MAS1 gene (Sequence Analysis-All Coding Exons)	MAS1	MAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARVELD3 geni (Tüm Gen Dizi Analizi)	MARVELD3 gene (Sequence Analysis-All Coding Exons)	MARVELD3	MARVELD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARVELD1 geni (Tüm Gen Dizi Analizi)	MARVELD1 gene (Sequence Analysis-All Coding Exons)	MARVELD1	MARVELD1, MARVD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARK4 geni (Tüm Gen Dizi Analizi)	MARK4 gene (Sequence Analysis-All Coding Exons)	MARK4	MARK4, KIAA1860, MARKL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARK3 geni (Tüm Gen Dizi Analizi)	MARK3 gene (Sequence Analysis-All Coding Exons)	MARK3	MARK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARK2 geni (Tüm Gen Dizi Analizi)	MARK2 gene (Sequence Analysis-All Coding Exons)	MARK2	MARK2, EMK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARK1 geni (Tüm Gen Dizi Analizi)	MARK1 gene (Sequence Analysis-All Coding Exons)	MARK1	MARK1, KIAA1477	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARF1 geni (Tüm Gen Dizi Analizi)	MARF1 gene (Sequence Analysis-All Coding Exons)	MARF1	KIAA0430, LKAP, MARF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARCO geni (Tüm Gen Dizi Analizi)	MARCO gene (Sequence Analysis-All Coding Exons)	MARCO	MARCO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARCKSL1 geni (Tüm Gen Dizi Analizi)	MARCKSL1 gene (Sequence Analysis-All Coding Exons)	MARCKSL1	MARCKSL1, MLP, MRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MARCKS geni (Tüm Gen Dizi Analizi)	MARCKS gene (Sequence Analysis-All Coding Exons)	MARCKS	MRACKS, MACS, PKCSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPRE3 geni (Tüm Gen Dizi Analizi)	MAPRE3 gene (Sequence Analysis-All Coding Exons)	MAPRE3	MAPRE3, EB3, RP3, EBF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPRE1 geni (Tüm Gen Dizi Analizi)	MAPRE1 gene (Sequence Analysis-All Coding Exons)	MAPRE1	MAPRE1, EB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPKAPK5 geni (Tüm Gen Dizi Analizi)	MAPKAPK5 gene (Sequence Analysis-All Coding Exons)	MAPKAPK5	MAPKAPK5, PRAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPKAPK2 geni (Tüm Gen Dizi Analizi)	MAPKAPK2 gene (Sequence Analysis-All Coding Exons)	MAPKAPK2	MAPKAPK2, MK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPKAP1 geni (Tüm Gen Dizi Analizi)	MAPKAP1 gene (Sequence Analysis-All Coding Exons)	MAPKAP1	MAPKAP1, SIN1, MIP1, JC310	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK9 geni (Tüm Gen Dizi Analizi)	MAPK9 gene (Sequence Analysis-All Coding Exons)	MAPK9	MAPK9, PRKM9, JNK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK8IP3 geni (Tüm Gen Dizi Analizi)	MAPK8IP3 gene (Sequence Analysis-All Coding Exons)	MAPK8IP3	MAPK8IP3, SYD2, JSAP1, JIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK8IP2 geni (Tüm Gen Dizi Analizi)	MAPK8IP2 gene (Sequence Analysis-All Coding Exons)	MAPK8IP2	MAPK8IP2, JIP2, IB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK8 geni (Tüm Gen Dizi Analizi)	MAPK8 gene (Sequence Analysis-All Coding Exons)	MAPK8	MAPK8, PRKM8, JNK1, SAPK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK7 geni (Tüm Gen Dizi Analizi)	MAPK7 gene (Sequence Analysis-All Coding Exons)	MAPK7	MAPK7, PRKM7, ERK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK4 geni (Tüm Gen Dizi Analizi)	MAPK4 gene (Sequence Analysis-All Coding Exons)	MAPK4	MAPK4, PRKM4, ERK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK3 geni (Tüm Gen Dizi Analizi)	MAPK3 gene (Sequence Analysis-All Coding Exons)	MAPK3	MAPK3, PRKM3, ERK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK1IP1L geni (Tüm Gen Dizi Analizi)	MAPK1IP1L gene (Sequence Analysis-All Coding Exons)	MAPK1IP1L	MAP1IP1L, MISS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK14 geni (Tüm Gen Dizi Analizi)	MAPK14 gene (Sequence Analysis-All Coding Exons)	MAPK14	MAPK14, CSBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK12 geni (Tüm Gen Dizi Analizi)	MAPK12 gene (Sequence Analysis-All Coding Exons)	MAPK12	MAPK12, SAPK3, ERK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK10 geni (Tüm Gen Dizi Analizi)	MAPK10 gene (Sequence Analysis-All Coding Exons)	MAPK10	MAPK10, PRKM10, JNK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK1 geni (Tüm Gen Dizi Analizi)	MAPK1 gene (Sequence Analysis-All Coding Exons)	MAPK1	MAPK1, PRKM1, ERK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP9 geni (Tüm Gen Dizi Analizi)	MAP9 gene (Sequence Analysis-All Coding Exons)	MAP9	ASAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP7D3 geni (Tüm Gen Dizi Analizi)	MAP7D3 gene (Sequence Analysis-All Coding Exons)	MAP7D3	MAP7D3, MDP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP7 geni (Tüm Gen Dizi Analizi)	MAP7 gene (Sequence Analysis-All Coding Exons)	MAP7	MAP7, EMAP115	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP6D1 geni (Tüm Gen Dizi Analizi)	MAP6D1 gene (Sequence Analysis-All Coding Exons)	MAP6D1	MAPO6D1, SL21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP4K5 geni (Tüm Gen Dizi Analizi)	MAP4K5 gene (Sequence Analysis-All Coding Exons)	MAP4K5	MAP4K5, MAPKKK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP4K4 geni (Tüm Gen Dizi Analizi)	MAP4K4 gene (Sequence Analysis-All Coding Exons)	MAP4K4	MAP4K4, HGK, NIK, KIAA0687	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCD4 geni (Tüm Gen Dizi Analizi)	PLCD4 gene (Sequence Analysis-All Coding Exons)	PLCD4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PLCD3 geni (Tüm Gen Dizi Analizi)	PLCD3 gene (Sequence Analysis-All Coding Exons)	PLCD3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP4K3 geni (Tüm Gen Dizi Analizi)	MAP4K3 gene (Sequence Analysis-All Coding Exons)	MAP4K3	MAP4K3, GLK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MAP4K2 geni (Tüm Gen Dizi Analizi)	MAP4K2 gene (Sequence Analysis-All Coding Exons)	MAP4K2	MAP4K2, RAB8IP, GCK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP4K1 geni (Tüm Gen Dizi Analizi)	MAP4K1 gene (Sequence Analysis-All Coding Exons)	MAP4K1	MAP4K1, HPK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP4 geni (Tüm Gen Dizi Analizi)	MAP4 gene (Sequence Analysis-All Coding Exons)	MAP4	MAP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K9 geni (Tüm Gen Dizi Analizi)	MAP3K9 gene (Sequence Analysis-All Coding Exons)	MAP3K9	MAP3K9, MLK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K7CL geni (Tüm Gen Dizi Analizi)	MAP3K7CL gene (Sequence Analysis-All Coding Exons)	MAP3K7CL	TAK1L, C21orf7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K6 geni (Tüm Gen Dizi Analizi)	MAP3K6 gene (Sequence Analysis-All Coding Exons)	MAP3K6	MAP3K6, MAPKKK6, ASK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K5 geni (Tüm Gen Dizi Analizi)	MAP3K5 gene (Sequence Analysis-All Coding Exons)	MAP3K5	MAP3K5, MEKK5, MAPKKK5, ASK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K4 geni (Tüm Gen Dizi Analizi)	MAP3K4 gene (Sequence Analysis-All Coding Exons)	MAP3K4	MAP3K4, MEKK4, MTK1, MAPKKK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K3 geni (Tüm Gen Dizi Analizi)	MAP3K3 gene (Sequence Analysis-All Coding Exons)	MAP3K3	MAP3K3, MEKK3, MAPKKK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K21 geni (Tüm Gen Dizi Analizi)	MAP3K21 gene (Sequence Analysis-All Coding Exons)	MAP3K21	MLK4, KIAA1804	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K2 geni (Tüm Gen Dizi Analizi)	MAP3K2 gene (Sequence Analysis-All Coding Exons)	MAP3K2	MAP3K2, MEKK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K15 geni (Tüm Gen Dizi Analizi)	MAP3K15 gene (Sequence Analysis-All Coding Exons)	MAP3K15	MAP3K15, ASK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K14 geni (Tüm Gen Dizi Analizi)	MAP3K14 gene (Sequence Analysis-All Coding Exons)	MAP3K14	MAP3K14, NIK, HSNIK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K13 geni (Tüm Gen Dizi Analizi)	MAP3K13 gene (Sequence Analysis-All Coding Exons)	MAP3K13	MAP3K13, LZK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K12 geni (Tüm Gen Dizi Analizi)	MAP3K12 gene (Sequence Analysis-All Coding Exons)	MAP3K12	MAP3K12, ZPK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K11 geni (Tüm Gen Dizi Analizi)	MAP3K11 gene (Sequence Analysis-All Coding Exons)	MAP3K11	MAP3K11, MLK3, PTK1, SPRK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP3K10 geni (Tüm Gen Dizi Analizi)	MAP3K10 gene (Sequence Analysis-All Coding Exons)	MAP3K10	MAP3K10, MLK2, MST	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2K7 geni (Tüm Gen Dizi Analizi)	MAP2K7 gene (Sequence Analysis-All Coding Exons)	MAP2K7	MAP2K7, PRKMK7, MKK7, JNKK2, MAPK7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2K6 geni (Tüm Gen Dizi Analizi)	MAP2K6 gene (Sequence Analysis-All Coding Exons)	MAP2K6	MAP2K6, PRKMK6, MKK6, MEK6, MAPK6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2K5 geni (Tüm Gen Dizi Analizi)	MAP2K5 gene (Sequence Analysis-All Coding Exons)	MAP2K5	MAP2K5, PRKMK5, MEK5, MAPK5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2K4 geni (Tüm Gen Dizi Analizi)	MAP2K4 gene (Sequence Analysis-All Coding Exons)	MAP2K4	SERK1, PRKMK4, MAPKK4, JNKK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2K3 geni (Tüm Gen Dizi Analizi)	MAP2K3 gene (Sequence Analysis-All Coding Exons)	MAP2K3	PRKMK3, MAPKK3, MEK3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP2 geni (Tüm Gen Dizi Analizi)	MAP2 gene (Sequence Analysis-All Coding Exons)	MAP2	MAP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1S geni (Tüm Gen Dizi Analizi)	MAP1S gene (Sequence Analysis-All Coding Exons)	MAP1S	MAP1S, BPY2IP1, VCY2IP1, C19ORF5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1LC3B geni (Tüm Gen Dizi Analizi)	MAP1LC3B gene (Sequence Analysis-All Coding Exons)	MAP1LC3B	MAP1LC3B, LC3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1LC3A geni (Tüm Gen Dizi Analizi)	MAP1LC3A gene (Sequence Analysis-All Coding Exons)	MAP1LC3A	MAP1ALC3, MAP1BLC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1B geni (Tüm Gen Dizi Analizi)	MAP1B gene (Sequence Analysis-All Coding Exons)	MAP1B	MAP1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1A geni (Tüm Gen Dizi Analizi)	MAP1A gene (Sequence Analysis-All Coding Exons)	MAP1A	MAP1A, MAP1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAOB geni (Tüm Gen Dizi Analizi)	MAOB gene (Sequence Analysis-All Coding Exons)	MAOB	MAOB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MANF geni (Tüm Gen Dizi Analizi)	MANF gene (Sequence Analysis-All Coding Exons)	MANF	ARMET, ARP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MANEA geni (Tüm Gen Dizi Analizi)	MANEA gene (Sequence Analysis-All Coding Exons)	MANEA	MANEA, ENDO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN2C1 geni (Tüm Gen Dizi Analizi)	MAN2C1 gene (Sequence Analysis-All Coding Exons)	MAN2C1	MAN2C1, MANA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN2A2 geni (Tüm Gen Dizi Analizi)	MAN2A2 gene (Sequence Analysis-All Coding Exons)	MAN2A2	MAN2A2, MANA2X	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN2A1 geni (Tüm Gen Dizi Analizi)	MAN2A1 gene (Sequence Analysis-All Coding Exons)	MAN2A1	MAN2A1, MANA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN1C1 geni (Tüm Gen Dizi Analizi)	MAN1C1 gene (Sequence Analysis-All Coding Exons)	MAN1C1	MAN1C1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN1A2 geni (Tüm Gen Dizi Analizi)	MAN1A2 gene (Sequence Analysis-All Coding Exons)	MAN1A2	MAN1A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAN1A1 geni (Tüm Gen Dizi Analizi)	MAN1A1 gene (Sequence Analysis-All Coding Exons)	MAN1A1	MAN1A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAMSTR geni (Tüm Gen Dizi Analizi)	MAMSTR gene (Sequence Analysis-All Coding Exons)	MAMSTR	MAMSTR, MASTR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAML3 geni (Tüm Gen Dizi Analizi)	MAML3 gene (Sequence Analysis-All Coding Exons)	MAML3	MAML3, MAM2, KIAA1816	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAML1 geni (Tüm Gen Dizi Analizi)	MAML1 gene (Sequence Analysis-All Coding Exons)	MAML1	MAML1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAMDC4 geni (Tüm Gen Dizi Analizi)	MAMDC4 gene (Sequence Analysis-All Coding Exons)	MAMDC4	MAMDC4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAMDC2 geni (Tüm Gen Dizi Analizi)	MAMDC2 gene (Sequence Analysis-All Coding Exons)	MAMDC2	MAMDC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MALSU1 geni (Tüm Gen Dizi Analizi)	MALSU1 gene (Sequence Analysis-All Coding Exons)	MALSU1	MALSU1, C7orf30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MALL geni (Tüm Gen Dizi Analizi)	MALL gene (Sequence Analysis-All Coding Exons)	MALL	MALL, BENE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MALAT1 geni (Tüm Gen Dizi Analizi)	MALAT1 gene (Sequence Analysis-All Coding Exons)	MALAT1	MALAT1, PRO1073	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAL geni (Tüm Gen Dizi Analizi)	MAL gene (Sequence Analysis-All Coding Exons)	MAL	MAL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAJIN geni (Tüm Gen Dizi Analizi)	MAJIN gene (Sequence Analysis-All Coding Exons)	MAJIN	MAJIN, C11orf85	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAIP1 geni (Tüm Gen Dizi Analizi)	MAIP1 gene (Sequence Analysis-All Coding Exons)	MAIP1	MAIP1, C2orf47	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGOH geni (Tüm Gen Dizi Analizi)	MAGOH gene (Sequence Analysis-All Coding Exons)	MAGOH	MAGOH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGI3 geni (Tüm Gen Dizi Analizi)	MAGI3 gene (Sequence Analysis-All Coding Exons)	MAGI3	MAGI3, KIAA1634	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGI2 geni (Tüm Gen Dizi Analizi)	MAGI2 gene (Sequence Analysis-All Coding Exons)	MAGI2	AIP1, KIAA0705	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGI1 geni (Tüm Gen Dizi Analizi)	MAGI1 gene (Sequence Analysis-All Coding Exons)	MAGI1	MAGI1, BAIAP1, WWP3, TNRC19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEH1 geni (Tüm Gen Dizi Analizi)	MAGEH1 gene (Sequence Analysis-All Coding Exons)	MAGEH1	MAGEH1, MAGEH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEF1 geni (Tüm Gen Dizi Analizi)	MAGEF1 gene (Sequence Analysis-All Coding Exons)	MAGEF1	MAGEF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEE2 geni (Tüm Gen Dizi Analizi)	MAGEE2 gene (Sequence Analysis-All Coding Exons)	MAGEE2	MAGEE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEE1 geni (Tüm Gen Dizi Analizi)	MAGEE1 gene (Sequence Analysis-All Coding Exons)	MAGEE1	MAGEE1, DAMAGE, KIAA1587	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGED4B geni (Tüm Gen Dizi Analizi)	MAGED4B gene (Sequence Analysis-All Coding Exons)	MAGED4B	MAGED4B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGED4 geni (Tüm Gen Dizi Analizi)	MAGED4 gene (Sequence Analysis-All Coding Exons)	MAGED4	MAGED4, MAGEE1, KIAA1958	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGED1 geni (Tüm Gen Dizi Analizi)	MAGED1 gene (Sequence Analysis-All Coding Exons)	MAGED1	MAGED1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEC3 geni (Tüm Gen Dizi Analizi)	MAGEC3 gene (Sequence Analysis-All Coding Exons)	MAGEC3	MAGEC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEC2 geni (Tüm Gen Dizi Analizi)	MAGEC2 gene (Sequence Analysis-All Coding Exons)	MAGEC2	MAGEC2, MAGEE1, MAGEC2, CT10, HCA587	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEC1 geni (Tüm Gen Dizi Analizi)	MAGEC1 gene (Sequence Analysis-All Coding Exons)	MAGEC1	MAGEC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB6 geni (Tüm Gen Dizi Analizi)	MAGEB6 gene (Sequence Analysis-All Coding Exons)	MAGEB6	MAGEB6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB5 geni (Tüm Gen Dizi Analizi)	MAGEB5 gene (Sequence Analysis-All Coding Exons)	MAGEB5	MAGEB5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB4 geni (Tüm Gen Dizi Analizi)	MAGEB4 gene (Sequence Analysis-All Coding Exons)	MAGEB4	MAGEB4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB3 geni (Tüm Gen Dizi Analizi)	MAGEB3 gene (Sequence Analysis-All Coding Exons)	MAGEB3	MAGEB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB2 geni (Tüm Gen Dizi Analizi)	MAGEB2 gene (Sequence Analysis-All Coding Exons)	MAGEB2	MAGEB2, DAM6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB17 geni (Tüm Gen Dizi Analizi)	MAGEB17 gene (Sequence Analysis-All Coding Exons)	MAGEB17	MAGEB17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB16 geni (Tüm Gen Dizi Analizi)	MAGEB16 gene (Sequence Analysis-All Coding Exons)	MAGEB16	MAGEB16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB10 geni (Tüm Gen Dizi Analizi)	MAGEB10 gene (Sequence Analysis-All Coding Exons)	MAGEB10	MAGEB10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEB1 geni (Tüm Gen Dizi Analizi)	MAGEB1 gene (Sequence Analysis-All Coding Exons)	MAGEB1	MAGEB1, MAGEL1, DAM10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA9B geni (Tüm Gen Dizi Analizi)	MAGEA9B gene (Sequence Analysis-All Coding Exons)	MAGEA9B	MAGEA9B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA9 geni (Tüm Gen Dizi Analizi)	MAGEA9 gene (Sequence Analysis-All Coding Exons)	MAGEA9	MAGEA9, MAGE9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA8 geni (Tüm Gen Dizi Analizi)	MAGEA8 gene (Sequence Analysis-All Coding Exons)	MAGEA8	MAGEA8, MAGE8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA6 geni (Tüm Gen Dizi Analizi)	MAGEA6 gene (Sequence Analysis-All Coding Exons)	MAGEA6	MAGEA6, MAGE6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA4 geni (Tüm Gen Dizi Analizi)	MAGEA4 gene (Sequence Analysis-All Coding Exons)	MAGEA4	MAGEA4, MAGE4A, MAGE4B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA3 geni (Tüm Gen Dizi Analizi)	MAGEA3 gene (Sequence Analysis-All Coding Exons)	MAGEA3	MAGEA3, MAGE3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA2B geni (Tüm Gen Dizi Analizi)	MAGEA2B gene (Sequence Analysis-All Coding Exons)	MAGEA2B	MAGEA2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA2 geni (Tüm Gen Dizi Analizi)	MAGEA2 gene (Sequence Analysis-All Coding Exons)	MAGEA2	MAGEA2, MAGE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA12 geni (Tüm Gen Dizi Analizi)	MAGEA12 gene (Sequence Analysis-All Coding Exons)	MAGEA12	MAGEA12, MAGE12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PITPNM2 geni (Tüm Gen Dizi Analizi)	PITPNM2 gene (Sequence Analysis-All Coding Exons)	PITPNM2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PITPNM1 geni (Tüm Gen Dizi Analizi)	PITPNM1 gene (Sequence Analysis-All Coding Exons)	PITPNM1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA11 geni (Tüm Gen Dizi Analizi)	MAGEA11 gene (Sequence Analysis-All Coding Exons)	MAGEA11	MAGEA11, MAGE11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA10 geni (Tüm Gen Dizi Analizi)	MAGEA10 gene (Sequence Analysis-All Coding Exons)	MAGEA10	MAGEA10, MAGE10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAGEA1 geni (Tüm Gen Dizi Analizi)	MAGEA1 gene (Sequence Analysis-All Coding Exons)	MAGEA1	MAGE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAFTRR geni (Tüm Gen Dizi Analizi)	MAFTRR gene (Sequence Analysis-All Coding Exons)	MAFTRR	MAFTRR, lincMAF4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAFK geni (Tüm Gen Dizi Analizi)	MAFK gene (Sequence Analysis-All Coding Exons)	MAFK	MAFK, NFE2U	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAFG geni (Tüm Gen Dizi Analizi)	MAFG gene (Sequence Analysis-All Coding Exons)	MAFG	MAFG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIPOX geni (Tüm Gen Dizi Analizi)	PIPOX gene (Sequence Analysis-All Coding Exons)	PIPOX		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAFF geni (Tüm Gen Dizi Analizi)	MAFF gene (Sequence Analysis-All Coding Exons)	MAFF	MAFF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAFA geni (Tüm Gen Dizi Analizi)	MAFA gene (Sequence Analysis-All Coding Exons)	MAFA	MAFA, RIPE3B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAF1 geni (Tüm Gen Dizi Analizi)	MAF1 gene (Sequence Analysis-All Coding Exons)	MAF1	MAF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAEL geni (Tüm Gen Dizi Analizi)	MAEL gene (Sequence Analysis-All Coding Exons)	MAEL	MAEL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MAEA geni (Tüm Gen Dizi Analizi)	MAEA gene (Sequence Analysis-All Coding Exons)	MAEA	MAEA, EMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MADD geni (Tüm Gen Dizi Analizi)	MADD gene (Sequence Analysis-All Coding Exons)	MADD	MADD, DENN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MADCAM1 geni (Tüm Gen Dizi Analizi)	MADCAM1 gene (Sequence Analysis-All Coding Exons)	MADCAM1	MACAM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAD2L1 geni (Tüm Gen Dizi Analizi)	MAD2L1 gene (Sequence Analysis-All Coding Exons)	MAD2L1	MAD2L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MACROD2 geni (Tüm Gen Dizi Analizi)	MACROD2 gene (Sequence Analysis-All Coding Exons)	MACROD2	MACROD2, C2orf133	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MACROD1 geni (Tüm Gen Dizi Analizi)	MACROD1 gene (Sequence Analysis-All Coding Exons)	MACROD1	MACROD1, LRP16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MACF1 geni (Tüm Gen Dizi Analizi)	MACF1 gene (Sequence Analysis-All Coding Exons)	MACF1	MACF1, ACF7, KIAA1251	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MACC1 geni (Tüm Gen Dizi Analizi)	MACC1 gene (Sequence Analysis-All Coding Exons)	MACC1	MACC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAB21L1 geni (Tüm Gen Dizi Analizi)	MAB21L1 gene (Sequence Analysis-All Coding Exons)	MAB21L1	MAB21L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAATS1 geni (Tüm Gen Dizi Analizi)	MAATS1 gene (Sequence Analysis-All Coding Exons)	MAATS1	MAATS1, AAT1, C3orf15	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
M6PR geni (Tüm Gen Dizi Analizi)	M6PR gene (Sequence Analysis-All Coding Exons)	M6PR	M6PR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LZTS3 geni (Tüm Gen Dizi Analizi)	LZTS3 gene (Sequence Analysis-All Coding Exons)	LZTS3	PROSAPIP1, KIAA0552	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LZTS2 geni (Tüm Gen Dizi Analizi)	LZTS2 gene (Sequence Analysis-All Coding Exons)	LZTS2	LZTS2, KIAA1813, LAPSER1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LZIC geni (Tüm Gen Dizi Analizi)	LZIC gene (Sequence Analysis-All Coding Exons)	LZIC	LZIC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYZL6 geni (Tüm Gen Dizi Analizi)	LYZL6 gene (Sequence Analysis-All Coding Exons)	LYZL6	LYZL6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3R6 geni (Tüm Gen Dizi Analizi)	PIK3R6 gene (Sequence Analysis-All Coding Exons)	PIK3R6		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYZL4 geni (Tüm Gen Dizi Analizi)	LYZL4 gene (Sequence Analysis-All Coding Exons)	LYZL4	LYZL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3R4 geni (Tüm Gen Dizi Analizi)	PIK3R4 gene (Sequence Analysis-All Coding Exons)	PIK3R4		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYZL2 geni (Tüm Gen Dizi Analizi)	LYZL2 gene (Sequence Analysis-All Coding Exons)	LYZL2	LYZL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYVE1 geni (Tüm Gen Dizi Analizi)	LYVE1 gene (Sequence Analysis-All Coding Exons)	LYVE1	LYVE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYRM1 geni (Tüm Gen Dizi Analizi)	LYRM1 gene (Sequence Analysis-All Coding Exons)	LYRM1	LYRM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPLAL1 geni (Tüm Gen Dizi Analizi)	LYPLAL1 gene (Sequence Analysis-All Coding Exons)	LYPLAL1	LYPLAL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPLA2 geni (Tüm Gen Dizi Analizi)	LYPLA2 gene (Sequence Analysis-All Coding Exons)	LYPLA2	LYPLA2, APT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPLA1 geni (Tüm Gen Dizi Analizi)	LYPLA1 gene (Sequence Analysis-All Coding Exons)	LYPLA1	LYPLA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPD8 geni (Tüm Gen Dizi Analizi)	LYPD8 gene (Sequence Analysis-All Coding Exons)	LYPD8	LYPD8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPD6 geni (Tüm Gen Dizi Analizi)	LYPD6 gene (Sequence Analysis-All Coding Exons)	LYPD6	LYPD6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPD3 geni (Tüm Gen Dizi Analizi)	LYPD3 gene (Sequence Analysis-All Coding Exons)	LYPD3	LYPD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYPD1 geni (Tüm Gen Dizi Analizi)	LYPD1 gene (Sequence Analysis-All Coding Exons)	LYPD1	LYPD1, PHTS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYNX1 geni (Tüm Gen Dizi Analizi)	LYNX1 gene (Sequence Analysis-All Coding Exons)	LYNX1	LYNX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYN geni (Tüm Gen Dizi Analizi)	LYN gene (Sequence Analysis-All Coding Exons)	LYN	LYN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LYG2 geni (Tüm Gen Dizi Analizi)	LYG2 gene (Sequence Analysis-All Coding Exons)	LYG2	LYG2, LYSG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY96 geni (Tüm Gen Dizi Analizi)	LY96 gene (Sequence Analysis-All Coding Exons)	LY96	LY96, MD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY9 geni (Tüm Gen Dizi Analizi)	LY9 gene (Sequence Analysis-All Coding Exons)	LY9	LY9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY86 geni (Tüm Gen Dizi Analizi)	LY86 gene (Sequence Analysis-All Coding Exons)	LY86	LY86, MD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6K geni (Tüm Gen Dizi Analizi)	LY6K gene (Sequence Analysis-All Coding Exons)	LY6K	LY6K	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIK3C3 geni (Tüm Gen Dizi Analizi)	PIK3C3 gene (Sequence Analysis-All Coding Exons)	PIK3C3		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6H geni (Tüm Gen Dizi Analizi)	LY6H gene (Sequence Analysis-All Coding Exons)	LY6H	LY6H	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G6D geni (Tüm Gen Dizi Analizi)	LY6G6D gene (Sequence Analysis-All Coding Exons)	LY6G6D	LY6G6D, MEGT1, G6D, C6orf23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G6C geni (Tüm Gen Dizi Analizi)	LY6G6C gene (Sequence Analysis-All Coding Exons)	LY6G6C	LY6G6C, G6C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G5C geni (Tüm Gen Dizi Analizi)	LY6G5C gene (Sequence Analysis-All Coding Exons)	LY6G5C	LY6G5C, G5C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G5B geni (Tüm Gen Dizi Analizi)	LY6G5B gene (Sequence Analysis-All Coding Exons)	LY6G5B	LY6G5B, G5B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6E geni (Tüm Gen Dizi Analizi)	LY6E gene (Sequence Analysis-All Coding Exons)	LY6E	LY6E, RIGE, SCA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6D geni (Tüm Gen Dizi Analizi)	LY6D gene (Sequence Analysis-All Coding Exons)	LY6D	LY6D, E48	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LXN geni (Tüm Gen Dizi Analizi)	LXN gene (Sequence Analysis-All Coding Exons)	LXN	LXN, ECI, TCI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LVRN geni (Tüm Gen Dizi Analizi)	LVRN gene (Sequence Analysis-All Coding Exons)	LVRN	LVRN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUZP6 geni (Tüm Gen Dizi Analizi)	LUZP6 gene (Sequence Analysis-All Coding Exons)	LUZP6	LUZP6, MPD6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUZP4 geni (Tüm Gen Dizi Analizi)	LUZP4 gene (Sequence Analysis-All Coding Exons)	LUZP4	LUZP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUZP2 geni (Tüm Gen Dizi Analizi)	LUZP2 gene (Sequence Analysis-All Coding Exons)	LUZP2	LUZP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUZP1 geni (Tüm Gen Dizi Analizi)	LUZP1 gene (Sequence Analysis-All Coding Exons)	LUZP1	LUZP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LURAP1L geni (Tüm Gen Dizi Analizi)	LURAP1L gene (Sequence Analysis-All Coding Exons)	LURAP1L	LURAP1L, LRAP35B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LURAP1 geni (Tüm Gen Dizi Analizi)	LURAP1 gene (Sequence Analysis-All Coding Exons)	LURAP1	LURAP1, LRAP35A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUM geni (Tüm Gen Dizi Analizi)	LUM gene (Sequence Analysis-All Coding Exons)	LUM	LUM, LDC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUC7L3 geni (Tüm Gen Dizi Analizi)	LUC7L3 gene (Sequence Analysis-All Coding Exons)	LUC7L3	LUC7L3, CROP, LUC7A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUC7L2 geni (Tüm Gen Dizi Analizi)	LUC7L2 gene (Sequence Analysis-All Coding Exons)	LUC7L2	LUC7L2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LUC7L geni (Tüm Gen Dizi Analizi)	LUC7L gene (Sequence Analysis-All Coding Exons)	LUC7L	LUC7L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTN1 geni (Tüm Gen Dizi Analizi)	LTN1 gene (Sequence Analysis-All Coding Exons)	LTN1	LTN1, RNF160, KIAA0714	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTK geni (Tüm Gen Dizi Analizi)	LTK gene (Sequence Analysis-All Coding Exons)	LTK	LTK, TYK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTF geni (Tüm Gen Dizi Analizi)	LTF gene (Sequence Analysis-All Coding Exons)	LTF	LTF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTBR geni (Tüm Gen Dizi Analizi)	LTBR gene (Sequence Analysis-All Coding Exons)	LTBR	LTBR, TNFCR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTBP1 geni (Tüm Gen Dizi Analizi)	LTBP1 gene (Sequence Analysis-All Coding Exons)	LTBP1	LTBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTB4R2 geni (Tüm Gen Dizi Analizi)	LTB4R2 gene (Sequence Analysis-All Coding Exons)	LTB4R2	LTB4R2, BLTR2, BLT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTB4R geni (Tüm Gen Dizi Analizi)	LTB4R gene (Sequence Analysis-All Coding Exons)	LTB4R	LTB4R, CMKRL1, P2RY7, BLTR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTB geni (Tüm Gen Dizi Analizi)	LTB gene (Sequence Analysis-All Coding Exons)	LTB	LTB, TNFC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LTA4H geni (Tüm Gen Dizi Analizi)	LTA4H gene (Sequence Analysis-All Coding Exons)	LTA4H	LTA4H	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LST1 geni (Tüm Gen Dizi Analizi)	LST1 gene (Sequence Analysis-All Coding Exons)	LST1	LST1, D6S49E, B144	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSR geni (Tüm Gen Dizi Analizi)	LSR gene (Sequence Analysis-All Coding Exons)	LSR	LSR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSP1 geni (Tüm Gen Dizi Analizi)	LSP1 gene (Sequence Analysis-All Coding Exons)	LSP1	LSP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM8 geni (Tüm Gen Dizi Analizi)	LSM8 gene (Sequence Analysis-All Coding Exons)	LSM8	LSM8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM7 geni (Tüm Gen Dizi Analizi)	LSM7 gene (Sequence Analysis-All Coding Exons)	LSM7	LSM7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM6 geni (Tüm Gen Dizi Analizi)	LSM6 gene (Sequence Analysis-All Coding Exons)	LSM6	LSM6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM5 geni (Tüm Gen Dizi Analizi)	LSM5 gene (Sequence Analysis-All Coding Exons)	LSM5	LSM5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM4 geni (Tüm Gen Dizi Analizi)	LSM4 gene (Sequence Analysis-All Coding Exons)	LSM4	LSM4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM3 geni (Tüm Gen Dizi Analizi)	LSM3 gene (Sequence Analysis-All Coding Exons)	LSM3	LSM3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM2 geni (Tüm Gen Dizi Analizi)	LSM2 gene (Sequence Analysis-All Coding Exons)	LSM2	LSM2, C6orf28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM14A geni (Tüm Gen Dizi Analizi)	LSM14A gene (Sequence Analysis-All Coding Exons)	LSM14A	LSM14A, RAP55	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM12 geni (Tüm Gen Dizi Analizi)	LSM12 gene (Sequence Analysis-All Coding Exons)	LSM12	LSM12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSM1 geni (Tüm Gen Dizi Analizi)	LSM1 gene (Sequence Analysis-All Coding Exons)	LSM1	LSM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSG1 geni (Tüm Gen Dizi Analizi)	LSG1 gene (Sequence Analysis-All Coding Exons)	LSG1	LSG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LSAMP geni (Tüm Gen Dizi Analizi)	LSAMP gene (Sequence Analysis-All Coding Exons)	LSAMP	LSAMP, LAMP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRWD1 geni (Tüm Gen Dizi Analizi)	LRWD1 gene (Sequence Analysis-All Coding Exons)	LRWD1	LRWD1, ORCA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRTM4 geni (Tüm Gen Dizi Analizi)	LRRTM4 gene (Sequence Analysis-All Coding Exons)	LRRTM4	LRRTM4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRTM3 geni (Tüm Gen Dizi Analizi)	LRRTM3 gene (Sequence Analysis-All Coding Exons)	LRRTM3	LRRTM3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PIAS2 geni (Tüm Gen Dizi Analizi)	PIAS2 gene (Sequence Analysis-All Coding Exons)	PIAS2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRTM2 geni (Tüm Gen Dizi Analizi)	LRRTM2 gene (Sequence Analysis-All Coding Exons)	LRRTM2	LRRTM2, KIAA0416	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRTM1 geni (Tüm Gen Dizi Analizi)	LRRTM1 gene (Sequence Analysis-All Coding Exons)	LRRTM1	LRRTM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRN2 geni (Tüm Gen Dizi Analizi)	LRRN2 gene (Sequence Analysis-All Coding Exons)	LRRN2	GAC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRK1 geni (Tüm Gen Dizi Analizi)	LRRK1 gene (Sequence Analysis-All Coding Exons)	LRRK1	LRRK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRFIP2 geni (Tüm Gen Dizi Analizi)	LRRFIP2 gene (Sequence Analysis-All Coding Exons)	LRRFIP2	LRRFIP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRFIP1 geni (Tüm Gen Dizi Analizi)	LRRFIP1 gene (Sequence Analysis-All Coding Exons)	LRRFIP1	LRRFIP1, GCF2, TRIP, FLAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC8E geni (Tüm Gen Dizi Analizi)	LRRC8E gene (Sequence Analysis-All Coding Exons)	LRRC8E	LRRC8E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC8D geni (Tüm Gen Dizi Analizi)	LRRC8D gene (Sequence Analysis-All Coding Exons)	LRRC8D	LRRC8D, LRRC5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC8C geni (Tüm Gen Dizi Analizi)	LRRC8C gene (Sequence Analysis-All Coding Exons)	LRRC8C	LRRC8C, FAD1S8, AD158	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHYHIP geni (Tüm Gen Dizi Analizi)	PHYHIP gene (Sequence Analysis-All Coding Exons)	PHYHIP		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

LRRC8B geni (Tüm Gen Dizi Analizi)	LRRC8B gene (Sequence Analysis-All Coding Exons)	LRRC8B	LRRC8B, TALRRP, KIAA0231	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC7 geni (Tüm Gen Dizi Analizi)	LRRC7 gene (Sequence Analysis-All Coding Exons)	LRRC7	LRRC7, DENSIN, KIAA1365	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC59 geni (Tüm Gen Dizi Analizi)	LRRC59 gene (Sequence Analysis-All Coding Exons)	LRRC59	LRRC59, p34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC55 geni (Tüm Gen Dizi Analizi)	LRRC55 gene (Sequence Analysis-All Coding Exons)	LRRC55	LRRC55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC52 geni (Tüm Gen Dizi Analizi)	LRRC52 gene (Sequence Analysis-All Coding Exons)	LRRC52	LRRC52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC4C geni (Tüm Gen Dizi Analizi)	LRRC4C gene (Sequence Analysis-All Coding Exons)	LRRC4C	LRRC4C, NGL1, KIAA1580	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC4 geni (Tüm Gen Dizi Analizi)	LRRC4 gene (Sequence Analysis-All Coding Exons)	LRRC4	LRRC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC38 geni (Tüm Gen Dizi Analizi)	LRRC38 gene (Sequence Analysis-All Coding Exons)	LRRC38	LRRC38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC37B geni (Tüm Gen Dizi Analizi)	LRRC37B gene (Sequence Analysis-All Coding Exons)	LRRC37B	LRRC37B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC37A3 geni (Tüm Gen Dizi Analizi)	LRRC37A3 gene (Sequence Analysis-All Coding Exons)	LRRC37A3	LRRC37A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC37A2 geni (Tüm Gen Dizi Analizi)	LRRC37A2 gene (Sequence Analysis-All Coding Exons)	LRRC37A2	LRRC37A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC37A geni (Tüm Gen Dizi Analizi)	LRRC37A gene (Sequence Analysis-All Coding Exons)	LRRC37A	LRRC37A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC32 geni (Tüm Gen Dizi Analizi)	LRRC32 gene (Sequence Analysis-All Coding Exons)	LRRC32	LRRC32, GARP, D11S833E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC26 geni (Tüm Gen Dizi Analizi)	LRRC26 gene (Sequence Analysis-All Coding Exons)	LRRC26	LRRC26, CAPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC25 geni (Tüm Gen Dizi Analizi)	LRRC25 gene (Sequence Analysis-All Coding Exons)	LRRC25	LRRC25, MAPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC2 geni (Tüm Gen Dizi Analizi)	LRRC2 gene (Sequence Analysis-All Coding Exons)	LRRC2	LRRC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC10 geni (Tüm Gen Dizi Analizi)	LRRC10 gene (Sequence Analysis-All Coding Exons)	LRRC10	LRRC10, HRLRRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRRC1 geni (Tüm Gen Dizi Analizi)	LRRC1 gene (Sequence Analysis-All Coding Exons)	LRRC1	LRRC1, LANO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRP3 geni (Tüm Gen Dizi Analizi)	LRP3 gene (Sequence Analysis-All Coding Exons)	LRP3	LRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRP10 geni (Tüm Gen Dizi Analizi)	LRP10 gene (Sequence Analysis-All Coding Exons)	LRP10	LRP10, LRP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRMP geni (Tüm Gen Dizi Analizi)	LRMP gene (Sequence Analysis-All Coding Exons)	LRMP	LRMP, JAW1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRIT1 geni (Tüm Gen Dizi Analizi)	LRIT1 gene (Sequence Analysis-All Coding Exons)	LRIT1	LRIT1, PAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRIG3 geni (Tüm Gen Dizi Analizi)	LRIG3 gene (Sequence Analysis-All Coding Exons)	LRIG3	LIG3, LIG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRIG1 geni (Tüm Gen Dizi Analizi)	LRIG1 gene (Sequence Analysis-All Coding Exons)	LRIG1	LRIG1, LIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRIF1 geni (Tüm Gen Dizi Analizi)	LRIF1 gene (Sequence Analysis-All Coding Exons)	LRIF1	LRIF1, RIF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRGUK geni (Tüm Gen Dizi Analizi)	LRGUK gene (Sequence Analysis-All Coding Exons)	LRGUK	LRGUK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRG1 geni (Tüm Gen Dizi Analizi)	LRG1 gene (Sequence Analysis-All Coding Exons)	LRG1	LRG1, LRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRFN5 geni (Tüm Gen Dizi Analizi)	LRFN5 gene (Sequence Analysis-All Coding Exons)	LRFN5	LRFN5, SALM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRFN4 geni (Tüm Gen Dizi Analizi)	LRFN4 gene (Sequence Analysis-All Coding Exons)	LRFN4	LRFN4, SALM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRFN3 geni (Tüm Gen Dizi Analizi)	LRFN3 gene (Sequence Analysis-All Coding Exons)	LRFN3	LRFN3, SALM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRFN2 geni (Tüm Gen Dizi Analizi)	LRFN2 gene (Sequence Analysis-All Coding Exons)	LRFN2	LRFN2, SALM1, KIAA1246	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRFN1 geni (Tüm Gen Dizi Analizi)	LRFN1 gene (Sequence Analysis-All Coding Exons)	LRFN1	LRFN1, SALM2, KIAA1484	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRCH1 geni (Tüm Gen Dizi Analizi)	LRCH1 gene (Sequence Analysis-All Coding Exons)	LRCH1	LRCH1, KIAA1016	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPXN geni (Tüm Gen Dizi Analizi)	LPXN gene (Sequence Analysis-All Coding Exons)	LPXN	LPXN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPO geni (Tüm Gen Dizi Analizi)	LPO gene (Sequence Analysis-All Coding Exons)	LPO	LPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPIN3 geni (Tüm Gen Dizi Analizi)	LPIN3 gene (Sequence Analysis-All Coding Exons)	LPIN3	LPIN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPCAT4 geni (Tüm Gen Dizi Analizi)	LPCAT4 gene (Sequence Analysis-All Coding Exons)	LPCAT4	AGPAT7, AYTL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPCAT3 geni (Tüm Gen Dizi Analizi)	LPCAT3 gene (Sequence Analysis-All Coding Exons)	LPCAT3	LPCAT3, MBOAT5, NESSY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPCAT2 geni (Tüm Gen Dizi Analizi)	LPCAT2 gene (Sequence Analysis-All Coding Exons)	LPCAT2	LPCAT1, AYTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPCAT1 geni (Tüm Gen Dizi Analizi)	LPCAT1 gene (Sequence Analysis-All Coding Exons)	LPCAT1	AYTL2, LPCAT, LPCAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PHC2 geni (Tüm Gen Dizi Analizi)	PHC2 gene (Sequence Analysis-All Coding Exons)	PHC2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAR5 geni (Tüm Gen Dizi Analizi)	LPAR5 gene (Sequence Analysis-All Coding Exons)	LPAR5	LPAR5, GPR92, GPR93, LPA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAR4 geni (Tüm Gen Dizi Analizi)	LPAR4 gene (Sequence Analysis-All Coding Exons)	LPAR4	LPAR4, LPA4, P2RY9, P2Y9, GPR23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAR3 geni (Tüm Gen Dizi Analizi)	LPAR3 gene (Sequence Analysis-All Coding Exons)	LPAR3	LPAR3, EDG7, LPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAR2 geni (Tüm Gen Dizi Analizi)	LPAR2 gene (Sequence Analysis-All Coding Exons)	LPAR2	LPAR2, EDG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAR1 geni (Tüm Gen Dizi Analizi)	LPAR1 gene (Sequence Analysis-All Coding Exons)	LPAR1	LPAR1, EDG2, LPA1, VZG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPAL2 geni (Tüm Gen Dizi Analizi)	LPAL2 gene (Sequence Analysis-All Coding Exons)	LPAL2	LPAL2, APOARGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LOXL4 geni (Tüm Gen Dizi Analizi)	LOXL4 gene (Sequence Analysis-All Coding Exons)	LOXL4	LOXL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LOXL3 geni (Tüm Gen Dizi Analizi)	LOXL3 gene (Sequence Analysis-All Coding Exons)	LOXL3	LOXL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LOXL2 geni (Tüm Gen Dizi Analizi)	LOXL2 gene (Sequence Analysis-All Coding Exons)	LOXL2	LOXL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LOXL1-AS1 geni (Tüm Gen Dizi Analizi)	LOXL1-AS1 gene (Sequence Analysis-All Coding Exons)	LOXL1-AS1	LOXL1AS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LNX2 geni (Tüm Gen Dizi Analizi)	LNX2 gene (Sequence Analysis-All Coding Exons)	LNX2	LNX2, PDZRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LNX1 geni (Tüm Gen Dizi Analizi)	LNX1 gene (Sequence Analysis-All Coding Exons)	LNX1	LNX1, LNX, PDZRN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LNPK geni (Tüm Gen Dizi Analizi)	LNPK gene (Sequence Analysis-All Coding Exons)	LNPK	LNP, KIAA1715	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LNPEP geni (Tüm Gen Dizi Analizi)	LNPEP gene (Sequence Analysis-All Coding Exons)	LNPEP	LNPEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMX1A geni (Tüm Gen Dizi Analizi)	LMX1A gene (Sequence Analysis-All Coding Exons)	LMX1A	LMX1A, LMX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMTK2 geni (Tüm Gen Dizi Analizi)	LMTK2 gene (Sequence Analysis-All Coding Exons)	LMTK2	LMTK2, KPI2, BREK, KIAA1079	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMOD2 geni (Tüm Gen Dizi Analizi)	LMOD2 gene (Sequence Analysis-All Coding Exons)	LMOD2	LMOD2, CLMOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMOD1 geni (Tüm Gen Dizi Analizi)	LMOD1 gene (Sequence Analysis-All Coding Exons)	LMOD1	LMOD1, SMLMOD, 1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMO7 geni (Tüm Gen Dizi Analizi)	LMO7 gene (Sequence Analysis-All Coding Exons)	LMO7	LMO7, FBXO20, FBX20, KIAA0858	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMO4 geni (Tüm Gen Dizi Analizi)	LMO4 gene (Sequence Analysis-All Coding Exons)	LMO4	LMO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMO3 geni (Tüm Gen Dizi Analizi)	LMO3 gene (Sequence Analysis-All Coding Exons)	LMO3	LMO3, RBTNL2, RHOM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMNTD1 geni (Tüm Gen Dizi Analizi)	LMNTD1 gene (Sequence Analysis-All Coding Exons)	LMNTD1	LMNTD1, IFLTD1, LMNARS1, PAS1C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMLN geni (Tüm Gen Dizi Analizi)	LMLN gene (Sequence Analysis-All Coding Exons)	LMLN	LMLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMCD1 geni (Tüm Gen Dizi Analizi)	LMCD1 gene (Sequence Analysis-All Coding Exons)	LMCD1	LMCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMBR1L geni (Tüm Gen Dizi Analizi)	LMBR1L gene (Sequence Analysis-All Coding Exons)	LMBR1L	LMBR1L, LIMR, KIAA1174	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LLPH geni (Tüm Gen Dizi Analizi)	LLPH gene (Sequence Analysis-All Coding Exons)	LLPH	LLPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LLGL1 geni (Tüm Gen Dizi Analizi)	LLGL1 gene (Sequence Analysis-All Coding Exons)	LLGL1	LLGL1, DLG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIX1 geni (Tüm Gen Dizi Analizi)	LIX1 gene (Sequence Analysis-All Coding Exons)	LIX1	LIX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIPM geni (Tüm Gen Dizi Analizi)	LIPM gene (Sequence Analysis-All Coding Exons)	LIPM	LIPM, LIPL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIPK geni (Tüm Gen Dizi Analizi)	LIPK gene (Sequence Analysis-All Coding Exons)	LIPK	LIPK, LIPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIPJ geni (Tüm Gen Dizi Analizi)	LIPJ gene (Sequence Analysis-All Coding Exons)	LIPJ	LIPJ, LIPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIPG geni (Tüm Gen Dizi Analizi)	LIPG gene (Sequence Analysis-All Coding Exons)	LIPG	LIPG, EL, EDL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINGO4 geni (Tüm Gen Dizi Analizi)	LINGO4 gene (Sequence Analysis-All Coding Exons)	LINGO4	LINGO4, LRRN6D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINGO3 geni (Tüm Gen Dizi Analizi)	LINGO3 gene (Sequence Analysis-All Coding Exons)	LINGO3	LRRN6B, LINGO3, LERN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINGO2 geni (Tüm Gen Dizi Analizi)	LINGO2 gene (Sequence Analysis-All Coding Exons)	LINGO2	LRRN6C, LINGO2, LERN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINGO1 geni (Tüm Gen Dizi Analizi)	LINGO1 gene (Sequence Analysis-All Coding Exons)	LINGO1	LRRN6A, LERN1, LINGO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC01370 geni (Tüm Gen Dizi Analizi)	LINC01370 gene (Sequence Analysis-All Coding Exons)	LINC01370	LINC01370, HLINC25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC01082 geni (Tüm Gen Dizi Analizi)	LINC01082 gene (Sequence Analysis-All Coding Exons)	LINC01082	TCONS00024492	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC01018 geni (Tüm Gen Dizi Analizi)	LINC01018 gene (Sequence Analysis-All Coding Exons)	LINC01018	LINC01018, SRHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00850 geni (Tüm Gen Dizi Analizi)	LINC00850 gene (Sequence Analysis-All Coding Exons)	LINC00850	LINC00850, KUCG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00538 geni (Tüm Gen Dizi Analizi)	LINC00538 gene (Sequence Analysis-All Coding Exons)	LINC00538	LINC00538, YIYA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00461 geni (Tüm Gen Dizi Analizi)	LINC00461 gene (Sequence Analysis-All Coding Exons)	LINC00461	LINC00461, VISC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00294 geni (Tüm Gen Dizi Analizi)	LINC00294 gene (Sequence Analysis-All Coding Exons)	LINC00294	LOC283267	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00293 geni (Tüm Gen Dizi Analizi)	LINC00293 gene (Sequence Analysis-All Coding Exons)	LINC00293	BEYLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00163 geni (Tüm Gen Dizi Analizi)	LINC00163 gene (Sequence Analysis-All Coding Exons)	LINC00163	LINC00163, NCRNA00163, NLC1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC00114 geni (Tüm Gen Dizi Analizi)	LINC00114 gene (Sequence Analysis-All Coding Exons)	LINC00114	C21orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LINC-ROR geni (Tüm Gen Dizi Analizi)	LINC-ROR gene (Sequence Analysis-All Coding Exons)	LINC-ROR	LINC-ROR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN9 geni (Tüm Gen Dizi Analizi)	LIN9 gene (Sequence Analysis-All Coding Exons)	LIN9	LIN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN7C geni (Tüm Gen Dizi Analizi)	LIN7C gene (Sequence Analysis-All Coding Exons)	LIN7C	LIN7C, VELI3, MALS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN7B geni (Tüm Gen Dizi Analizi)	LIN7B gene (Sequence Analysis-All Coding Exons)	LIN7B	LIN7B, VELI2, MALS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN54 geni (Tüm Gen Dizi Analizi)	LIN54 gene (Sequence Analysis-All Coding Exons)	LIN54	LIN54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN28B geni (Tüm Gen Dizi Analizi)	LIN28B gene (Sequence Analysis-All Coding Exons)	LIN28B	LIN28B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

LIN28A geni (Tüm Gen Dizi Analizi)	LIN28A gene (Sequence Analysis-All Coding Exons)	LIN28A	LIN28, LIN28A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PFDN6 geni (Tüm Gen Dizi Analizi)	PFDN6 gene (Sequence Analysis-All Coding Exons)	PFDN6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIMK2 geni (Tüm Gen Dizi Analizi)	LIMK2 gene (Sequence Analysis-All Coding Exons)	LIMK2	LIMK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIMK1 geni (Tüm Gen Dizi Analizi)	LIMK1 gene (Sequence Analysis-All Coding Exons)	LIMK1	LIMK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIME1 geni (Tüm Gen Dizi Analizi)	LIME1 gene (Sequence Analysis-All Coding Exons)	LIME1	LIME1, LIME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIMD1 geni (Tüm Gen Dizi Analizi)	LIMD1 gene (Sequence Analysis-All Coding Exons)	LIMD1	LIMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIMA1 geni (Tüm Gen Dizi Analizi)	LIMA1 gene (Sequence Analysis-All Coding Exons)	LIMA1	LIMA1, EPLIN, SREBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRB5 geni (Tüm Gen Dizi Analizi)	LILRB5 gene (Sequence Analysis-All Coding Exons)	LILRB5	LILRB5, LIR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRB4 geni (Tüm Gen Dizi Analizi)	LILRB4 gene (Sequence Analysis-All Coding Exons)	LILRB4	LILRB4, LIR5, ILT3, HM18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRB3 geni (Tüm Gen Dizi Analizi)	LILRB3 gene (Sequence Analysis-All Coding Exons)	LILRB3	LILRB3, LIR3, ILT5, HL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRB2 geni (Tüm Gen Dizi Analizi)	LILRB2 gene (Sequence Analysis-All Coding Exons)	LILRB2	LILRB2, LIR2, ILT4, MIR10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRB1 geni (Tüm Gen Dizi Analizi)	LILRB1 gene (Sequence Analysis-All Coding Exons)	LILRB1	LILRB1, LIR1, ILT2, MIR7, CD85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRA5 geni (Tüm Gen Dizi Analizi)	LILRA5 gene (Sequence Analysis-All Coding Exons)	LILRA5	ILT11, CD85F, LIR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRA4 geni (Tüm Gen Dizi Analizi)	LILRA4 gene (Sequence Analysis-All Coding Exons)	LILRA4	LILRA4, ILT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRA2 geni (Tüm Gen Dizi Analizi)	LILRA2 gene (Sequence Analysis-All Coding Exons)	LILRA2	LILRA2, LIR7, ILT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRA1 geni (Tüm Gen Dizi Analizi)	LILRA1 gene (Sequence Analysis-All Coding Exons)	LILRA1	LILRA1, LIR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIG3 geni (Tüm Gen Dizi Analizi)	LIG3 gene (Sequence Analysis-All Coding Exons)	LIG3	LIG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIF geni (Tüm Gen Dizi Analizi)	LIF gene (Sequence Analysis-All Coding Exons)	LIF	LIF, HILDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX9 geni (Tüm Gen Dizi Analizi)	LHX9 gene (Sequence Analysis-All Coding Exons)	LHX9	LHX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX8 geni (Tüm Gen Dizi Analizi)	LHX8 gene (Sequence Analysis-All Coding Exons)	LHX8	LHX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX6 geni (Tüm Gen Dizi Analizi)	LHX6 gene (Sequence Analysis-All Coding Exons)	LHX6	LHX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX5 geni (Tüm Gen Dizi Analizi)	LHX5 gene (Sequence Analysis-All Coding Exons)	LHX5	LHX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX2 geni (Tüm Gen Dizi Analizi)	LHX2 gene (Sequence Analysis-All Coding Exons)	LHX2	LHX2, LH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHX1 geni (Tüm Gen Dizi Analizi)	LHX1 gene (Sequence Analysis-All Coding Exons)	LHX1	LHX1, LIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHPP geni (Tüm Gen Dizi Analizi)	LHPP gene (Sequence Analysis-All Coding Exons)	LHPP	LHPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHFPL4 geni (Tüm Gen Dizi Analizi)	LHFPL4 gene (Sequence Analysis-All Coding Exons)	LHFPL4	LHFPL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHFPL3 geni (Tüm Gen Dizi Analizi)	LHFPL3 gene (Sequence Analysis-All Coding Exons)	LHFPL3	LHFPL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHFPL2 geni (Tüm Gen Dizi Analizi)	LHFPL2 gene (Sequence Analysis-All Coding Exons)	LHFPL2	LHFPL2, KIAA0206	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHFPL1 geni (Tüm Gen Dizi Analizi)	LHFPL1 gene (Sequence Analysis-All Coding Exons)	LHFPL1	LHFPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LHFP geni (Tüm Gen Dizi Analizi)	LHFP gene (Sequence Analysis-All Coding Exons)	LHFP	LHFP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGR6 geni (Tüm Gen Dizi Analizi)	LGR6 gene (Sequence Analysis-All Coding Exons)	LGR6	LGR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGR5 geni (Tüm Gen Dizi Analizi)	LGR5 gene (Sequence Analysis-All Coding Exons)	LGR5	GPR49, LGR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGMN geni (Tüm Gen Dizi Analizi)	LGMN gene (Sequence Analysis-All Coding Exons)	LGMN	LGMN, PRSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGI4 geni (Tüm Gen Dizi Analizi)	LGI4 gene (Sequence Analysis-All Coding Exons)	LGI4	LGI4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGI3 geni (Tüm Gen Dizi Analizi)	LGI3 gene (Sequence Analysis-All Coding Exons)	LGI3	LGI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGI2 geni (Tüm Gen Dizi Analizi)	LGI2 gene (Sequence Analysis-All Coding Exons)	LGI2	LGI2, KIAA1916	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS9 geni (Tüm Gen Dizi Analizi)	LGALS9 gene (Sequence Analysis-All Coding Exons)	LGALS9	LGALS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS8 geni (Tüm Gen Dizi Analizi)	LGALS8 gene (Sequence Analysis-All Coding Exons)	LGALS8	LGALS8, PCTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS7B geni (Tüm Gen Dizi Analizi)	LGALS7B gene (Sequence Analysis-All Coding Exons)	LGALS7B	LGALS7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS7 geni (Tüm Gen Dizi Analizi)	LGALS7 gene (Sequence Analysis-All Coding Exons)	LGALS7	LGALS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS4 geni (Tüm Gen Dizi Analizi)	LGALS4 gene (Sequence Analysis-All Coding Exons)	LGALS4	LGALS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS3BP geni (Tüm Gen Dizi Analizi)	LGALS3BP gene (Sequence Analysis-All Coding Exons)	LGALS3BP	LGALS3BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS3 geni (Tüm Gen Dizi Analizi)	LGALS3 gene (Sequence Analysis-All Coding Exons)	LGALS3	LGALS3, MAC2, GALBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS14 geni (Tüm Gen Dizi Analizi)	LGALS14 gene (Sequence Analysis-All Coding Exons)	LGALS14	PPL13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS13 geni (Tüm Gen Dizi Analizi)	LGALS13 gene (Sequence Analysis-All Coding Exons)	LGALS13	LGALS13, GAL13, PP13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS12 geni (Tüm Gen Dizi Analizi)	LGALS12 gene (Sequence Analysis-All Coding Exons)	LGALS12	LGALS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGALS1 geni (Tüm Gen Dizi Analizi)	LGALS1 gene (Sequence Analysis-All Coding Exons)	LGALS1	LGALS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEXM geni (Tüm Gen Dizi Analizi)	LEXM gene (Sequence Analysis-All Coding Exons)	LEXM	C1orf177, LEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LETM1 geni (Tüm Gen Dizi Analizi)	LETM1 gene (Sequence Analysis-All Coding Exons)	LETM1	LETM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEPROTL1 geni (Tüm Gen Dizi Analizi)	LEPROTL1 gene (Sequence Analysis-All Coding Exons)	LEPROTL1	LEPROTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEPROT geni (Tüm Gen Dizi Analizi)	LEPROT gene (Sequence Analysis-All Coding Exons)	LEPROT	LEPROT, OBRGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEO1 geni (Tüm Gen Dizi Analizi)	LEO1 gene (Sequence Analysis-All Coding Exons)	LEO1	LEO1, RDL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LENG8 geni (Tüm Gen Dizi Analizi)	LENG8 gene (Sequence Analysis-All Coding Exons)	LENG8	LENG8, KIAA1932	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LENEP geni (Tüm Gen Dizi Analizi)	LENEP gene (Sequence Analysis-All Coding Exons)	LENEP	LENEP, LEPS03	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEMD1 geni (Tüm Gen Dizi Analizi)	LEMD1 gene (Sequence Analysis-All Coding Exons)	LEMD1	LEMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LELP1 geni (Tüm Gen Dizi Analizi)	LELP1 gene (Sequence Analysis-All Coding Exons)	LELP1	LELP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEKR1 geni (Tüm Gen Dizi Analizi)	LEKR1 gene (Sequence Analysis-All Coding Exons)	LEKR1	LEKR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEFTY1 geni (Tüm Gen Dizi Analizi)	LEFTY1 gene (Sequence Analysis-All Coding Exons)	LEFTY1	LEFTB, LEFTY1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LECT2 geni (Tüm Gen Dizi Analizi)	LECT2 gene (Sequence Analysis-All Coding Exons)	LECT2	LECT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LEAP2 geni (Tüm Gen Dizi Analizi)	LEAP2 gene (Sequence Analysis-All Coding Exons)	LEAP2	LEAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDOC1 geni (Tüm Gen Dizi Analizi)	LDOC1 gene (Sequence Analysis-All Coding Exons)	LDOC1	LDOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDLRAD4 geni (Tüm Gen Dizi Analizi)	LDLRAD4 gene (Sequence Analysis-All Coding Exons)	LDLRAD4	C18orf1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDHD geni (Tüm Gen Dizi Analizi)	LDHD gene (Sequence Analysis-All Coding Exons)	LDHD	LDHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDHC geni (Tüm Gen Dizi Analizi)	LDHC gene (Sequence Analysis-All Coding Exons)	LDHC	LDHC, LDH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDB2 geni (Tüm Gen Dizi Analizi)	LDB2 gene (Sequence Analysis-All Coding Exons)	LDB2	LDB2, CLIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDB1 geni (Tüm Gen Dizi Analizi)	LDB1 gene (Sequence Analysis-All Coding Exons)	LDB1	LDB1, CLIM2, NLI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCTL geni (Tüm Gen Dizi Analizi)	LCTL gene (Sequence Analysis-All Coding Exons)	LCTL	LCTL, KLG, KLPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCP2 geni (Tüm Gen Dizi Analizi)	LCP2 gene (Sequence Analysis-All Coding Exons)	LCP2	LCP2, SLP76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCP1 geni (Tüm Gen Dizi Analizi)	LCP1 gene (Sequence Analysis-All Coding Exons)	LCP1	LCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCORL geni (Tüm Gen Dizi Analizi)	LCORL gene (Sequence Analysis-All Coding Exons)	LCORL	LCORL, MLR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCOR geni (Tüm Gen Dizi Analizi)	LCOR gene (Sequence Analysis-All Coding Exons)	LCOR	LCOR, MLR2, KIAA1795	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN9 geni (Tüm Gen Dizi Analizi)	LCN9 gene (Sequence Analysis-All Coding Exons)	LCN9	LCN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN8 geni (Tüm Gen Dizi Analizi)	LCN8 gene (Sequence Analysis-All Coding Exons)	LCN8	LCN8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN6 geni (Tüm Gen Dizi Analizi)	LCN6 gene (Sequence Analysis-All Coding Exons)	LCN6	LCN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN2 geni (Tüm Gen Dizi Analizi)	LCN2 gene (Sequence Analysis-All Coding Exons)	LCN2	LCN2, NGAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN12 geni (Tüm Gen Dizi Analizi)	LCN12 gene (Sequence Analysis-All Coding Exons)	LCN12	LCN12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN10 geni (Tüm Gen Dizi Analizi)	LCN10 gene (Sequence Analysis-All Coding Exons)	LCN10	LCN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCN1 geni (Tüm Gen Dizi Analizi)	LCN1 gene (Sequence Analysis-All Coding Exons)	LCN1	LCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PDXP geni (Tüm Gen Dizi Analizi)	PDXP gene (Sequence Analysis-All Coding Exons)	PDXP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCMT2 geni (Tüm Gen Dizi Analizi)	LCMT2 gene (Sequence Analysis-All Coding Exons)	LCMT2	LCMT2, TYW4, KIAA0547	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCMT1 geni (Tüm Gen Dizi Analizi)	LCMT1 gene (Sequence Analysis-All Coding Exons)	LCMT1	LCMT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCLAT1 geni (Tüm Gen Dizi Analizi)	LCLAT1 gene (Sequence Analysis-All Coding Exons)	LCLAT1	LCLAT1, ALCAT1, AGPAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE5A geni (Tüm Gen Dizi Analizi)	LCE5A gene (Sequence Analysis-All Coding Exons)	LCE5A	LCE5A, LEP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE4A geni (Tüm Gen Dizi Analizi)	LCE4A gene (Sequence Analysis-All Coding Exons)	LCE4A	LCE4A, LEP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE3E geni (Tüm Gen Dizi Analizi)	LCE3E gene (Sequence Analysis-All Coding Exons)	LCE3E	LCE3E, LEP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE3D geni (Tüm Gen Dizi Analizi)	LCE3D gene (Sequence Analysis-All Coding Exons)	LCE3D	LCE3D, LEP16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE3C geni (Tüm Gen Dizi Analizi)	LCE3C gene (Sequence Analysis-All Coding Exons)	LCE3C	LCE3C, LEP15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE3B geni (Tüm Gen Dizi Analizi)	LCE3B gene (Sequence Analysis-All Coding Exons)	LCE3B	LCE3B, LEP14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE3A geni (Tüm Gen Dizi Analizi)	LCE3A gene (Sequence Analysis-All Coding Exons)	LCE3A	LCE3A, LEP13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE2D geni (Tüm Gen Dizi Analizi)	LCE2D gene (Sequence Analysis-All Coding Exons)	LCE2D	LCE2D, LEP12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE2C geni (Tüm Gen Dizi Analizi)	LCE2C gene (Sequence Analysis-All Coding Exons)	LCE2C	LCE2C, LEP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE2B geni (Tüm Gen Dizi Analizi)	LCE2B gene (Sequence Analysis-All Coding Exons)	LCE2B	LCE2B, LEP10, XP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE2A geni (Tüm Gen Dizi Analizi)	LCE2A gene (Sequence Analysis-All Coding Exons)	LCE2A	LCE2A, LEP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

LCE1F geni (Tüm Gen Dizi Analizi)	LCE1F gene (Sequence Analysis-All Coding Exons)	LCE1F	LCE1F, LEP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE1E geni (Tüm Gen Dizi Analizi)	LCE1E gene (Sequence Analysis-All Coding Exons)	LCE1E	LCE1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE1D geni (Tüm Gen Dizi Analizi)	LCE1D gene (Sequence Analysis-All Coding Exons)	LCE1D	LCE1D, LEP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE1C geni (Tüm Gen Dizi Analizi)	LCE1C gene (Sequence Analysis-All Coding Exons)	LCE1C	LCE1C, LEP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE1B geni (Tüm Gen Dizi Analizi)	LCE1B gene (Sequence Analysis-All Coding Exons)	LCE1B	LCE1B, LEP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LCE1A geni (Tüm Gen Dizi Analizi)	LCE1A gene (Sequence Analysis-All Coding Exons)	LCE1A	LCE1A, LEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LBX2 geni (Tüm Gen Dizi Analizi)	LBX2 gene (Sequence Analysis-All Coding Exons)	LBX2	LBX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LBX1 geni (Tüm Gen Dizi Analizi)	LBX1 gene (Sequence Analysis-All Coding Exons)	LBX1	LBX1, LBX1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LBP geni (Tüm Gen Dizi Analizi)	LBP gene (Sequence Analysis-All Coding Exons)	LBP	LBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LBH geni (Tüm Gen Dizi Analizi)	LBH gene (Sequence Analysis-All Coding Exons)	LBH	LBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LATS2 geni (Tüm Gen Dizi Analizi)	LATS2 gene (Sequence Analysis-All Coding Exons)	LATS2	LATS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LATS1 geni (Tüm Gen Dizi Analizi)	LATS1 gene (Sequence Analysis-All Coding Exons)	LATS1	LATS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAT2 geni (Tüm Gen Dizi Analizi)	LAT2 gene (Sequence Analysis-All Coding Exons)	LAT2	LAT2, WBSCR5, LAB, NTAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAT geni (Tüm Gen Dizi Analizi)	LAT gene (Sequence Analysis-All Coding Exons)	LAT	LAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LASP1 geni (Tüm Gen Dizi Analizi)	LASP1 gene (Sequence Analysis-All Coding Exons)	LASP1	LASP1, MLN50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LARP6 geni (Tüm Gen Dizi Analizi)	LARP6 gene (Sequence Analysis-All Coding Exons)	LARP6	LARP6, ACHN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LARP4B geni (Tüm Gen Dizi Analizi)	LARP4B gene (Sequence Analysis-All Coding Exons)	LARP4B	LARP4B, LARP5, KIAA0217	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LARP1 geni (Tüm Gen Dizi Analizi)	LARP1 gene (Sequence Analysis-All Coding Exons)	LARP1	LARP1, LARP, KIAA0731	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LARGE2 geni (Tüm Gen Dizi Analizi)	LARGE2 gene (Sequence Analysis-All Coding Exons)	LARGE2	GYLTL1B, LARGE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAPTM5 geni (Tüm Gen Dizi Analizi)	LAPTM5 gene (Sequence Analysis-All Coding Exons)	LAPTM5	LAPTM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAPTM4B geni (Tüm Gen Dizi Analizi)	LAPTM4B gene (Sequence Analysis-All Coding Exons)	LAPTM4B	LAPTM4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAP3 geni (Tüm Gen Dizi Analizi)	LAP3 gene (Sequence Analysis-All Coding Exons)	LAP3	PEPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LANCL2 geni (Tüm Gen Dizi Analizi)	LANCL2 gene (Sequence Analysis-All Coding Exons)	LANCL2	LANCL2, TASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LANCL1 geni (Tüm Gen Dizi Analizi)	LANCL1 gene (Sequence Analysis-All Coding Exons)	LANCL1	LANCL1, GPR69A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMTOR5 geni (Tüm Gen Dizi Analizi)	LAMTOR5 gene (Sequence Analysis-All Coding Exons)	LAMTOR5	LAMTOR5, HBXIP, XIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMTOR3 geni (Tüm Gen Dizi Analizi)	LAMTOR3 gene (Sequence Analysis-All Coding Exons)	LAMTOR3	LAMTOR3, MP1, MAPBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMTOR1 geni (Tüm Gen Dizi Analizi)	LAMTOR1 gene (Sequence Analysis-All Coding Exons)	LAMTOR1	LAMTOR1, C11orf59, PDRO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMP5 geni (Tüm Gen Dizi Analizi)	LAMP5 gene (Sequence Analysis-All Coding Exons)	LAMP5	LAMP5, BAD.LAMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMP3 geni (Tüm Gen Dizi Analizi)	LAMP3 gene (Sequence Analysis-All Coding Exons)	LAMP3	DC.LAMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMP1 geni (Tüm Gen Dizi Analizi)	LAMP1 gene (Sequence Analysis-All Coding Exons)	LAMP1	LAMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMC1 geni (Tüm Gen Dizi Analizi)	LAMC1 gene (Sequence Analysis-All Coding Exons)	LAMC1	LAMC1, LAMB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMB4 geni (Tüm Gen Dizi Analizi)	LAMB4 gene (Sequence Analysis-All Coding Exons)	LAMB4	LAMB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAMA5 geni (Tüm Gen Dizi Analizi)	LAMA5 gene (Sequence Analysis-All Coding Exons)	LAMA5	LAMA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LALBA geni (Tüm Gen Dizi Analizi)	LALBA gene (Sequence Analysis-All Coding Exons)	LALBA	LALBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAIR2 geni (Tüm Gen Dizi Analizi)	LAIR2 gene (Sequence Analysis-All Coding Exons)	LAIR2	LIAR2, CD306	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAIR1 geni (Tüm Gen Dizi Analizi)	LAIR1 gene (Sequence Analysis-All Coding Exons)	LAIR1	LAIR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAGE3 geni (Tüm Gen Dizi Analizi)	LAGE3 gene (Sequence Analysis-All Coding Exons)	LAGE3	LAGE3, ITBA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAG3 geni (Tüm Gen Dizi Analizi)	LAG3 gene (Sequence Analysis-All Coding Exons)	LAG3	LAG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LAD1 geni (Tüm Gen Dizi Analizi)	LAD1 gene (Sequence Analysis-All Coding Exons)	LAD1	LAD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LACTB geni (Tüm Gen Dizi Analizi)	LACTB gene (Sequence Analysis-All Coding Exons)	LACTB	LACTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LACRT geni (Tüm Gen Dizi Analizi)	LACRT gene (Sequence Analysis-All Coding Exons)	LACRT	LACRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LACC1 geni (Tüm Gen Dizi Analizi)	LACC1 gene (Sequence Analysis-All Coding Exons)	LACC1	LACC1, C13orf31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L3MBTL4 geni (Tüm Gen Dizi Analizi)	L3MBTL4 gene (Sequence Analysis-All Coding Exons)	L3MBTL4	L3MBTL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L3MBTL2 geni (Tüm Gen Dizi Analizi)	L3MBTL2 gene (Sequence Analysis-All Coding Exons)	L3MBTL2	L3MBTL2, L3MBT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L3MBTL1 geni (Tüm Gen Dizi Analizi)	L3MBTL1 gene (Sequence Analysis-All Coding Exons)	L3MBTL1	L3MBTL, L3MBTL1, KIAA0681	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
L3HYPDH geni (Tüm Gen Dizi Analizi)	L3HYPDH gene (Sequence Analysis-All Coding Exons)	L3HYPDH	L3HYPDH, C14orf149	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KYAT3 geni (Tüm Gen Dizi Analizi)	KYAT3 gene (Sequence Analysis-All Coding Exons)	KYAT3	CCBL2, KAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KYAT1 geni (Tüm Gen Dizi Analizi)	KYAT1 gene (Sequence Analysis-All Coding Exons)	KYAT1	CCBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KXD1 geni (Tüm Gen Dizi Analizi)	KXD1 gene (Sequence Analysis-All Coding Exons)	KXD1	KXD1, C10orf50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KTN1 geni (Tüm Gen Dizi Analizi)	KTN1 gene (Sequence Analysis-All Coding Exons)	KTN1	KTN1, CG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KSR2 geni (Tüm Gen Dizi Analizi)	KSR2 gene (Sequence Analysis-All Coding Exons)	KSR2	KSR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KSR1 geni (Tüm Gen Dizi Analizi)	KSR1 gene (Sequence Analysis-All Coding Exons)	KSR1	KSR, KSR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTDAP geni (Tüm Gen Dizi Analizi)	KRTDAP gene (Sequence Analysis-All Coding Exons)	KRTDAP	KDAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP5-9 geni (Tüm Gen Dizi Analizi)	KRTAP5-9 gene (Sequence Analysis-All Coding Exons)	KRTAP5-9	KRTAP5-9, KRN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP5-1 geni (Tüm Gen Dizi Analizi)	KRTAP5-1 gene (Sequence Analysis-All Coding Exons)	KRTAP5-1	KRTAP5-1, KRN1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP13-1 geni (Tüm Gen Dizi Analizi)	KRTAP13-1 gene (Sequence Analysis-All Coding Exons)	KRTAP13-1	KRTAP13-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP11-1 geni (Tüm Gen Dizi Analizi)	KRTAP11-1 gene (Sequence Analysis-All Coding Exons)	KRTAP11-1	KRTAP11-1, HACL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP1-5 geni (Tüm Gen Dizi Analizi)	KRTAP1-5 gene (Sequence Analysis-All Coding Exons)	KRTAP1-5	KRTAP-15, KAP1.5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP1-4 geni (Tüm Gen Dizi Analizi)	KRTAP1-4 gene (Sequence Analysis-All Coding Exons)	KRTAP1-4	KRTAP1-4, KAP1.4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP1-3 geni (Tüm Gen Dizi Analizi)	KRTAP1-3 gene (Sequence Analysis-All Coding Exons)	KRTAP1-3	KRTAP1-3, KAP1.3, KAP1.2, KAP1.9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTAP1-1 geni (Tüm Gen Dizi Analizi)	KRTAP1-1 gene (Sequence Analysis-All Coding Exons)	KRTAP1-1	KRTAP1-1, KAP1.1, KAP1.7, KAP1.6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTB4 geni (Tüm Gen Dizi Analizi)	KRTB4 gene (Sequence Analysis-All Coding Exons)	KRTB4	KRTB4, KRTHB4, HB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTB2 geni (Tüm Gen Dizi Analizi)	KRTB2 gene (Sequence Analysis-All Coding Exons)	KRTB2	KRTB2, KRTHB2, HB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRTB0 geni (Tüm Gen Dizi Analizi)	KRTB0 gene (Sequence Analysis-All Coding Exons)	KRTB0	KRTB0, KB20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT79 geni (Tüm Gen Dizi Analizi)	KRT79 gene (Sequence Analysis-All Coding Exons)	KRT79	KRT79, KRT6L, K6L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT78 geni (Tüm Gen Dizi Analizi)	KRT78 gene (Sequence Analysis-All Coding Exons)	KRT78	KRT78, KSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT77 geni (Tüm Gen Dizi Analizi)	KRT77 gene (Sequence Analysis-All Coding Exons)	KRT77	KRT77, K1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT76 geni (Tüm Gen Dizi Analizi)	KRT76 gene (Sequence Analysis-All Coding Exons)	KRT76	KRT76, K76, KRT2P	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT73 geni (Tüm Gen Dizi Analizi)	KRT73 gene (Sequence Analysis-All Coding Exons)	KRT73	KRT73, K6IRS3, KRT6IRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT72 geni (Tüm Gen Dizi Analizi)	KRT72 gene (Sequence Analysis-All Coding Exons)	KRT72	KRT72, K6IRS2, KRT6IRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT7 geni (Tüm Gen Dizi Analizi)	KRT7 gene (Sequence Analysis-All Coding Exons)	KRT7	KRT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT40 geni (Tüm Gen Dizi Analizi)	KRT40 gene (Sequence Analysis-All Coding Exons)	KRT40	KRT40, KA36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT39 geni (Tüm Gen Dizi Analizi)	KRT39 gene (Sequence Analysis-All Coding Exons)	KRT39	KRT39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT38 geni (Tüm Gen Dizi Analizi)	KRT38 gene (Sequence Analysis-All Coding Exons)	KRT38	KRT38, KRTHA8, HA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT37 geni (Tüm Gen Dizi Analizi)	KRT37 gene (Sequence Analysis-All Coding Exons)	KRT37	KRT37, KRTHA7, HA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT36 geni (Tüm Gen Dizi Analizi)	KRT36 gene (Sequence Analysis-All Coding Exons)	KRT36	KRT36, KRTHA6, HA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT35 geni (Tüm Gen Dizi Analizi)	KRT35 gene (Sequence Analysis-All Coding Exons)	KRT35	KRT35, KRTHA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT34 geni (Tüm Gen Dizi Analizi)	KRT34 gene (Sequence Analysis-All Coding Exons)	KRT34	KRT34, KRTHA4, HA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT33B geni (Tüm Gen Dizi Analizi)	KRT33B gene (Sequence Analysis-All Coding Exons)	KRT33B	KRT33B, KRTHA3A, HA3II	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT33A geni (Tüm Gen Dizi Analizi)	KRT33A gene (Sequence Analysis-All Coding Exons)	KRT33A	KRT33A, KRTHA3A, HA3I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT32 geni (Tüm Gen Dizi Analizi)	KRT32 gene (Sequence Analysis-All Coding Exons)	KRT32	KRT32, KRTHA2, HA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT31 geni (Tüm Gen Dizi Analizi)	KRT31 gene (Sequence Analysis-All Coding Exons)	KRT31	KRT31, KRTHA1, HA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCYT2 geni (Tüm Gen Dizi Analizi)	PCYT2 gene (Sequence Analysis-All Coding Exons)	PCYT2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT28 geni (Tüm Gen Dizi Analizi)	KRT28 gene (Sequence Analysis-All Coding Exons)	KRT28	KRT28, K2SD, K2SIRS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT27 geni (Tüm Gen Dizi Analizi)	KRT27 gene (Sequence Analysis-All Coding Exons)	KRT27	KRT27, KRT2SC, K2SIRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT26 geni (Tüm Gen Dizi Analizi)	KRT26 gene (Sequence Analysis-All Coding Exons)	KRT26	KRT26, K2S, K2SB, K2SIRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT24 geni (Tüm Gen Dizi Analizi)	KRT24 gene (Sequence Analysis-All Coding Exons)	KRT24	KRT24, FLJ20261	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT23 geni (Tüm Gen Dizi Analizi)	KRT23 gene (Sequence Analysis-All Coding Exons)	KRT23	KRT23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT20 geni (Tüm Gen Dizi Analizi)	KRT20 gene (Sequence Analysis-All Coding Exons)	KRT20	KRT20, CD20, KRT21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT19 geni (Tüm Gen Dizi Analizi)	KRT19 gene (Sequence Analysis-All Coding Exons)	KRT19	KRT19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRT15 geni (Tüm Gen Dizi Analizi)	KRT15 gene (Sequence Analysis-All Coding Exons)	KRT15	KRT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KRR1 geni (Tüm Gen Dizi Analizi)	KRR1 gene (Sequence Analysis-All Coding Exons)	KRR1	KRR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KREMEN2 geni (Tüm Gen Dizi Analizi)	KREMEN2 gene (Sequence Analysis-All Coding Exons)	KREMEN2	KREMEN2, KRM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

KRBOX4 geni (Tüm Gen Dizi Analizi)	KRBOX4 gene (Sequence Analysis-All Coding Exons)	KRBOX4	ZNF673, FLJ20344	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPRP geni (Tüm Gen Dizi Analizi)	KPRP gene (Sequence Analysis-All Coding Exons)	KPRP	KPRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNB1 geni (Tüm Gen Dizi Analizi)	KPNB1 gene (Sequence Analysis-All Coding Exons)	KPNB1	KPNB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA7 geni (Tüm Gen Dizi Analizi)	KPNA7 gene (Sequence Analysis-All Coding Exons)	KPNA7	KPNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA6 geni (Tüm Gen Dizi Analizi)	KPNA6 gene (Sequence Analysis-All Coding Exons)	KPNA6	KPNA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA5 geni (Tüm Gen Dizi Analizi)	KPNA5 gene (Sequence Analysis-All Coding Exons)	KPNA5	KPNA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA4 geni (Tüm Gen Dizi Analizi)	KPNA4 gene (Sequence Analysis-All Coding Exons)	KPNA4	KPNA4, QIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA3 geni (Tüm Gen Dizi Analizi)	KPNA3 gene (Sequence Analysis-All Coding Exons)	KPNA3	KPNA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA2 geni (Tüm Gen Dizi Analizi)	KPNA2 gene (Sequence Analysis-All Coding Exons)	KPNA2	KPNA2, RCH1, QIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KPNA1 geni (Tüm Gen Dizi Analizi)	KPNA1 gene (Sequence Analysis-All Coding Exons)	KPNA1	KPNA1, RCH2, SRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KNTC1 geni (Tüm Gen Dizi Analizi)	KNTC1 gene (Sequence Analysis-All Coding Exons)	KNTC1	KNTC1, ROD, KIAA0166	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KNSTRN geni (Tüm Gen Dizi Analizi)	KNSTRN gene (Sequence Analysis-All Coding Exons)	KNSTRN	KNSTRN, C15orf23, SKAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KNDC1 geni (Tüm Gen Dizi Analizi)	KNDC1 gene (Sequence Analysis-All Coding Exons)	KNDC1	KNDC1, RASGEF2, KIAA1768	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KNCN geni (Tüm Gen Dizi Analizi)	KNCN gene (Sequence Analysis-All Coding Exons)	KNCN	KNCN, KIN0, LS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMT5C geni (Tüm Gen Dizi Analizi)	KMT5C gene (Sequence Analysis-All Coding Exons)	KMT5C	SUV420H2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMT5B geni (Tüm Gen Dizi Analizi)	KMT5B gene (Sequence Analysis-All Coding Exons)	KMT5B	KMT5B, SUV420H1, CGI85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMT2E geni (Tüm Gen Dizi Analizi)	KMT2E gene (Sequence Analysis-All Coding Exons)	KMT2E	KMT2E, MLL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMT2C geni (Tüm Gen Dizi Analizi)	KMT2C gene (Sequence Analysis-All Coding Exons)	KMT2C	KMT2C, MLL3, KIAA1506	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMO geni (Tüm Gen Dizi Analizi)	KMO gene (Sequence Analysis-All Coding Exons)	KMO	KMO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRK1 geni (Tüm Gen Dizi Analizi)	KLRK1 gene (Sequence Analysis-All Coding Exons)	KLRK1	KLRK1, NKG2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PCGF1 geni (Tüm Gen Dizi Analizi)	PCGF1 gene (Sequence Analysis-All Coding Exons)	PCGF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRG1 geni (Tüm Gen Dizi Analizi)	KLRG1 gene (Sequence Analysis-All Coding Exons)	KLRG1	KLRG1, MAFA, MAFAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRF1 geni (Tüm Gen Dizi Analizi)	KLRF1 gene (Sequence Analysis-All Coding Exons)	KLRF1	KLRF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRD1 geni (Tüm Gen Dizi Analizi)	KLRD1 gene (Sequence Analysis-All Coding Exons)	KLRD1	KLRD1, CD94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRC4 geni (Tüm Gen Dizi Analizi)	KLRC4 gene (Sequence Analysis-All Coding Exons)	KLRC4	KLRC4, NKG2F, D12S2489E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRC3 geni (Tüm Gen Dizi Analizi)	KLRC3 gene (Sequence Analysis-All Coding Exons)	KLRC3	KLRC3, NKG2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRC2 geni (Tüm Gen Dizi Analizi)	KLRC2 gene (Sequence Analysis-All Coding Exons)	KLRC2	KLRC2, NKG2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRC1 geni (Tüm Gen Dizi Analizi)	KLRC1 gene (Sequence Analysis-All Coding Exons)	KLRC1	KLRC1, NKG2, NKG2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLRB1 geni (Tüm Gen Dizi Analizi)	KLRB1 gene (Sequence Analysis-All Coding Exons)	KLRB1	KLRB1, NKRP1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK9 geni (Tüm Gen Dizi Analizi)	KLK9 gene (Sequence Analysis-All Coding Exons)	KLK9	KLK9, KLKL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK8 geni (Tüm Gen Dizi Analizi)	KLK8 gene (Sequence Analysis-All Coding Exons)	KLK8	KLK8, TADG14, PRSS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK7 geni (Tüm Gen Dizi Analizi)	KLK7 gene (Sequence Analysis-All Coding Exons)	KLK7	LKL7, PRSS6, SCCE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK6 geni (Tüm Gen Dizi Analizi)	KLK6 gene (Sequence Analysis-All Coding Exons)	KLK6	KLK6, PRSS9, ZYME	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK5 geni (Tüm Gen Dizi Analizi)	KLK5 gene (Sequence Analysis-All Coding Exons)	KLK5	KLK5, SCTE, KLKL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK3 geni (Tüm Gen Dizi Analizi)	KLK3 gene (Sequence Analysis-All Coding Exons)	KLK3	KLK3, APS, PSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK2 geni (Tüm Gen Dizi Analizi)	KLK2 gene (Sequence Analysis-All Coding Exons)	KLK2	KLK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK15 geni (Tüm Gen Dizi Analizi)	KLK15 gene (Sequence Analysis-All Coding Exons)	KLK15	KLK15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK14 geni (Tüm Gen Dizi Analizi)	KLK14 gene (Sequence Analysis-All Coding Exons)	KLK14	KLK14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK13 geni (Tüm Gen Dizi Analizi)	KLK13 gene (Sequence Analysis-All Coding Exons)	KLK13	KLK13, KLKL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK12 geni (Tüm Gen Dizi Analizi)	KLK12 gene (Sequence Analysis-All Coding Exons)	KLK12	KLK12, KLKL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK11 geni (Tüm Gen Dizi Analizi)	KLK11 gene (Sequence Analysis-All Coding Exons)	KLK11	KLK11, PRSS20, TLSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLK10 geni (Tüm Gen Dizi Analizi)	KLK10 gene (Sequence Analysis-All Coding Exons)	KLK10	KLK10, PRSSL1, NES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL9 geni (Tüm Gen Dizi Analizi)	KLHL9 gene (Sequence Analysis-All Coding Exons)	KLHL9	KLHL9, KIAA1354	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL8 geni (Tüm Gen Dizi Analizi)	KLHL8 gene (Sequence Analysis-All Coding Exons)	KLHL8	KLHL8, KIAA1378	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL6 geni (Tüm Gen Dizi Analizi)	KLHL6 gene (Sequence Analysis-All Coding Exons)	KLHL6	KLHL6, FLJ00029	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL5 geni (Tüm Gen Dizi Analizi)	KLHL5 gene (Sequence Analysis-All Coding Exons)	KLHL5	KLHL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL4 geni (Tüm Gen Dizi Analizi)	KLHL4 gene (Sequence Analysis-All Coding Exons)	KLHL4	KLHL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL31 geni (Tüm Gen Dizi Analizi)	KLHL31 gene (Sequence Analysis-All Coding Exons)	KLHL31	KLHL31, KLHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL21 geni (Tüm Gen Dizi Analizi)	KLHL21 gene (Sequence Analysis-All Coding Exons)	KLHL21	KLHL21, KIAA0469	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL2 geni (Tüm Gen Dizi Analizi)	KLHL2 gene (Sequence Analysis-All Coding Exons)	KLHL2	KLHL2, MAYVEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL14 geni (Tüm Gen Dizi Analizi)	KLHL14 gene (Sequence Analysis-All Coding Exons)	KLHL14	KLHL14, PRINTOR, KIAA1384	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL13 geni (Tüm Gen Dizi Analizi)	KLHL13 gene (Sequence Analysis-All Coding Exons)	KLHL13	KLHL13, KIAA1309	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL12 geni (Tüm Gen Dizi Analizi)	KLHL12 gene (Sequence Analysis-All Coding Exons)	KLHL12	KLHL12, DKIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHL1 geni (Tüm Gen Dizi Analizi)	KLHL1 gene (Sequence Analysis-All Coding Exons)	KLHL1	KLHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC9 geni (Tüm Gen Dizi Analizi)	KLHDC9 gene (Sequence Analysis-All Coding Exons)	KLHDC9	KLHDC9, KARCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC8A geni (Tüm Gen Dizi Analizi)	KLHDC8A gene (Sequence Analysis-All Coding Exons)	KLHDC8A	KLHDC8A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC3 geni (Tüm Gen Dizi Analizi)	KLHDC3 gene (Sequence Analysis-All Coding Exons)	KLHDC3	KLHDC3, PEAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC2 geni (Tüm Gen Dizi Analizi)	KLHDC2 gene (Sequence Analysis-All Coding Exons)	KLHDC2	KLHDC2, HCLP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC10 geni (Tüm Gen Dizi Analizi)	KLHDC10 gene (Sequence Analysis-All Coding Exons)	KLHDC10	KLHDC10, SLIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLHDC1 geni (Tüm Gen Dizi Analizi)	KLHDC1 gene (Sequence Analysis-All Coding Exons)	KLHDC1	KLHDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF9 geni (Tüm Gen Dizi Analizi)	KLF9 gene (Sequence Analysis-All Coding Exons)	KLF9	BTEB1, BTEB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF8 geni (Tüm Gen Dizi Analizi)	KLF8 gene (Sequence Analysis-All Coding Exons)	KLF8	KLF8, ZNF741, BKLF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF7 geni (Tüm Gen Dizi Analizi)	KLF7 gene (Sequence Analysis-All Coding Exons)	KLF7	KLF7, UKLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF4 geni (Tüm Gen Dizi Analizi)	KLF4 gene (Sequence Analysis-All Coding Exons)	KLF4	KLF4, EZF, GKLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF3 geni (Tüm Gen Dizi Analizi)	KLF3 gene (Sequence Analysis-All Coding Exons)	KLF3	KLF3, BKLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF2 geni (Tüm Gen Dizi Analizi)	KLF2 gene (Sequence Analysis-All Coding Exons)	KLF2	KLF2, LKLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF17 geni (Tüm Gen Dizi Analizi)	KLF17 gene (Sequence Analysis-All Coding Exons)	KLF17	ZNF393	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF16 geni (Tüm Gen Dizi Analizi)	KLF16 gene (Sequence Analysis-All Coding Exons)	KLF16	KLF16, BTEB4, DRRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF15 geni (Tüm Gen Dizi Analizi)	KLF15 gene (Sequence Analysis-All Coding Exons)	KLF15	KLF15, KKLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF14 geni (Tüm Gen Dizi Analizi)	KLF14 gene (Sequence Analysis-All Coding Exons)	KLF14	LKF14, BTEB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF13 geni (Tüm Gen Dizi Analizi)	KLF13 gene (Sequence Analysis-All Coding Exons)	KLF13	KLF13, RFLAT1, FKLF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF12 geni (Tüm Gen Dizi Analizi)	KLF12 gene (Sequence Analysis-All Coding Exons)	KLF12	KLF12, AP2REP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF10 geni (Tüm Gen Dizi Analizi)	KLF10 gene (Sequence Analysis-All Coding Exons)	KLF10	KLF10, TIEG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLC3 geni (Tüm Gen Dizi Analizi)	KLC3 gene (Sequence Analysis-All Coding Exons)	KLC3	KLC3, KLC2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLC1 geni (Tüm Gen Dizi Analizi)	KLC1 gene (Sequence Analysis-All Coding Exons)	KLC1	KLC1, KNS2, KNS2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLB geni (Tüm Gen Dizi Analizi)	KLB gene (Sequence Analysis-All Coding Exons)	KLB	KLB, BKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIRREL2 geni (Tüm Gen Dizi Analizi)	KIRREL2 gene (Sequence Analysis-All Coding Exons)	KIRREL2	KIRREL2, NEPH3, NLG1, FILTRIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIRREL geni (Tüm Gen Dizi Analizi)	KIRREL gene (Sequence Analysis-All Coding Exons)	KIRREL	KIRREL, NEPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR3DL3 geni (Tüm Gen Dizi Analizi)	KIR3DL3 gene (Sequence Analysis-All Coding Exons)	KIR3DL3	KIR3KL3, KIRC1, KIR3DL7, KIR44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR3DL2 geni (Tüm Gen Dizi Analizi)	KIR3DL2 gene (Sequence Analysis-All Coding Exons)	KIR3DL2	KIR3DL2, NKAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DS4 geni (Tüm Gen Dizi Analizi)	KIR2DS4 gene (Sequence Analysis-All Coding Exons)	KIR2DS4	KIR2DS4, NKAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL4 geni (Tüm Gen Dizi Analizi)	KIR2DL4 gene (Sequence Analysis-All Coding Exons)	KIR2DL4	KIR2DL4, KIR103AS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL3 geni (Tüm Gen Dizi Analizi)	KIR2DL3 gene (Sequence Analysis-All Coding Exons)	KIR2DL3	KIR2DL3, NKAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL1 geni (Tüm Gen Dizi Analizi)	KIR2DL1 gene (Sequence Analysis-All Coding Exons)	KIR2DL1	KIR2DL1, NKAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIN geni (Tüm Gen Dizi Analizi)	KIN gene (Sequence Analysis-All Coding Exons)	KIN	KIN, KIN17, BTCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIFC3 geni (Tüm Gen Dizi Analizi)	KIFC3 gene (Sequence Analysis-All Coding Exons)	KIFC3	KIFC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIFC2 geni (Tüm Gen Dizi Analizi)	KIFC2 gene (Sequence Analysis-All Coding Exons)	KIFC2	KIFC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIFC1 geni (Tüm Gen Dizi Analizi)	KIFC1 gene (Sequence Analysis-All Coding Exons)	KIFC1	KIFC1, KNSL2, HSET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIFAP3 geni (Tüm Gen Dizi Analizi)	KIFAP3 gene (Sequence Analysis-All Coding Exons)	KIFAP3	KIFAP3, SMAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF9 geni (Tüm Gen Dizi Analizi)	KIF9 gene (Sequence Analysis-All Coding Exons)	KIF9	KIF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF6 geni (Tüm Gen Dizi Analizi)	KIF6 gene (Sequence Analysis-All Coding Exons)	KIF6	KIF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF5B geni (Tüm Gen Dizi Analizi)	KIF5B gene (Sequence Analysis-All Coding Exons)	KIF5B	KIF5B, KNS1, UKHC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF4B geni (Tüm Gen Dizi Analizi)	KIF4B gene (Sequence Analysis-All Coding Exons)	KIF4B	KIF4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

KIF3C geni (Tüm Gen Dizi Analizi)	KIF3C gene (Sequence Analysis-All Coding Exons)	KIF3C	KIF3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF3B geni (Tüm Gen Dizi Analizi)	KIF3B gene (Sequence Analysis-All Coding Exons)	KIF3B	KIF3B, KIAA0359	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF3A geni (Tüm Gen Dizi Analizi)	KIF3A gene (Sequence Analysis-All Coding Exons)	KIF3A	KIF3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF2C geni (Tüm Gen Dizi Analizi)	KIF2C gene (Sequence Analysis-All Coding Exons)	KIF2C	KIF2C, KNSL6, MCAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF2B geni (Tüm Gen Dizi Analizi)	KIF2B gene (Sequence Analysis-All Coding Exons)	KIF2B	KIF2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF27 geni (Tüm Gen Dizi Analizi)	KIF27 gene (Sequence Analysis-All Coding Exons)	KIF27	KIF27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF26B geni (Tüm Gen Dizi Analizi)	KIF26B gene (Sequence Analysis-All Coding Exons)	KIF26B	KIF26B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF26A geni (Tüm Gen Dizi Analizi)	KIF26A gene (Sequence Analysis-All Coding Exons)	KIF26A	KIF26A, KIAA1236	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF25 geni (Tüm Gen Dizi Analizi)	KIF25 gene (Sequence Analysis-All Coding Exons)	KIF25	KIF25, KNSL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF24 geni (Tüm Gen Dizi Analizi)	KIF24 gene (Sequence Analysis-All Coding Exons)	KIF24	KIF24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF23 geni (Tüm Gen Dizi Analizi)	KIF23 gene (Sequence Analysis-All Coding Exons)	KIF23	KIF23, KNSL5, MKLP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF21B geni (Tüm Gen Dizi Analizi)	KIF21B gene (Sequence Analysis-All Coding Exons)	KIF21B	KIF21B, KIAA0449	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF20B geni (Tüm Gen Dizi Analizi)	KIF20B gene (Sequence Analysis-All Coding Exons)	KIF20B	MPHOSPH1, MPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF20A geni (Tüm Gen Dizi Analizi)	KIF20A gene (Sequence Analysis-All Coding Exons)	KIF20A	KIF20A, RAB6KIFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF18B geni (Tüm Gen Dizi Analizi)	KIF18B gene (Sequence Analysis-All Coding Exons)	KIF18B	KIF18B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF18A geni (Tüm Gen Dizi Analizi)	KIF18A gene (Sequence Analysis-All Coding Exons)	KIF18A	KIF18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF17 geni (Tüm Gen Dizi Analizi)	KIF17 gene (Sequence Analysis-All Coding Exons)	KIF17	KIF17, KIAA1405	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF13B geni (Tüm Gen Dizi Analizi)	KIF13B gene (Sequence Analysis-All Coding Exons)	KIF13B	KIF13B, GAKIN, KIAA0639	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF13A geni (Tüm Gen Dizi Analizi)	KIF13A gene (Sequence Analysis-All Coding Exons)	KIF13A	KIF13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIF12 geni (Tüm Gen Dizi Analizi)	KIF12 gene (Sequence Analysis-All Coding Exons)	KIF12	KIF12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1958 geni (Tüm Gen Dizi Analizi)	KIAA1958 gene (Sequence Analysis-All Coding Exons)	KIAA1958	KIAA1958	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1549L geni (Tüm Gen Dizi Analizi)	KIAA1549L gene (Sequence Analysis-All Coding Exons)	KIAA1549L	C11orf41, G2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1549 geni (Tüm Gen Dizi Analizi)	KIAA1549 gene (Sequence Analysis-All Coding Exons)	KIAA1549	KIAA1549	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1524 geni (Tüm Gen Dizi Analizi)	KIAA1524 gene (Sequence Analysis-All Coding Exons)	KIAA1524	KIAA1524, p90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1462 geni (Tüm Gen Dizi Analizi)	KIAA1462 gene (Sequence Analysis-All Coding Exons)	KIAA1462	KIAA1462, JCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1456 geni (Tüm Gen Dizi Analizi)	KIAA1456 gene (Sequence Analysis-All Coding Exons)	KIAA1456	KIAA1456, TRM9L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1429 geni (Tüm Gen Dizi Analizi)	KIAA1429 gene (Sequence Analysis-All Coding Exons)	KIAA1429	KIAA1429	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1328 geni (Tüm Gen Dizi Analizi)	KIAA1328 gene (Sequence Analysis-All Coding Exons)	KIAA1328	KIAA1328, HINDERIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1324L geni (Tüm Gen Dizi Analizi)	KIAA1324L gene (Sequence Analysis-All Coding Exons)	KIAA1324L	KIAA1324L, EIG121L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1324 geni (Tüm Gen Dizi Analizi)	KIAA1324 gene (Sequence Analysis-All Coding Exons)	KIAA1324	KIAA1324, EIG121	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1217 geni (Tüm Gen Dizi Analizi)	KIAA1217 gene (Sequence Analysis-All Coding Exons)	KIAA1217	KIAA1217, SKT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA1109 geni (Tüm Gen Dizi Analizi)	KIAA1109 gene (Sequence Analysis-All Coding Exons)	KIAA1109	KIAA1109	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0922 geni (Tüm Gen Dizi Analizi)	KIAA0922 gene (Sequence Analysis-All Coding Exons)	KIAA0922	KIAA0922, TMEM131L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0825 geni (Tüm Gen Dizi Analizi)	KIAA0825 gene (Sequence Analysis-All Coding Exons)	KIAA0825	KIAA0825	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0513 geni (Tüm Gen Dizi Analizi)	KIAA0513 gene (Sequence Analysis-All Coding Exons)	KIAA0513	KIAA0513	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0391 geni (Tüm Gen Dizi Analizi)	KIAA0391 gene (Sequence Analysis-All Coding Exons)	KIAA0391	KIAA0391	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0368 geni (Tüm Gen Dizi Analizi)	KIAA0368 gene (Sequence Analysis-All Coding Exons)	KIAA0368	KIAA0368, ECM29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0319L geni (Tüm Gen Dizi Analizi)	KIAA0319L gene (Sequence Analysis-All Coding Exons)	KIAA0319L	KIAA0319L, KIAA1837	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0141 geni (Tüm Gen Dizi Analizi)	KIAA0141 gene (Sequence Analysis-All Coding Exons)	KIAA0141	KIAA0141, DELE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0040 geni (Tüm Gen Dizi Analizi)	KIAA0040 gene (Sequence Analysis-All Coding Exons)	KIAA0040	KIAA0040	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KHSRP geni (Tüm Gen Dizi Analizi)	KHSRP gene (Sequence Analysis-All Coding Exons)	KHSRP	KHSRP, KSRP, FUBP2, FBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KHDRBS3 geni (Tüm Gen Dizi Analizi)	KHDRBS3 gene (Sequence Analysis-All Coding Exons)	KHDRBS3	KHDRBS3, TSTAR, SALP, SLM2, ETOILE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KHDRBS2 geni (Tüm Gen Dizi Analizi)	KHDRBS2 gene (Sequence Analysis-All Coding Exons)	KHDRBS2	KHDRBS2, SLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KHDRBS1 geni (Tüm Gen Dizi Analizi)	KHDRBS1 gene (Sequence Analysis-All Coding Exons)	KHDRBS1	KHDRBS1, SAM68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KHDC1 geni (Tüm Gen Dizi Analizi)	KHDC1 gene (Sequence Analysis-All Coding Exons)	KHDC1	KHDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KEAP1 geni (Tüm Gen Dizi Analizi)	KEAP1 gene (Sequence Analysis-All Coding Exons)	KEAP1	KEAP1, KIAA0132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM8 geni (Tüm Gen Dizi Analizi)	KDM8 gene (Sequence Analysis-All Coding Exons)	KDM8	JMJD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM6B geni (Tüm Gen Dizi Analizi)	KDM6B gene (Sequence Analysis-All Coding Exons)	KDM6B	KDM6B, JMJD3, KIAA0346	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM5D geni (Tüm Gen Dizi Analizi)	KDM5D gene (Sequence Analysis-All Coding Exons)	KDM5D	KDM5D, JARID1D, SMCY, HY4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM5B geni (Tüm Gen Dizi Analizi)	KDM5B gene (Sequence Analysis-All Coding Exons)	KDM5B	KDM5B, JARID1B, PUT1, PLU1, RBBP2H1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM5A geni (Tüm Gen Dizi Analizi)	KDM5A gene (Sequence Analysis-All Coding Exons)	KDM5A	KDM5A, JARID1A, RBP2, RBBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM4E geni (Tüm Gen Dizi Analizi)	KDM4E gene (Sequence Analysis-All Coding Exons)	KDM4E	KDM4E, JMJD2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM4D geni (Tüm Gen Dizi Analizi)	KDM4D gene (Sequence Analysis-All Coding Exons)	KDM4D	JMJD2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM4C geni (Tüm Gen Dizi Analizi)	KDM4C gene (Sequence Analysis-All Coding Exons)	KDM4C	KDM4C, JMJD2C, GASC1, KIA0780	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM4B geni (Tüm Gen Dizi Analizi)	KDM4B gene (Sequence Analysis-All Coding Exons)	KDM4B	KDM4B, JMJD2B, KIAA0876	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM4A geni (Tüm Gen Dizi Analizi)	KDM4A gene (Sequence Analysis-All Coding Exons)	KDM4A	KDM4A, JMJD2A, JHDM3A, KIAA0677	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM3B geni (Tüm Gen Dizi Analizi)	KDM3B gene (Sequence Analysis-All Coding Exons)	KDM3B	KDM3B, C5orf7, KIAA1082, JMJD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM3A geni (Tüm Gen Dizi Analizi)	KDM3A gene (Sequence Analysis-All Coding Exons)	KDM3A	KDM3A, JMJD1A, JHDM2A, TSGA, KIAA0742	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM2B geni (Tüm Gen Dizi Analizi)	KDM2B gene (Sequence Analysis-All Coding Exons)	KDM2B	KDM2B, FBXL10, FBL10, CXXC2, JHDM1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM2A geni (Tüm Gen Dizi Analizi)	KDM2A gene (Sequence Analysis-All Coding Exons)	KDM2A	FBXL11, FBL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDM1B geni (Tüm Gen Dizi Analizi)	KDM1B gene (Sequence Analysis-All Coding Exons)	KDM1B	KDM1B, LSD2, AOF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDELR1 geni (Tüm Gen Dizi Analizi)	KDELR1 gene (Sequence Analysis-All Coding Exons)	KDELR1	KDELR1, ERD2, HDEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDELC1 geni (Tüm Gen Dizi Analizi)	KDELC1 gene (Sequence Analysis-All Coding Exons)	KDELC1	KDELC1, EP58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARP10 geni (Tüm Gen Dizi Analizi)	PARP10 gene (Sequence Analysis-All Coding Exons)	PARP10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD9 geni (Tüm Gen Dizi Analizi)	KCTD9 gene (Sequence Analysis-All Coding Exons)	KCTD9	KCTD9, BTBD27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD5 geni (Tüm Gen Dizi Analizi)	KCTD5 gene (Sequence Analysis-All Coding Exons)	KCTD5	KCTD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD3 geni (Tüm Gen Dizi Analizi)	KCTD3 gene (Sequence Analysis-All Coding Exons)	KCTD3	KCTD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD20 geni (Tüm Gen Dizi Analizi)	KCTD20 gene (Sequence Analysis-All Coding Exons)	KCTD20	KCTD20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD2 geni (Tüm Gen Dizi Analizi)	KCTD2 gene (Sequence Analysis-All Coding Exons)	KCTD2	KCTD2, KIAA0176	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD16 geni (Tüm Gen Dizi Analizi)	KCTD16 gene (Sequence Analysis-All Coding Exons)	KCTD16	KCTD16, KIAA1317	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD15 geni (Tüm Gen Dizi Analizi)	KCTD15 gene (Sequence Analysis-All Coding Exons)	KCTD15	KCTD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD13 geni (Tüm Gen Dizi Analizi)	KCTD13 gene (Sequence Analysis-All Coding Exons)	KCTD13	KCTD13, POLDIP1, FKSG86	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PARD6A geni (Tüm Gen Dizi Analizi)	PARD6A gene (Sequence Analysis-All Coding Exons)	PARD6A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD12 geni (Tüm Gen Dizi Analizi)	KCTD12 gene (Sequence Analysis-All Coding Exons)	KCTD12	KCTD12, PFET1, KIAA1778, C13orf2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD11 geni (Tüm Gen Dizi Analizi)	KCTD11 gene (Sequence Analysis-All Coding Exons)	KCTD11	KCTD11, REn	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCTD10 geni (Tüm Gen Dizi Analizi)	KCTD10 gene (Sequence Analysis-All Coding Exons)	KCTD10	KCTD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCP geni (Tüm Gen Dizi Analizi)	KCP gene (Sequence Analysis-All Coding Exons)	KCP	KCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNV1 geni (Tüm Gen Dizi Analizi)	KCNV1 gene (Sequence Analysis-All Coding Exons)	KCNV1	KCNV1, KV8.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNU1 geni (Tüm Gen Dizi Analizi)	KCNU1 gene (Sequence Analysis-All Coding Exons)	KCNU1	KCNU1, SLO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNT2 geni (Tüm Gen Dizi Analizi)	KCNT2 gene (Sequence Analysis-All Coding Exons)	KCNT2	KCNT2, SLICK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNS3 geni (Tüm Gen Dizi Analizi)	KCNS3 gene (Sequence Analysis-All Coding Exons)	KCNS3	KCNS3, KV9.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNS2 geni (Tüm Gen Dizi Analizi)	KCNS2 gene (Sequence Analysis-All Coding Exons)	KCNS2	KCNS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNS1 geni (Tüm Gen Dizi Analizi)	KCNS1 gene (Sequence Analysis-All Coding Exons)	KCNS1	KCNS1, KV9.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNRG geni (Tüm Gen Dizi Analizi)	KCNRG gene (Sequence Analysis-All Coding Exons)	KCNRG	KCNRG, DLTET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNQ5 geni (Tüm Gen Dizi Analizi)	KCNQ5 gene (Sequence Analysis-All Coding Exons)	KCNQ5	KCNQ5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNQ1DN geni (Tüm Gen Dizi Analizi)	KCNQ1DN gene (Sequence Analysis-All Coding Exons)	KCNQ1DN	KCNQ1DN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNN3 geni (Tüm Gen Dizi Analizi)	KCNN3 gene (Sequence Analysis-All Coding Exons)	KCNN3	KCNN3, SK3, SKCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNN2 geni (Tüm Gen Dizi Analizi)	KCNN2 gene (Sequence Analysis-All Coding Exons)	KCNN2	KCNN2, SK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNMB4 geni (Tüm Gen Dizi Analizi)	KCNMB4 gene (Sequence Analysis-All Coding Exons)	KCNMB4	KCNMB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAPD5 geni (Tüm Gen Dizi Analizi)	PAPD5 gene (Sequence Analysis-All Coding Exons)	PAPD5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNMB3 geni (Tüm Gen Dizi Analizi)	KCNMB3 gene (Sequence Analysis-All Coding Exons)	KCNMB3	KCNMB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNMB2 geni (Tüm Gen Dizi Analizi)	KCNMB2 gene (Sequence Analysis-All Coding Exons)	KCNMB2	KCNMB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK7 geni (Tüm Gen Dizi Analizi)	KCNK7 gene (Sequence Analysis-All Coding Exons)	KCNK7	KCNK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

KCNK6 geni (Tüm Gen Dizi Analizi)	KCNK6 gene (Sequence Analysis-All Coding Exons)	KCNK6	KCNK6, TWIK2, TOSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK5 geni (Tüm Gen Dizi Analizi)	KCNK5 gene (Sequence Analysis-All Coding Exons)	KCNK5	KCNK5, TASK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK4 geni (Tüm Gen Dizi Analizi)	KCNK4 gene (Sequence Analysis-All Coding Exons)	KCNK4	KCNK4, TRAAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK2 geni (Tüm Gen Dizi Analizi)	KCNK2 gene (Sequence Analysis-All Coding Exons)	KCNK2	KCNK2, TREK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK17 geni (Tüm Gen Dizi Analizi)	KCNK17 gene (Sequence Analysis-All Coding Exons)	KCNK17	KCNK17, TASK4, TALK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK16 geni (Tüm Gen Dizi Analizi)	KCNK16 gene (Sequence Analysis-All Coding Exons)	KCNK16	KCNK16, TALK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK15 geni (Tüm Gen Dizi Analizi)	KCNK15 gene (Sequence Analysis-All Coding Exons)	KCNK15	KCNK15, TASK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK13 geni (Tüm Gen Dizi Analizi)	KCNK13 gene (Sequence Analysis-All Coding Exons)	KCNK13	KCNK13, THIK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK12 geni (Tüm Gen Dizi Analizi)	KCNK12 gene (Sequence Analysis-All Coding Exons)	KCNK12	KCNK12, THIK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK10 geni (Tüm Gen Dizi Analizi)	KCNK10 gene (Sequence Analysis-All Coding Exons)	KCNK10	KCNK10, TREK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNK1 geni (Tüm Gen Dizi Analizi)	KCNK1 gene (Sequence Analysis-All Coding Exons)	KCNK1	KCNK1, TWIK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ9 geni (Tüm Gen Dizi Analizi)	KCNJ9 gene (Sequence Analysis-All Coding Exons)	KCNJ9	KCNJ9, GIRK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ8 geni (Tüm Gen Dizi Analizi)	KCNJ8 gene (Sequence Analysis-All Coding Exons)	KCNJ8	KCNJ8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ8 geni (Tüm Gen Dizi Analizi)	KCNJ8 gene (Sequence Analysis-All Coding Exons)	KCNJ8	KCNJ8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ4 geni (Tüm Gen Dizi Analizi)	KCNJ4 gene (Sequence Analysis-All Coding Exons)	KCNJ4	KCNJ4, HIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ3 geni (Tüm Gen Dizi Analizi)	KCNJ3 gene (Sequence Analysis-All Coding Exons)	KCNJ3	KCNJ3, GIRK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ16 geni (Tüm Gen Dizi Analizi)	KCNJ16 gene (Sequence Analysis-All Coding Exons)	KCNJ16	KCNJ16, KIR5.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ15 geni (Tüm Gen Dizi Analizi)	KCNJ15 gene (Sequence Analysis-All Coding Exons)	KCNJ15	KCNJ15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PAK5 geni (Tüm Gen Dizi Analizi)	PAK5 gene (Sequence Analysis-All Coding Exons)	PAK5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ12 geni (Tüm Gen Dizi Analizi)	KCNJ12 gene (Sequence Analysis-All Coding Exons)	KCNJ12	KCNJ12, KCNJN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNIP4 geni (Tüm Gen Dizi Analizi)	KCNIP4 gene (Sequence Analysis-All Coding Exons)	KCNIP4	KCNIP4, KCHIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNIP3 geni (Tüm Gen Dizi Analizi)	KCNIP3 gene (Sequence Analysis-All Coding Exons)	KCNIP3	KCNIP3, KCHIP3, CSEN, DREAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH7 geni (Tüm Gen Dizi Analizi)	KCNH7 gene (Sequence Analysis-All Coding Exons)	KCNH7	KCNH7, ERG3, HERG3, Kv11.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH6 geni (Tüm Gen Dizi Analizi)	KCNH6 gene (Sequence Analysis-All Coding Exons)	KCNH6	KCNH6, HERG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH5 geni (Tüm Gen Dizi Analizi)	KCNH5 gene (Sequence Analysis-All Coding Exons)	KCNH5	KCNH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH3 geni (Tüm Gen Dizi Analizi)	KCNH3 gene (Sequence Analysis-All Coding Exons)	KCNH3	KCNH3, BEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNG4 geni (Tüm Gen Dizi Analizi)	KCNG4 gene (Sequence Analysis-All Coding Exons)	KCNG4	KCNG4, KV6.3, KV6.4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNG3 geni (Tüm Gen Dizi Analizi)	KCNG3 gene (Sequence Analysis-All Coding Exons)	KCNG3	KCNG3, KV6.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNG1 geni (Tüm Gen Dizi Analizi)	KCNG1 gene (Sequence Analysis-All Coding Exons)	KCNG1	KCNG1, KH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNF1 geni (Tüm Gen Dizi Analizi)	KCNF1 gene (Sequence Analysis-All Coding Exons)	KCNF1	KCNF1, KH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNE5 geni (Tüm Gen Dizi Analizi)	KCNE5 gene (Sequence Analysis-All Coding Exons)	KCNE5	KCNE1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNE4 geni (Tüm Gen Dizi Analizi)	KCNE4 gene (Sequence Analysis-All Coding Exons)	KCNE4	KCNE4, MIRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCND2 geni (Tüm Gen Dizi Analizi)	KCND2 gene (Sequence Analysis-All Coding Exons)	KCND2	KCND2, KIAA1044	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCND1 geni (Tüm Gen Dizi Analizi)	KCND1 gene (Sequence Analysis-All Coding Exons)	KCND1	KCND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNC4 geni (Tüm Gen Dizi Analizi)	KCNC4 gene (Sequence Analysis-All Coding Exons)	KCNC4	KCNC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNC2 geni (Tüm Gen Dizi Analizi)	KCNC2 gene (Sequence Analysis-All Coding Exons)	KCNC2	KCNC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNB2 geni (Tüm Gen Dizi Analizi)	KCNB2 gene (Sequence Analysis-All Coding Exons)	KCNB2	KCNB2, KV2.2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNAB3 geni (Tüm Gen Dizi Analizi)	KCNAB3 gene (Sequence Analysis-All Coding Exons)	KCNAB3	KCNAB3, KCNA3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNAB2 geni (Tüm Gen Dizi Analizi)	KCNAB2 gene (Sequence Analysis-All Coding Exons)	KCNAB2	KCNAB2, KCNA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNAB1 geni (Tüm Gen Dizi Analizi)	KCNAB1 gene (Sequence Analysis-All Coding Exons)	KCNAB1	KCNAB1, KCNA1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNA7 geni (Tüm Gen Dizi Analizi)	KCNA7 gene (Sequence Analysis-All Coding Exons)	KCNA7	KCNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNA6 geni (Tüm Gen Dizi Analizi)	KCNA6 gene (Sequence Analysis-All Coding Exons)	KCNA6	KCNA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNA4 geni (Tüm Gen Dizi Analizi)	KCNA4 gene (Sequence Analysis-All Coding Exons)	KCNA4	KCNA4, KCNA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNA3 geni (Tüm Gen Dizi Analizi)	KCNA3 gene (Sequence Analysis-All Coding Exons)	KCNA3	KCNA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNA10 geni (Tüm Gen Dizi Analizi)	KCNA10 gene (Sequence Analysis-All Coding Exons)	KCNA10	KCNA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCMF1 geni (Tüm Gen Dizi Analizi)	KCMF1 gene (Sequence Analysis-All Coding Exons)	KCMF1	KCMF1, DEBT91, FIGC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KBTBD8 geni (Tüm Gen Dizi Analizi)	KBTBD8 gene (Sequence Analysis-All Coding Exons)	KBTBD8	KBTBD8, TAKRP, KIAA1842	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KATNBL1 geni (Tüm Gen Dizi Analizi)	KATNBL1 gene (Sequence Analysis-All Coding Exons)	KATNBL1	KATNBL1, C15orf29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KATNAL2 geni (Tüm Gen Dizi Analizi)	KATNAL2 gene (Sequence Analysis-All Coding Exons)	KATNAL2	KATNAL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KATNAL1 geni (Tüm Gen Dizi Analizi)	KATNAL1 gene (Sequence Analysis-All Coding Exons)	KATNAL1	KATNAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KATNA1 geni (Tüm Gen Dizi Analizi)	KATNA1 gene (Sequence Analysis-All Coding Exons)	KATNA1	KATNA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAT8 geni (Tüm Gen Dizi Analizi)	KAT8 gene (Sequence Analysis-All Coding Exons)	KAT8	KAT8, MYST1, MOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAT7 geni (Tüm Gen Dizi Analizi)	KAT7 gene (Sequence Analysis-All Coding Exons)	KAT7	KAT7, MYST2, HBO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAT5 geni (Tüm Gen Dizi Analizi)	KAT5 gene (Sequence Analysis-All Coding Exons)	KAT5	KAT5, HTATIP, TIP60, ESA1, PLIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAT2B geni (Tüm Gen Dizi Analizi)	KAT2B gene (Sequence Analysis-All Coding Exons)	KAT2B	CAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAT2A geni (Tüm Gen Dizi Analizi)	KAT2A gene (Sequence Analysis-All Coding Exons)	KAT2A	KAT2A, GCN5L2, GCN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
PABPC3 geni (Tüm Gen Dizi Analizi)	PABPC3 gene (Sequence Analysis-All Coding Exons)	PABPC3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KANSL2 geni (Tüm Gen Dizi Analizi)	KANSL2 gene (Sequence Analysis-All Coding Exons)	KANSL2	KANSL2, NSL2, C12orf41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KANSL1L geni (Tüm Gen Dizi Analizi)	KANSL1L gene (Sequence Analysis-All Coding Exons)	KANSL1L	KANSL1L, C2orf67, MSL1V2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KANK4 geni (Tüm Gen Dizi Analizi)	KANK4 gene (Sequence Analysis-All Coding Exons)	KANK4	KANK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KANK3 geni (Tüm Gen Dizi Analizi)	KANK3 gene (Sequence Analysis-All Coding Exons)	KANK3	KANK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAAG1 geni (Tüm Gen Dizi Analizi)	KAAG1 gene (Sequence Analysis-All Coding Exons)	KAAG1	KAAG1, RUSAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JUND geni (Tüm Gen Dizi Analizi)	JUND gene (Sequence Analysis-All Coding Exons)	JUND	JUND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JUNB geni (Tüm Gen Dizi Analizi)	JUNB gene (Sequence Analysis-All Coding Exons)	JUNB	JUNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JUN geni (Tüm Gen Dizi Analizi)	JUN gene (Sequence Analysis-All Coding Exons)	JUN	JUN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JTB geni (Tüm Gen Dizi Analizi)	JTB gene (Sequence Analysis-All Coding Exons)	JTB	JTB, PAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JSRP1 geni (Tüm Gen Dizi Analizi)	JSRP1 gene (Sequence Analysis-All Coding Exons)	JSRP1	JSRP1, JP45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JRKL geni (Tüm Gen Dizi Analizi)	JRKL gene (Sequence Analysis-All Coding Exons)	JRKL	JRKL, HHMJG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JRK geni (Tüm Gen Dizi Analizi)	JRK gene (Sequence Analysis-All Coding Exons)	JRK	JRK, JH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JRK geni (Tüm Gen Dizi Analizi)	JRK gene (Sequence Analysis-All Coding Exons)	JRK	JRK, JH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JPX geni (Tüm Gen Dizi Analizi)	JPX gene (Sequence Analysis-All Coding Exons)	JPX	NCRNA00183, JPX, ENOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JOSD2 geni (Tüm Gen Dizi Analizi)	JOSD2 gene (Sequence Analysis-All Coding Exons)	JOSD2	JOSD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JOSD1 geni (Tüm Gen Dizi Analizi)	JOSD1 gene (Sequence Analysis-All Coding Exons)	JOSD1	JOSD1, KIAA0063	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JMY geni (Tüm Gen Dizi Analizi)	JMY gene (Sequence Analysis-All Coding Exons)	JMY	JMY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JMJD6 geni (Tüm Gen Dizi Analizi)	JMJD6 gene (Sequence Analysis-All Coding Exons)	JMJD6	JMJD6, PSR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JMJD1C geni (Tüm Gen Dizi Analizi)	JMJD1C gene (Sequence Analysis-All Coding Exons)	JMJD1C	JMJD1C, TRIP8, KIAA1380	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JMJD1C geni (Tüm Gen Dizi Analizi)	JMJD1C gene (Sequence Analysis-All Coding Exons)	JMJD1C	JMJD1C, TRIP8, KIAA1380	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JKAMP geni (Tüm Gen Dizi Analizi)	JKAMP gene (Sequence Analysis-All Coding Exons)	JKAMP	JKAMP, C24orf100, JAMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JDP2 geni (Tüm Gen Dizi Analizi)	JDP2 gene (Sequence Analysis-All Coding Exons)	JDP2	JDP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
P2RX5 geni (Tüm Gen Dizi Analizi)	P2RX5 gene (Sequence Analysis-All Coding Exons)	P2RX5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JCHAIN geni (Tüm Gen Dizi Analizi)	JCHAIN gene (Sequence Analysis-All Coding Exons)	JCHAIN	IGJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAZF1 geni (Tüm Gen Dizi Analizi)	JAZF1 gene (Sequence Analysis-All Coding Exons)	JAZF1	JAZF1, TIP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JARID2 geni (Tüm Gen Dizi Analizi)	JARID2 gene (Sequence Analysis-All Coding Exons)	JARID2	JMJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAML geni (Tüm Gen Dizi Analizi)	JAML gene (Sequence Analysis-All Coding Exons)	JAML	AMICA1, JAML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAM2 geni (Tüm Gen Dizi Analizi)	JAM2 gene (Sequence Analysis-All Coding Exons)	JAM2	JAM2, VEJAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAKMIP3 geni (Tüm Gen Dizi Analizi)	JAKMIP3 gene (Sequence Analysis-All Coding Exons)	JAKMIP3	JAKMIP3, C10orf59, NECC2, JAMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAKMIP2 geni (Tüm Gen Dizi Analizi)	JAKMIP2 gene (Sequence Analysis-All Coding Exons)	JAKMIP2	JAKMIP2, JAMP2, NECC1, KIAA0555	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAKMIP1 geni (Tüm Gen Dizi Analizi)	JAKMIP1 gene (Sequence Analysis-All Coding Exons)	JAKMIP1	JAKMIP1, JAMP1, MARLIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAK1 geni (Tüm Gen Dizi Analizi)	JAK1 gene (Sequence Analysis-All Coding Exons)	JAK1	JAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JAG2 geni (Tüm Gen Dizi Analizi)	JAG2 gene (Sequence Analysis-All Coding Exons)	JAG2	JAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JADE3 geni (Tüm Gen Dizi Analizi)	JADE3 gene (Sequence Analysis-All Coding Exons)	JADE3	PHF16, JADE3, KIAA0215	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JADE2 geni (Tüm Gen Dizi Analizi)	JADE2 gene (Sequence Analysis-All Coding Exons)	JADE2	PHF15, JADE2, KIAA0239	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
JADE1 geni (Tüm Gen Dizi Analizi)	JADE1 gene (Sequence Analysis-All Coding Exons)	JADE1	PHF17, JADE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

IZUMO1R geni (Tüm Gen Dizi Analizi)	IZUMO1R gene (Sequence Analysis-All Coding Exons)	IZUMO1R	FOLR4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IZUMO1 geni (Tüm Gen Dizi Analizi)	IZUMO1 gene (Sequence Analysis-All Coding Exons)	IZUMO1	IZUMO1, MGC34799	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IVNS1ABP geni (Tüm Gen Dizi Analizi)	IVNS1ABP gene (Sequence Analysis-All Coding Exons)	IVNS1ABP	IVNS1ABP, NS1BP, ND1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IVL geni (Tüm Gen Dizi Analizi)	IVL gene (Sequence Analysis-All Coding Exons)	IVL	IVL	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITSN2 geni (Tüm Gen Dizi Analizi)	ITSN2 gene (Sequence Analysis-All Coding Exons)	ITSN2	ITSN2, SH3D1B, SWAP, KIAA1256	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITSN1 geni (Tüm Gen Dizi Analizi)	ITSN1 gene (Sequence Analysis-All Coding Exons)	ITSN1	ITSN1, SH3D1A, SH3P17	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITPKB geni (Tüm Gen Dizi Analizi)	ITPKB gene (Sequence Analysis-All Coding Exons)	ITPKB	ITPKB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITPKA geni (Tüm Gen Dizi Analizi)	ITPKA gene (Sequence Analysis-All Coding Exons)	ITPKA	ITPKA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITPK1 geni (Tüm Gen Dizi Analizi)	ITPK1 gene (Sequence Analysis-All Coding Exons)	ITPK1	ITPK1, ITRPK1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITM2C geni (Tüm Gen Dizi Analizi)	ITM2C gene (Sequence Analysis-All Coding Exons)	ITM2C	ITM2C, BRI3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITM2A geni (Tüm Gen Dizi Analizi)	ITM2A gene (Sequence Analysis-All Coding Exons)	ITM2A	ITM2A, E25A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITLN2 geni (Tüm Gen Dizi Analizi)	ITLN2 gene (Sequence Analysis-All Coding Exons)	ITLN2	ITLN2, HL2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITLN1 geni (Tüm Gen Dizi Analizi)	ITLN1 gene (Sequence Analysis-All Coding Exons)	ITLN1	ITLN1, INTL, LFR, HL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITIH5 geni (Tüm Gen Dizi Analizi)	ITIH5 gene (Sequence Analysis-All Coding Exons)	ITIH5	ITIH5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITIH3 geni (Tüm Gen Dizi Analizi)	ITIH3 gene (Sequence Analysis-All Coding Exons)	ITIH3	ITIH3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITIH2 geni (Tüm Gen Dizi Analizi)	ITIH2 gene (Sequence Analysis-All Coding Exons)	ITIH2	ITIH2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITIH1 geni (Tüm Gen Dizi Analizi)	ITIH1 gene (Sequence Analysis-All Coding Exons)	ITIH1	ITIH1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGBL1 geni (Tüm Gen Dizi Analizi)	ITGBL1 gene (Sequence Analysis-All Coding Exons)	ITGBL1	ITGBL1, TIED	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB7 geni (Tüm Gen Dizi Analizi)	ITGB7 gene (Sequence Analysis-All Coding Exons)	ITGB7	ITGB7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB5 geni (Tüm Gen Dizi Analizi)	ITGB5 gene (Sequence Analysis-All Coding Exons)	ITGB5	ITGB5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB3BP geni (Tüm Gen Dizi Analizi)	ITGB3BP gene (Sequence Analysis-All Coding Exons)	ITGB3BP	ITGB3BP, NRIF3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB1BP2 geni (Tüm Gen Dizi Analizi)	ITGB1BP2 gene (Sequence Analysis-All Coding Exons)	ITGB1BP2	ITGB1BP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB1BP1 geni (Tüm Gen Dizi Analizi)	ITGB1BP1 gene (Sequence Analysis-All Coding Exons)	ITGB1BP1	ITGB1BP1, ICAP1, ICAP1A, ICAP1B	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB1 geni (Tüm Gen Dizi Analizi)	ITGB1 gene (Sequence Analysis-All Coding Exons)	ITGB1	ITGB1, FNRB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGAX geni (Tüm Gen Dizi Analizi)	ITGAX gene (Sequence Analysis-All Coding Exons)	ITGAX	ITGAX, CD11C	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGAV geni (Tüm Gen Dizi Analizi)	ITGAV gene (Sequence Analysis-All Coding Exons)	ITGAV	ITGAV, VNRA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGAL geni (Tüm Gen Dizi Analizi)	ITGAL gene (Sequence Analysis-All Coding Exons)	ITGAL	ITGAL, CD11A, LFA1A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGAE geni (Tüm Gen Dizi Analizi)	ITGAE gene (Sequence Analysis-All Coding Exons)	ITGAE	ITGAE, CD103	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGAD geni (Tüm Gen Dizi Analizi)	ITGAD gene (Sequence Analysis-All Coding Exons)	ITGAD	ITGAD, CD11D	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA9 geni (Tüm Gen Dizi Analizi)	ITGA9 gene (Sequence Analysis-All Coding Exons)	ITGA9	ITGA9	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA5 geni (Tüm Gen Dizi Analizi)	ITGA5 gene (Sequence Analysis-All Coding Exons)	ITGA5	ITGA5, FNRA, VLA5A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA4 geni (Tüm Gen Dizi Analizi)	ITGA4 gene (Sequence Analysis-All Coding Exons)	ITGA4	ITGA4, CD49D	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA11 geni (Tüm Gen Dizi Analizi)	ITGA11 gene (Sequence Analysis-All Coding Exons)	ITGA11	ITGA11	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA10 geni (Tüm Gen Dizi Analizi)	ITGA10 gene (Sequence Analysis-All Coding Exons)	ITGA10	ITGA10	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGA1 geni (Tüm Gen Dizi Analizi)	ITGA1 gene (Sequence Analysis-All Coding Exons)	ITGA1	ITGA1, VLA1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITFG1 geni (Tüm Gen Dizi Analizi)	ITFG1 gene (Sequence Analysis-All Coding Exons)	ITFG1	ITFG1, TIP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISYNA1 geni (Tüm Gen Dizi Analizi)	ISYNA1 gene (Sequence Analysis-All Coding Exons)	ISYNA1	ISYNA1, IPS	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISY1 geni (Tüm Gen Dizi Analizi)	ISY1 gene (Sequence Analysis-All Coding Exons)	ISY1	ISY1, KIAA1160	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISX geni (Tüm Gen Dizi Analizi)	ISX gene (Sequence Analysis-All Coding Exons)	ISX	ISX, RAXLX	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IST1 geni (Tüm Gen Dizi Analizi)	IST1 gene (Sequence Analysis-All Coding Exons)	IST1	IST1, KIAA0174	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISOC2 geni (Tüm Gen Dizi Analizi)	ISOC2 gene (Sequence Analysis-All Coding Exons)	ISOC2	ISOC2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISM2 geni (Tüm Gen Dizi Analizi)	ISM2 gene (Sequence Analysis-All Coding Exons)	ISM2	ISM2, THSD3, TAIL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISM1 geni (Tüm Gen Dizi Analizi)	ISM1 gene (Sequence Analysis-All Coding Exons)	ISM1	ISM1, ISM	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISLR2 geni (Tüm Gen Dizi Analizi)	ISLR2 gene (Sequence Analysis-All Coding Exons)	ISLR2	ISLR2, LINX, KIAA1464	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISLR geni (Tüm Gen Dizi Analizi)	ISLR gene (Sequence Analysis-All Coding Exons)	ISLR	ISLR	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISL1 geni (Tüm Gen Dizi Analizi)	ISL1 gene (Sequence Analysis-All Coding Exons)	ISL1	ISL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISG20L2 geni (Tüm Gen Dizi Analizi)	ISG20L2 gene (Sequence Analysis-All Coding Exons)	ISG20L2	ISG20L2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISG20 geni (Tüm Gen Dizi Analizi)	ISG20 gene (Sequence Analysis-All Coding Exons)	ISG20	ISG20, HEM45	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISCA1 geni (Tüm Gen Dizi Analizi)	ISCA1 gene (Sequence Analysis-All Coding Exons)	ISCA1	ISCA1, HBLD2, HSCA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRX6 geni (Tüm Gen Dizi Analizi)	IRX6 gene (Sequence Analysis-All Coding Exons)	IRX6	IRX6, IRX7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRX4 geni (Tüm Gen Dizi Analizi)	IRX4 gene (Sequence Analysis-All Coding Exons)	IRX4	IRX4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRX3 geni (Tüm Gen Dizi Analizi)	IRX3 gene (Sequence Analysis-All Coding Exons)	IRX3	IRX3, IRXB1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRX2 geni (Tüm Gen Dizi Analizi)	IRX2 gene (Sequence Analysis-All Coding Exons)	IRX2	IRX2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRX1 geni (Tüm Gen Dizi Analizi)	IRX1 gene (Sequence Analysis-All Coding Exons)	IRX1	IRX1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRS4 geni (Tüm Gen Dizi Analizi)	IRS4 gene (Sequence Analysis-All Coding Exons)	IRS4	IRS4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRF9 geni (Tüm Gen Dizi Analizi)	IRF9 gene (Sequence Analysis-All Coding Exons)	IRF9	ISGF3, ISGF3G	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRF2BPL geni (Tüm Gen Dizi Analizi)	IRF2BPL gene (Sequence Analysis-All Coding Exons)	IRF2BPL	C14orf4, EAP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRF2BP2 geni (Tüm Gen Dizi Analizi)	IRF2BP2 gene (Sequence Analysis-All Coding Exons)	IRF2BP2	IRF2BP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRF2BP1 geni (Tüm Gen Dizi Analizi)	IRF2BP1 gene (Sequence Analysis-All Coding Exons)	IRF2BP1	IRF2BP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRF2 geni (Tüm Gen Dizi Analizi)	IRF2 gene (Sequence Analysis-All Coding Exons)	IRF2	IRF2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IREB2 geni (Tüm Gen Dizi Analizi)	IREB2 gene (Sequence Analysis-All Coding Exons)	IREB2	IREB2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRAK2 geni (Tüm Gen Dizi Analizi)	IRAK2 gene (Sequence Analysis-All Coding Exons)	IRAK2	IRAK2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRAK1BP1 geni (Tüm Gen Dizi Analizi)	IRAK1BP1 gene (Sequence Analysis-All Coding Exons)	IRAK1BP1	IRAK1BP1, SIMPL	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IRAK1 geni (Tüm Gen Dizi Analizi)	IRAK1 gene (Sequence Analysis-All Coding Exons)	IRAK1	IRAK1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQSEC3 geni (Tüm Gen Dizi Analizi)	IQSEC3 gene (Sequence Analysis-All Coding Exons)	IQSEC3	IQSEC3, KIAA1110	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQSEC1 geni (Tüm Gen Dizi Analizi)	IQSEC1 gene (Sequence Analysis-All Coding Exons)	IQSEC1	IQSEC1, KIAA0763	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQGAP2 geni (Tüm Gen Dizi Analizi)	IQGAP2 gene (Sequence Analysis-All Coding Exons)	IQGAP2	IQGAP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQGAP1 geni (Tüm Gen Dizi Analizi)	IQGAP1 gene (Sequence Analysis-All Coding Exons)	IQGAP1	IQGAP1, SAR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQCJ geni (Tüm Gen Dizi Analizi)	IQCJ gene (Sequence Analysis-All Coding Exons)	IQCJ	IQCJ	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQCH geni (Tüm Gen Dizi Analizi)	IQCH gene (Sequence Analysis-All Coding Exons)	IQCH	IQCH, NYDSP5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IQCG geni (Tüm Gen Dizi Analizi)	IQCG gene (Sequence Analysis-All Coding Exons)	IQCG	IQCG	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPP geni (Tüm Gen Dizi Analizi)	IPP gene (Sequence Analysis-All Coding Exons)	IPP	IPP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPO8 geni (Tüm Gen Dizi Analizi)	IPO8 gene (Sequence Analysis-All Coding Exons)	IPO8	IPO8, RANBP8	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPO7 geni (Tüm Gen Dizi Analizi)	IPO7 gene (Sequence Analysis-All Coding Exons)	IPO7	IPO7, RANBP7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPO13 geni (Tüm Gen Dizi Analizi)	IPO13 gene (Sequence Analysis-All Coding Exons)	IPO13	IPO13, IMP13, KIAA0724, RANBP13	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPO11 geni (Tüm Gen Dizi Analizi)	IPO11 gene (Sequence Analysis-All Coding Exons)	IPO11	IPO11, RANBP11, SLRN	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPMK geni (Tüm Gen Dizi Analizi)	IPMK gene (Sequence Analysis-All Coding Exons)	IPMK	IPMK	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IP6K3 geni (Tüm Gen Dizi Analizi)	IP6K3 gene (Sequence Analysis-All Coding Exons)	IP6K3	IHPK3, INSP6K3, IP6K3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IP6K2 geni (Tüm Gen Dizi Analizi)	IP6K2 gene (Sequence Analysis-All Coding Exons)	IP6K2	IHPK2, IP6K2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IP6K1 geni (Tüm Gen Dizi Analizi)	IP6K1 gene (Sequence Analysis-All Coding Exons)	IP6K1	IHPK1, IP6K1, KIAA0263	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTU geni (Tüm Gen Dizi Analizi)	INTU gene (Sequence Analysis-All Coding Exons)	INTU	INTU, KIAA1284, PDZK6	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS9 geni (Tüm Gen Dizi Analizi)	INTS9 gene (Sequence Analysis-All Coding Exons)	INTS9	INTS9, INT9, RC74	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS8 geni (Tüm Gen Dizi Analizi)	INTS8 gene (Sequence Analysis-All Coding Exons)	INTS8	INTS8, INT8	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS7 geni (Tüm Gen Dizi Analizi)	INTS7 gene (Sequence Analysis-All Coding Exons)	INTS7	INTS7, INT7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS6 geni (Tüm Gen Dizi Analizi)	INTS6 gene (Sequence Analysis-All Coding Exons)	INTS6	INTS6, DDX26, DICE1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS5 geni (Tüm Gen Dizi Analizi)	INTS5 gene (Sequence Analysis-All Coding Exons)	INTS5	INTS5, INT5, KIAA1698	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS4 geni (Tüm Gen Dizi Analizi)	INTS4 gene (Sequence Analysis-All Coding Exons)	INTS4	INTS4, INT4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS3 geni (Tüm Gen Dizi Analizi)	INTS3 gene (Sequence Analysis-All Coding Exons)	INTS3	INTS3, INT3, SOSSA	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS2 geni (Tüm Gen Dizi Analizi)	INTS2 gene (Sequence Analysis-All Coding Exons)	INTS2	INTS2, INT2, KIAA1287	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS13 geni (Tüm Gen Dizi Analizi)	INTS13 gene (Sequence Analysis-All Coding Exons)	INTS13	ASUN, MAT89BB, GCT1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS12 geni (Tüm Gen Dizi Analizi)	INTS12 gene (Sequence Analysis-All Coding Exons)	INTS12	INTS12, INT12	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPTC geni (Tüm Gen Dizi Analizi)	OPTC gene (Sequence Analysis-All Coding Exons)	OPTC		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS11 geni (Tüm Gen Dizi Analizi)	INTS11 gene (Sequence Analysis-All Coding Exons)	INTS11	CPSF3L, INTS11, RC68	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

INTS10 geni (Tüm Gen Dizi Analizi)	INTS10 gene (Sequence Analysis-All Coding Exons)	INTS10	INTS10, INT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OPRL1 geni (Tüm Gen Dizi Analizi)	OPRL1 gene (Sequence Analysis-All Coding Exons)	OPRL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INTS1 geni (Tüm Gen Dizi Analizi)	INTS1 gene (Sequence Analysis-All Coding Exons)	INTS1	INTS1, INT1, KIAA1440	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSRR geni (Tüm Gen Dizi Analizi)	INSRR gene (Sequence Analysis-All Coding Exons)	INSRR	INSRR, IRR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSM2 geni (Tüm Gen Dizi Analizi)	INSM2 gene (Sequence Analysis-All Coding Exons)	INSM2	INSM2, IA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSM1 geni (Tüm Gen Dizi Analizi)	INSM1 gene (Sequence Analysis-All Coding Exons)	INSM1	INSM1, IA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSL6 geni (Tüm Gen Dizi Analizi)	INSL6 gene (Sequence Analysis-All Coding Exons)	INSL6	INSL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSL5 geni (Tüm Gen Dizi Analizi)	INSL5 gene (Sequence Analysis-All Coding Exons)	INSL5	INSL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSL4 geni (Tüm Gen Dizi Analizi)	INSL4 gene (Sequence Analysis-All Coding Exons)	INSL4	INSL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSIG2 geni (Tüm Gen Dizi Analizi)	INSIG2 gene (Sequence Analysis-All Coding Exons)	INSIG2	INSIG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSIG1 geni (Tüm Gen Dizi Analizi)	INSIG1 gene (Sequence Analysis-All Coding Exons)	INSIG1	INSIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INSC geni (Tüm Gen Dizi Analizi)	INSC gene (Sequence Analysis-All Coding Exons)	INSC	INSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5K geni (Tüm Gen Dizi Analizi)	INPP5K gene (Sequence Analysis-All Coding Exons)	INPP5K	INPP5K, SKIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5J geni (Tüm Gen Dizi Analizi)	INPP5J gene (Sequence Analysis-All Coding Exons)	INPP5J	INPP5J, PIB5PA, PIPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5F geni (Tüm Gen Dizi Analizi)	INPP5F gene (Sequence Analysis-All Coding Exons)	INPP5F	INPP5F, SAC2, KIAA0966	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5D geni (Tüm Gen Dizi Analizi)	INPP5D gene (Sequence Analysis-All Coding Exons)	INPP5D	INPP5D, SHIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5B geni (Tüm Gen Dizi Analizi)	INPP5B gene (Sequence Analysis-All Coding Exons)	INPP5B	INPP5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP5A geni (Tüm Gen Dizi Analizi)	INPP5A gene (Sequence Analysis-All Coding Exons)	INPP5A	INPP5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP4A geni (Tüm Gen Dizi Analizi)	INPP4A gene (Sequence Analysis-All Coding Exons)	INPP4A	INPP4A, INPP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP1 geni (Tüm Gen Dizi Analizi)	INPP1 gene (Sequence Analysis-All Coding Exons)	INPP1	INPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INO80B geni (Tüm Gen Dizi Analizi)	INO80B gene (Sequence Analysis-All Coding Exons)	INO80B	INO80B, PAPA1, HMGA1L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INO80 geni (Tüm Gen Dizi Analizi)	INO80 gene (Sequence Analysis-All Coding Exons)	INO80	INO80, INOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INMT geni (Tüm Gen Dizi Analizi)	INMT gene (Sequence Analysis-All Coding Exons)	INMT	INMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INIP geni (Tüm Gen Dizi Analizi)	INIP gene (Sequence Analysis-All Coding Exons)	INIP	C9orf80, SOSSC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INHBE geni (Tüm Gen Dizi Analizi)	INHBE gene (Sequence Analysis-All Coding Exons)	INHBE	INHBE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INHBC geni (Tüm Gen Dizi Analizi)	INHBC gene (Sequence Analysis-All Coding Exons)	INHBC	INHBC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INHBB geni (Tüm Gen Dizi Analizi)	INHBB gene (Sequence Analysis-All Coding Exons)	INHBB	INHBB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INHBA geni (Tüm Gen Dizi Analizi)	INHBA gene (Sequence Analysis-All Coding Exons)	INHBA	INHBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INHA geni (Tüm Gen Dizi Analizi)	INHA gene (Sequence Analysis-All Coding Exons)	INHA	INHA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INGX geni (Tüm Gen Dizi Analizi)	INGX gene (Sequence Analysis-All Coding Exons)	INGX	INGX, ING2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ING5 geni (Tüm Gen Dizi Analizi)	ING5 gene (Sequence Analysis-All Coding Exons)	ING5	ING5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ING4 geni (Tüm Gen Dizi Analizi)	ING4 gene (Sequence Analysis-All Coding Exons)	ING4	ING4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ING3 geni (Tüm Gen Dizi Analizi)	ING3 gene (Sequence Analysis-All Coding Exons)	ING3	ING3, P47ING3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ING2 geni (Tüm Gen Dizi Analizi)	ING2 gene (Sequence Analysis-All Coding Exons)	ING2	ING2, ING1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INE2 geni (Tüm Gen Dizi Analizi)	INE2 gene (Sequence Analysis-All Coding Exons)	INE2	INE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INE1 geni (Tüm Gen Dizi Analizi)	INE1 gene (Sequence Analysis-All Coding Exons)	INE1	INE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INCENP geni (Tüm Gen Dizi Analizi)	INCENP gene (Sequence Analysis-All Coding Exons)	INCENP	INCENP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INCA1 geni (Tüm Gen Dizi Analizi)	INCA1 gene (Sequence Analysis-All Coding Exons)	INCA1	INCA1, HSD45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMPACT geni (Tüm Gen Dizi Analizi)	IMPACT gene (Sequence Analysis-All Coding Exons)	IMPACT	IMPACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMPA2 geni (Tüm Gen Dizi Analizi)	IMPA2 gene (Sequence Analysis-All Coding Exons)	IMPA2	IMPA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMP4 geni (Tüm Gen Dizi Analizi)	IMP4 gene (Sequence Analysis-All Coding Exons)	IMP4	IMP4, BXDC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OIP5 geni (Tüm Gen Dizi Analizi)	OIP5 gene (Sequence Analysis-All Coding Exons)	OIP5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMP3 geni (Tüm Gen Dizi Analizi)	IMP3 gene (Sequence Analysis-All Coding Exons)	IMP3	IMP3, MRPS4, BRMS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMMT geni (Tüm Gen Dizi Analizi)	IMMT gene (Sequence Analysis-All Coding Exons)	IMMT	IMMT, HMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMMP2L geni (Tüm Gen Dizi Analizi)	IMMP2L gene (Sequence Analysis-All Coding Exons)	IMMP2L	IMMP2L, IMP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OGFR geni (Tüm Gen Dizi Analizi)	OGFR gene (Sequence Analysis-All Coding Exons)	OGFR		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IMMP1L geni (Tüm Gen Dizi Analizi)	IMMP1L gene (Sequence Analysis-All Coding Exons)	IMMP1L	IMMP1L, IMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ILVBL geni (Tüm Gen Dizi Analizi)	ILVBL gene (Sequence Analysis-All Coding Exons)	ILVBL	ILVBL, AHAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ILK geni (Tüm Gen Dizi Analizi)	ILK gene (Sequence Analysis-All Coding Exons)	ILK	ILK, PS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ILF3 geni (Tüm Gen Dizi Analizi)	ILF3 gene (Sequence Analysis-All Coding Exons)	ILF3	ILF3, NF90, DRBP76, MPHOSPH4, NFAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL9R geni (Tüm Gen Dizi Analizi)	IL9R gene (Sequence Analysis-All Coding Exons)	IL9R	IL9R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL9 geni (Tüm Gen Dizi Analizi)	IL9 gene (Sequence Analysis-All Coding Exons)	IL9	IL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OFCC1 geni (Tüm Gen Dizi Analizi)	OFCC1 gene (Sequence Analysis-All Coding Exons)	OFCC1	OFCC1, MRDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL7 geni (Tüm Gen Dizi Analizi)	IL7 gene (Sequence Analysis-All Coding Exons)	IL7	IL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL6ST geni (Tüm Gen Dizi Analizi)	IL6ST gene (Sequence Analysis-All Coding Exons)	IL6ST	IL6ST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL5RA geni (Tüm Gen Dizi Analizi)	IL5RA gene (Sequence Analysis-All Coding Exons)	IL5RA	IL5RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL5 geni (Tüm Gen Dizi Analizi)	IL5 gene (Sequence Analysis-All Coding Exons)	IL5	IL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL4I1 geni (Tüm Gen Dizi Analizi)	IL4I1 gene (Sequence Analysis-All Coding Exons)	IL4I1	IL4I1, FIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL4 geni (Tüm Gen Dizi Analizi)	IL4 gene (Sequence Analysis-All Coding Exons)	IL4	IL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL3RA geni (Tüm Gen Dizi Analizi)	IL3RA gene (Sequence Analysis-All Coding Exons)	IL3RA	IL3RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL3RA geni (Tüm Gen Dizi Analizi)	IL3RA gene (Sequence Analysis-All Coding Exons)	IL3RA	IL3RA, IL3RY, IL3RAY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL37 geni (Tüm Gen Dizi Analizi)	IL37 gene (Sequence Analysis-All Coding Exons)	IL37	IL37, IL1F7, FIL1Z, IL1H4, IL1RP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL36G geni (Tüm Gen Dizi Analizi)	IL36G gene (Sequence Analysis-All Coding Exons)	IL36G	IL36G, IL1F9, IL1H1, IL1RP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OCLM geni (Tüm Gen Dizi Analizi)	OCLM gene (Sequence Analysis-All Coding Exons)	OCLM		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL36B geni (Tüm Gen Dizi Analizi)	IL36B gene (Sequence Analysis-All Coding Exons)	IL36B	IL36B, IL1F8, IL1H2, FIL1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL36A geni (Tüm Gen Dizi Analizi)	IL36A gene (Sequence Analysis-All Coding Exons)	IL36A	IL36A, IL1F6, FIL1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL34 geni (Tüm Gen Dizi Analizi)	IL34 gene (Sequence Analysis-All Coding Exons)	IL34	IL34, C16orf77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL33 geni (Tüm Gen Dizi Analizi)	IL33 gene (Sequence Analysis-All Coding Exons)	IL33	IL33, C9orf26, NFEHEV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL32 geni (Tüm Gen Dizi Analizi)	IL32 gene (Sequence Analysis-All Coding Exons)	IL32	IL32, NK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL31 geni (Tüm Gen Dizi Analizi)	IL31 gene (Sequence Analysis-All Coding Exons)	IL31	IL31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
OBSCN geni (Tüm Gen Dizi Analizi)	OBSCN gene (Sequence Analysis-All Coding Exons)	OBSCN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL3 geni (Tüm Gen Dizi Analizi)	IL3 gene (Sequence Analysis-All Coding Exons)	IL3	IL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL2RB geni (Tüm Gen Dizi Analizi)	IL2RB gene (Sequence Analysis-All Coding Exons)	IL2RB	IL2RB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL2RB geni (Tüm Gen Dizi Analizi)	IL2RB gene (Sequence Analysis-All Coding Exons)	IL2RB	IL2RB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL27RA geni (Tüm Gen Dizi Analizi)	IL27RA gene (Sequence Analysis-All Coding Exons)	IL27RA	TCCR, WSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL27 geni (Tüm Gen Dizi Analizi)	IL27 gene (Sequence Analysis-All Coding Exons)	IL27	IL27, IL30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL26 geni (Tüm Gen Dizi Analizi)	IL26 gene (Sequence Analysis-All Coding Exons)	IL26	IL26, AK155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL25 geni (Tüm Gen Dizi Analizi)	IL25 gene (Sequence Analysis-All Coding Exons)	IL25	IL25, IL17E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL24 geni (Tüm Gen Dizi Analizi)	IL24 gene (Sequence Analysis-All Coding Exons)	IL24	IL24, ST16, MDA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL23A geni (Tüm Gen Dizi Analizi)	IL23A gene (Sequence Analysis-All Coding Exons)	IL23A	IL23A, SGRF, P19, IL23P19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL22RA2 geni (Tüm Gen Dizi Analizi)	IL22RA2 gene (Sequence Analysis-All Coding Exons)	IL22RA2	IL22BP, IL22RA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL22 geni (Tüm Gen Dizi Analizi)	IL22 gene (Sequence Analysis-All Coding Exons)	IL22	IL22, IL21, ILTIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL20RA geni (Tüm Gen Dizi Analizi)	IL20RA gene (Sequence Analysis-All Coding Exons)	IL20RA	IL20RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL20 geni (Tüm Gen Dizi Analizi)	IL20 gene (Sequence Analysis-All Coding Exons)	IL20	IL20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL2 geni (Tüm Gen Dizi Analizi)	IL2 gene (Sequence Analysis-All Coding Exons)	IL2	IL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving IL2 is found in a form of T-cell acute lymphoblastic leukemia (T-ALL). Translocation t(4;16)(q26;p13) with involves TNFRSF17. [ECO:0000269|PubMed:1396583].	EDTA Blood Sample
IL1RL2 geni (Tüm Gen Dizi Analizi)	IL1RL2 gene (Sequence Analysis-All Coding Exons)	IL1RL2	IL1RL2, IL1RRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1RL1 geni (Tüm Gen Dizi Analizi)	IL1RL1 gene (Sequence Analysis-All Coding Exons)	IL1RL1	IL1RL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1RAPL2 geni (Tüm Gen Dizi Analizi)	IL1RAPL2 gene (Sequence Analysis-All Coding Exons)	IL1RAPL2	IL1RAPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1RAP geni (Tüm Gen Dizi Analizi)	IL1RAP gene (Sequence Analysis-All Coding Exons)	IL1RAP	IL1RAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1R2 geni (Tüm Gen Dizi Analizi)	IL1R2 gene (Sequence Analysis-All Coding Exons)	IL1R2	IL1R2, IL1RB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1R1 geni (Tüm Gen Dizi Analizi)	IL1R1 gene (Sequence Analysis-All Coding Exons)	IL1R1	IL1R1, IL1RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL1F10 geni (Tüm Gen Dizi Analizi)	IL1F10 gene (Sequence Analysis-All Coding Exons)	IL1F10	IL1F10, IL1HY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

IL1A geni (Tüm Gen Dizi Analizi)	IL1A gene (Sequence Analysis-All Coding Exons)	IL1A	IL1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL19 geni (Tüm Gen Dizi Analizi)	IL19 gene (Sequence Analysis-All Coding Exons)	IL19	IL19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL18RAP geni (Tüm Gen Dizi Analizi)	IL18RAP gene (Sequence Analysis-All Coding Exons)	IL18RAP	IL18RAP, ACPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL18R1 geni (Tüm Gen Dizi Analizi)	IL18R1 gene (Sequence Analysis-All Coding Exons)	IL18R1	IL18R1, IL1RRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL18BP geni (Tüm Gen Dizi Analizi)	IL18BP gene (Sequence Analysis-All Coding Exons)	IL18BP	IL18BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL18 geni (Tüm Gen Dizi Analizi)	IL18 gene (Sequence Analysis-All Coding Exons)	IL18	IL18, IGIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17REL geni (Tüm Gen Dizi Analizi)	IL17REL gene (Sequence Analysis-All Coding Exons)	IL17REL	IL17REL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17RE geni (Tüm Gen Dizi Analizi)	IL17RE gene (Sequence Analysis-All Coding Exons)	IL17RE	IL17RE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17RB geni (Tüm Gen Dizi Analizi)	IL17RB gene (Sequence Analysis-All Coding Exons)	IL17RB	IL17RB, IL17BR, IL17RH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17D geni (Tüm Gen Dizi Analizi)	IL17D gene (Sequence Analysis-All Coding Exons)	IL17D	IL17D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUTM1 geni (Tüm Gen Dizi Analizi)	NUTM1 gene (Sequence Analysis-All Coding Exons)	NUTM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUTF2 geni (Tüm Gen Dizi Analizi)	NUTF2 gene (Sequence Analysis-All Coding Exons)	NUTF2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17C geni (Tüm Gen Dizi Analizi)	IL17C gene (Sequence Analysis-All Coding Exons)	IL17C	IL17C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17B geni (Tüm Gen Dizi Analizi)	IL17B gene (Sequence Analysis-All Coding Exons)	IL17B	IL17B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL17A geni (Tüm Gen Dizi Analizi)	IL17A gene (Sequence Analysis-All Coding Exons)	IL17A	IL17A, IL17, CTLA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL16 geni (Tüm Gen Dizi Analizi)	IL16 gene (Sequence Analysis-All Coding Exons)	IL16	IL16, LCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL15RA geni (Tüm Gen Dizi Analizi)	IL15RA gene (Sequence Analysis-All Coding Exons)	IL15RA	IL15RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL15 geni (Tüm Gen Dizi Analizi)	IL15 gene (Sequence Analysis-All Coding Exons)	IL15	IL15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL13RA2 geni (Tüm Gen Dizi Analizi)	IL13RA2 gene (Sequence Analysis-All Coding Exons)	IL13RA2	IL13RA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL13RA1 geni (Tüm Gen Dizi Analizi)	IL13RA1 gene (Sequence Analysis-All Coding Exons)	IL13RA1	IL13RA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL12RB2 geni (Tüm Gen Dizi Analizi)	IL12RB2 gene (Sequence Analysis-All Coding Exons)	IL12RB2	IL12RB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL12A geni (Tüm Gen Dizi Analizi)	IL12A gene (Sequence Analysis-All Coding Exons)	IL12A	IL12A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL12A geni (Tüm Gen Dizi Analizi)	IL12A gene (Sequence Analysis-All Coding Exons)	IL12A	IL12A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IL11 geni (Tüm Gen Dizi Analizi)	IL11 gene (Sequence Analysis-All Coding Exons)	IL11	IL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKZF4 geni (Tüm Gen Dizi Analizi)	IKZF4 gene (Sequence Analysis-All Coding Exons)	IKZF4	IKZF4, ZNFN1A4, EOS, KIAA1782	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKZF3 geni (Tüm Gen Dizi Analizi)	IKZF3 gene (Sequence Analysis-All Coding Exons)	IKZF3	IKZF3, ZNFN1A3, AIOLOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKZF2 geni (Tüm Gen Dizi Analizi)	IKZF2 gene (Sequence Analysis-All Coding Exons)	IKZF2	IKZF2, ANF1A2, HELIOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKBKE geni (Tüm Gen Dizi Analizi)	IKBKE gene (Sequence Analysis-All Coding Exons)	IKBKE	IKBKE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKBIP geni (Tüm Gen Dizi Analizi)	IKBIP gene (Sequence Analysis-All Coding Exons)	IKBIP	IKIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IK geni (Tüm Gen Dizi Analizi)	IK gene (Sequence Analysis-All Coding Exons)	IK	IK, RED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF9B geni (Tüm Gen Dizi Analizi)	IGSF9B gene (Sequence Analysis-All Coding Exons)	IGSF9B	IGSF9B, KIAA1030	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF9 geni (Tüm Gen Dizi Analizi)	IGSF9 gene (Sequence Analysis-All Coding Exons)	IGSF9	IGSF9, KIAA1355	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF8 geni (Tüm Gen Dizi Analizi)	IGSF8 gene (Sequence Analysis-All Coding Exons)	IGSF8	IGSF8, PGRL, CD81P3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF6 geni (Tüm Gen Dizi Analizi)	IGSF6 gene (Sequence Analysis-All Coding Exons)	IGSF6	IGSF6, DORA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF5 geni (Tüm Gen Dizi Analizi)	IGSF5 gene (Sequence Analysis-All Coding Exons)	IGSF5	JAM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF10 geni (Tüm Gen Dizi Analizi)	IGSF10 gene (Sequence Analysis-All Coding Exons)	IGSF10	IGSF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFN1 geni (Tüm Gen Dizi Analizi)	IGFN1 gene (Sequence Analysis-All Coding Exons)	IGFN1	IGFN1, EEF1A2BP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFLR1 geni (Tüm Gen Dizi Analizi)	IGFLR1 gene (Sequence Analysis-All Coding Exons)	IGFLR1	IGFLR1, TMEM149	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFL4 geni (Tüm Gen Dizi Analizi)	IGFL4 gene (Sequence Analysis-All Coding Exons)	IGFL4	IGFL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFL3 geni (Tüm Gen Dizi Analizi)	IGFL3 gene (Sequence Analysis-All Coding Exons)	IGFL3	IGFL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFL2 geni (Tüm Gen Dizi Analizi)	IGFL2 gene (Sequence Analysis-All Coding Exons)	IGFL2	IGFL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFL1 geni (Tüm Gen Dizi Analizi)	IGFL1 gene (Sequence Analysis-All Coding Exons)	IGFL1	IGFL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT9 geni (Tüm Gen Dizi Analizi)	NUDT9 gene (Sequence Analysis-All Coding Exons)	NUDT9		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT7 geni (Tüm Gen Dizi Analizi)	NUDT7 gene (Sequence Analysis-All Coding Exons)	NUDT7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT6 geni (Tüm Gen Dizi Analizi)	NUDT6 gene (Sequence Analysis-All Coding Exons)	NUDT6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT5 geni (Tüm Gen Dizi Analizi)	NUDT5 gene (Sequence Analysis-All Coding Exons)	NUDT5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT4 geni (Tüm Gen Dizi Analizi)	NUDT4 gene (Sequence Analysis-All Coding Exons)	NUDT4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT3 geni (Tüm Gen Dizi Analizi)	NUDT3 gene (Sequence Analysis-All Coding Exons)	NUDT3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBPL1 geni (Tüm Gen Dizi Analizi)	IGFBPL1 gene (Sequence Analysis-All Coding Exons)	IGFBPL1	IGFBPL1, IGFBPRP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP6 geni (Tüm Gen Dizi Analizi)	IGFBP6 gene (Sequence Analysis-All Coding Exons)	IGFBP6	IGFBP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP5 geni (Tüm Gen Dizi Analizi)	IGFBP5 gene (Sequence Analysis-All Coding Exons)	IGFBP5	IGFBP5, IBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP4 geni (Tüm Gen Dizi Analizi)	IGFBP4 gene (Sequence Analysis-All Coding Exons)	IGFBP4	IGFBP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP3 geni (Tüm Gen Dizi Analizi)	IGFBP3 gene (Sequence Analysis-All Coding Exons)	IGFBP3	IGFBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP2 geni (Tüm Gen Dizi Analizi)	IGFBP2 gene (Sequence Analysis-All Coding Exons)	IGFBP2	IGFBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT14 geni (Tüm Gen Dizi Analizi)	NUDT14 gene (Sequence Analysis-All Coding Exons)	NUDT14		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT13 geni (Tüm Gen Dizi Analizi)	NUDT13 gene (Sequence Analysis-All Coding Exons)	NUDT13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NUDT12 geni (Tüm Gen Dizi Analizi)	NUDT12 gene (Sequence Analysis-All Coding Exons)	NUDT12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGFBP1 geni (Tüm Gen Dizi Analizi)	IGFBP1 gene (Sequence Analysis-All Coding Exons)	IGFBP1	IGFBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGF2BP3 geni (Tüm Gen Dizi Analizi)	IGF2BP3 gene (Sequence Analysis-All Coding Exons)	IGF2BP3	IGF2BP3, IMP3, KOC1, MGC71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGF2BP1 geni (Tüm Gen Dizi Analizi)	IGF2BP1 gene (Sequence Analysis-All Coding Exons)	IGF2BP1	IGF2BP1, IMP1, CRDBP, ZBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGF2-AS geni (Tüm Gen Dizi Analizi)	IGF2-AS gene (Sequence Analysis-All Coding Exons)	IGF2-AS	IGF2AS, PEG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGDCC4 geni (Tüm Gen Dizi Analizi)	IGDCC4 gene (Sequence Analysis-All Coding Exons)	IGDCC4	IGDCC4, NOPE, KIAA1628	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGDCC3 geni (Tüm Gen Dizi Analizi)	IGDCC3 gene (Sequence Analysis-All Coding Exons)	IGDCC3	PUNC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFT88 geni (Tüm Gen Dizi Analizi)	IFT88 gene (Sequence Analysis-All Coding Exons)	IFT88	IFT88, D13S1056E, TG737	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFTB1 geni (Tüm Gen Dizi Analizi)	IFTB1 gene (Sequence Analysis-All Coding Exons)	IFTB1	IFTB1, DV1, CDV1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFT57 geni (Tüm Gen Dizi Analizi)	IFT57 gene (Sequence Analysis-All Coding Exons)	IFT57	IFT57, ESRRBL1, HIPPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFT20 geni (Tüm Gen Dizi Analizi)	IFT20 gene (Sequence Analysis-All Coding Exons)	IFT20	IFT20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFRD2 geni (Tüm Gen Dizi Analizi)	IFRD2 gene (Sequence Analysis-All Coding Exons)	IFRD2	IFRD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFRD1 geni (Tüm Gen Dizi Analizi)	IFRD1 gene (Sequence Analysis-All Coding Exons)	IFRD1	IFRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNW1 geni (Tüm Gen Dizi Analizi)	IFNW1 gene (Sequence Analysis-All Coding Exons)	IFNW1	IFNW1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNLR1 geni (Tüm Gen Dizi Analizi)	IFNLR1 gene (Sequence Analysis-All Coding Exons)	IFNLR1	IFNLR1, IL28RA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNL4 geni (Tüm Gen Dizi Analizi)	IFNL4 gene (Sequence Analysis-All Coding Exons)	IFNL4	IFNL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNL2 geni (Tüm Gen Dizi Analizi)	IFNL2 gene (Sequence Analysis-All Coding Exons)	IFNL2	IFNL2, IL28A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNL1 geni (Tüm Gen Dizi Analizi)	IFNL1 gene (Sequence Analysis-All Coding Exons)	IFNL1	IFNL1, IL29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNK geni (Tüm Gen Dizi Analizi)	IFNK gene (Sequence Analysis-All Coding Exons)	IFNK	IFNK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNE geni (Tüm Gen Dizi Analizi)	IFNE gene (Sequence Analysis-All Coding Exons)	IFNE	IFNE, INFE1, IFNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNB1 geni (Tüm Gen Dizi Analizi)	IFNB1 gene (Sequence Analysis-All Coding Exons)	IFNB1	IFNB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNAR1 geni (Tüm Gen Dizi Analizi)	IFNAR1 gene (Sequence Analysis-All Coding Exons)	IFNAR1	IFNAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA8 geni (Tüm Gen Dizi Analizi)	IFNA8 gene (Sequence Analysis-All Coding Exons)	IFNA8	IFNA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA7 geni (Tüm Gen Dizi Analizi)	IFNA7 gene (Sequence Analysis-All Coding Exons)	IFNA7	IFNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA6 geni (Tüm Gen Dizi Analizi)	IFNA6 gene (Sequence Analysis-All Coding Exons)	IFNA6	IFNA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA5 geni (Tüm Gen Dizi Analizi)	IFNA5 gene (Sequence Analysis-All Coding Exons)	IFNA5	IFNA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA4 geni (Tüm Gen Dizi Analizi)	IFNA4 gene (Sequence Analysis-All Coding Exons)	IFNA4	IFNA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA21 geni (Tüm Gen Dizi Analizi)	IFNA21 gene (Sequence Analysis-All Coding Exons)	IFNA21	IFNA21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA2 geni (Tüm Gen Dizi Analizi)	IFNA2 gene (Sequence Analysis-All Coding Exons)	IFNA2	IFNA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA17 geni (Tüm Gen Dizi Analizi)	IFNA17 gene (Sequence Analysis-All Coding Exons)	IFNA17	IFNA17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA16 geni (Tüm Gen Dizi Analizi)	IFNA16 gene (Sequence Analysis-All Coding Exons)	IFNA16	IFNA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA14 geni (Tüm Gen Dizi Analizi)	IFNA14 gene (Sequence Analysis-All Coding Exons)	IFNA14	IFNA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA13 geni (Tüm Gen Dizi Analizi)	IFNA13 gene (Sequence Analysis-All Coding Exons)	IFNA13	IFNA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFNA10 geni (Tüm Gen Dizi Analizi)	IFNA10 gene (Sequence Analysis-All Coding Exons)	IFNA10	IFNA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFITM2 geni (Tüm Gen Dizi Analizi)	IFITM2 gene (Sequence Analysis-All Coding Exons)	IFITM2	IFITM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFITM1 geni (Tüm Gen Dizi Analizi)	IFITM1 gene (Sequence Analysis-All Coding Exons)	IFITM1	IFITM1, IFI17, LEU13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFIT5 geni (Tüm Gen Dizi Analizi)	IFIT5 gene (Sequence Analysis-All Coding Exons)	IFIT5	IFIT5, RI58, ISG58	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

IFIT3 geni (Tüm Gen Dizi Analizi)	IFIT3 gene (Sequence Analysis-All Coding Exons)	IFIT3	IFIT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFIT2 geni (Tüm Gen Dizi Analizi)	IFIT2 gene (Sequence Analysis-All Coding Exons)	IFIT2	IFIT2, IFI54, G10P2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFIT1 geni (Tüm Gen Dizi Analizi)	IFIT1 gene (Sequence Analysis-All Coding Exons)	IFIT1	IFIT1, IFI56, G10P1, IFNAI1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI6 geni (Tüm Gen Dizi Analizi)	IFI6 gene (Sequence Analysis-All Coding Exons)	IFI6	G1P3, IFI616	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI44L geni (Tüm Gen Dizi Analizi)	IFI44L gene (Sequence Analysis-All Coding Exons)	IFI44L	IFI44L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI44 geni (Tüm Gen Dizi Analizi)	IFI44 gene (Sequence Analysis-All Coding Exons)	IFI44	IFI44, P44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI35 geni (Tüm Gen Dizi Analizi)	IFI35 gene (Sequence Analysis-All Coding Exons)	IFI35	IFI35	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI30 geni (Tüm Gen Dizi Analizi)	IFI30 gene (Sequence Analysis-All Coding Exons)	IFI30	IFI30, GILT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI27L2 geni (Tüm Gen Dizi Analizi)	IFI27L2 gene (Sequence Analysis-All Coding Exons)	IFI27L2	IFI27L2, FAM14A, ISG12B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI27 geni (Tüm Gen Dizi Analizi)	IFI27 gene (Sequence Analysis-All Coding Exons)	IFI27	IFI27	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI16 geni (Tüm Gen Dizi Analizi)	IFI16 gene (Sequence Analysis-All Coding Exons)	IFI16	IFI16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFFO1 geni (Tüm Gen Dizi Analizi)	IFFO1 gene (Sequence Analysis-All Coding Exons)	IFFO1	IFFO1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IER5 geni (Tüm Gen Dizi Analizi)	IER5 gene (Sequence Analysis-All Coding Exons)	IER5	IER5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IER3 geni (Tüm Gen Dizi Analizi)	IER3 gene (Sequence Analysis-All Coding Exons)	IER3	IER3, DIF2, IEX1, PRG1, GLY96	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDO2 geni (Tüm Gen Dizi Analizi)	IDO2 gene (Sequence Analysis-All Coding Exons)	IDO2	IDO2, INDOL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDO1 geni (Tüm Gen Dizi Analizi)	IDO1 gene (Sequence Analysis-All Coding Exons)	IDO1	IDO1, INDO, IDO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDNK geni (Tüm Gen Dizi Analizi)	IDNK gene (Sequence Analysis-All Coding Exons)	IDNK	C9orf103	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDI2-AS1 geni (Tüm Gen Dizi Analizi)	IDI2-AS1 gene (Sequence Analysis-All Coding Exons)	IDI2-AS1	IDI2AS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDI2 geni (Tüm Gen Dizi Analizi)	IDI2 gene (Sequence Analysis-All Coding Exons)	IDI2	IDI2, IPP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDI1 geni (Tüm Gen Dizi Analizi)	IDI1 gene (Sequence Analysis-All Coding Exons)	IDI1	IDI1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDH3G geni (Tüm Gen Dizi Analizi)	IDH3G gene (Sequence Analysis-All Coding Exons)	IDH3G	IDH3G	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDH3A geni (Tüm Gen Dizi Analizi)	IDH3A gene (Sequence Analysis-All Coding Exons)	IDH3A	IDH3A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IDE geni (Tüm Gen Dizi Analizi)	IDE gene (Sequence Analysis-All Coding Exons)	IDE	IDE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ID4 geni (Tüm Gen Dizi Analizi)	ID4 gene (Sequence Analysis-All Coding Exons)	ID4	ID4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ID3 geni (Tüm Gen Dizi Analizi)	ID3 gene (Sequence Analysis-All Coding Exons)	ID3	ID3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ID2 geni (Tüm Gen Dizi Analizi)	ID2 gene (Sequence Analysis-All Coding Exons)	ID2	ID2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ID1 geni (Tüm Gen Dizi Analizi)	ID1 gene (Sequence Analysis-All Coding Exons)	ID1	ID1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICOSLG geni (Tüm Gen Dizi Analizi)	ICOSLG gene (Sequence Analysis-All Coding Exons)	ICOSLG	ICOSLG, B7H2, GL50, B7RP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICMT geni (Tüm Gen Dizi Analizi)	ICMT gene (Sequence Analysis-All Coding Exons)	ICMT	ICMT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICE2 geni (Tüm Gen Dizi Analizi)	ICE2 gene (Sequence Analysis-All Coding Exons)	ICE2	NARG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICAM5 geni (Tüm Gen Dizi Analizi)	ICAM5 gene (Sequence Analysis-All Coding Exons)	ICAM5	ICAM5, TLCN, TLN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICAM3 geni (Tüm Gen Dizi Analizi)	ICAM3 gene (Sequence Analysis-All Coding Exons)	ICAM3	ICAM3, CDW50	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICAM2 geni (Tüm Gen Dizi Analizi)	ICAM2 gene (Sequence Analysis-All Coding Exons)	ICAM2	ICAM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ICA1 geni (Tüm Gen Dizi Analizi)	ICA1 gene (Sequence Analysis-All Coding Exons)	ICA1	ICA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IBTK geni (Tüm Gen Dizi Analizi)	IBTK gene (Sequence Analysis-All Coding Exons)	IBTK	IBTK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IBSP geni (Tüm Gen Dizi Analizi)	IBSP gene (Sequence Analysis-All Coding Exons)	IBSP	IBSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IAPP geni (Tüm Gen Dizi Analizi)	IAPP gene (Sequence Analysis-All Coding Exons)	IAPP	IAPP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYPK geni (Tüm Gen Dizi Analizi)	HYPK gene (Sequence Analysis-All Coding Exons)	HYPK	HYPK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYOU1 geni (Tüm Gen Dizi Analizi)	HYOU1 gene (Sequence Analysis-All Coding Exons)	HYOU1	HYOU1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYKK geni (Tüm Gen Dizi Analizi)	HYKK gene (Sequence Analysis-All Coding Exons)	HYKK	AGPHD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYAL4 geni (Tüm Gen Dizi Analizi)	HYAL4 gene (Sequence Analysis-All Coding Exons)	HYAL4	HYAL4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYAL3 geni (Tüm Gen Dizi Analizi)	HYAL3 gene (Sequence Analysis-All Coding Exons)	HYAL3	HYAL3, LUCA3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HYAL2 geni (Tüm Gen Dizi Analizi)	HYAL2 gene (Sequence Analysis-All Coding Exons)	HYAL2	HYAL2, LUCA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HVCN1 geni (Tüm Gen Dizi Analizi)	HVCN1 gene (Sequence Analysis-All Coding Exons)	HVCN1	HVCN1, HV1, VSOP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HUS1B geni (Tüm Gen Dizi Analizi)	HUS1B gene (Sequence Analysis-All Coding Exons)	HUS1B	HUS1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HUS1 geni (Tüm Gen Dizi Analizi)	HUS1 gene (Sequence Analysis-All Coding Exons)	HUS1	HUS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HUNK geni (Tüm Gen Dizi Analizi)	HUNK gene (Sequence Analysis-All Coding Exons)	HUNK	HUNK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTRA4 geni (Tüm Gen Dizi Analizi)	HTRA4 gene (Sequence Analysis-All Coding Exons)	HTRA4	HTRA4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTRA3 geni (Tüm Gen Dizi Analizi)	HTRA3 gene (Sequence Analysis-All Coding Exons)	HTRA3	HTRA3, PRSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR7 geni (Tüm Gen Dizi Analizi)	HTR7 gene (Sequence Analysis-All Coding Exons)	HTR7	HTR7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR6 geni (Tüm Gen Dizi Analizi)	HTR6 gene (Sequence Analysis-All Coding Exons)	HTR6	HTR6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR5A geni (Tüm Gen Dizi Analizi)	HTR5A gene (Sequence Analysis-All Coding Exons)	HTR5A	HTR5A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR4 geni (Tüm Gen Dizi Analizi)	HTR4 gene (Sequence Analysis-All Coding Exons)	HTR4	HTR4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR3E geni (Tüm Gen Dizi Analizi)	HTR3E gene (Sequence Analysis-All Coding Exons)	HTR3E	HTR3E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR3D geni (Tüm Gen Dizi Analizi)	HTR3D gene (Sequence Analysis-All Coding Exons)	HTR3D	HTR3D	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR3C geni (Tüm Gen Dizi Analizi)	HTR3C gene (Sequence Analysis-All Coding Exons)	HTR3C	HTR3C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR3B geni (Tüm Gen Dizi Analizi)	HTR3B gene (Sequence Analysis-All Coding Exons)	HTR3B	HTR3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR3A geni (Tüm Gen Dizi Analizi)	HTR3A gene (Sequence Analysis-All Coding Exons)	HTR3A	HTR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR2C geni (Tüm Gen Dizi Analizi)	HTR2C gene (Sequence Analysis-All Coding Exons)	HTR2C	HTR2C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR2B geni (Tüm Gen Dizi Analizi)	HTR2B gene (Sequence Analysis-All Coding Exons)	HTR2B	HTR2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR1F geni (Tüm Gen Dizi Analizi)	HTR1F gene (Sequence Analysis-All Coding Exons)	HTR1F	HTR1F, HTR1EL, MR77	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR1E geni (Tüm Gen Dizi Analizi)	HTR1E gene (Sequence Analysis-All Coding Exons)	HTR1E	HTR1E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR1D geni (Tüm Gen Dizi Analizi)	HTR1D gene (Sequence Analysis-All Coding Exons)	HTR1D	HTR1D	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTR1B geni (Tüm Gen Dizi Analizi)	HTR1B gene (Sequence Analysis-All Coding Exons)	HTR1B	HTR1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTN3 geni (Tüm Gen Dizi Analizi)	HTN3 gene (Sequence Analysis-All Coding Exons)	HTN3	HTN3, HTN2, HIS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTN1 geni (Tüm Gen Dizi Analizi)	HTN1 gene (Sequence Analysis-All Coding Exons)	HTN1	HTN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTATSF1 geni (Tüm Gen Dizi Analizi)	HTATSF1 gene (Sequence Analysis-All Coding Exons)	HTATSF1	HTATSF1, TATSF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HTATIP2 geni (Tüm Gen Dizi Analizi)	HTATIP2 gene (Sequence Analysis-All Coding Exons)	HTATIP2	HTATIP2, TIP30, CC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPH1 geni (Tüm Gen Dizi Analizi)	HSPH1 gene (Sequence Analysis-All Coding Exons)	HSPH1	HSPH1, HSP105, KIAA0201	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPE1 geni (Tüm Gen Dizi Analizi)	HSPE1 gene (Sequence Analysis-All Coding Exons)	HSPE1	HSPE1, HSP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPBP1 geni (Tüm Gen Dizi Analizi)	HSPBP1 gene (Sequence Analysis-All Coding Exons)	HSPBP1	HSPBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPB9 geni (Tüm Gen Dizi Analizi)	HSPB9 gene (Sequence Analysis-All Coding Exons)	HSPB9	HSPB9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPB7 geni (Tüm Gen Dizi Analizi)	HSPB7 gene (Sequence Analysis-All Coding Exons)	HSPB7	HSPB7, CVHSP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPB6 geni (Tüm Gen Dizi Analizi)	HSPB6 gene (Sequence Analysis-All Coding Exons)	HSPB6	HSPB6, HSP20	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPB2 geni (Tüm Gen Dizi Analizi)	HSPB2 gene (Sequence Analysis-All Coding Exons)	HSPB2	HSPB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA8 geni (Tüm Gen Dizi Analizi)	HSPA8 gene (Sequence Analysis-All Coding Exons)	HSPA8	HSPA8, HSP73	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA6 geni (Tüm Gen Dizi Analizi)	HSPA6 gene (Sequence Analysis-All Coding Exons)	HSPA6	HSPA6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA5 geni (Tüm Gen Dizi Analizi)	HSPA5 gene (Sequence Analysis-All Coding Exons)	HSPA5	HSPA5, GRP78	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA4 geni (Tüm Gen Dizi Analizi)	HSPA4 gene (Sequence Analysis-All Coding Exons)	HSPA4	HSPA4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA2 geni (Tüm Gen Dizi Analizi)	HSPA2 gene (Sequence Analysis-All Coding Exons)	HSPA2	HSPA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA1L geni (Tüm Gen Dizi Analizi)	HSPA1L gene (Sequence Analysis-All Coding Exons)	HSPA1L	HSPA1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA1B geni (Tüm Gen Dizi Analizi)	HSPA1B gene (Sequence Analysis-All Coding Exons)	HSPA1B	HSPA1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA1A geni (Tüm Gen Dizi Analizi)	HSPA1A gene (Sequence Analysis-All Coding Exons)	HSPA1A	HSPA1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA14 geni (Tüm Gen Dizi Analizi)	HSPA14 gene (Sequence Analysis-All Coding Exons)	HSPA14	HSPA14, HSP70L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA13 geni (Tüm Gen Dizi Analizi)	HSPA13 gene (Sequence Analysis-All Coding Exons)	HSPA13	STCH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA12B geni (Tüm Gen Dizi Analizi)	HSPA12B gene (Sequence Analysis-All Coding Exons)	HSPA12B	HSPA12B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR2C2AP geni (Tüm Gen Dizi Analizi)	NR2C2AP gene (Sequence Analysis-All Coding Exons)	NR2C2AP		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPA12A geni (Tüm Gen Dizi Analizi)	HSPA12A gene (Sequence Analysis-All Coding Exons)	HSPA12A	HSPA12A, KIAA0417	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NR2C1 geni (Tüm Gen Dizi Analizi)	NR2C1 gene (Sequence Analysis-All Coding Exons)	NR2C1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSP90B1 geni (Tüm Gen Dizi Analizi)	HSP90B1 gene (Sequence Analysis-All Coding Exons)	HSP90B1	HSP90B1, TRA1, GRP94, GP96	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSP90AB1 geni (Tüm Gen Dizi Analizi)	HSP90AB1 gene (Sequence Analysis-All Coding Exons)	HSP90AB1	HSP90AB1, HSPC8, HSPC2, HSP90B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSP90AA1 geni (Tüm Gen Dizi Analizi)	HSP90AA1 gene (Sequence Analysis-All Coding Exons)	HSP90AA1	HSP90AA1, HSPC4, HSPC1, HSP90A, HSP89A, HSPCAL4, LAP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

NR1H3 geni (Tüm Gen Dizi Analizi)	NR1H3 gene (Sequence Analysis-All Coding Exons)	NR1H3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSFY1 geni (Tüm Gen Dizi Analizi)	HSFY1 gene (Sequence Analysis-All Coding Exons)	HSFY1	HSFY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSF2BP geni (Tüm Gen Dizi Analizi)	HSF2BP gene (Sequence Analysis-All Coding Exons)	HSF2BP	HSF2BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSF2 geni (Tüm Gen Dizi Analizi)	HSF2 gene (Sequence Analysis-All Coding Exons)	HSF2	HSF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSF1 geni (Tüm Gen Dizi Analizi)	HSF1 gene (Sequence Analysis-All Coding Exons)	HSF1	HSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD3B1 geni (Tüm Gen Dizi Analizi)	HSD3B1 gene (Sequence Analysis-All Coding Exons)	HSD3B1	HSD3B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B8 geni (Tüm Gen Dizi Analizi)	HSD17B8 gene (Sequence Analysis-All Coding Exons)	HSD17B8	HSD17B8, FABGL, D6S234E, HEPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B7 geni (Tüm Gen Dizi Analizi)	HSD17B7 gene (Sequence Analysis-All Coding Exons)	HSD17B7	HSD17B7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B6 geni (Tüm Gen Dizi Analizi)	HSD17B6 gene (Sequence Analysis-All Coding Exons)	HSD17B6	HSE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B2 geni (Tüm Gen Dizi Analizi)	HSD17B2 gene (Sequence Analysis-All Coding Exons)	HSD17B2	HSD17B2, EDH17B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B14 geni (Tüm Gen Dizi Analizi)	HSD17B14 gene (Sequence Analysis-All Coding Exons)	HSD17B14	HSD17B14, DHRS10, RETSDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B13 geni (Tüm Gen Dizi Analizi)	HSD17B13 gene (Sequence Analysis-All Coding Exons)	HSD17B13	HSD17B13, SCDR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B11 geni (Tüm Gen Dizi Analizi)	HSD17B11 gene (Sequence Analysis-All Coding Exons)	HSD17B11	HSD17B11, RETSDR2, PAN18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B1 geni (Tüm Gen Dizi Analizi)	HSD17B1 gene (Sequence Analysis-All Coding Exons)	HSD17B1	HSD17B1, EDH17B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSCB geni (Tüm Gen Dizi Analizi)	HSCB gene (Sequence Analysis-All Coding Exons)	HSCB	HSC20, HSCB, JAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSBP1 geni (Tüm Gen Dizi Analizi)	HSBP1 gene (Sequence Analysis-All Coding Exons)	HSBP1	HSBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS6ST3 geni (Tüm Gen Dizi Analizi)	HS6ST3 gene (Sequence Analysis-All Coding Exons)	HS6ST3	HS6ST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS6ST2 geni (Tüm Gen Dizi Analizi)	HS6ST2 gene (Sequence Analysis-All Coding Exons)	HS6ST2	HS6ST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST5 geni (Tüm Gen Dizi Analizi)	HS3ST5 gene (Sequence Analysis-All Coding Exons)	HS3ST5	HS3ST5, 3OST5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPTXR geni (Tüm Gen Dizi Analizi)	NPTXR gene (Sequence Analysis-All Coding Exons)	NPTXR		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST4 geni (Tüm Gen Dizi Analizi)	HS3ST4 gene (Sequence Analysis-All Coding Exons)	HS3ST4	HS3ST4, 3OST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST3B1 geni (Tüm Gen Dizi Analizi)	HS3ST3B1 gene (Sequence Analysis-All Coding Exons)	HS3ST3B1	HS3ST3B1, 3OST3B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST3A1 geni (Tüm Gen Dizi Analizi)	HS3ST3A1 gene (Sequence Analysis-All Coding Exons)	HS3ST3A1	HS3ST3A1, 3OST3A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST2 geni (Tüm Gen Dizi Analizi)	HS3ST2 gene (Sequence Analysis-All Coding Exons)	HS3ST2	HS3ST2, 3OST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS3ST1 geni (Tüm Gen Dizi Analizi)	HS3ST1 gene (Sequence Analysis-All Coding Exons)	HS3ST1	HS3ST1, 3OST, 3OST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS2ST1 geni (Tüm Gen Dizi Analizi)	HS2ST1 gene (Sequence Analysis-All Coding Exons)	HS2ST1	HS2ST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HS1BP3 geni (Tüm Gen Dizi Analizi)	HS1BP3 gene (Sequence Analysis-All Coding Exons)	HS1BP3	HS1BP3, FLJ14249, ETM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRNR geni (Tüm Gen Dizi Analizi)	HRNR gene (Sequence Analysis-All Coding Exons)	HRNR	HRNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRK geni (Tüm Gen Dizi Analizi)	HRK gene (Sequence Analysis-All Coding Exons)	HRK	HRK, DP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRH4 geni (Tüm Gen Dizi Analizi)	HRH4 gene (Sequence Analysis-All Coding Exons)	HRH4	HRH4, H4R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRH3 geni (Tüm Gen Dizi Analizi)	HRH3 gene (Sequence Analysis-All Coding Exons)	HRH3	HRH3, GPCR97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRH1 geni (Tüm Gen Dizi Analizi)	HRH1 gene (Sequence Analysis-All Coding Exons)	HRH1	HRH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRC geni (Tüm Gen Dizi Analizi)	HRC gene (Sequence Analysis-All Coding Exons)	HRC	HRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRASLS2 geni (Tüm Gen Dizi Analizi)	HRASLS2 gene (Sequence Analysis-All Coding Exons)	HRASLS2	HRASLS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRASLS geni (Tüm Gen Dizi Analizi)	HRASLS gene (Sequence Analysis-All Coding Exons)	HRASLS	HRASLS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPX geni (Tüm Gen Dizi Analizi)	HPX gene (Sequence Analysis-All Coding Exons)	HPX	HPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPVC1 geni (Tüm Gen Dizi Analizi)	HPVC1 gene (Sequence Analysis-All Coding Exons)	HPVC1	HPVC1, PESL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPSE geni (Tüm Gen Dizi Analizi)	HPSE gene (Sequence Analysis-All Coding Exons)	HPSE	HPSE, HSE1, HPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPR geni (Tüm Gen Dizi Analizi)	HPR gene (Sequence Analysis-All Coding Exons)	HPR	HPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPN geni (Tüm Gen Dizi Analizi)	HPN gene (Sequence Analysis-All Coding Exons)	HPN	HPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPGDS geni (Tüm Gen Dizi Analizi)	HPGDS gene (Sequence Analysis-All Coding Exons)	HPGDS	HPGDS, PGDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPF1 geni (Tüm Gen Dizi Analizi)	HPF1 gene (Sequence Analysis-All Coding Exons)	HPF1	HPF1, C4orf27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HPCAL1 geni (Tüm Gen Dizi Analizi)	HPCAL1 gene (Sequence Analysis-All Coding Exons)	HPCAL1	HPCAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HP1BP3 geni (Tüm Gen Dizi Analizi)	HP1BP3 gene (Sequence Analysis-All Coding Exons)	HP1BP3	HP1BP3, HP1BP74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD9 geni (Tüm Gen Dizi Analizi)	HOXD9 gene (Sequence Analysis-All Coding Exons)	HOXD9	HOXD9, HOX4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD8 geni (Tüm Gen Dizi Analizi)	HOXD8 gene (Sequence Analysis-All Coding Exons)	HOXD8	HOXD8, HOX4E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD4 geni (Tüm Gen Dizi Analizi)	HOXD4 gene (Sequence Analysis-All Coding Exons)	HOXD4	HOXD4, HOX4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD3 geni (Tüm Gen Dizi Analizi)	HOXD3 gene (Sequence Analysis-All Coding Exons)	HOXD3	HOXD3, HOX4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD12 geni (Tüm Gen Dizi Analizi)	HOXD12 gene (Sequence Analysis-All Coding Exons)	HOXD12	HOXD12, HOX4H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD11 geni (Tüm Gen Dizi Analizi)	HOXD11 gene (Sequence Analysis-All Coding Exons)	HOXD11	HOXD11, HOX4F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXD1 geni (Tüm Gen Dizi Analizi)	HOXD1 gene (Sequence Analysis-All Coding Exons)	HOXD1	HOXD1, HOX4G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC9 geni (Tüm Gen Dizi Analizi)	HOXC9 gene (Sequence Analysis-All Coding Exons)	HOXC9	HOXC9, HOX3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC8 geni (Tüm Gen Dizi Analizi)	HOXC8 gene (Sequence Analysis-All Coding Exons)	HOXC8	HOXC8, HOX3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC6 geni (Tüm Gen Dizi Analizi)	HOXC6 gene (Sequence Analysis-All Coding Exons)	HOXC6	HOXC6, HOX3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC5 geni (Tüm Gen Dizi Analizi)	HOXC5 gene (Sequence Analysis-All Coding Exons)	HOXC5	HOXC5, HOX3D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC4 geni (Tüm Gen Dizi Analizi)	HOXC4 gene (Sequence Analysis-All Coding Exons)	HOXC4	HOXC4, HOX3E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NPFF geni (Tüm Gen Dizi Analizi)	NPFF gene (Sequence Analysis-All Coding Exons)	NPFF		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC12 geni (Tüm Gen Dizi Analizi)	HOXC12 gene (Sequence Analysis-All Coding Exons)	HOXC12	HOXC12, HOX3F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC11 geni (Tüm Gen Dizi Analizi)	HOXC11 gene (Sequence Analysis-All Coding Exons)	HOXC11	HOXC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXC10 geni (Tüm Gen Dizi Analizi)	HOXC10 gene (Sequence Analysis-All Coding Exons)	HOXC10	HOXC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB9 geni (Tüm Gen Dizi Analizi)	HOXB9 gene (Sequence Analysis-All Coding Exons)	HOXB9	HOXB9, HOX2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB8 geni (Tüm Gen Dizi Analizi)	HOXB8 gene (Sequence Analysis-All Coding Exons)	HOXB8	HOXB8, HOX2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB7 geni (Tüm Gen Dizi Analizi)	HOXB7 gene (Sequence Analysis-All Coding Exons)	HOXB7	HOXB7, HOX2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB6 geni (Tüm Gen Dizi Analizi)	HOXB6 gene (Sequence Analysis-All Coding Exons)	HOXB6	HOXB6, HOX2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB5 geni (Tüm Gen Dizi Analizi)	HOXB5 gene (Sequence Analysis-All Coding Exons)	HOXB5	HOXB5, HOX2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB4 geni (Tüm Gen Dizi Analizi)	HOXB4 gene (Sequence Analysis-All Coding Exons)	HOXB4	HOXB4, HOX2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB3 geni (Tüm Gen Dizi Analizi)	HOXB3 gene (Sequence Analysis-All Coding Exons)	HOXB3	HOXB3, HOX2G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB2 geni (Tüm Gen Dizi Analizi)	HOXB2 gene (Sequence Analysis-All Coding Exons)	HOXB2	HOXB2, HOX2H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXB13 geni (Tüm Gen Dizi Analizi)	HOXB13 gene (Sequence Analysis-All Coding Exons)	HOXB13	HOXB13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA9 geni (Tüm Gen Dizi Analizi)	HOXA9 gene (Sequence Analysis-All Coding Exons)	HOXA9	HOXA9, HOX1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA7 geni (Tüm Gen Dizi Analizi)	HOXA7 gene (Sequence Analysis-All Coding Exons)	HOXA7	HOXA7, HOX1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA6 geni (Tüm Gen Dizi Analizi)	HOXA6 gene (Sequence Analysis-All Coding Exons)	HOXA6	HOXA6, HOX1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA5 geni (Tüm Gen Dizi Analizi)	HOXA5 gene (Sequence Analysis-All Coding Exons)	HOXA5	HOXA5, HOX1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA4 geni (Tüm Gen Dizi Analizi)	HOXA4 gene (Sequence Analysis-All Coding Exons)	HOXA4	HOXA4, HOX1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA3 geni (Tüm Gen Dizi Analizi)	HOXA3 gene (Sequence Analysis-All Coding Exons)	HOXA3	HOXA3, HOX1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA11-AS geni (Tüm Gen Dizi Analizi)	HOXA11-AS gene (Sequence Analysis-All Coding Exons)	HOXA11-AS	HOXA11AS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOXA10 geni (Tüm Gen Dizi Analizi)	HOXA10 gene (Sequence Analysis-All Coding Exons)	HOXA10	HOXA10, HOX1H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOTAIR geni (Tüm Gen Dizi Analizi)	HOTAIR gene (Sequence Analysis-All Coding Exons)	HOTAIR	HOTAIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HORMAD1 geni (Tüm Gen Dizi Analizi)	HORMAD1 gene (Sequence Analysis-All Coding Exons)	HORMAD1	HORMAD1, NOHMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOPX geni (Tüm Gen Dizi Analizi)	HOPX gene (Sequence Analysis-All Coding Exons)	HOPX	HOPX, HOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOOK3 geni (Tüm Gen Dizi Analizi)	HOOK3 gene (Sequence Analysis-All Coding Exons)	HOOK3	HOOK3, HK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOOK2 geni (Tüm Gen Dizi Analizi)	HOOK2 gene (Sequence Analysis-All Coding Exons)	HOOK2	HOOK2, HK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOOK1 geni (Tüm Gen Dizi Analizi)	HOOK1 gene (Sequence Analysis-All Coding Exons)	HOOK1	HOOK1, HK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOMEZ geni (Tüm Gen Dizi Analizi)	HOMEZ gene (Sequence Analysis-All Coding Exons)	HOMEZ	HOMEZ, KIAA1443	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOMER3 geni (Tüm Gen Dizi Analizi)	HOMER3 gene (Sequence Analysis-All Coding Exons)	HOMER3	HOMER3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HOMER1 geni (Tüm Gen Dizi Analizi)	HOMER1 gene (Sequence Analysis-All Coding Exons)	HOMER1	HOMER1, HOMER1B, HOMER1C, HOMER1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPUL1 geni (Tüm Gen Dizi Analizi)	HNRNPUL1 gene (Sequence Analysis-All Coding Exons)	HNRNPUL1	HNRNPUL1, HNRPUL1, E1B-AP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPR geni (Tüm Gen Dizi Analizi)	HNRNPR gene (Sequence Analysis-All Coding Exons)	HNRNPR	HNRPR, HNRNPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPM geni (Tüm Gen Dizi Analizi)	HNRNPM gene (Sequence Analysis-All Coding Exons)	HNRNPM	HNRPM, HNRPM4, NAGR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPLL geni (Tüm Gen Dizi Analizi)	HNRNPLL gene (Sequence Analysis-All Coding Exons)	HNRNPLL	HNRPLL, HNRPLL, SRRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPL geni (Tüm Gen Dizi Analizi)	HNRNPL gene (Sequence Analysis-All Coding Exons)	HNRNPL	HNRNPL, HNRPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPH3 geni (Tüm Gen Dizi Analizi)	HNRNPH3 gene (Sequence Analysis-All Coding Exons)	HNRNPH3	HNRPH3, 2H9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPH1 geni (Tüm Gen Dizi Analizi)	HNRNPH1 gene (Sequence Analysis-All Coding Exons)	HNRNPH1	HNRPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

HNRNPF geni (Tüm Gen Dizi Analizi)	HNRNPF gene (Sequence Analysis-All Coding Exons)	HNRNPF	HNRPF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPD geni (Tüm Gen Dizi Analizi)	HNRNPD gene (Sequence Analysis-All Coding Exons)	HNRNPD	HNRPD, AUF1, AUF1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPC geni (Tüm Gen Dizi Analizi)	HNRNPC gene (Sequence Analysis-All Coding Exons)	HNRNPC	HNRNPC, HNRPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPA3 geni (Tüm Gen Dizi Analizi)	HNRNPA3 gene (Sequence Analysis-All Coding Exons)	HNRNPA3	HNRPA3, D10S102, FBRNP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPA0 geni (Tüm Gen Dizi Analizi)	HNRNPA0 gene (Sequence Analysis-All Coding Exons)	HNRNPA0	HNRPA0	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNF4G geni (Tüm Gen Dizi Analizi)	HNF4G gene (Sequence Analysis-All Coding Exons)	HNF4G	HNF4G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMX3 geni (Tüm Gen Dizi Analizi)	HMX3 gene (Sequence Analysis-All Coding Exons)	HMX3	HMX3, NKX5.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMX2 geni (Tüm Gen Dizi Analizi)	HMX2 gene (Sequence Analysis-All Coding Exons)	HMX2	HMX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMSD geni (Tüm Gen Dizi Analizi)	HMSD gene (Sequence Analysis-All Coding Exons)	HMSD	HMSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMOX2 geni (Tüm Gen Dizi Analizi)	HMOX2 gene (Sequence Analysis-All Coding Exons)	HMOX2	HMOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMHB1 geni (Tüm Gen Dizi Analizi)	HMHB1 gene (Sequence Analysis-All Coding Exons)	HMHB1	HLA-HB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGXB4 geni (Tüm Gen Dizi Analizi)	HMGXB4 gene (Sequence Analysis-All Coding Exons)	HMGXB4	HMGXB4, HMG2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGN5 geni (Tüm Gen Dizi Analizi)	HMGN5 gene (Sequence Analysis-All Coding Exons)	HMGN5	NSBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGN3 geni (Tüm Gen Dizi Analizi)	HMGN3 gene (Sequence Analysis-All Coding Exons)	HMGN3	HMGN3, TRIP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGN2 geni (Tüm Gen Dizi Analizi)	HMGN2 gene (Sequence Analysis-All Coding Exons)	HMGN2	HMGN2, HMG17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGN1 geni (Tüm Gen Dizi Analizi)	HMGN1 gene (Sequence Analysis-All Coding Exons)	HMGN1	HMGN1, HMG14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGCS1 geni (Tüm Gen Dizi Analizi)	HMGCS1 gene (Sequence Analysis-All Coding Exons)	HMGCS1	HMGCS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGB4 geni (Tüm Gen Dizi Analizi)	HMGB4 gene (Sequence Analysis-All Coding Exons)	HMGB4	HMGB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGB2 geni (Tüm Gen Dizi Analizi)	HMGB2 gene (Sequence Analysis-All Coding Exons)	HMGB2	HMGB2, HMG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMGB1 geni (Tüm Gen Dizi Analizi)	HMGB1 gene (Sequence Analysis-All Coding Exons)	HMGB1	HMGB1, HMG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMG20B geni (Tüm Gen Dizi Analizi)	HMG20B gene (Sequence Analysis-All Coding Exons)	HMG20B	HMG20B, BRAF35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HMG20A geni (Tüm Gen Dizi Analizi)	HMG20A gene (Sequence Analysis-All Coding Exons)	HMG20A	HMG20A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HM13 geni (Tüm Gen Dizi Analizi)	HM13 gene (Sequence Analysis-All Coding Exons)	HM13	HM13, SPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLX geni (Tüm Gen Dizi Analizi)	HLX gene (Sequence Analysis-All Coding Exons)	HLX	HLX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLTF geni (Tüm Gen Dizi Analizi)	HLTF gene (Sequence Analysis-All Coding Exons)	HLTF	SMARCA3, SNF2L3, HIP116	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLF geni (Tüm Gen Dizi Analizi)	HLF gene (Sequence Analysis-All Coding Exons)	HLF	HLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-F geni (Tüm Gen Dizi Analizi)	HLA-F gene (Sequence Analysis-All Coding Exons)	HLA-F	HLA-F, HLA-CDA12, HLAF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-E geni (Tüm Gen Dizi Analizi)	HLA-E gene (Sequence Analysis-All Coding Exons)	HLA-E	HLA-E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DRB5 geni (Tüm Gen Dizi Analizi)	HLA-DRB5 gene (Sequence Analysis-All Coding Exons)	HLA-DRB5	HLA-DRB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DRA geni (Tüm Gen Dizi Analizi)	HLA-DRA gene (Sequence Analysis-All Coding Exons)	HLA-DRA	HLA-DRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DQB2 geni (Tüm Gen Dizi Analizi)	HLA-DQB2 gene (Sequence Analysis-All Coding Exons)	HLA-DQB2	HLA-DQB2, HLA-DXB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DQA2 geni (Tüm Gen Dizi Analizi)	HLA-DQA2 gene (Sequence Analysis-All Coding Exons)	HLA-DQA2	HLA-DQA2, HLA-DXA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DPA1 geni (Tüm Gen Dizi Analizi)	HLA-DPA1 gene (Sequence Analysis-All Coding Exons)	HLA-DPA1	HLA-DPA1, HLADP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DOB geni (Tüm Gen Dizi Analizi)	HLA-DOB gene (Sequence Analysis-All Coding Exons)	HLA-DOB	HLA-DOB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DOA geni (Tüm Gen Dizi Analizi)	HLA-DOA gene (Sequence Analysis-All Coding Exons)	HLA-DOA	HLA-DNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DMB geni (Tüm Gen Dizi Analizi)	HLA-DMB gene (Sequence Analysis-All Coding Exons)	HLA-DMB	HLA-DMB, RING7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DMA geni (Tüm Gen Dizi Analizi)	HLA-DMA gene (Sequence Analysis-All Coding Exons)	HLA-DMA	HLA-DMA, RING6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HKR1 geni (Tüm Gen Dizi Analizi)	HKR1 gene (Sequence Analysis-All Coding Exons)	HKR1	HKR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HKDC1 geni (Tüm Gen Dizi Analizi)	HKDC1 gene (Sequence Analysis-All Coding Exons)	HKDC1	HKDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HK3 geni (Tüm Gen Dizi Analizi)	HK3 gene (Sequence Analysis-All Coding Exons)	HK3	HK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HK2 geni (Tüm Gen Dizi Analizi)	HK2 gene (Sequence Analysis-All Coding Exons)	HK2	HK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HJURP geni (Tüm Gen Dizi Analizi)	HJURP gene (Sequence Analysis-All Coding Exons)	HJURP	HJURP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIVEP3 geni (Tüm Gen Dizi Analizi)	HIVEP3 gene (Sequence Analysis-All Coding Exons)	HIVEP3	HIVEP3, KRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIVEP1 geni (Tüm Gen Dizi Analizi)	HIVEP1 gene (Sequence Analysis-All Coding Exons)	HIVEP1	HIVEP1, ZNF40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST4H4 geni (Tüm Gen Dizi Analizi)	HIST4H4 gene (Sequence Analysis-All Coding Exons)	HIST4H4	HIST4H4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST3H3 geni (Tüm Gen Dizi Analizi)	HIST3H3 gene (Sequence Analysis-All Coding Exons)	HIST3H3	HIST3H3, H3F1, H3T	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NOB1 geni (Tüm Gen Dizi Analizi)	NOB1 gene (Sequence Analysis-All Coding Exons)	NOB1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST3H2BB geni (Tüm Gen Dizi Analizi)	HIST3H2BB gene (Sequence Analysis-All Coding Exons)	HIST3H2BB	HIST3H2BB, H2Bb	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST3H2A geni (Tüm Gen Dizi Analizi)	HIST3H2A gene (Sequence Analysis-All Coding Exons)	HIST3H2A	HIST3H2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H4A geni (Tüm Gen Dizi Analizi)	HIST2H4A gene (Sequence Analysis-All Coding Exons)	HIST2H4A	H4FN, H4F2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H3C geni (Tüm Gen Dizi Analizi)	HIST2H3C gene (Sequence Analysis-All Coding Exons)	HIST2H3C	HIST2H2C, H3F2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H2BE geni (Tüm Gen Dizi Analizi)	HIST2H2BE gene (Sequence Analysis-All Coding Exons)	HIST2H2BE	HIST2H2BE, H2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H2AC geni (Tüm Gen Dizi Analizi)	HIST2H2AC gene (Sequence Analysis-All Coding Exons)	HIST2H2AC	HIST2H2AC, H2AFQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMU geni (Tüm Gen Dizi Analizi)	NMU gene (Sequence Analysis-All Coding Exons)	NMU		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H2AB geni (Tüm Gen Dizi Analizi)	HIST2H2AB gene (Sequence Analysis-All Coding Exons)	HIST2H2AB	HIST2H2AB, H2AB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST2H2AA3 geni (Tüm Gen Dizi Analizi)	HIST2H2AA3 gene (Sequence Analysis-All Coding Exons)	HIST2H2AA3	HIST2H2AA, H2AFO, H2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMRK2 geni (Tüm Gen Dizi Analizi)	NMRK2 gene (Sequence Analysis-All Coding Exons)	NMRK2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NMRK1 geni (Tüm Gen Dizi Analizi)	NMRK1 gene (Sequence Analysis-All Coding Exons)	NMRK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4L geni (Tüm Gen Dizi Analizi)	HIST1H4L gene (Sequence Analysis-All Coding Exons)	HIST1H4L	HIST1H4L, H4FK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4K geni (Tüm Gen Dizi Analizi)	HIST1H4K gene (Sequence Analysis-All Coding Exons)	HIST1H4K	HIST1H4K, H4FD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4J geni (Tüm Gen Dizi Analizi)	HIST1H4J gene (Sequence Analysis-All Coding Exons)	HIST1H4J	HIST1H4J, H4FE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4I geni (Tüm Gen Dizi Analizi)	HIST1H4I gene (Sequence Analysis-All Coding Exons)	HIST1H4I	HIST1H4I, H4FM, H4M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4H geni (Tüm Gen Dizi Analizi)	HIST1H4H gene (Sequence Analysis-All Coding Exons)	HIST1H4H	HIST1H4H, H4FH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4G geni (Tüm Gen Dizi Analizi)	HIST1H4G gene (Sequence Analysis-All Coding Exons)	HIST1H4G	HIST1H4G, H4FL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4F geni (Tüm Gen Dizi Analizi)	HIST1H4F gene (Sequence Analysis-All Coding Exons)	HIST1H4F	HIST1H4F, H4FC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4E geni (Tüm Gen Dizi Analizi)	HIST1H4E gene (Sequence Analysis-All Coding Exons)	HIST1H4E	HIST1H4E, H4FJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4D geni (Tüm Gen Dizi Analizi)	HIST1H4D gene (Sequence Analysis-All Coding Exons)	HIST1H4D	HIST1H4D, H4FB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4C geni (Tüm Gen Dizi Analizi)	HIST1H4C gene (Sequence Analysis-All Coding Exons)	HIST1H4C	HIST1H4C, H4FG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4B geni (Tüm Gen Dizi Analizi)	HIST1H4B gene (Sequence Analysis-All Coding Exons)	HIST1H4B	HIST1H4B, H4FI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H4A geni (Tüm Gen Dizi Analizi)	HIST1H4A gene (Sequence Analysis-All Coding Exons)	HIST1H4A	HIST1H4A, H4FA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3J geni (Tüm Gen Dizi Analizi)	HIST1H3J gene (Sequence Analysis-All Coding Exons)	HIST1H3J	HIST1H3J, H3FJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3I geni (Tüm Gen Dizi Analizi)	HIST1H3I gene (Sequence Analysis-All Coding Exons)	HIST1H3I	HIST1H3I, H3FF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3H geni (Tüm Gen Dizi Analizi)	HIST1H3H gene (Sequence Analysis-All Coding Exons)	HIST1H3H	HIST1H3H, H3FK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3G geni (Tüm Gen Dizi Analizi)	HIST1H3G gene (Sequence Analysis-All Coding Exons)	HIST1H3G	HIST1H3G, H3FH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3F geni (Tüm Gen Dizi Analizi)	HIST1H3F gene (Sequence Analysis-All Coding Exons)	HIST1H3F	HIST1H3F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3E geni (Tüm Gen Dizi Analizi)	HIST1H3E gene (Sequence Analysis-All Coding Exons)	HIST1H3E	HIST1H3E, H3FD, H3.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3D geni (Tüm Gen Dizi Analizi)	HIST1H3D gene (Sequence Analysis-All Coding Exons)	HIST1H3D	HIST1H3D, H3FB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3C geni (Tüm Gen Dizi Analizi)	HIST1H3C gene (Sequence Analysis-All Coding Exons)	HIST1H3C	HIST1H3C, H3FC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3B geni (Tüm Gen Dizi Analizi)	HIST1H3B gene (Sequence Analysis-All Coding Exons)	HIST1H3B	HIST1H3B, H3FL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H3A geni (Tüm Gen Dizi Analizi)	HIST1H3A gene (Sequence Analysis-All Coding Exons)	HIST1H3A	HIST1H3A, H3FA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BO geni (Tüm Gen Dizi Analizi)	HIST1H2BO gene (Sequence Analysis-All Coding Exons)	HIST1H2BO	HIST1H2BO, H2BFN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BN geni (Tüm Gen Dizi Analizi)	HIST1H2BN gene (Sequence Analysis-All Coding Exons)	HIST1H2BN	HIST1H2BN, H2BFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BM geni (Tüm Gen Dizi Analizi)	HIST1H2BM gene (Sequence Analysis-All Coding Exons)	HIST1H2BM	HIST1H2BM, H2BFE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BL geni (Tüm Gen Dizi Analizi)	HIST1H2BL gene (Sequence Analysis-All Coding Exons)	HIST1H2BL	HIST1H2BL, H2BFC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BK geni (Tüm Gen Dizi Analizi)	HIST1H2BK gene (Sequence Analysis-All Coding Exons)	HIST1H2BK	HIST1H2BK, H2BK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BJ geni (Tüm Gen Dizi Analizi)	HIST1H2BJ gene (Sequence Analysis-All Coding Exons)	HIST1H2BJ	HIST1H2BJ, H2BJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BI geni (Tüm Gen Dizi Analizi)	HIST1H2BI gene (Sequence Analysis-All Coding Exons)	HIST1H2BI	HIST1H2BI, H2BFK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BH geni (Tüm Gen Dizi Analizi)	HIST1H2BH gene (Sequence Analysis-All Coding Exons)	HIST1H2BH	HIST1H2BH, H2BFJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BG geni (Tüm Gen Dizi Analizi)	HIST1H2BG gene (Sequence Analysis-All Coding Exons)	HIST1H2BG	HIST1H2BG, H2BFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BF geni (Tüm Gen Dizi Analizi)	HIST1H2BF gene (Sequence Analysis-All Coding Exons)	HIST1H2BF	HIST1H2BF, H2BFG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BE geni (Tüm Gen Dizi Analizi)	HIST1H2BE gene (Sequence Analysis-All Coding Exons)	HIST1H2BE	HIST1H2BE, H2BFH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BD geni (Tüm Gen Dizi Analizi)	HIST1H2BD gene (Sequence Analysis-All Coding Exons)	HIST1H2BD	HIST1H2BD, H2BFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BC geni (Tüm Gen Dizi Analizi)	HIST1H2BC gene (Sequence Analysis-All Coding Exons)	HIST1H2BC	HIST1H2BC, H2BFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

HIST1H2BB geni (Tüm Gen Dizi Analizi)	HIST1H2BB gene (Sequence Analysis-All Coding Exons)	HIST1H2BB	HIST1H2BB, H2BFF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2BA geni (Tüm Gen Dizi Analizi)	HIST1H2BA gene (Sequence Analysis-All Coding Exons)	HIST1H2BA	HIST1H2BA, TSH2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AM geni (Tüm Gen Dizi Analizi)	HIST1H2AM gene (Sequence Analysis-All Coding Exons)	HIST1H2AM	HIST1H2AM, H2AFN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AL geni (Tüm Gen Dizi Analizi)	HIST1H2AL gene (Sequence Analysis-All Coding Exons)	HIST1H2AL	HIST1H2AL, H2AFI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NLK geni (Tüm Gen Dizi Analizi)	NLK gene (Sequence Analysis-All Coding Exons)	NLK		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AK geni (Tüm Gen Dizi Analizi)	HIST1H2AK gene (Sequence Analysis-All Coding Exons)	HIST1H2AK	HIST1H2AK, HIST1H2AI, H2AFD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AJ geni (Tüm Gen Dizi Analizi)	HIST1H2AJ gene (Sequence Analysis-All Coding Exons)	HIST1H2AJ	HIST1H2AJ, HIST1H2AK, H2AFF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AI geni (Tüm Gen Dizi Analizi)	HIST1H2AI gene (Sequence Analysis-All Coding Exons)	HIST1H2AI	HIST1H2AI, H2AFC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AH geni (Tüm Gen Dizi Analizi)	HIST1H2AH gene (Sequence Analysis-All Coding Exons)	HIST1H2AH	HIST1H2AH, H2AH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AG geni (Tüm Gen Dizi Analizi)	HIST1H2AG gene (Sequence Analysis-All Coding Exons)	HIST1H2AG	HIST1H2AG, H2AG, H2AFP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AE geni (Tüm Gen Dizi Analizi)	HIST1H2AE gene (Sequence Analysis-All Coding Exons)	HIST1H2AE	HIST1H2AE, H2AFA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AD geni (Tüm Gen Dizi Analizi)	HIST1H2AD gene (Sequence Analysis-All Coding Exons)	HIST1H2AD	HIST1H2AD, H2AFG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AC geni (Tüm Gen Dizi Analizi)	HIST1H2AC gene (Sequence Analysis-All Coding Exons)	HIST1H2AC	HIST1H2AC, H2AFL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AB geni (Tüm Gen Dizi Analizi)	HIST1H2AB gene (Sequence Analysis-All Coding Exons)	HIST1H2AB	HIST1H2AB, H2AFM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H2AA geni (Tüm Gen Dizi Analizi)	HIST1H2AA gene (Sequence Analysis-All Coding Exons)	HIST1H2AA	HIST2H2AA, H2AFR, H2AA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1T geni (Tüm Gen Dizi Analizi)	HIST1H1T gene (Sequence Analysis-All Coding Exons)	HIST1H1T	HIST1H1T, H1FT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1E geni (Tüm Gen Dizi Analizi)	HIST1H1E gene (Sequence Analysis-All Coding Exons)	HIST1H1E	HIST1H1E, H1F4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1D geni (Tüm Gen Dizi Analizi)	HIST1H1D gene (Sequence Analysis-All Coding Exons)	HIST1H1D	HIST1H1D, H1F3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1C geni (Tüm Gen Dizi Analizi)	HIST1H1C gene (Sequence Analysis-All Coding Exons)	HIST1H1C	HIST1H1C, H1F2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1B geni (Tüm Gen Dizi Analizi)	HIST1H1B gene (Sequence Analysis-All Coding Exons)	HIST1H1B	HIST1H1B, H1F5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIST1H1A geni (Tüm Gen Dizi Analizi)	HIST1H1A gene (Sequence Analysis-All Coding Exons)	HIST1H1A	HIST1H1A, H1F1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIRIP3 geni (Tüm Gen Dizi Analizi)	HIRIP3 gene (Sequence Analysis-All Coding Exons)	HIRIP3	HIRIP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIRA geni (Tüm Gen Dizi Analizi)	HIRA gene (Sequence Analysis-All Coding Exons)	HIRA	HIRA, TUPLE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIPK4 geni (Tüm Gen Dizi Analizi)	HIPK4 gene (Sequence Analysis-All Coding Exons)	HIPK4	HIPK4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIPK3 geni (Tüm Gen Dizi Analizi)	HIPK3 gene (Sequence Analysis-All Coding Exons)	HIPK3	HIPK3, PKY, DYRK6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIPK2 geni (Tüm Gen Dizi Analizi)	HIPK2 gene (Sequence Analysis-All Coding Exons)	HIPK2	HIPK2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIP1R geni (Tüm Gen Dizi Analizi)	HIP1R gene (Sequence Analysis-All Coding Exons)	HIP1R	HIP1R, HIP12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HINT3 geni (Tüm Gen Dizi Analizi)	HINT3 gene (Sequence Analysis-All Coding Exons)	HINT3	HINT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HINT2 geni (Tüm Gen Dizi Analizi)	HINT2 gene (Sequence Analysis-All Coding Exons)	HINT2	HINT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HINFP geni (Tüm Gen Dizi Analizi)	HINFP gene (Sequence Analysis-All Coding Exons)	HINFP	MIZF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIF3A geni (Tüm Gen Dizi Analizi)	HIF3A gene (Sequence Analysis-All Coding Exons)	HIF3A	HIF3A, IPAS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIF1AN geni (Tüm Gen Dizi Analizi)	HIF1AN gene (Sequence Analysis-All Coding Exons)	HIF1AN	HIF1AN, FIH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIF1A-AS1 geni (Tüm Gen Dizi Analizi)	HIF1A-AS1 gene (Sequence Analysis-All Coding Exons)	HIF1A-AS1	HIF1AS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIF1A geni (Tüm Gen Dizi Analizi)	HIF1A gene (Sequence Analysis-All Coding Exons)	HIF1A	HIF1A, MOP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HID1 geni (Tüm Gen Dizi Analizi)	HID1 gene (Sequence Analysis-All Coding Exons)	HID1	C17orf28, DMC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIC2 geni (Tüm Gen Dizi Analizi)	HIC2 gene (Sequence Analysis-All Coding Exons)	HIC2	HIC2, HRG22, KIAA1020	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKIRAS1 geni (Tüm Gen Dizi Analizi)	NKIRAS1 gene (Sequence Analysis-All Coding Exons)	NKIRAS1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKG7 geni (Tüm Gen Dizi Analizi)	NKG7 gene (Sequence Analysis-All Coding Exons)	NKG7		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIC1 geni (Tüm Gen Dizi Analizi)	HIC1 gene (Sequence Analysis-All Coding Exons)	HIC1	HIC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIBADH geni (Tüm Gen Dizi Analizi)	HIBADH gene (Sequence Analysis-All Coding Exons)	HIBADH	HIBADH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHLA3 geni (Tüm Gen Dizi Analizi)	HHLA3 gene (Sequence Analysis-All Coding Exons)	HHLA3	HHLA3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHLA2 geni (Tüm Gen Dizi Analizi)	HHLA2 gene (Sequence Analysis-All Coding Exons)	HHLA2	HHLA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NKAIN3 geni (Tüm Gen Dizi Analizi)	NKAIN3 gene (Sequence Analysis-All Coding Exons)	NKAIN3	NKAIN3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHLA1 geni (Tüm Gen Dizi Analizi)	HHLA1 gene (Sequence Analysis-All Coding Exons)	HHLA1	HHLA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHIP geni (Tüm Gen Dizi Analizi)	HHIP gene (Sequence Analysis-All Coding Exons)	HHIP	HHIP, HIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHEX geni (Tüm Gen Dizi Analizi)	HHEX gene (Sequence Analysis-All Coding Exons)	HHEX	HHEX, PRHX, PRH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHATL geni (Tüm Gen Dizi Analizi)	HHATL gene (Sequence Analysis-All Coding Exons)	HHATL	HHATL, MBOAT3, C3orf3, KIAA1173	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HHAT geni (Tüm Gen Dizi Analizi)	HHAT gene (Sequence Analysis-All Coding Exons)	HHAT	HHAT, MART2, SKI1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NIPSNAP3B geni (Tüm Gen Dizi Analizi)	NIPSNAP3B gene (Sequence Analysis-All Coding Exons)	NIPSNAP3B		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HGS geni (Tüm Gen Dizi Analizi)	HGS gene (Sequence Analysis-All Coding Exons)	HGS	HGS, HRS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HGFAC geni (Tüm Gen Dizi Analizi)	HGFAC gene (Sequence Analysis-All Coding Exons)	HGFAC	HGFAC, HGFA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEYL geni (Tüm Gen Dizi Analizi)	HEYL gene (Sequence Analysis-All Coding Exons)	HEYL	HEYL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEY2 geni (Tüm Gen Dizi Analizi)	HEY2 gene (Sequence Analysis-All Coding Exons)	HEY2	HEY2, HERP1, GRL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEXIM2 geni (Tüm Gen Dizi Analizi)	HEXIM2 gene (Sequence Analysis-All Coding Exons)	HEXIM2	HEXIM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEXIM1 geni (Tüm Gen Dizi Analizi)	HEXIM1 gene (Sequence Analysis-All Coding Exons)	HEXIM1	CLP1, HIS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES6 geni (Tüm Gen Dizi Analizi)	HES6 gene (Sequence Analysis-All Coding Exons)	HES6	HES6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NINL geni (Tüm Gen Dizi Analizi)	NINL gene (Sequence Analysis-All Coding Exons)	NINL		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES5 geni (Tüm Gen Dizi Analizi)	HES5 gene (Sequence Analysis-All Coding Exons)	HES5	HES5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES4 geni (Tüm Gen Dizi Analizi)	HES4 gene (Sequence Analysis-All Coding Exons)	HES4	HES4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES3 geni (Tüm Gen Dizi Analizi)	HES3 gene (Sequence Analysis-All Coding Exons)	HES3	HES3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES2 geni (Tüm Gen Dizi Analizi)	HES2 gene (Sequence Analysis-All Coding Exons)	HES2	HES2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HES1 geni (Tüm Gen Dizi Analizi)	HES1 gene (Sequence Analysis-All Coding Exons)	HES1	HES1, HRY	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HERPUD1 geni (Tüm Gen Dizi Analizi)	HERPUD1 gene (Sequence Analysis-All Coding Exons)	HERPUD1	HERPUD1, MIF1, HERP, KIAA0025	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HERC5 geni (Tüm Gen Dizi Analizi)	HERC5 gene (Sequence Analysis-All Coding Exons)	HERC5	HERC5, CEB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HERC3 geni (Tüm Gen Dizi Analizi)	HERC3 gene (Sequence Analysis-All Coding Exons)	HERC3	HERC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEPN1 geni (Tüm Gen Dizi Analizi)	HEPN1 gene (Sequence Analysis-All Coding Exons)	HEPN1	HEPN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEPH geni (Tüm Gen Dizi Analizi)	HEPH gene (Sequence Analysis-All Coding Exons)	HEPH	HEPH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEPACAM2 geni (Tüm Gen Dizi Analizi)	HEPACAM2 gene (Sequence Analysis-All Coding Exons)	HEPACAM2	HEPACAM2, MIKI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HENMT1 geni (Tüm Gen Dizi Analizi)	HENMT1 gene (Sequence Analysis-All Coding Exons)	HENMT1	HENMT1, C1orf59, HEN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEMGN geni (Tüm Gen Dizi Analizi)	HEMGN gene (Sequence Analysis-All Coding Exons)	HEMGN	HEMGN, EDAG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HELZ2 geni (Tüm Gen Dizi Analizi)	HELZ2 gene (Sequence Analysis-All Coding Exons)	HELZ2	PRIC285, PDIP1, KIAA1769	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HELZ geni (Tüm Gen Dizi Analizi)	HELZ gene (Sequence Analysis-All Coding Exons)	HELZ	HELZ, KIAA0054, HUMORFS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HELQ geni (Tüm Gen Dizi Analizi)	HELQ gene (Sequence Analysis-All Coding Exons)	HELQ	HELQ, HEL308	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HELB geni (Tüm Gen Dizi Analizi)	HELB gene (Sequence Analysis-All Coding Exons)	HELB	HELB, HDHB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEG1 geni (Tüm Gen Dizi Analizi)	HEG1 gene (Sequence Analysis-All Coding Exons)	HEG1	HEG1, KIAA1237	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HECW1 geni (Tüm Gen Dizi Analizi)	HECW1 gene (Sequence Analysis-All Coding Exons)	HECW1	HECW1, NEDL1, KIAA0322	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HECA geni (Tüm Gen Dizi Analizi)	HECA gene (Sequence Analysis-All Coding Exons)	HECA	HECA, HDC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEBP2 geni (Tüm Gen Dizi Analizi)	HEBP2 gene (Sequence Analysis-All Coding Exons)	HEBP2	C6orf34, SOUL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEBP1 geni (Tüm Gen Dizi Analizi)	HEBP1 gene (Sequence Analysis-All Coding Exons)	HEBP1	HEBP1, HBP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEATR3 geni (Tüm Gen Dizi Analizi)	HEATR3 gene (Sequence Analysis-All Coding Exons)	HEATR3	HEATR3, SYO1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDLBP geni (Tüm Gen Dizi Analizi)	HDLBP gene (Sequence Analysis-All Coding Exons)	HDLBP	HDLBP, VGL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDGF geni (Tüm Gen Dizi Analizi)	HDGF gene (Sequence Analysis-All Coding Exons)	HDGF	HDGF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC9 geni (Tüm Gen Dizi Analizi)	HDAC9 gene (Sequence Analysis-All Coding Exons)	HDAC9	HDAC9, MITR, HDAC7B, KIAA0744	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC7 geni (Tüm Gen Dizi Analizi)	HDAC7 gene (Sequence Analysis-All Coding Exons)	HDAC7	HDAC7A, HDAC7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC5 geni (Tüm Gen Dizi Analizi)	HDAC5 gene (Sequence Analysis-All Coding Exons)	HDAC5	HDAC5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC4 geni (Tüm Gen Dizi Analizi)	HDAC4 gene (Sequence Analysis-All Coding Exons)	HDAC4	HDAC4, HDACA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC3 geni (Tüm Gen Dizi Analizi)	HDAC3 gene (Sequence Analysis-All Coding Exons)	HDAC3	HDAC3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC2 geni (Tüm Gen Dizi Analizi)	HDAC2 gene (Sequence Analysis-All Coding Exons)	HDAC2	HDAC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC1 geni (Tüm Gen Dizi Analizi)	HDAC1 gene (Sequence Analysis-All Coding Exons)	HDAC1	HDAC1, RPD3L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCST geni (Tüm Gen Dizi Analizi)	HCST gene (Sequence Analysis-All Coding Exons)	HCST	DAP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCRTR1 geni (Tüm Gen Dizi Analizi)	HCRTR1 gene (Sequence Analysis-All Coding Exons)	HCRTR1	HCRTR1, OX1R	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCP5 geni (Tüm Gen Dizi Analizi)	HCP5 gene (Sequence Analysis-All Coding Exons)	HCP5	HCP5, 6S2650E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCN3 geni (Tüm Gen Dizi Analizi)	HCN3 gene (Sequence Analysis-All Coding Exons)	HCN3	HCN3, KIAA1535	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

HCN2 geni (Tüm Gen Dizi Analizi)	HCN2 gene (Sequence Analysis-All Coding Exons)	HCN2	HCN2, BCNG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCLS1 geni (Tüm Gen Dizi Analizi)	HCLS1 gene (Sequence Analysis-All Coding Exons)	HCLS1	HCLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCK geni (Tüm Gen Dizi Analizi)	HCK gene (Sequence Analysis-All Coding Exons)	HCK	HCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCG9 geni (Tüm Gen Dizi Analizi)	HCG9 gene (Sequence Analysis-All Coding Exons)	HCG9	HCG9, HCGIX, HCGIX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCG22 geni (Tüm Gen Dizi Analizi)	HCG22 gene (Sequence Analysis-All Coding Exons)	HCG22	HCG22, PBMUCL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCFC2 geni (Tüm Gen Dizi Analizi)	HCFC2 gene (Sequence Analysis-All Coding Exons)	HCFC2	HCFC2, HCF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NFE4 geni (Tüm Gen Dizi Analizi)	NFE4 gene (Sequence Analysis-All Coding Exons)	NFE4	NFE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCCAT5 geni (Tüm Gen Dizi Analizi)	HCCAT5 gene (Sequence Analysis-All Coding Exons)	HCCAT5	HCCAT5, HTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCAR3 geni (Tüm Gen Dizi Analizi)	HCAR3 gene (Sequence Analysis-All Coding Exons)	HCAR3	HCAR3, GPR109B, HM74, PUMAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCAR2 geni (Tüm Gen Dizi Analizi)	HCAR2 gene (Sequence Analysis-All Coding Exons)	HCAR2	HCAR2, GPR109A, HM74A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCAR1 geni (Tüm Gen Dizi Analizi)	HCAR1 gene (Sequence Analysis-All Coding Exons)	HCAR1	HCAR1, GPR81, TAGPCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBZ geni (Tüm Gen Dizi Analizi)	HBZ gene (Sequence Analysis-All Coding Exons)	HBZ	HBZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBS1L geni (Tüm Gen Dizi Analizi)	HBS1L gene (Sequence Analysis-All Coding Exons)	HBS1L	HBS1L, KIAA1038, ERFS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBQ1 geni (Tüm Gen Dizi Analizi)	HBQ1 gene (Sequence Analysis-All Coding Exons)	HBQ1	HBQ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBP1 geni (Tüm Gen Dizi Analizi)	HBP1 gene (Sequence Analysis-All Coding Exons)	HBP1	HBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBM geni (Tüm Gen Dizi Analizi)	HBM gene (Sequence Analysis-All Coding Exons)	HBM	HBM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBE1 geni (Tüm Gen Dizi Analizi)	HBE1 gene (Sequence Analysis-All Coding Exons)	HBE1	HBE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBD geni (Tüm Gen Dizi Analizi)	HBD gene (Sequence Analysis-All Coding Exons)	HBD	HBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HBD geni (Tüm Gen Dizi Analizi)	HBD gene (Sequence Analysis-All Coding Exons)	HBD	HBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAVCR2 geni (Tüm Gen Dizi Analizi)	HAVCR2 gene (Sequence Analysis-All Coding Exons)	HAVCR2	TIM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS8 geni (Tüm Gen Dizi Analizi)	HAUS8 gene (Sequence Analysis-All Coding Exons)	HAUS8	HAUS8, DGT4, HCE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS7 geni (Tüm Gen Dizi Analizi)	HAUS7 gene (Sequence Analysis-All Coding Exons)	HAUS7	HAUS7, UCHL5IP, UIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS6 geni (Tüm Gen Dizi Analizi)	HAUS6 gene (Sequence Analysis-All Coding Exons)	HAUS6	HAUS6, DGT6, FAM29A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS5 geni (Tüm Gen Dizi Analizi)	HAUS5 gene (Sequence Analysis-All Coding Exons)	HAUS5	HAUS5, DGT5, KIAA0841	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS4 geni (Tüm Gen Dizi Analizi)	HAUS4 gene (Sequence Analysis-All Coding Exons)	HAUS4	HAUS4, C14orf94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS3 geni (Tüm Gen Dizi Analizi)	HAUS3 gene (Sequence Analysis-All Coding Exons)	HAUS3	HAUS3, DGT3, C4orf15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS2 geni (Tüm Gen Dizi Analizi)	HAUS2 gene (Sequence Analysis-All Coding Exons)	HAUS2	HAUS2, CEP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAUS1 geni (Tüm Gen Dizi Analizi)	HAUS1 gene (Sequence Analysis-All Coding Exons)	HAUS1	HAUS1, CCDC5, HEIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAT1 geni (Tüm Gen Dizi Analizi)	HAT1 gene (Sequence Analysis-All Coding Exons)	HAT1	HAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAS3 geni (Tüm Gen Dizi Analizi)	HAS3 gene (Sequence Analysis-All Coding Exons)	HAS3	HAS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAS2 geni (Tüm Gen Dizi Analizi)	HAS2 gene (Sequence Analysis-All Coding Exons)	HAS2	HAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAS1 geni (Tüm Gen Dizi Analizi)	HAS1 gene (Sequence Analysis-All Coding Exons)	HAS1	HAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HARBI1 geni (Tüm Gen Dizi Analizi)	HARBI1 gene (Sequence Analysis-All Coding Exons)	HARBI1	HARBI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAR1B geni (Tüm Gen Dizi Analizi)	HAR1B gene (Sequence Analysis-All Coding Exons)	HAR1B	HAR1B, HAR1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAPLN1 geni (Tüm Gen Dizi Analizi)	HAPLN1 gene (Sequence Analysis-All Coding Exons)	HAPLN1	HAPLN1, CRTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAP1 geni (Tüm Gen Dizi Analizi)	HAP1 gene (Sequence Analysis-All Coding Exons)	HAP1	HAP1, HLP, HAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAO2 geni (Tüm Gen Dizi Analizi)	HAO2 gene (Sequence Analysis-All Coding Exons)	HAO2	HAO2, HAOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAO1 geni (Tüm Gen Dizi Analizi)	HAO1 gene (Sequence Analysis-All Coding Exons)	HAO1	HAO1, GOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAND2-AS1 geni (Tüm Gen Dizi Analizi)	HAND2-AS1 gene (Sequence Analysis-All Coding Exons)	HAND2-AS1	HAND2AS1, UPH, DEIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEURL2 geni (Tüm Gen Dizi Analizi)	NEURL2 gene (Sequence Analysis-All Coding Exons)	NEURL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAND2 geni (Tüm Gen Dizi Analizi)	HAND2 gene (Sequence Analysis-All Coding Exons)	HAND2	HAND2, DHAND2, DHAND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HACL1 geni (Tüm Gen Dizi Analizi)	HACL1 gene (Sequence Analysis-All Coding Exons)	HACL1	HACL1, HPCL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HACD4 geni (Tüm Gen Dizi Analizi)	HACD4 gene (Sequence Analysis-All Coding Exons)	HACD4	PTPLAD2, HACD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HACD3 geni (Tüm Gen Dizi Analizi)	HACD3 gene (Sequence Analysis-All Coding Exons)	HACD3	PTPLAD1, BIND1, HACD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HACD2 geni (Tüm Gen Dizi Analizi)	HACD2 gene (Sequence Analysis-All Coding Exons)	HACD2	PTPLB, HACD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HACD1 geni (Tüm Gen Dizi Analizi)	HACD1 gene (Sequence Analysis-All Coding Exons)	HACD1	HACD1, PTPLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAAO geni (Tüm Gen Dizi Analizi)	HAAO gene (Sequence Analysis-All Coding Exons)	HAAO	HAAO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H3F3C geni (Tüm Gen Dizi Analizi)	H3F3C gene (Sequence Analysis-All Coding Exons)	H3F3C	H3F3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H3F3B geni (Tüm Gen Dizi Analizi)	H3F3B gene (Sequence Analysis-All Coding Exons)	H3F3B	H3F3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NET1 geni (Tüm Gen Dizi Analizi)	NET1 gene (Sequence Analysis-All Coding Exons)	NET1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H3F3A geni (Tüm Gen Dizi Analizi)	H3F3A gene (Sequence Analysis-All Coding Exons)	H3F3A	H3F3A, H3F3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2BFWT geni (Tüm Gen Dizi Analizi)	H2BFWT gene (Sequence Analysis-All Coding Exons)	H2BFWT	H2BFWT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2AFZ geni (Tüm Gen Dizi Analizi)	H2AFZ gene (Sequence Analysis-All Coding Exons)	H2AFZ	H2AZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2AFY2 geni (Tüm Gen Dizi Analizi)	H2AFY2 gene (Sequence Analysis-All Coding Exons)	H2AFY2	H2AFY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2AFY geni (Tüm Gen Dizi Analizi)	H2AFY gene (Sequence Analysis-All Coding Exons)	H2AFY	H2AFY, MH2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2AFX geni (Tüm Gen Dizi Analizi)	H2AFX gene (Sequence Analysis-All Coding Exons)	H2AFX	H2AX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H2AFB3 geni (Tüm Gen Dizi Analizi)	H2AFB3 gene (Sequence Analysis-All Coding Exons)	H2AFB3	H2AFB, H2ABBD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H1FX geni (Tüm Gen Dizi Analizi)	H1FX gene (Sequence Analysis-All Coding Exons)	H1FX	H1FX, H1X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
H1F0 geni (Tüm Gen Dizi Analizi)	H1F0 gene (Sequence Analysis-All Coding Exons)	H1F0	H1F0, H1FV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZMM geni (Tüm Gen Dizi Analizi)	GZMM gene (Sequence Analysis-All Coding Exons)	GZMM	GZMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZMH geni (Tüm Gen Dizi Analizi)	GZMH gene (Sequence Analysis-All Coding Exons)	GZMH	GZMH, CTSGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZMB geni (Tüm Gen Dizi Analizi)	GZMB gene (Sequence Analysis-All Coding Exons)	GZMB	GZMB, CTLA1, CSPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZMA geni (Tüm Gen Dizi Analizi)	GZMA gene (Sequence Analysis-All Coding Exons)	GZMA	GZMA, CTLA3, HFSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZF1 geni (Tüm Gen Dizi Analizi)	GZF1 gene (Sequence Analysis-All Coding Exons)	GZF1	GZF1, ZNF336	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GYPE geni (Tüm Gen Dizi Analizi)	GYPE gene (Sequence Analysis-All Coding Exons)	GYPE	GYPE, GPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GYG2 geni (Tüm Gen Dizi Analizi)	GYG2 gene (Sequence Analysis-All Coding Exons)	GYG2	GYG2, GN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GXYLT2 geni (Tüm Gen Dizi Analizi)	GXYLT2 gene (Sequence Analysis-All Coding Exons)	GXYLT2	GXYLT2, GLT8D4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GXYLT1 geni (Tüm Gen Dizi Analizi)	GXYLT1 gene (Sequence Analysis-All Coding Exons)	GXYLT1	GXYLT1, GLT8D3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GULP1 geni (Tüm Gen Dizi Analizi)	GULP1 gene (Sequence Analysis-All Coding Exons)	GULP1	GULP1, CED6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUK1 geni (Tüm Gen Dizi Analizi)	GUK1 gene (Sequence Analysis-All Coding Exons)	GUK1	GUK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCY2F geni (Tüm Gen Dizi Analizi)	GUCY2F gene (Sequence Analysis-All Coding Exons)	GUCY2F	GUCY2F, GUC2F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCY1B3 geni (Tüm Gen Dizi Analizi)	GUCY1B3 gene (Sequence Analysis-All Coding Exons)	GUCY1B3	GUCY1B3, GUC1B3, GUCSB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCY1B2 geni (Tüm Gen Dizi Analizi)	GUCY1B2 gene (Sequence Analysis-All Coding Exons)	GUCY1B2	GUCY1B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCY1A2 geni (Tüm Gen Dizi Analizi)	GUCY1A2 gene (Sequence Analysis-All Coding Exons)	GUCY1A2	GUCY1A2, GUC1A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCA2B geni (Tüm Gen Dizi Analizi)	GUCA2B gene (Sequence Analysis-All Coding Exons)	GUCA2B	GUCA2B, UGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCA2A geni (Tüm Gen Dizi Analizi)	GUCA2A gene (Sequence Analysis-All Coding Exons)	GUCA2A	GUCA2A, GUCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEIL3 geni (Tüm Gen Dizi Analizi)	NEIL3 gene (Sequence Analysis-All Coding Exons)	NEIL3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NEIL2 geni (Tüm Gen Dizi Analizi)	NEIL2 gene (Sequence Analysis-All Coding Exons)	NEIL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GUCA1C geni (Tüm Gen Dizi Analizi)	GUCA1C gene (Sequence Analysis-All Coding Exons)	GUCA1C	GUCA1C, GCAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTPBP6 geni (Tüm Gen Dizi Analizi)	GTPBP6 gene (Sequence Analysis-All Coding Exons)	GTPBP6	GTPBP6, PGPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTPBP2 geni (Tüm Gen Dizi Analizi)	GTPBP2 gene (Sequence Analysis-All Coding Exons)	GTPBP2	GTPBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTPBP10 geni (Tüm Gen Dizi Analizi)	GTPBP10 gene (Sequence Analysis-All Coding Exons)	GTPBP10	GTPBP10, OBGH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTPBP1 geni (Tüm Gen Dizi Analizi)	GTPBP1 gene (Sequence Analysis-All Coding Exons)	GTPBP1	GTPBP1, GP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C6 geni (Tüm Gen Dizi Analizi)	GTF3C6 gene (Sequence Analysis-All Coding Exons)	GTF3C6	GTF3C6, C6orf51	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C5 geni (Tüm Gen Dizi Analizi)	GTF3C5 gene (Sequence Analysis-All Coding Exons)	GTF3C5	GTF3C5, TFIIIC63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C3 geni (Tüm Gen Dizi Analizi)	GTF3C3 gene (Sequence Analysis-All Coding Exons)	GTF3C3	GTF3C3, TFIIIC102	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C1 geni (Tüm Gen Dizi Analizi)	GTF3C1 gene (Sequence Analysis-All Coding Exons)	GTF3C1	GTF3C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3A geni (Tüm Gen Dizi Analizi)	GTF3A gene (Sequence Analysis-All Coding Exons)	GTF3A	GTF3A, TFIIIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2IRD1 geni (Tüm Gen Dizi Analizi)	GTF2IRD1 gene (Sequence Analysis-All Coding Exons)	GTF2IRD1	GTF2IRD1, GTF3, MUSTRD1, WBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2I geni (Tüm Gen Dizi Analizi)	GTF2I gene (Sequence Analysis-All Coding Exons)	GTF2I	GTF2I, BAP135, WBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2H4 geni (Tüm Gen Dizi Analizi)	GTF2H4 gene (Sequence Analysis-All Coding Exons)	GTF2H4	GTF2H4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2H2 geni (Tüm Gen Dizi Analizi)	GTF2H2 gene (Sequence Analysis-All Coding Exons)	GTF2H2	GTF2H2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2H1 geni (Tüm Gen Dizi Analizi)	GTF2H1 gene (Sequence Analysis-All Coding Exons)	GTF2H1	GTF2H1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

GTF2F2 geni (Tüm Gen Dizi Analizi)	GTF2F2 gene (Sequence Analysis-All Coding Exons)	GTF2F2	GTF2F2, TF2F2, RAP30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2F1 geni (Tüm Gen Dizi Analizi)	GTF2F1 gene (Sequence Analysis-All Coding Exons)	GTF2F1	GTF2F1, RAP74	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2E1 geni (Tüm Gen Dizi Analizi)	GTF2E1 gene (Sequence Analysis-All Coding Exons)	GTF2E1	GTF2E1, TF2E1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2B geni (Tüm Gen Dizi Analizi)	GTF2B gene (Sequence Analysis-All Coding Exons)	GTF2B	GTF2B, TFIIB, TF2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2A2 geni (Tüm Gen Dizi Analizi)	GTF2A2 gene (Sequence Analysis-All Coding Exons)	GTF2A2	GTF2A2, TF2A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2A1L geni (Tüm Gen Dizi Analizi)	GTF2A1L gene (Sequence Analysis-All Coding Exons)	GTF2A1L	GTF2A1L, ALF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2A1 geni (Tüm Gen Dizi Analizi)	GTF2A1 gene (Sequence Analysis-All Coding Exons)	GTF2A1	GTF2A1, TF2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTDC1 geni (Tüm Gen Dizi Analizi)	GTDC1 gene (Sequence Analysis-All Coding Exons)	GTDC1	GTDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSX2 geni (Tüm Gen Dizi Analizi)	GSX2 gene (Sequence Analysis-All Coding Exons)	GSX2	GSX2, GSH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSX1 geni (Tüm Gen Dizi Analizi)	GSX1 gene (Sequence Analysis-All Coding Exons)	GSX1	GSX1, GSH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTZ1 geni (Tüm Gen Dizi Analizi)	GSTZ1 gene (Sequence Analysis-All Coding Exons)	GSTZ1	GSTZ1, MAAI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTT2 geni (Tüm Gen Dizi Analizi)	GSTT2 gene (Sequence Analysis-All Coding Exons)	GSTT2	GSTT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTP1 geni (Tüm Gen Dizi Analizi)	GSTP1 gene (Sequence Analysis-All Coding Exons)	GSTP1	GSTP1, GST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTO2 geni (Tüm Gen Dizi Analizi)	GSTO2 gene (Sequence Analysis-All Coding Exons)	GSTO2	GSTO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTO1 geni (Tüm Gen Dizi Analizi)	GSTO1 gene (Sequence Analysis-All Coding Exons)	GSTO1	GSTO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTM5 geni (Tüm Gen Dizi Analizi)	GSTM5 gene (Sequence Analysis-All Coding Exons)	GSTM5	GSTM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTM4 geni (Tüm Gen Dizi Analizi)	GSTM4 gene (Sequence Analysis-All Coding Exons)	GSTM4	GSTM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTM3 geni (Tüm Gen Dizi Analizi)	GSTM3 gene (Sequence Analysis-All Coding Exons)	GSTM3	GSTM3, GST5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTM2 geni (Tüm Gen Dizi Analizi)	GSTM2 gene (Sequence Analysis-All Coding Exons)	GSTM2	GSTM2, GST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTM1 geni (Tüm Gen Dizi Analizi)	GSTM1 gene (Sequence Analysis-All Coding Exons)	GSTM1	GSTM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTK1 geni (Tüm Gen Dizi Analizi)	GSTK1 gene (Sequence Analysis-All Coding Exons)	GSTK1	GSTK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTCD geni (Tüm Gen Dizi Analizi)	GSTCD gene (Sequence Analysis-All Coding Exons)	GSTCD	GSTCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTA5 geni (Tüm Gen Dizi Analizi)	GSTA5 gene (Sequence Analysis-All Coding Exons)	GSTA5	GSTA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTA4 geni (Tüm Gen Dizi Analizi)	GSTA4 gene (Sequence Analysis-All Coding Exons)	GSTA4	GSTA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTA3 geni (Tüm Gen Dizi Analizi)	GSTA3 gene (Sequence Analysis-All Coding Exons)	GSTA3	GSTA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTA2 geni (Tüm Gen Dizi Analizi)	GSTA2 gene (Sequence Analysis-All Coding Exons)	GSTA2	GSTA2, GST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB6 geni (Tüm Gen Dizi Analizi)	NDUFB6 gene (Sequence Analysis-All Coding Exons)	NDUFB6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB5 geni (Tüm Gen Dizi Analizi)	NDUFB5 gene (Sequence Analysis-All Coding Exons)	NDUFB5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTA1 geni (Tüm Gen Dizi Analizi)	GSTA1 gene (Sequence Analysis-All Coding Exons)	GSTA1	GSTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSPT2 geni (Tüm Gen Dizi Analizi)	GSPT2 gene (Sequence Analysis-All Coding Exons)	GSPT2	GSPT2, GST2, ERF3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDUFB2 geni (Tüm Gen Dizi Analizi)	NDUFB2 gene (Sequence Analysis-All Coding Exons)	NDUFB2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSPT1 geni (Tüm Gen Dizi Analizi)	GSPT1 gene (Sequence Analysis-All Coding Exons)	GSPT1	GSPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSKIP geni (Tüm Gen Dizi Analizi)	GSKIP gene (Sequence Analysis-All Coding Exons)	GSKIP	GSKIP, C14orf129	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSK3B geni (Tüm Gen Dizi Analizi)	GSK3B gene (Sequence Analysis-All Coding Exons)	GSK3B	GSK3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSG1L geni (Tüm Gen Dizi Analizi)	GSG1L gene (Sequence Analysis-All Coding Exons)	GSG1L	GSG1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSE1 geni (Tüm Gen Dizi Analizi)	GSE1 gene (Sequence Analysis-All Coding Exons)	GSE1	GSE1, KIAA0182	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSDMD geni (Tüm Gen Dizi Analizi)	GSDMD gene (Sequence Analysis-All Coding Exons)	GSDMD	GSDMD, GSDMDC1, DFNA5L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSDMC geni (Tüm Gen Dizi Analizi)	GSDMC gene (Sequence Analysis-All Coding Exons)	GSDMC	GSDMC, MLZE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSDMB geni (Tüm Gen Dizi Analizi)	GSDMB gene (Sequence Analysis-All Coding Exons)	GSDMB	GSDMB, GSDML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSDMA geni (Tüm Gen Dizi Analizi)	GSDMA gene (Sequence Analysis-All Coding Exons)	GSDMA	GSDMA, GSDM1, GSDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSC2 geni (Tüm Gen Dizi Analizi)	GSC2 gene (Sequence Analysis-All Coding Exons)	GSC2	GSC2, GSCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSAP geni (Tüm Gen Dizi Analizi)	GSAP gene (Sequence Analysis-All Coding Exons)	GSAP	GSAP, PION	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRWD1 geni (Tüm Gen Dizi Analizi)	GRWD1 gene (Sequence Analysis-All Coding Exons)	GRWD1	GRWD1, GRWD, KIAA1942	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRSF1 geni (Tüm Gen Dizi Analizi)	GRSF1 gene (Sequence Analysis-All Coding Exons)	GRSF1	GRSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRPR geni (Tüm Gen Dizi Analizi)	GRPR gene (Sequence Analysis-All Coding Exons)	GRPR	GRPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRPEL1 geni (Tüm Gen Dizi Analizi)	GRPEL1 gene (Sequence Analysis-All Coding Exons)	GRPEL1	GRPEL1, HMGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRP geni (Tüm Gen Dizi Analizi)	GRP gene (Sequence Analysis-All Coding Exons)	GRP	GRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM8 geni (Tüm Gen Dizi Analizi)	GRM8 gene (Sequence Analysis-All Coding Exons)	GRM8	GRM8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM7 geni (Tüm Gen Dizi Analizi)	GRM7 gene (Sequence Analysis-All Coding Exons)	GRM7	GRM7, MGLUR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM5 geni (Tüm Gen Dizi Analizi)	GRM5 gene (Sequence Analysis-All Coding Exons)	GRM5	GRM5, MGLUR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM4 geni (Tüm Gen Dizi Analizi)	GRM4 gene (Sequence Analysis-All Coding Exons)	GRM4	GRM4, MGLUR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM3 geni (Tüm Gen Dizi Analizi)	GRM3 gene (Sequence Analysis-All Coding Exons)	GRM3	GRM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRM2 geni (Tüm Gen Dizi Analizi)	GRM2 gene (Sequence Analysis-All Coding Exons)	GRM2	GRM2, MGLUR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK7 geni (Tüm Gen Dizi Analizi)	GRK7 gene (Sequence Analysis-All Coding Exons)	GRK7	GRK7, GPRK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK6 geni (Tüm Gen Dizi Analizi)	GRK6 gene (Sequence Analysis-All Coding Exons)	GRK6	GRK6, GPRK6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK5 geni (Tüm Gen Dizi Analizi)	GRK5 gene (Sequence Analysis-All Coding Exons)	GRK5	GRK5, GPRK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK4 geni (Tüm Gen Dizi Analizi)	GRK4 gene (Sequence Analysis-All Coding Exons)	GRK4	GRK4, GPRK2L, GPRK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK3 geni (Tüm Gen Dizi Analizi)	GRK3 gene (Sequence Analysis-All Coding Exons)	GRK3	ADRBK2, BARK2, GRK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDST3 geni (Tüm Gen Dizi Analizi)	NDST3 gene (Sequence Analysis-All Coding Exons)	NDST3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRK2 geni (Tüm Gen Dizi Analizi)	GRK2 gene (Sequence Analysis-All Coding Exons)	GRK2	ADRBK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIPAP1 geni (Tüm Gen Dizi Analizi)	GRIPAP1 gene (Sequence Analysis-All Coding Exons)	GRIPAP1	GRIPAP1, GRASP1, KIAA1167	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRINA geni (Tüm Gen Dizi Analizi)	GRINA gene (Sequence Analysis-All Coding Exons)	GRINA	GRINA, NMDARA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIN3A geni (Tüm Gen Dizi Analizi)	GRIN3A gene (Sequence Analysis-All Coding Exons)	GRIN3A	GRIN3A, NR3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIN2C geni (Tüm Gen Dizi Analizi)	GRIN2C gene (Sequence Analysis-All Coding Exons)	GRIN2C	GRIN2C, NMDAR2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIK5 geni (Tüm Gen Dizi Analizi)	GRIK5 gene (Sequence Analysis-All Coding Exons)	GRIK5	GRIK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIK4 geni (Tüm Gen Dizi Analizi)	GRIK4 gene (Sequence Analysis-All Coding Exons)	GRIK4	GRIK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIK3 geni (Tüm Gen Dizi Analizi)	GRIK3 gene (Sequence Analysis-All Coding Exons)	GRIK3	GRIK3, GLUR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIK1 geni (Tüm Gen Dizi Analizi)	GRIK1 gene (Sequence Analysis-All Coding Exons)	GRIK1	GRIK1, GLUR5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRID1 geni (Tüm Gen Dizi Analizi)	GRID1 gene (Sequence Analysis-All Coding Exons)	GRID1	GRID1, KIAA1220	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIA4 geni (Tüm Gen Dizi Analizi)	GRIA4 gene (Sequence Analysis-All Coding Exons)	GRIA4	GRIA4, GLUR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIA2 geni (Tüm Gen Dizi Analizi)	GRIA2 gene (Sequence Analysis-All Coding Exons)	GRIA2	GRIA2, GLUR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIA1 geni (Tüm Gen Dizi Analizi)	GRIA1 gene (Sequence Analysis-All Coding Exons)	GRIA1	GRIA1, GLUR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRHL1 geni (Tüm Gen Dizi Analizi)	GRHL1 gene (Sequence Analysis-All Coding Exons)	GRHL1	GRHL1, LBP32, MGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GREM1 geni (Tüm Gen Dizi Analizi)	GREM1 gene (Sequence Analysis-All Coding Exons)	GREM1	GREM1, CKTSF1B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GREB1 geni (Tüm Gen Dizi Analizi)	GREB1 gene (Sequence Analysis-All Coding Exons)	GREB1	GREB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NDC80 geni (Tüm Gen Dizi Analizi)	NDC80 gene (Sequence Analysis-All Coding Exons)	NDC80		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRB7 geni (Tüm Gen Dizi Analizi)	GRB7 gene (Sequence Analysis-All Coding Exons)	GRB7	GRB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRB2 geni (Tüm Gen Dizi Analizi)	GRB2 gene (Sequence Analysis-All Coding Exons)	GRB2	GRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRB14 geni (Tüm Gen Dizi Analizi)	GRB14 gene (Sequence Analysis-All Coding Exons)	GRB14	GRB14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRB10 geni (Tüm Gen Dizi Analizi)	GRB10 gene (Sequence Analysis-All Coding Exons)	GRB10	GRB10, RSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRASP geni (Tüm Gen Dizi Analizi)	GRASP gene (Sequence Analysis-All Coding Exons)	GRASP	GRASP, TAMALIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRAP2 geni (Tüm Gen Dizi Analizi)	GRAP2 gene (Sequence Analysis-All Coding Exons)	GRAP2	GRAP2, GRID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRAMD4 geni (Tüm Gen Dizi Analizi)	GRAMD4 gene (Sequence Analysis-All Coding Exons)	GRAMD4	GRAMD4, KIAA0767, DIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX8 geni (Tüm Gen Dizi Analizi)	GPX8 gene (Sequence Analysis-All Coding Exons)	GPX8	GPX8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX7 geni (Tüm Gen Dizi Analizi)	GPX7 gene (Sequence Analysis-All Coding Exons)	GPX7	GPX7, NPGPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX6 geni (Tüm Gen Dizi Analizi)	GPX6 gene (Sequence Analysis-All Coding Exons)	GPX6	GPX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX5 geni (Tüm Gen Dizi Analizi)	GPX5 gene (Sequence Analysis-All Coding Exons)	GPX5	GPX5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX3 geni (Tüm Gen Dizi Analizi)	GPX3 gene (Sequence Analysis-All Coding Exons)	GPX3	GPX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPX2 geni (Tüm Gen Dizi Analizi)	GPX2 gene (Sequence Analysis-All Coding Exons)	GPX2	GPX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPT geni (Tüm Gen Dizi Analizi)	GPT gene (Sequence Analysis-All Coding Exons)	GPT	GPT, GPT1, AAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPS1 geni (Tüm Gen Dizi Analizi)	GPS1 gene (Sequence Analysis-All Coding Exons)	GPS1	GPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRIN3 geni (Tüm Gen Dizi Analizi)	GPRIN3 gene (Sequence Analysis-All Coding Exons)	GPRIN3	GPRIN3, GRIN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NCLN geni (Tüm Gen Dizi Analizi)	NCLN gene (Sequence Analysis-All Coding Exons)	NCLN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

GPRIN2 geni (Tüm Gen Dizi Analizi)	GPRIN2 gene (Sequence Analysis-All Coding Exons)	GPRIN2	GPRIN2, GRIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRIN1 geni (Tüm Gen Dizi Analizi)	GPRIN1 gene (Sequence Analysis-All Coding Exons)	GPRIN1	GPRIN1, GRIN1, KIAA1893	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRC5D geni (Tüm Gen Dizi Analizi)	GPRC5D gene (Sequence Analysis-All Coding Exons)	GPRC5D	GPRC5D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRC5C geni (Tüm Gen Dizi Analizi)	GPRC5C gene (Sequence Analysis-All Coding Exons)	GPRC5C	GPRC5C, RAIG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRC5B geni (Tüm Gen Dizi Analizi)	GPRC5B gene (Sequence Analysis-All Coding Exons)	GPRC5B	GPRC5B, RAIG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRC5A geni (Tüm Gen Dizi Analizi)	GPRC5A gene (Sequence Analysis-All Coding Exons)	GPRC5A	GPRC5A, RAI3, RAIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRASP2 geni (Tüm Gen Dizi Analizi)	GPRASP2 gene (Sequence Analysis-All Coding Exons)	GPRASP2	GPRASP2, GASP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRASP1 geni (Tüm Gen Dizi Analizi)	GPRASP1 gene (Sequence Analysis-All Coding Exons)	GPRASP1	GPRASP1, GASP, KIAA0443	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR89B geni (Tüm Gen Dizi Analizi)	GPR89B gene (Sequence Analysis-All Coding Exons)	GPR89B	GPR89B, GPHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR89A geni (Tüm Gen Dizi Analizi)	GPR89A gene (Sequence Analysis-All Coding Exons)	GPR89A	GPR89A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR87 geni (Tüm Gen Dizi Analizi)	GPR87 gene (Sequence Analysis-All Coding Exons)	GPR87	GPR87	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR85 geni (Tüm Gen Dizi Analizi)	GPR85 gene (Sequence Analysis-All Coding Exons)	GPR85	GPR85, SREB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR84 geni (Tüm Gen Dizi Analizi)	GPR84 gene (Sequence Analysis-All Coding Exons)	GPR84	GPR84	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR83 geni (Tüm Gen Dizi Analizi)	GPR83 gene (Sequence Analysis-All Coding Exons)	GPR83	GPR83, GIR, GPR72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR82 geni (Tüm Gen Dizi Analizi)	GPR82 gene (Sequence Analysis-All Coding Exons)	GPR82	GPR82	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR78 geni (Tüm Gen Dizi Analizi)	GPR78 gene (Sequence Analysis-All Coding Exons)	GPR78	GPR78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR75 geni (Tüm Gen Dizi Analizi)	GPR75 gene (Sequence Analysis-All Coding Exons)	GPR75	GPR75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR65 geni (Tüm Gen Dizi Analizi)	GPR65 gene (Sequence Analysis-All Coding Exons)	GPR65	GPR65, TDAG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR63 geni (Tüm Gen Dizi Analizi)	GPR63 gene (Sequence Analysis-All Coding Exons)	GPR63	GPR63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR62 geni (Tüm Gen Dizi Analizi)	GPR62 gene (Sequence Analysis-All Coding Exons)	GPR62	GPR62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR61 geni (Tüm Gen Dizi Analizi)	GPR61 gene (Sequence Analysis-All Coding Exons)	GPR61	GPR61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR6 geni (Tüm Gen Dizi Analizi)	GPR6 gene (Sequence Analysis-All Coding Exons)	GPR6	GPR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR55 geni (Tüm Gen Dizi Analizi)	GPR55 gene (Sequence Analysis-All Coding Exons)	GPR55	GPR55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR52 geni (Tüm Gen Dizi Analizi)	GPR52 gene (Sequence Analysis-All Coding Exons)	GPR52	GPR52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR50 geni (Tüm Gen Dizi Analizi)	GPR50 gene (Sequence Analysis-All Coding Exons)	GPR50	GPR50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR45 geni (Tüm Gen Dizi Analizi)	GPR45 gene (Sequence Analysis-All Coding Exons)	GPR45	GPR45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR4 geni (Tüm Gen Dizi Analizi)	GPR4 gene (Sequence Analysis-All Coding Exons)	GPR4	GPR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR39 geni (Tüm Gen Dizi Analizi)	GPR39 gene (Sequence Analysis-All Coding Exons)	GPR39	GPR39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR37 geni (Tüm Gen Dizi Analizi)	GPR37 gene (Sequence Analysis-All Coding Exons)	GPR37	GPR37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR35 geni (Tüm Gen Dizi Analizi)	GPR35 gene (Sequence Analysis-All Coding Exons)	GPR35	GPR35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR34 geni (Tüm Gen Dizi Analizi)	GPR34 gene (Sequence Analysis-All Coding Exons)	GPR34	GPR34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR33 geni (Tüm Gen Dizi Analizi)	GPR33 gene (Sequence Analysis-All Coding Exons)	GPR33	GPR33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR32 geni (Tüm Gen Dizi Analizi)	GPR32 gene (Sequence Analysis-All Coding Exons)	GPR32	GPR32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR31 geni (Tüm Gen Dizi Analizi)	GPR31 gene (Sequence Analysis-All Coding Exons)	GPR31	GPR31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF19 geni (Tüm Gen Dizi Analizi)	NBPF19 gene (Sequence Analysis-All Coding Exons)	NBPF19	NBPF19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR3 geni (Tüm Gen Dizi Analizi)	GPR3 gene (Sequence Analysis-All Coding Exons)	GPR3	GPR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR27 geni (Tüm Gen Dizi Analizi)	GPR27 gene (Sequence Analysis-All Coding Exons)	GPR27	GPR27, SREB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR26 geni (Tüm Gen Dizi Analizi)	GPR26 gene (Sequence Analysis-All Coding Exons)	GPR26	GPR26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR25 geni (Tüm Gen Dizi Analizi)	GPR25 gene (Sequence Analysis-All Coding Exons)	GPR25	GPR25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR22 geni (Tüm Gen Dizi Analizi)	GPR22 gene (Sequence Analysis-All Coding Exons)	GPR22	GPR22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR21 geni (Tüm Gen Dizi Analizi)	GPR21 gene (Sequence Analysis-All Coding Exons)	GPR21	GPR21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBPF1 geni (Tüm Gen Dizi Analizi)	NBPF1 gene (Sequence Analysis-All Coding Exons)	NBPF1	NBPF1, KIAA1693	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR20 geni (Tüm Gen Dizi Analizi)	GPR20 gene (Sequence Analysis-All Coding Exons)	GPR20	GPR20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR19 geni (Tüm Gen Dizi Analizi)	GPR19 gene (Sequence Analysis-All Coding Exons)	GPR19	GPR19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR183 geni (Tüm Gen Dizi Analizi)	GPR183 gene (Sequence Analysis-All Coding Exons)	GPR183	GPR183, EBI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR180 geni (Tüm Gen Dizi Analizi)	GPR180 gene (Sequence Analysis-All Coding Exons)	GPR180	GPR180, ITR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR18 geni (Tüm Gen Dizi Analizi)	GPR18 gene (Sequence Analysis-All Coding Exons)	GPR18	GPR18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR176 geni (Tüm Gen Dizi Analizi)	GPR176 gene (Sequence Analysis-All Coding Exons)	GPR176	GPR176	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR174 geni (Tüm Gen Dizi Analizi)	GPR174 gene (Sequence Analysis-All Coding Exons)	GPR174	GPR174, GPCR17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NBDY geni (Tüm Gen Dizi Analizi)	NBDY gene (Sequence Analysis-All Coding Exons)	NBDY	NBDY, LINC01420, NOBODY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR173 geni (Tüm Gen Dizi Analizi)	GPR173 gene (Sequence Analysis-All Coding Exons)	GPR173	GPR173, SREB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR17 geni (Tüm Gen Dizi Analizi)	GPR17 gene (Sequence Analysis-All Coding Exons)	GPR17	GPR17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR161 geni (Tüm Gen Dizi Analizi)	GPR161 gene (Sequence Analysis-All Coding Exons)	GPR161	GPR161	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR158 geni (Tüm Gen Dizi Analizi)	GPR158 gene (Sequence Analysis-All Coding Exons)	GPR158	GPR158, KIAA1136	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR156 geni (Tüm Gen Dizi Analizi)	GPR156 gene (Sequence Analysis-All Coding Exons)	GPR156	GPR156, GABABL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR153 geni (Tüm Gen Dizi Analizi)	GPR153 gene (Sequence Analysis-All Coding Exons)	GPR153	GPR153	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR15 geni (Tüm Gen Dizi Analizi)	GPR15 gene (Sequence Analysis-All Coding Exons)	GPR15	GPR15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR142 geni (Tüm Gen Dizi Analizi)	GPR142 gene (Sequence Analysis-All Coding Exons)	GPR142	GPR142, PGR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR141 geni (Tüm Gen Dizi Analizi)	GPR141 gene (Sequence Analysis-All Coding Exons)	GPR141	GPR141, PGR13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR137B geni (Tüm Gen Dizi Analizi)	GPR137B gene (Sequence Analysis-All Coding Exons)	GPR137B	TM7SF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR135 geni (Tüm Gen Dizi Analizi)	GPR135 gene (Sequence Analysis-All Coding Exons)	GPR135	GPR135	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR132 geni (Tüm Gen Dizi Analizi)	GPR132 gene (Sequence Analysis-All Coding Exons)	GPR132	GPR132, G2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR12 geni (Tüm Gen Dizi Analizi)	GPR12 gene (Sequence Analysis-All Coding Exons)	GPR12	GPR12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR119 geni (Tüm Gen Dizi Analizi)	GPR119 gene (Sequence Analysis-All Coding Exons)	GPR119	GPR119, GPCR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR1 geni (Tüm Gen Dizi Analizi)	GPR1 gene (Sequence Analysis-All Coding Exons)	GPR1	GPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPN1 geni (Tüm Gen Dizi Analizi)	GPN1 gene (Sequence Analysis-All Coding Exons)	GPN1	XAB1, MBDIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPM6B geni (Tüm Gen Dizi Analizi)	GPM6B gene (Sequence Analysis-All Coding Exons)	GPM6B	GPM6B, M6B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPM6A geni (Tüm Gen Dizi Analizi)	GPM6A gene (Sequence Analysis-All Coding Exons)	GPM6A	GPM6A, M6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPLD1 geni (Tüm Gen Dizi Analizi)	GPLD1 gene (Sequence Analysis-All Coding Exons)	GPLD1	GPLD1, PIGPLD, GPIPLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NARF geni (Tüm Gen Dizi Analizi)	NARF gene (Sequence Analysis-All Coding Exons)	NARF		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPER1 geni (Tüm Gen Dizi Analizi)	GPER1 gene (Sequence Analysis-All Coding Exons)	GPER1	GPER, CMKRL2, GPR30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPCPD1 geni (Tüm Gen Dizi Analizi)	GPCPD1 gene (Sequence Analysis-All Coding Exons)	GPCPD1	GPCPD1, GDE5, KIAA1434	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPC5 geni (Tüm Gen Dizi Analizi)	GPC5 gene (Sequence Analysis-All Coding Exons)	GPC5	GPC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPC4 geni (Tüm Gen Dizi Analizi)	GPC4 gene (Sequence Analysis-All Coding Exons)	GPC4	GPC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPC1 geni (Tüm Gen Dizi Analizi)	GPC1 gene (Sequence Analysis-All Coding Exons)	GPC1	GPC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPBP1 geni (Tüm Gen Dizi Analizi)	GPBP1 gene (Sequence Analysis-All Coding Exons)	GPBP1	GPBP1, GPBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPBAR1 geni (Tüm Gen Dizi Analizi)	GPBAR1 gene (Sequence Analysis-All Coding Exons)	GPBAR1	GPBAR1, BG37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPATCH8 geni (Tüm Gen Dizi Analizi)	GPATCH8 gene (Sequence Analysis-All Coding Exons)	GPATCH8	GPATCH8, KIAA0553	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPATCH2 geni (Tüm Gen Dizi Analizi)	GPATCH2 gene (Sequence Analysis-All Coding Exons)	GPATCH2	GPATCH2, GPATC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPAT4 geni (Tüm Gen Dizi Analizi)	GPAT4 gene (Sequence Analysis-All Coding Exons)	GPAT4	AGPAT6, LPAATZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPAT3 geni (Tüm Gen Dizi Analizi)	GPAT3 gene (Sequence Analysis-All Coding Exons)	GPAT3	AGPAT9, GPAT3, MAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPAT2 geni (Tüm Gen Dizi Analizi)	GPAT2 gene (Sequence Analysis-All Coding Exons)	GPAT2	GPAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPANK1 geni (Tüm Gen Dizi Analizi)	GPANK1 gene (Sequence Analysis-All Coding Exons)	GPANK1	GPANK1, BAT4, D6S54E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPAM geni (Tüm Gen Dizi Analizi)	GPAM gene (Sequence Analysis-All Coding Exons)	GPAM	GPAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPAA1 geni (Tüm Gen Dizi Analizi)	GPAA1 gene (Sequence Analysis-All Coding Exons)	GPAA1	GPAA1, GAA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GP5 geni (Tüm Gen Dizi Analizi)	GP5 gene (Sequence Analysis-All Coding Exons)	GP5	GP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GP2 geni (Tüm Gen Dizi Analizi)	GP2 gene (Sequence Analysis-All Coding Exons)	GP2	GP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOT2 geni (Tüm Gen Dizi Analizi)	GOT2 gene (Sequence Analysis-All Coding Exons)	GOT2	GOT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOSR1 geni (Tüm Gen Dizi Analizi)	GOSR1 gene (Sequence Analysis-All Coding Exons)	GOSR1	GOSR1, GOS28, GS28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GORASP1 geni (Tüm Gen Dizi Analizi)	GORASP1 gene (Sequence Analysis-All Coding Exons)	GORASP1	GORASP1, GOLPH5, GRASP65, P65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOPC geni (Tüm Gen Dizi Analizi)	GOPC gene (Sequence Analysis-All Coding Exons)	GOPC	GOPC, PIST, FIG, CAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GON4L geni (Tüm Gen Dizi Analizi)	GON4L gene (Sequence Analysis-All Coding Exons)	GON4L	GON4L, KIAA1606	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLT1B geni (Tüm Gen Dizi Analizi)	GOLT1B gene (Sequence Analysis-All Coding Exons)	GOLT1B	GOLT1B, GOT1B, GCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

GOLPH3L geni (Tüm Gen Dizi Analizi)	GOLPH3L gene (Sequence Analysis-All Coding Exons)	GOLPH3L	GOLPH3L, GPP34R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLPH3 geni (Tüm Gen Dizi Analizi)	GOLPH3 gene (Sequence Analysis-All Coding Exons)	GOLPH3	GOLPH3, GPP34, MIDAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLM1 geni (Tüm Gen Dizi Analizi)	GOLM1 gene (Sequence Analysis-All Coding Exons)	GOLM1	GOLM1, GOLPH2, GP73, C9orf155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLIM4 geni (Tüm Gen Dizi Analizi)	GOLIM4 gene (Sequence Analysis-All Coding Exons)	GOLIM4	GOLIM4, GOLPH4, GPP130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGB1 geni (Tüm Gen Dizi Analizi)	GOLGB1 gene (Sequence Analysis-All Coding Exons)	GOLGB1	GOLGB1, GCP372	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA8B geni (Tüm Gen Dizi Analizi)	GOLGA8B gene (Sequence Analysis-All Coding Exons)	GOLGA8B	GOLGA8B, KIAA0855	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA8A geni (Tüm Gen Dizi Analizi)	GOLGA8A gene (Sequence Analysis-All Coding Exons)	GOLGA8A	GOLGA8A, KIAA0855	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA7B geni (Tüm Gen Dizi Analizi)	GOLGA7B gene (Sequence Analysis-All Coding Exons)	GOLGA7B	GOLGA7B, C10orf132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA6A geni (Tüm Gen Dizi Analizi)	GOLGA6A gene (Sequence Analysis-All Coding Exons)	GOLGA6A	GOLGA6, GLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA5 geni (Tüm Gen Dizi Analizi)	GOLGA5 gene (Sequence Analysis-All Coding Exons)	GOLGA5	GOLGA5, RFG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA4 geni (Tüm Gen Dizi Analizi)	GOLGA4 gene (Sequence Analysis-All Coding Exons)	GOLGA4	GOLGA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA3 geni (Tüm Gen Dizi Analizi)	GOLGA3 gene (Sequence Analysis-All Coding Exons)	GOLGA3	GOLGA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA2P2Y geni (Tüm Gen Dizi Analizi)	GOLGA2P2Y gene (Sequence Analysis-All Coding Exons)	GOLGA2P2Y	GOLGA2L Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA2 geni (Tüm Gen Dizi Analizi)	GOLGA2 gene (Sequence Analysis-All Coding Exons)	GOLGA2	GOLGA2, GM130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NACC1 geni (Tüm Gen Dizi Analizi)	NACC1 gene (Sequence Analysis-All Coding Exons)	NACC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
NACA2 geni (Tüm Gen Dizi Analizi)	NACA2 gene (Sequence Analysis-All Coding Exons)	NACA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA1 geni (Tüm Gen Dizi Analizi)	GOLGA1 gene (Sequence Analysis-All Coding Exons)	GOLGA1	GOLGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNRH2 geni (Tüm Gen Dizi Analizi)	GNRH2 gene (Sequence Analysis-All Coding Exons)	GNRH2	GNRH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNPNAT1 geni (Tüm Gen Dizi Analizi)	GNPNAT1 gene (Sequence Analysis-All Coding Exons)	GNPNAT1	GNPNAT1, GNA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNPDA2 geni (Tüm Gen Dizi Analizi)	GNPDA2 gene (Sequence Analysis-All Coding Exons)	GNPDA2	GNPDA2, GNP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNPDA1 geni (Tüm Gen Dizi Analizi)	GNPDA1 gene (Sequence Analysis-All Coding Exons)	GNPDA1	GNPDA1, GNP1, GNPI, KIAA0060	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNLY geni (Tüm Gen Dizi Analizi)	GNLY gene (Sequence Analysis-All Coding Exons)	GNLY	GNLY, D2S69E, TLA519	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNL3L geni (Tüm Gen Dizi Analizi)	GNL3L gene (Sequence Analysis-All Coding Exons)	GNL3L	GNL3L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNL3 geni (Tüm Gen Dizi Analizi)	GNL3 gene (Sequence Analysis-All Coding Exons)	GNL3	GNL3, NS, E2IG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNL2 geni (Tüm Gen Dizi Analizi)	GNL2 gene (Sequence Analysis-All Coding Exons)	GNL2	GNL2, NGP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNL1 geni (Tüm Gen Dizi Analizi)	GNL1 gene (Sequence Analysis-All Coding Exons)	GNL1	GNL1, HSR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNGT2 geni (Tüm Gen Dizi Analizi)	GNGT2 gene (Sequence Analysis-All Coding Exons)	GNGT2	GNGT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNGT1 geni (Tüm Gen Dizi Analizi)	GNGT1 gene (Sequence Analysis-All Coding Exons)	GNGT1	GNGT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG5 geni (Tüm Gen Dizi Analizi)	GNG5 gene (Sequence Analysis-All Coding Exons)	GNG5	GNG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG4 geni (Tüm Gen Dizi Analizi)	GNG4 gene (Sequence Analysis-All Coding Exons)	GNG4	GNG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG2 geni (Tüm Gen Dizi Analizi)	GNG2 gene (Sequence Analysis-All Coding Exons)	GNG2	GNG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG13 geni (Tüm Gen Dizi Analizi)	GNG13 gene (Sequence Analysis-All Coding Exons)	GNG13	GNG13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG12-AS1 geni (Tüm Gen Dizi Analizi)	GNG12-AS1 gene (Sequence Analysis-All Coding Exons)	GNG12-AS1	GNG12AS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG12 geni (Tüm Gen Dizi Analizi)	GNG12 gene (Sequence Analysis-All Coding Exons)	GNG12	GNG12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG11 geni (Tüm Gen Dizi Analizi)	GNG11 gene (Sequence Analysis-All Coding Exons)	GNG11	GNG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG10 geni (Tüm Gen Dizi Analizi)	GNG10 gene (Sequence Analysis-All Coding Exons)	GNG10	GNG10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNB2 geni (Tüm Gen Dizi Analizi)	GNB2 gene (Sequence Analysis-All Coding Exons)	GNB2	GNB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNB1L geni (Tüm Gen Dizi Analizi)	GNB1L gene (Sequence Analysis-All Coding Exons)	GNB1L	GNB1L, WDR14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNAZ geni (Tüm Gen Dizi Analizi)	GNAZ gene (Sequence Analysis-All Coding Exons)	GNAZ	GNAZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNAT3 geni (Tüm Gen Dizi Analizi)	GNAT3 gene (Sequence Analysis-All Coding Exons)	GNAT3	GNAT3, GDCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNAI1 geni (Tüm Gen Dizi Analizi)	GNAI1 gene (Sequence Analysis-All Coding Exons)	GNAI1	GNAI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNA15 geni (Tüm Gen Dizi Analizi)	GNA15 gene (Sequence Analysis-All Coding Exons)	GNA15	GNA15, GNA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNA14 geni (Tüm Gen Dizi Analizi)	GNA14 gene (Sequence Analysis-All Coding Exons)	GNA14	GNA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNA13 geni (Tüm Gen Dizi Analizi)	GNA13 gene (Sequence Analysis-All Coding Exons)	GNA13	GNA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNA12 geni (Tüm Gen Dizi Analizi)	GNA12 gene (Sequence Analysis-All Coding Exons)	GNA12	GNA12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMPS geni (Tüm Gen Dizi Analizi)	GMPS gene (Sequence Analysis-All Coding Exons)	GMPS	GMPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMPR2 geni (Tüm Gen Dizi Analizi)	GMPR2 gene (Sequence Analysis-All Coding Exons)	GMPR2	GMPR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMPR geni (Tüm Gen Dizi Analizi)	GMPR gene (Sequence Analysis-All Coding Exons)	GMPR	GMPR, GMPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMNC geni (Tüm Gen Dizi Analizi)	GMNC gene (Sequence Analysis-All Coding Exons)	GMNC	GMNC, GEMC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GML geni (Tüm Gen Dizi Analizi)	GML gene (Sequence Analysis-All Coding Exons)	GML	GML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMIP geni (Tüm Gen Dizi Analizi)	GMIP gene (Sequence Analysis-All Coding Exons)	GMIP	GMIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMFG geni (Tüm Gen Dizi Analizi)	GMFG gene (Sequence Analysis-All Coding Exons)	GMFG	GMFG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMEB2 geni (Tüm Gen Dizi Analizi)	GMEB2 gene (Sequence Analysis-All Coding Exons)	GMEB2	GMEB2, KIAA1269	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMEB1 geni (Tüm Gen Dizi Analizi)	GMEB1 gene (Sequence Analysis-All Coding Exons)	GMEB1	GMEB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMDS geni (Tüm Gen Dizi Analizi)	GMDS gene (Sequence Analysis-All Coding Exons)	GMDS	GMDS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLYR1 geni (Tüm Gen Dizi Analizi)	GLYR1 gene (Sequence Analysis-All Coding Exons)	GLYR1	NP60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLYATL3 geni (Tüm Gen Dizi Analizi)	GLYATL3 gene (Sequence Analysis-All Coding Exons)	GLYATL3	GLYATL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLYATL2 geni (Tüm Gen Dizi Analizi)	GLYATL2 gene (Sequence Analysis-All Coding Exons)	GLYATL2	GLYATL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLYATL1 geni (Tüm Gen Dizi Analizi)	GLYATL1 gene (Sequence Analysis-All Coding Exons)	GLYATL1	GLYATL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLYAT geni (Tüm Gen Dizi Analizi)	GLYAT gene (Sequence Analysis-All Coding Exons)	GLYAT	GLYAT, ACGNAT, CAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLTSCR1 geni (Tüm Gen Dizi Analizi)	GLTSCR1 gene (Sequence Analysis-All Coding Exons)	GLTSCR1	GLTSCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLTP geni (Tüm Gen Dizi Analizi)	GLTP gene (Sequence Analysis-All Coding Exons)	GLTP	GLTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLT6D1 geni (Tüm Gen Dizi Analizi)	GLT6D1 gene (Sequence Analysis-All Coding Exons)	GLT6D1	GLT6D1, GT6M7, GLTDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLS2 geni (Tüm Gen Dizi Analizi)	GLS2 gene (Sequence Analysis-All Coding Exons)	GLS2	GLS2, GA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLS geni (Tüm Gen Dizi Analizi)	GLS gene (Sequence Analysis-All Coding Exons)	GLS	GLS, GLS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLRX3 geni (Tüm Gen Dizi Analizi)	GLRX3 gene (Sequence Analysis-All Coding Exons)	GLRX3	GLRX3, PICOT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLRX2 geni (Tüm Gen Dizi Analizi)	GLRX2 gene (Sequence Analysis-All Coding Exons)	GLRX2	GLRX2, GRX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLRX geni (Tüm Gen Dizi Analizi)	GLRX gene (Sequence Analysis-All Coding Exons)	GLRX	GLRX, GRX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLRA3 geni (Tüm Gen Dizi Analizi)	GLRA3 gene (Sequence Analysis-All Coding Exons)	GLRA3	GLRA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLRA2 geni (Tüm Gen Dizi Analizi)	GLRA2 gene (Sequence Analysis-All Coding Exons)	GLRA2	GLRA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLP2R geni (Tüm Gen Dizi Analizi)	GLP2R gene (Sequence Analysis-All Coding Exons)	GLP2R	GLP2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLP1R geni (Tüm Gen Dizi Analizi)	GLP1R gene (Sequence Analysis-All Coding Exons)	GLP1R	GLP1R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLO1 geni (Tüm Gen Dizi Analizi)	GLO1 gene (Sequence Analysis-All Coding Exons)	GLO1	GLO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLIS1 geni (Tüm Gen Dizi Analizi)	GLIS1 gene (Sequence Analysis-All Coding Exons)	GLIS1	GLIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLIPR2 geni (Tüm Gen Dizi Analizi)	GLIPR2 gene (Sequence Analysis-All Coding Exons)	GLIPR2	GLIPR2, GAPR1, C9orf19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLIPR1L2 geni (Tüm Gen Dizi Analizi)	GLIPR1L2 gene (Sequence Analysis-All Coding Exons)	GLIPR1L2	GLIPR1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLIPR1L1 geni (Tüm Gen Dizi Analizi)	GLIPR1L1 gene (Sequence Analysis-All Coding Exons)	GLIPR1L1	GLIPR1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLIPR1 geni (Tüm Gen Dizi Analizi)	GLIPR1 gene (Sequence Analysis-All Coding Exons)	GLIPR1	GLIPR1, GLIPR, RTVP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLI4 geni (Tüm Gen Dizi Analizi)	GLI4 gene (Sequence Analysis-All Coding Exons)	GLI4	GLI4, HKR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLI1 geni (Tüm Gen Dizi Analizi)	GLI1 gene (Sequence Analysis-All Coding Exons)	GLI1	GLI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLG1 geni (Tüm Gen Dizi Analizi)	GLG1 gene (Sequence Analysis-All Coding Exons)	GLG1	GLG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GLCE geni (Tüm Gen Dizi Analizi)	GLCE gene (Sequence Analysis-All Coding Exons)	GLCE	GLCE, HSEPI, KIAA0836	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GKN1 geni (Tüm Gen Dizi Analizi)	GKN1 gene (Sequence Analysis-All Coding Exons)	GKN1	GKN1, CA11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GKAP1 geni (Tüm Gen Dizi Analizi)	GKAP1 gene (Sequence Analysis-All Coding Exons)	GKAP1	GKAP1, GKAP42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJD4 geni (Tüm Gen Dizi Analizi)	GJD4 gene (Sequence Analysis-All Coding Exons)	GJD4	GJD4, CX40.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJD3 geni (Tüm Gen Dizi Analizi)	GJD3 gene (Sequence Analysis-All Coding Exons)	GJD3	GJD3, GJC1, CX31.9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJD2 geni (Tüm Gen Dizi Analizi)	GJD2 gene (Sequence Analysis-All Coding Exons)	GJD2	GJD2, GJA9, CX36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJC3 geni (Tüm Gen Dizi Analizi)	GJC3 gene (Sequence Analysis-All Coding Exons)	GJC3	GJC3, CX30.2, CX31.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJB7 geni (Tüm Gen Dizi Analizi)	GJB7 gene (Sequence Analysis-All Coding Exons)	GJB7	GJB7, CX25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJB5 geni (Tüm Gen Dizi Analizi)	GJB5 gene (Sequence Analysis-All Coding Exons)	GJB5	GJB5, CX31.1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJA9 geni (Tüm Gen Dizi Analizi)	GJA9 gene (Sequence Analysis-All Coding Exons)	GJA9	GJA9, CX59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJA4 geni (Tüm Gen Dizi Analizi)	GJA4 gene (Sequence Analysis-All Coding Exons)	GJA4	GJA4, CX37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJA10 geni (Tüm Gen Dizi Analizi)	GJA10 gene (Sequence Analysis-All Coding Exons)	GJA10	GJA10, CX62	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

GIT1 geni (Tüm Gen Dizi Analizi)	GIT1 gene (Sequence Analysis-All Coding Exons)	GIT1	GIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIPC1 geni (Tüm Gen Dizi Analizi)	GIPC1 gene (Sequence Analysis-All Coding Exons)	GIPC1	RGS19IP1, C19orf3, GIPC,	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIP geni (Tüm Gen Dizi Analizi)	GIP gene (Sequence Analysis-All Coding Exons)	GIP	GIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GINS4 geni (Tüm Gen Dizi Analizi)	GINS4 gene (Sequence Analysis-All Coding Exons)	GINS4	GINS4, SLD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GINS3 geni (Tüm Gen Dizi Analizi)	GINS3 gene (Sequence Analysis-All Coding Exons)	GINS3	GINS3, PSF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GINS2 geni (Tüm Gen Dizi Analizi)	GINS2 gene (Sequence Analysis-All Coding Exons)	GINS2	GINS2, PSF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GINS1 geni (Tüm Gen Dizi Analizi)	GINS1 gene (Sequence Analysis-All Coding Exons)	GINS1	GINS1, PSF1, KIAA0186	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP8 geni (Tüm Gen Dizi Analizi)	GIMAP8 gene (Sequence Analysis-All Coding Exons)	GIMAP8	GIMAP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP7 geni (Tüm Gen Dizi Analizi)	GIMAP7 gene (Sequence Analysis-All Coding Exons)	GIMAP7	GIMAP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP6 geni (Tüm Gen Dizi Analizi)	GIMAP6 gene (Sequence Analysis-All Coding Exons)	GIMAP6	GIMAP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP5 geni (Tüm Gen Dizi Analizi)	GIMAP5 gene (Sequence Analysis-All Coding Exons)	GIMAP5	GIMAP5, IAN4L1, IMAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP4 geni (Tüm Gen Dizi Analizi)	GIMAP4 gene (Sequence Analysis-All Coding Exons)	GIMAP4	GIMAP4, IMAP4, IAN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP2 geni (Tüm Gen Dizi Analizi)	GIMAP2 gene (Sequence Analysis-All Coding Exons)	GIMAP2	GIMAP2, IMAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIMAP1 geni (Tüm Gen Dizi Analizi)	GIMAP1 gene (Sequence Analysis-All Coding Exons)	GIMAP1	GIMAP1, IMAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIGYF1 geni (Tüm Gen Dizi Analizi)	GIGYF1 gene (Sequence Analysis-All Coding Exons)	GIGYF1	GIGYF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GID8 geni (Tüm Gen Dizi Analizi)	GID8 gene (Sequence Analysis-All Coding Exons)	GID8	C20orf11, TWA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GH2 geni (Tüm Gen Dizi Analizi)	GH2 gene (Sequence Analysis-All Coding Exons)	GH2	GH2, GHV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGTLC2 geni (Tüm Gen Dizi Analizi)	GGTLC2 gene (Sequence Analysis-All Coding Exons)	GGTLC2	GGTLC2, GGTL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGTLC1 geni (Tüm Gen Dizi Analizi)	GGTLC1 gene (Sequence Analysis-All Coding Exons)	GGTLC1	GGTLC1, GGTL6, GGTLA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGTA1P geni (Tüm Gen Dizi Analizi)	GGTA1P gene (Sequence Analysis-All Coding Exons)	GGTA1P	GGTA1, GLYT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGT7 geni (Tüm Gen Dizi Analizi)	GGT7 gene (Sequence Analysis-All Coding Exons)	GGT7	GGT7, GGTL3, GGTL5, GGT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGT6 geni (Tüm Gen Dizi Analizi)	GGT6 gene (Sequence Analysis-All Coding Exons)	GGT6	GGT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGT5 geni (Tüm Gen Dizi Analizi)	GGT5 gene (Sequence Analysis-All Coding Exons)	GGT5	GGT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGT1 geni (Tüm Gen Dizi Analizi)	GGT1 gene (Sequence Analysis-All Coding Exons)	GGT1	GGT1, GGT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGPS1 geni (Tüm Gen Dizi Analizi)	GGPS1 gene (Sequence Analysis-All Coding Exons)	GGPS1	GGPS1, GGPPS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGNBP2 geni (Tüm Gen Dizi Analizi)	GGNBP2 gene (Sequence Analysis-All Coding Exons)	GGNBP2	GGNBP2, DIF3, LCRG1, L2H1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGN geni (Tüm Gen Dizi Analizi)	GGN gene (Sequence Analysis-All Coding Exons)	GGN	GGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGH geni (Tüm Gen Dizi Analizi)	GGH gene (Sequence Analysis-All Coding Exons)	GGH	GGH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGCT geni (Tüm Gen Dizi Analizi)	GGCT gene (Sequence Analysis-All Coding Exons)	GGCT	GGCT, GCTG, C7orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGACT geni (Tüm Gen Dizi Analizi)	GGACT gene (Sequence Analysis-All Coding Exons)	GGACT	A2LD1, GGACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGA3 geni (Tüm Gen Dizi Analizi)	GGA3 gene (Sequence Analysis-All Coding Exons)	GGA3	GGA3, KIAA0154	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGA1 geni (Tüm Gen Dizi Analizi)	GGA1 gene (Sequence Analysis-All Coding Exons)	GGA1	GGA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GFRA3 geni (Tüm Gen Dizi Analizi)	GFRA3 gene (Sequence Analysis-All Coding Exons)	GFRA3	GFRA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GFRA2 geni (Tüm Gen Dizi Analizi)	GFRA2 gene (Sequence Analysis-All Coding Exons)	GFRA2	GFRA2, GDNFRB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GFRA1 geni (Tüm Gen Dizi Analizi)	GFRA1 gene (Sequence Analysis-All Coding Exons)	GFRA1	GFRA1, GDNFRA, GDNFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GFPT2 geni (Tüm Gen Dizi Analizi)	GFPT2 gene (Sequence Analysis-All Coding Exons)	GFPT2	GFPT2, GFAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GFM2 geni (Tüm Gen Dizi Analizi)	GFM2 gene (Sequence Analysis-All Coding Exons)	GFM2	EFG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GET4 geni (Tüm Gen Dizi Analizi)	GET4 gene (Sequence Analysis-All Coding Exons)	GET4	GET4, C7orf20, CEE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEN1 geni (Tüm Gen Dizi Analizi)	GEN1 gene (Sequence Analysis-All Coding Exons)	GEN1	GEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN8 geni (Tüm Gen Dizi Analizi)	GEMIN8 gene (Sequence Analysis-All Coding Exons)	GEMIN8	GEMIN8, FAM51A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN7 geni (Tüm Gen Dizi Analizi)	GEMIN7 gene (Sequence Analysis-All Coding Exons)	GEMIN7	GEMIN7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN6 geni (Tüm Gen Dizi Analizi)	GEMIN6 gene (Sequence Analysis-All Coding Exons)	GEMIN6	GEMIN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN5 geni (Tüm Gen Dizi Analizi)	GEMIN5 gene (Sequence Analysis-All Coding Exons)	GEMIN5	GEMIN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN2 geni (Tüm Gen Dizi Analizi)	GEMIN2 gene (Sequence Analysis-All Coding Exons)	GEMIN2	GEMIN2, SIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEM geni (Tüm Gen Dizi Analizi)	GEM gene (Sequence Analysis-All Coding Exons)	GEM	GEM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDPD5 geni (Tüm Gen Dizi Analizi)	GDPD5 gene (Sequence Analysis-All Coding Exons)	GDPD5	GDPD5, GDE2, PP1665	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDPD3 geni (Tüm Gen Dizi Analizi)	GDPD3 gene (Sequence Analysis-All Coding Exons)	GDPD3	GDPD3, GDE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDPD2 geni (Tüm Gen Dizi Analizi)	GDPD2 gene (Sequence Analysis-All Coding Exons)	GDPD2	GDPD2, GDE3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDPD1 geni (Tüm Gen Dizi Analizi)	GDPD1 gene (Sequence Analysis-All Coding Exons)	GDPD1	GDPD1, GDE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDI2 geni (Tüm Gen Dizi Analizi)	GDI2 gene (Sequence Analysis-All Coding Exons)	GDI2	GDI2, RABGDIB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDF9 geni (Tüm Gen Dizi Analizi)	GDF9 gene (Sequence Analysis-All Coding Exons)	GDF9	GDF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDF7 geni (Tüm Gen Dizi Analizi)	GDF7 gene (Sequence Analysis-All Coding Exons)	GDF7	GDF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDF15 geni (Tüm Gen Dizi Analizi)	GDF15 gene (Sequence Analysis-All Coding Exons)	GDF15	GDF15, PLAB, PDF, MIC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDF11 geni (Tüm Gen Dizi Analizi)	GDF11 gene (Sequence Analysis-All Coding Exons)	GDF11	GDF11, BMP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDE1 geni (Tüm Gen Dizi Analizi)	GDE1 gene (Sequence Analysis-All Coding Exons)	GDE1	GDE1, MIR16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDA geni (Tüm Gen Dizi Analizi)	GDA gene (Sequence Analysis-All Coding Exons)	GDA	GDA, CYPIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCSAM geni (Tüm Gen Dizi Analizi)	GCSAM gene (Sequence Analysis-All Coding Exons)	GCSAM	GCET2, HGAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCNT4 geni (Tüm Gen Dizi Analizi)	GCNT4 gene (Sequence Analysis-All Coding Exons)	GCNT4	GCNT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCNT3 geni (Tüm Gen Dizi Analizi)	GCNT3 gene (Sequence Analysis-All Coding Exons)	GCNT3	GCNT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCNT1 geni (Tüm Gen Dizi Analizi)	GCNT1 gene (Sequence Analysis-All Coding Exons)	GCNT1	GCNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCNA geni (Tüm Gen Dizi Analizi)	GCNA gene (Sequence Analysis-All Coding Exons)	GCNA	ACRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCN1 geni (Tüm Gen Dizi Analizi)	GCN1 gene (Sequence Analysis-All Coding Exons)	GCN1	GCN1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCM1 geni (Tüm Gen Dizi Analizi)	GCM1 gene (Sequence Analysis-All Coding Exons)	GCM1	GCM1, GCMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCHFR geni (Tüm Gen Dizi Analizi)	GCHFR gene (Sequence Analysis-All Coding Exons)	GCHFR	GCHFR, GFRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCFC2 geni (Tüm Gen Dizi Analizi)	GCFC2 gene (Sequence Analysis-All Coding Exons)	GCFC2	C2orf3, TCF9, GCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCC2 geni (Tüm Gen Dizi Analizi)	GCC2 gene (Sequence Analysis-All Coding Exons)	GCC2	GCC2, GCC185, KIAA0336	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCC1 geni (Tüm Gen Dizi Analizi)	GCC1 gene (Sequence Analysis-All Coding Exons)	GCC1	GCC1, GCC88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCAT geni (Tüm Gen Dizi Analizi)	GCAT gene (Sequence Analysis-All Coding Exons)	GCAT	GCAT, KBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GCA geni (Tüm Gen Dizi Analizi)	GCA gene (Sequence Analysis-All Coding Exons)	GCA	GCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GC geni (Tüm Gen Dizi Analizi)	GC gene (Sequence Analysis-All Coding Exons)	GC	GC, DBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBX2 geni (Tüm Gen Dizi Analizi)	GBX2 gene (Sequence Analysis-All Coding Exons)	GBX2	GBX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBX1 geni (Tüm Gen Dizi Analizi)	GBX1 gene (Sequence Analysis-All Coding Exons)	GBX1	GBX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP7 geni (Tüm Gen Dizi Analizi)	GBP7 gene (Sequence Analysis-All Coding Exons)	GBP7	GBP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP6 geni (Tüm Gen Dizi Analizi)	GBP6 gene (Sequence Analysis-All Coding Exons)	GBP6	GBP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP5 geni (Tüm Gen Dizi Analizi)	GBP5 gene (Sequence Analysis-All Coding Exons)	GBP5	GBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP4 geni (Tüm Gen Dizi Analizi)	GBP4 gene (Sequence Analysis-All Coding Exons)	GBP4	GBP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP3 geni (Tüm Gen Dizi Analizi)	GBP3 gene (Sequence Analysis-All Coding Exons)	GBP3	GBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP2 geni (Tüm Gen Dizi Analizi)	GBP2 gene (Sequence Analysis-All Coding Exons)	GBP2	GBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBP1 geni (Tüm Gen Dizi Analizi)	GBP1 gene (Sequence Analysis-All Coding Exons)	GBP1	GBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBGT1 geni (Tüm Gen Dizi Analizi)	GBGT1 gene (Sequence Analysis-All Coding Exons)	GBGT1	GBGT1, FS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBF1 geni (Tüm Gen Dizi Analizi)	GBF1 gene (Sequence Analysis-All Coding Exons)	GBF1	GBF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GBA3 geni (Tüm Gen Dizi Analizi)	GBA3 gene (Sequence Analysis-All Coding Exons)	GBA3	GBA3, CBGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATC geni (Tüm Gen Dizi Analizi)	GATC gene (Sequence Analysis-All Coding Exons)	GATC	GATC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATB geni (Tüm Gen Dizi Analizi)	GATB gene (Sequence Analysis-All Coding Exons)	GATB	GATB, PET112	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATAD2A geni (Tüm Gen Dizi Analizi)	GATAD2A gene (Sequence Analysis-All Coding Exons)	GATAD2A	GATAD2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATA5 geni (Tüm Gen Dizi Analizi)	GATA5 gene (Sequence Analysis-All Coding Exons)	GATA5	GATA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATA5 geni (Tüm Gen Dizi Analizi)	GATA5 gene (Sequence Analysis-All Coding Exons)	GATA5	GATA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GATA5 geni (Tüm Gen Dizi Analizi)	GATA5 gene (Sequence Analysis-All Coding Exons)	GATA5	GATA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAST geni (Tüm Gen Dizi Analizi)	GAST gene (Sequence Analysis-All Coding Exons)	GAST	GAS, GAST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS7 geni (Tüm Gen Dizi Analizi)	GAS7 gene (Sequence Analysis-All Coding Exons)	GAS7	GAS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS6 geni (Tüm Gen Dizi Analizi)	GAS6 gene (Sequence Analysis-All Coding Exons)	GAS6	GAS6, AXLLG, AXSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS5 geni (Tüm Gen Dizi Analizi)	GAS5 gene (Sequence Analysis-All Coding Exons)	GAS5	GAS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS2L3 geni (Tüm Gen Dizi Analizi)	GAS2L3 gene (Sequence Analysis-All Coding Exons)	GAS2L3	GAS2L3, G2L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

GAS2L2 geni (Tüm Gen Dizi Analizi)	GAS2L2 gene (Sequence Analysis-All Coding Exons)	GAS2L2	GAS2L2, GAR17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS2L1 geni (Tüm Gen Dizi Analizi)	GAS2L1 gene (Sequence Analysis-All Coding Exons)	GAS2L1	GAS2L1, GAR22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS2 geni (Tüm Gen Dizi Analizi)	GAS2 gene (Sequence Analysis-All Coding Exons)	GAS2	GAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAS1 geni (Tüm Gen Dizi Analizi)	GAS1 gene (Sequence Analysis-All Coding Exons)	GAS1	GAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GART geni (Tüm Gen Dizi Analizi)	GART gene (Sequence Analysis-All Coding Exons)	GART	GART, PRGS, PGFT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAR1 geni (Tüm Gen Dizi Analizi)	GAR1 gene (Sequence Analysis-All Coding Exons)	GAR1	NOLA1, GAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAPVD1 geni (Tüm Gen Dizi Analizi)	GAPVD1 gene (Sequence Analysis-All Coding Exons)	GAPVD1	GAPVD1, RAP6, GAPEX5, KIAA1521	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAPDHS geni (Tüm Gen Dizi Analizi)	GAPDHS gene (Sequence Analysis-All Coding Exons)	GAPDHS	GAPDHS, GAPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAPDH geni (Tüm Gen Dizi Analizi)	GAPDH gene (Sequence Analysis-All Coding Exons)	GAPDH	GAPDH, GAPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MUC15 geni (Tüm Gen Dizi Analizi)	MUC15 gene (Sequence Analysis-All Coding Exons)	MUC15		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAP43 geni (Tüm Gen Dizi Analizi)	GAP43 gene (Sequence Analysis-All Coding Exons)	GAP43	GAP43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GANC geni (Tüm Gen Dizi Analizi)	GANC gene (Sequence Analysis-All Coding Exons)	GANC	GANC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALR3 geni (Tüm Gen Dizi Analizi)	GALR3 gene (Sequence Analysis-All Coding Exons)	GALR3	GALR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALR2 geni (Tüm Gen Dizi Analizi)	GALR2 gene (Sequence Analysis-All Coding Exons)	GALR2	GALR2, GALNR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALR1 geni (Tüm Gen Dizi Analizi)	GALR1 gene (Sequence Analysis-All Coding Exons)	GALR1	GALR1, GALNR1, GALNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTUS1 geni (Tüm Gen Dizi Analizi)	MTUS1 gene (Sequence Analysis-All Coding Exons)	MTUS1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALP geni (Tüm Gen Dizi Analizi)	GALP gene (Sequence Analysis-All Coding Exons)	GALP	GALP, ALARIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNTL6 geni (Tüm Gen Dizi Analizi)	GALNTL6 gene (Sequence Analysis-All Coding Exons)	GALNTL6	GALNTL6, GALNACT20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNTL5 geni (Tüm Gen Dizi Analizi)	GALNTL5 gene (Sequence Analysis-All Coding Exons)	GALNTL5	GALNTL5, GALNACT19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT9 geni (Tüm Gen Dizi Analizi)	GALNT9 gene (Sequence Analysis-All Coding Exons)	GALNT9	GALNT9, GALNAC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT8 geni (Tüm Gen Dizi Analizi)	GALNT8 gene (Sequence Analysis-All Coding Exons)	GALNT8	GALNT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT7 geni (Tüm Gen Dizi Analizi)	GALNT7 gene (Sequence Analysis-All Coding Exons)	GALNT7	GALNT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT6 geni (Tüm Gen Dizi Analizi)	GALNT6 gene (Sequence Analysis-All Coding Exons)	GALNT6	GALNT6, GalNAc6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTRF1L geni (Tüm Gen Dizi Analizi)	MTRF1L gene (Sequence Analysis-All Coding Exons)	MTRF1L		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT5 geni (Tüm Gen Dizi Analizi)	GALNT5 gene (Sequence Analysis-All Coding Exons)	GALNT5	GALNT5, GALNACT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT4 geni (Tüm Gen Dizi Analizi)	GALNT4 gene (Sequence Analysis-All Coding Exons)	GALNT4	GALNT4, GalNAc4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT2 geni (Tüm Gen Dizi Analizi)	GALNT2 gene (Sequence Analysis-All Coding Exons)	GALNT2	GALNT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT18 geni (Tüm Gen Dizi Analizi)	GALNT18 gene (Sequence Analysis-All Coding Exons)	GALNT18	GALNT18, GALNACT18, GALNT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT16 geni (Tüm Gen Dizi Analizi)	GALNT16 gene (Sequence Analysis-All Coding Exons)	GALNT16	GALNT16, GALNACT16, GalNT7.1, KIAA1130	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT15 geni (Tüm Gen Dizi Analizi)	GALNT15 gene (Sequence Analysis-All Coding Exons)	GALNT15	GALNT15, GALNACT15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT14 geni (Tüm Gen Dizi Analizi)	GALNT14 gene (Sequence Analysis-All Coding Exons)	GALNT14	GALNT14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT13 geni (Tüm Gen Dizi Analizi)	GALNT13 gene (Sequence Analysis-All Coding Exons)	GALNT13	GALNT13, KIAA1918	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT11 geni (Tüm Gen Dizi Analizi)	GALNT11 gene (Sequence Analysis-All Coding Exons)	GALNT11	GALNT11, GALNACT11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT10 geni (Tüm Gen Dizi Analizi)	GALNT10 gene (Sequence Analysis-All Coding Exons)	GALNT10	GALNT10, GalNAcT10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALNT1 geni (Tüm Gen Dizi Analizi)	GALNT1 gene (Sequence Analysis-All Coding Exons)	GALNT1	GALNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALM geni (Tüm Gen Dizi Analizi)	GALM gene (Sequence Analysis-All Coding Exons)	GALM	GALM, GLAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GALK2 geni (Tüm Gen Dizi Analizi)	GALK2 gene (Sequence Analysis-All Coding Exons)	GALK2	GALK2, GK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAL3ST4 geni (Tüm Gen Dizi Analizi)	GAL3ST4 gene (Sequence Analysis-All Coding Exons)	GAL3ST4	GAL3ST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAL3ST3 geni (Tüm Gen Dizi Analizi)	GAL3ST3 gene (Sequence Analysis-All Coding Exons)	GAL3ST3	GAL3ST3, GAL3ST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAL3ST2 geni (Tüm Gen Dizi Analizi)	GAL3ST2 gene (Sequence Analysis-All Coding Exons)	GAL3ST2	GAL3ST2, GP3ST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAL3ST1 geni (Tüm Gen Dizi Analizi)	GAL3ST1 gene (Sequence Analysis-All Coding Exons)	GAL3ST1	GAL3ST1, CST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAK geni (Tüm Gen Dizi Analizi)	GAK gene (Sequence Analysis-All Coding Exons)	GAK	GAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE8 geni (Tüm Gen Dizi Analizi)	GAGE8 gene (Sequence Analysis-All Coding Exons)	GAGE8	GAGE8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE7 geni (Tüm Gen Dizi Analizi)	GAGE7 gene (Sequence Analysis-All Coding Exons)	GAGE7	GAGE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE6 geni (Tüm Gen Dizi Analizi)	GAGE6 gene (Sequence Analysis-All Coding Exons)	GAGE6	GAGE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE5 geni (Tüm Gen Dizi Analizi)	GAGE5 gene (Sequence Analysis-All Coding Exons)	GAGE5	GAGE5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE4 geni (Tüm Gen Dizi Analizi)	GAGE4 gene (Sequence Analysis-All Coding Exons)	GAGE4	GAGE4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE2E geni (Tüm Gen Dizi Analizi)	GAGE2E gene (Sequence Analysis-All Coding Exons)	GAGE2E	GAGE2E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE2D geni (Tüm Gen Dizi Analizi)	GAGE2D gene (Sequence Analysis-All Coding Exons)	GAGE2D	GAGE2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE2C geni (Tüm Gen Dizi Analizi)	GAGE2C gene (Sequence Analysis-All Coding Exons)	GAGE2C	GAGE2C, GAGE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE2B geni (Tüm Gen Dizi Analizi)	GAGE2B gene (Sequence Analysis-All Coding Exons)	GAGE2B	GAGE2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTHFD2 geni (Tüm Gen Dizi Analizi)	MTHFD2 gene (Sequence Analysis-All Coding Exons)	MTHFD2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE2A geni (Tüm Gen Dizi Analizi)	GAGE2A gene (Sequence Analysis-All Coding Exons)	GAGE2A	GAGE2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE13 geni (Tüm Gen Dizi Analizi)	GAGE13 gene (Sequence Analysis-All Coding Exons)	GAGE13	GAGE13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12J geni (Tüm Gen Dizi Analizi)	GAGE12J gene (Sequence Analysis-All Coding Exons)	GAGE12J	GAGE12J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12I geni (Tüm Gen Dizi Analizi)	GAGE12I gene (Sequence Analysis-All Coding Exons)	GAGE12I	GAGE7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12H geni (Tüm Gen Dizi Analizi)	GAGE12H gene (Sequence Analysis-All Coding Exons)	GAGE12H	GAGE12H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12G geni (Tüm Gen Dizi Analizi)	GAGE12G gene (Sequence Analysis-All Coding Exons)	GAGE12G	GAGE12G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12F geni (Tüm Gen Dizi Analizi)	GAGE12F gene (Sequence Analysis-All Coding Exons)	GAGE12F	GAGE12F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12E geni (Tüm Gen Dizi Analizi)	GAGE12E gene (Sequence Analysis-All Coding Exons)	GAGE12E	GAGE12E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12D geni (Tüm Gen Dizi Analizi)	GAGE12D gene (Sequence Analysis-All Coding Exons)	GAGE12D	GAGE12D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE12C geni (Tüm Gen Dizi Analizi)	GAGE12C gene (Sequence Analysis-All Coding Exons)	GAGE12C	GAGE12C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE10 geni (Tüm Gen Dizi Analizi)	GAGE10 gene (Sequence Analysis-All Coding Exons)	GAGE10	GAGE10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAGE1 geni (Tüm Gen Dizi Analizi)	GAGE1 gene (Sequence Analysis-All Coding Exons)	GAGE1	GAGE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GADL1 geni (Tüm Gen Dizi Analizi)	GADL1 gene (Sequence Analysis-All Coding Exons)	GADL1	GADL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GADD45GIP1 geni (Tüm Gen Dizi Analizi)	GADD45GIP1 gene (Sequence Analysis-All Coding Exons)	GADD45GIP1	GADD45GIP1, PRG6, CRIF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GADD45G geni (Tüm Gen Dizi Analizi)	GADD45G gene (Sequence Analysis-All Coding Exons)	GADD45G	GADD45G, GRP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GADD45B geni (Tüm Gen Dizi Analizi)	GADD45B gene (Sequence Analysis-All Coding Exons)	GADD45B	GADD45B, MYD118	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GADD45A geni (Tüm Gen Dizi Analizi)	GADD45A gene (Sequence Analysis-All Coding Exons)	GADD45A	DDIT1, GADD45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAD2 geni (Tüm Gen Dizi Analizi)	GAD2 gene (Sequence Analysis-All Coding Exons)	GAD2	GAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GACAT2 geni (Tüm Gen Dizi Analizi)	GACAT2 gene (Sequence Analysis-All Coding Exons)	GACAT2	GACAT2, MTCL1AS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MTA3 geni (Tüm Gen Dizi Analizi)	MTA3 gene (Sequence Analysis-All Coding Exons)	MTA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRR2 geni (Tüm Gen Dizi Analizi)	GABRR2 gene (Sequence Analysis-All Coding Exons)	GABRR2	GABRR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRR1 geni (Tüm Gen Dizi Analizi)	GABRR1 gene (Sequence Analysis-All Coding Exons)	GABRR1	GABRR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRQ geni (Tüm Gen Dizi Analizi)	GABRQ gene (Sequence Analysis-All Coding Exons)	GABRQ	GABRQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRP geni (Tüm Gen Dizi Analizi)	GABRP gene (Sequence Analysis-All Coding Exons)	GABRP	GABRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRG3 geni (Tüm Gen Dizi Analizi)	GABRG3 gene (Sequence Analysis-All Coding Exons)	GABRG3	GABRG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRG1 geni (Tüm Gen Dizi Analizi)	GABRG1 gene (Sequence Analysis-All Coding Exons)	GABRG1	GABRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRE geni (Tüm Gen Dizi Analizi)	GABRE gene (Sequence Analysis-All Coding Exons)	GABRE	GABRE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRB2 geni (Tüm Gen Dizi Analizi)	GABRB2 gene (Sequence Analysis-All Coding Exons)	GABRB2	GABRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRA6 geni (Tüm Gen Dizi Analizi)	GABRA6 gene (Sequence Analysis-All Coding Exons)	GABRA6	GABRA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRA5 geni (Tüm Gen Dizi Analizi)	GABRA5 gene (Sequence Analysis-All Coding Exons)	GABRA5	GABRA5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRA4 geni (Tüm Gen Dizi Analizi)	GABRA4 gene (Sequence Analysis-All Coding Exons)	GABRA4	GABRA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABRA3 geni (Tüm Gen Dizi Analizi)	GABRA3 gene (Sequence Analysis-All Coding Exons)	GABRA3	GABRA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABPB1 geni (Tüm Gen Dizi Analizi)	GABPB1 gene (Sequence Analysis-All Coding Exons)	GABPB1	GABPB, BABPB2, GABPB1, E4TF1B, NRF2B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABPA geni (Tüm Gen Dizi Analizi)	GABPA gene (Sequence Analysis-All Coding Exons)	GABPA	GABPA, E4TF1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABARAPL2 geni (Tüm Gen Dizi Analizi)	GABARAPL2 gene (Sequence Analysis-All Coding Exons)	GABARAPL2	GABARAPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND6 geni (Tüm Gen Dizi Analizi)	MT-ND6 gene (Sequence Analysis-All Coding Exons)	MT-ND6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

MT-ND5 geni (Tüm Gen Dizi Analizi)	MT-ND5 gene (Sequence Analysis-All Coding Exons)	MT-ND5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND4L geni (Tüm Gen Dizi Analizi)	MT-ND4L gene (Sequence Analysis-All Coding Exons)	MT-ND4L		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND4 geni (Tüm Gen Dizi Analizi)	MT-ND4 gene (Sequence Analysis-All Coding Exons)	MT-ND4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND3 geni (Tüm Gen Dizi Analizi)	MT-ND3 gene (Sequence Analysis-All Coding Exons)	MT-ND3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND2 geni (Tüm Gen Dizi Analizi)	MT-ND2 gene (Sequence Analysis-All Coding Exons)	MT-ND2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-ND1 geni (Tüm Gen Dizi Analizi)	MT-ND1 gene (Sequence Analysis-All Coding Exons)	MT-ND1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-CYB geni (Tüm Gen Dizi Analizi)	MT-CYB gene (Sequence Analysis-All Coding Exons)	MT-CYB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-CO3 geni (Tüm Gen Dizi Analizi)	MT-CO3 gene (Sequence Analysis-All Coding Exons)	MT-CO3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-CO2 geni (Tüm Gen Dizi Analizi)	MT-CO2 gene (Sequence Analysis-All Coding Exons)	MT-CO2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MT-CO1 geni (Tüm Gen Dizi Analizi)	MT-CO1 gene (Sequence Analysis-All Coding Exons)	MT-CO1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABARAPL1 geni (Tüm Gen Dizi Analizi)	GABARAPL1 gene (Sequence Analysis-All Coding Exons)	GABARAPL1	GABARAPL1, GEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABARAP geni (Tüm Gen Dizi Analizi)	GABARAP gene (Sequence Analysis-All Coding Exons)	GABARAP	GABARAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAB3 geni (Tüm Gen Dizi Analizi)	GAB3 gene (Sequence Analysis-All Coding Exons)	GAB3	GAB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAB2 geni (Tüm Gen Dizi Analizi)	GAB2 gene (Sequence Analysis-All Coding Exons)	GAB2	GAB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GAB1 geni (Tüm Gen Dizi Analizi)	GAB1 gene (Sequence Analysis-All Coding Exons)	GAB1	GAB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
G6PC2 geni (Tüm Gen Dizi Analizi)	G6PC2 gene (Sequence Analysis-All Coding Exons)	G6PC2	G6PC2, IGRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
G3BP1 geni (Tüm Gen Dizi Analizi)	G3BP1 gene (Sequence Analysis-All Coding Exons)	G3BP1	G3BP1, G3BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
G2E3 geni (Tüm Gen Dizi Analizi)	G2E3 gene (Sequence Analysis-All Coding Exons)	G2E3	G2E3, KIAA1333	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
G0S2 geni (Tüm Gen Dizi Analizi)	G0S2 gene (Sequence Analysis-All Coding Exons)	G0S2	G0S2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZR1 geni (Tüm Gen Dizi Analizi)	FZR1 gene (Sequence Analysis-All Coding Exons)	FZR1	HCDH, FZR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD9 geni (Tüm Gen Dizi Analizi)	FZD9 gene (Sequence Analysis-All Coding Exons)	FZD9	FZD9, FZD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD8 geni (Tüm Gen Dizi Analizi)	FZD8 gene (Sequence Analysis-All Coding Exons)	FZD8	FZD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD7 geni (Tüm Gen Dizi Analizi)	FZD7 gene (Sequence Analysis-All Coding Exons)	FZD7	FZD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSRA geni (Tüm Gen Dizi Analizi)	MSRA gene (Sequence Analysis-All Coding Exons)	MSRA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD5 geni (Tüm Gen Dizi Analizi)	FZD5 gene (Sequence Analysis-All Coding Exons)	FZD5	FZD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD3 geni (Tüm Gen Dizi Analizi)	FZD3 gene (Sequence Analysis-All Coding Exons)	FZD3	FZD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD2 geni (Tüm Gen Dizi Analizi)	FZD2 gene (Sequence Analysis-All Coding Exons)	FZD2	FZD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD10 geni (Tüm Gen Dizi Analizi)	FZD10 gene (Sequence Analysis-All Coding Exons)	FZD10	FZD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FZD1 geni (Tüm Gen Dizi Analizi)	FZD1 gene (Sequence Analysis-All Coding Exons)	FZD1	FZD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FYTTD1 geni (Tüm Gen Dizi Analizi)	FYTTD1 gene (Sequence Analysis-All Coding Exons)	FYTTD1	FYTTD1, UIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FYN geni (Tüm Gen Dizi Analizi)	FYN gene (Sequence Analysis-All Coding Exons)	FYN	FYN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FYB1 geni (Tüm Gen Dizi Analizi)	FYB gene (Sequence Analysis-All Coding Exons)	FYB	FYB, SLAP130, ADAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD7 geni (Tüm Gen Dizi Analizi)	FXYD7 gene (Sequence Analysis-All Coding Exons)	FXYD7	FXYD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD6 geni (Tüm Gen Dizi Analizi)	FXYD6 gene (Sequence Analysis-All Coding Exons)	FXYD6	FXYD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD5 geni (Tüm Gen Dizi Analizi)	FXYD5 gene (Sequence Analysis-All Coding Exons)	FXYD5	FXYD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD4 geni (Tüm Gen Dizi Analizi)	FXYD4 gene (Sequence Analysis-All Coding Exons)	FXYD4	FXYD4, CHIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MSH5 geni (Tüm Gen Dizi Analizi)	MSH5 gene (Sequence Analysis-All Coding Exons)	MSH5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD3 geni (Tüm Gen Dizi Analizi)	FXYD3 gene (Sequence Analysis-All Coding Exons)	FXYD3	FXYD3, PLML, MAT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXYD1 geni (Tüm Gen Dizi Analizi)	FXYD1 gene (Sequence Analysis-All Coding Exons)	FXYD1	FXYD1, PLM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXR2 geni (Tüm Gen Dizi Analizi)	FXR2 gene (Sequence Analysis-All Coding Exons)	FXR2	FXR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FXR1 geni (Tüm Gen Dizi Analizi)	FXR1 gene (Sequence Analysis-All Coding Exons)	FXR1	FXR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT9 geni (Tüm Gen Dizi Analizi)	FUT9 gene (Sequence Analysis-All Coding Exons)	FUT9	FUT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT8 geni (Tüm Gen Dizi Analizi)	FUT8 gene (Sequence Analysis-All Coding Exons)	FUT8	FUT8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT7 geni (Tüm Gen Dizi Analizi)	FUT7 gene (Sequence Analysis-All Coding Exons)	FUT7	FUT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT5 geni (Tüm Gen Dizi Analizi)	FUT5 gene (Sequence Analysis-All Coding Exons)	FUT5	FUT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT4 geni (Tüm Gen Dizi Analizi)	FUT4 gene (Sequence Analysis-All Coding Exons)	FUT4	FUT4, FCT3A, CD15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT10 geni (Tüm Gen Dizi Analizi)	FUT10 gene (Sequence Analysis-All Coding Exons)	FUT10	FUT10, FUCTX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FURIN geni (Tüm Gen Dizi Analizi)	FURIN gene (Sequence Analysis-All Coding Exons)	FURIN	FURIN, PACE, FUR, PCSK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUNDC1 geni (Tüm Gen Dizi Analizi)	FUNDC1 gene (Sequence Analysis-All Coding Exons)	FUNDC1	FUNDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUCA2 geni (Tüm Gen Dizi Analizi)	FUCA2 gene (Sequence Analysis-All Coding Exons)	FUCA2	FUCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUBP3 geni (Tüm Gen Dizi Analizi)	FUBP3 gene (Sequence Analysis-All Coding Exons)	FUBP3	FUBP3, FBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUBP1 geni (Tüm Gen Dizi Analizi)	FUBP1 gene (Sequence Analysis-All Coding Exons)	FUBP1	FUBP1, FUBP, FBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FTX geni (Tüm Gen Dizi Analizi)	FTX gene (Sequence Analysis-All Coding Exons)	FTX	FTX, LINC00182	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FTMT geni (Tüm Gen Dizi Analizi)	FTMT gene (Sequence Analysis-All Coding Exons)	FTMT	FTMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FTHL17 geni (Tüm Gen Dizi Analizi)	FTHL17 gene (Sequence Analysis-All Coding Exons)	FTHL17	FTHL17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSTL3 geni (Tüm Gen Dizi Analizi)	FSTL3 gene (Sequence Analysis-All Coding Exons)	FSTL3	FSTL3, FLRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSTL1 geni (Tüm Gen Dizi Analizi)	FSTL1 gene (Sequence Analysis-All Coding Exons)	FSTL1	FSTL1, FRP, MIR198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FST geni (Tüm Gen Dizi Analizi)	FST gene (Sequence Analysis-All Coding Exons)	FST	FST, FS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSIP2 geni (Tüm Gen Dizi Analizi)	FSIP2 gene (Sequence Analysis-All Coding Exons)	FSIP2	FSIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSIP1 geni (Tüm Gen Dizi Analizi)	FSIP1 gene (Sequence Analysis-All Coding Exons)	FSIP1	FSIP1, HSD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSD1L geni (Tüm Gen Dizi Analizi)	FSD1L gene (Sequence Analysis-All Coding Exons)	FSD1L	FSD1NL, FSD1L, MIR1, CODC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSD1 geni (Tüm Gen Dizi Analizi)	FSD1 gene (Sequence Analysis-All Coding Exons)	FSD1	FSD1, GLFND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSCN3 geni (Tüm Gen Dizi Analizi)	FSCN3 gene (Sequence Analysis-All Coding Exons)	FSCN3	FSCN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSCN1 geni (Tüm Gen Dizi Analizi)	FSCN1 gene (Sequence Analysis-All Coding Exons)	FSCN1	FSCN1, SNL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSCB geni (Tüm Gen Dizi Analizi)	FSCB gene (Sequence Analysis-All Coding Exons)	FSCB	FSCB, C14orf155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FSBP geni (Tüm Gen Dizi Analizi)	FSBP gene (Sequence Analysis-All Coding Exons)	FSBP	FSBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRY geni (Tüm Gen Dizi Analizi)	FRY gene (Sequence Analysis-All Coding Exons)	FRY	FRY, C13orf14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRS2 geni (Tüm Gen Dizi Analizi)	FRS2 gene (Sequence Analysis-All Coding Exons)	FRS2	FRS2, FRS1A, SNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRRS1 geni (Tüm Gen Dizi Analizi)	FRRS1 gene (Sequence Analysis-All Coding Exons)	FRRS1	FRRS1, SDR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRMPD2 geni (Tüm Gen Dizi Analizi)	FRMPD2 gene (Sequence Analysis-All Coding Exons)	FRMPD2	FRMPD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRMPD1 geni (Tüm Gen Dizi Analizi)	FRMPD1 gene (Sequence Analysis-All Coding Exons)	FRMPD1	FRMPD1, FRMD2, KIAA0967	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRMD6 geni (Tüm Gen Dizi Analizi)	FRMD6 gene (Sequence Analysis-All Coding Exons)	FRMD6	FRMD6, WILLIN, C14orf31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRMD5 geni (Tüm Gen Dizi Analizi)	FRMD5 gene (Sequence Analysis-All Coding Exons)	FRMD5	FRMD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRMD3 geni (Tüm Gen Dizi Analizi)	FRMD3 gene (Sequence Analysis-All Coding Exons)	FRMD3	FRMD3, EPB41LO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRK geni (Tüm Gen Dizi Analizi)	FRK gene (Sequence Analysis-All Coding Exons)	FRK	FRK, RAK, GTK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRG2 geni (Tüm Gen Dizi Analizi)	FRG2 gene (Sequence Analysis-All Coding Exons)	FRG2	FRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRG1 geni (Tüm Gen Dizi Analizi)	FRG1 gene (Sequence Analysis-All Coding Exons)	FRG1	FRG1, FSG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FREM3 geni (Tüm Gen Dizi Analizi)	FREM3 gene (Sequence Analysis-All Coding Exons)	FREM3	FREM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRAT2 geni (Tüm Gen Dizi Analizi)	FRAT2 gene (Sequence Analysis-All Coding Exons)	FRAT2	FRAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRA10AC1 geni (Tüm Gen Dizi Analizi)	FRA10AC1 gene (Sequence Analysis-All Coding Exons)	FRA10AC1	C10orf4, FRA10AC1, FRA10A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FPR3 geni (Tüm Gen Dizi Analizi)	FPR3 gene (Sequence Analysis-All Coding Exons)	FPR3	FPR3, FPRL2, FPRH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FPR2 geni (Tüm Gen Dizi Analizi)	FPR2 gene (Sequence Analysis-All Coding Exons)	FPR2	FPR2, FPRL1, FPRH1, LXA4R, HM63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FPR1 geni (Tüm Gen Dizi Analizi)	FPR1 gene (Sequence Analysis-All Coding Exons)	FPR1	FPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FPGT geni (Tüm Gen Dizi Analizi)	FPGT gene (Sequence Analysis-All Coding Exons)	FPGT	FPGT, GFPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FPGS geni (Tüm Gen Dizi Analizi)	FPGS gene (Sequence Analysis-All Coding Exons)	FPGS	FPGS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXS1 geni (Tüm Gen Dizi Analizi)	FOXS1 gene (Sequence Analysis-All Coding Exons)	FOXS1	FOXS1, FKHL18, FREAC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXRED2 geni (Tüm Gen Dizi Analizi)	FOXRED2 gene (Sequence Analysis-All Coding Exons)	FOXRED2	FOXRED2, ERFAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXR2 geni (Tüm Gen Dizi Analizi)	FOXR2 gene (Sequence Analysis-All Coding Exons)	FOXR2	FOXR2, FOXN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXR1 geni (Tüm Gen Dizi Analizi)	FOXR1 gene (Sequence Analysis-All Coding Exons)	FOXR1	FOXR1, FOXN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXQ1 geni (Tüm Gen Dizi Analizi)	FOXQ1 gene (Sequence Analysis-All Coding Exons)	FOXQ1	FOXQ1, HFH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXP4 geni (Tüm Gen Dizi Analizi)	FOXP4 gene (Sequence Analysis-All Coding Exons)	FOXP4	FOXP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXO6 geni (Tüm Gen Dizi Analizi)	FOXO6 gene (Sequence Analysis-All Coding Exons)	FOXO6	FOXO6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXO4 geni (Tüm Gen Dizi Analizi)	FOXO4 gene (Sequence Analysis-All Coding Exons)	FOXO4	FOXO4, MLLT7, AFX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

FOXO3 geni (Tüm Gen Dizi Analizi)	FOXO3 gene (Sequence Analysis-All Coding Exons)	FOXO3	FOXO3A, FOXO3, FKHRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXN4 geni (Tüm Gen Dizi Analizi)	FOXN4 gene (Sequence Analysis-All Coding Exons)	FOXN4	FOXN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXN3 geni (Tüm Gen Dizi Analizi)	FOXN3 gene (Sequence Analysis-All Coding Exons)	FOXN3	FOXN3, CHES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXN2 geni (Tüm Gen Dizi Analizi)	FOXN2 gene (Sequence Analysis-All Coding Exons)	FOXN2	HTLF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXM1 geni (Tüm Gen Dizi Analizi)	FOXM1 gene (Sequence Analysis-All Coding Exons)	FOXM1	FOXM1, FKHL16, HFH11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXL1 geni (Tüm Gen Dizi Analizi)	FOXL1 gene (Sequence Analysis-All Coding Exons)	FOXL1	FOXL1, FKHL11, FREAC7, FKH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXK2 geni (Tüm Gen Dizi Analizi)	FOXK2 gene (Sequence Analysis-All Coding Exons)	FOXK2	FOXK2, ILF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXK1 geni (Tüm Gen Dizi Analizi)	FOXK1 gene (Sequence Analysis-All Coding Exons)	FOXK1	FOXK1, MNF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXJ3 geni (Tüm Gen Dizi Analizi)	FOXJ3 gene (Sequence Analysis-All Coding Exons)	FOXJ3	FOXJ3, KIAA1041	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXJ1 geni (Tüm Gen Dizi Analizi)	FOXJ1 gene (Sequence Analysis-All Coding Exons)	FOXJ1	FOXJ1, FKHL13, HFH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXI3 geni (Tüm Gen Dizi Analizi)	FOXI3 gene (Sequence Analysis-All Coding Exons)	FOXI3	FOXI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXI2 geni (Tüm Gen Dizi Analizi)	FOXI2 gene (Sequence Analysis-All Coding Exons)	FOXI2	FOXI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXH1 geni (Tüm Gen Dizi Analizi)	FOXH1 gene (Sequence Analysis-All Coding Exons)	FOXH1	FAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXH1 geni (Tüm Gen Dizi Analizi)	FOXH1 gene (Sequence Analysis-All Coding Exons)	FOXH1	FAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXH1 geni (Tüm Gen Dizi Analizi)	FOXH1 gene (Sequence Analysis-All Coding Exons)	FOXH1	FAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXH1 geni (Tüm Gen Dizi Analizi)	FOXH1 gene (Sequence Analysis-All Coding Exons)	FOXH1	FAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXH1 geni (Tüm Gen Dizi Analizi)	FOXH1 gene (Sequence Analysis-All Coding Exons)	FOXH1	FAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXF2 geni (Tüm Gen Dizi Analizi)	FOXF2 gene (Sequence Analysis-All Coding Exons)	FOXF2	FOXF2, FKHL6, FREAC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD4L4 geni (Tüm Gen Dizi Analizi)	FOXD4L4 gene (Sequence Analysis-All Coding Exons)	FOXD4L4	FOXD4L4, FOXD4L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD4L3 geni (Tüm Gen Dizi Analizi)	FOXD4L3 gene (Sequence Analysis-All Coding Exons)	FOXD4L3	FOXD4L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD4L1 geni (Tüm Gen Dizi Analizi)	FOXD4L1 gene (Sequence Analysis-All Coding Exons)	FOXD4L1	FOXD4L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD4 geni (Tüm Gen Dizi Analizi)	FOXD4 gene (Sequence Analysis-All Coding Exons)	FOXD4	FOXD4, FKHL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD2 geni (Tüm Gen Dizi Analizi)	FOXD2 gene (Sequence Analysis-All Coding Exons)	FOXD2	FOXD2, FKHL17, FREAC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXD1 geni (Tüm Gen Dizi Analizi)	FOXD1 gene (Sequence Analysis-All Coding Exons)	FOXD1	FOXD1, FKHL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXA3 geni (Tüm Gen Dizi Analizi)	FOXA3 gene (Sequence Analysis-All Coding Exons)	FOXA3	FOXA3, HNF3G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXA2 geni (Tüm Gen Dizi Analizi)	FOXA2 gene (Sequence Analysis-All Coding Exons)	FOXA2	FOXA2, HNF3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOXA1 geni (Tüm Gen Dizi Analizi)	FOXA1 gene (Sequence Analysis-All Coding Exons)	FOXA1	FOXA1, HNF3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOSL2 geni (Tüm Gen Dizi Analizi)	FOSL2 gene (Sequence Analysis-All Coding Exons)	FOSL2	FOSL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOSL1 geni (Tüm Gen Dizi Analizi)	FOSL1 gene (Sequence Analysis-All Coding Exons)	FOSL1	FOSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOSB geni (Tüm Gen Dizi Analizi)	FOSB gene (Sequence Analysis-All Coding Exons)	FOSB	FOSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOS geni (Tüm Gen Dizi Analizi)	FOS gene (Sequence Analysis-All Coding Exons)	FOS	FOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOPNL geni (Tüm Gen Dizi Analizi)	FOPNL gene (Sequence Analysis-All Coding Exons)	FOPNL	FOPNL, FOR20, C16orf63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOLR2 geni (Tüm Gen Dizi Analizi)	FOLR2 gene (Sequence Analysis-All Coding Exons)	FOLR2	FOLR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOLH1B geni (Tüm Gen Dizi Analizi)	FOLH1B gene (Sequence Analysis-All Coding Exons)	FOLH1B	FOLH1B, PSMAL, GCP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOLH1 geni (Tüm Gen Dizi Analizi)	FOLH1 gene (Sequence Analysis-All Coding Exons)	FOLH1	FOLH1, FOLH, PSM, PSMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOCAD geni (Tüm Gen Dizi Analizi)	FOCAD gene (Sequence Analysis-All Coding Exons)	FOCAD	FOCAD, KIAA1797	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNTB geni (Tüm Gen Dizi Analizi)	FNTB gene (Sequence Analysis-All Coding Exons)	FNTB	FNTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNTA geni (Tüm Gen Dizi Analizi)	FNTA gene (Sequence Analysis-All Coding Exons)	FNTA	FNTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNIP2 geni (Tüm Gen Dizi Analizi)	FNIP2 gene (Sequence Analysis-All Coding Exons)	FNIP2	FNIP2, FNIPL, KIAA1450	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNIP1 geni (Tüm Gen Dizi Analizi)	FNIP1 gene (Sequence Analysis-All Coding Exons)	FNIP1	FNIP1, KIAA1961	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNDC5 geni (Tüm Gen Dizi Analizi)	FNDC5 gene (Sequence Analysis-All Coding Exons)	FNDC5	FNDC5, FRCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNDC4 geni (Tüm Gen Dizi Analizi)	FNDC4 gene (Sequence Analysis-All Coding Exons)	FNDC4	FNDC4, FRCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNDC3B geni (Tüm Gen Dizi Analizi)	FNDC3B gene (Sequence Analysis-All Coding Exons)	FNDC3B	FNDC3B, FAD104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNDC3A geni (Tüm Gen Dizi Analizi)	FNDC3A gene (Sequence Analysis-All Coding Exons)	FNDC3A	FNDC3A, KIAA0970, HUGO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNDC1 geni (Tüm Gen Dizi Analizi)	FNDC1 gene (Sequence Analysis-All Coding Exons)	FNDC1	FNDC1, AG58, KIAA1866	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNBP4 geni (Tüm Gen Dizi Analizi)	FNBP4 gene (Sequence Analysis-All Coding Exons)	FNBP4	FNBP4, FBP30, KIAA1014	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNBP1L geni (Tüm Gen Dizi Analizi)	FNBP1L gene (Sequence Analysis-All Coding Exons)	FNBP1L	FNBP1L, TOCA1, C1orf39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FNBP1 geni (Tüm Gen Dizi Analizi)	FNBP1 gene (Sequence Analysis-All Coding Exons)	FNBP1	FNBP1, FBP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FN3KRP geni (Tüm Gen Dizi Analizi)	FN3KRP gene (Sequence Analysis-All Coding Exons)	FN3KRP	FN3KRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FN3K geni (Tüm Gen Dizi Analizi)	FN3K gene (Sequence Analysis-All Coding Exons)	FN3K	FN3K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMR1-AS1 geni (Tüm Gen Dizi Analizi)	FMR1-AS1 gene (Sequence Analysis-All Coding Exons)	FMR1-AS1	FMR1AS1, ASFMR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMOD geni (Tüm Gen Dizi Analizi)	FMOD gene (Sequence Analysis-All Coding Exons)	FMOD	FMOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMO5 geni (Tüm Gen Dizi Analizi)	FMO5 gene (Sequence Analysis-All Coding Exons)	FMO5	FMO5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMO4 geni (Tüm Gen Dizi Analizi)	FMO4 gene (Sequence Analysis-All Coding Exons)	FMO4	FMO4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMO2 geni (Tüm Gen Dizi Analizi)	FMO2 gene (Sequence Analysis-All Coding Exons)	FMO2	FMO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMO1 geni (Tüm Gen Dizi Analizi)	FMO1 gene (Sequence Analysis-All Coding Exons)	FMO1	FMO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMNL3 geni (Tüm Gen Dizi Analizi)	FMNL3 gene (Sequence Analysis-All Coding Exons)	FMNL3	FMNL3, FRL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMNL2 geni (Tüm Gen Dizi Analizi)	FMNL2 gene (Sequence Analysis-All Coding Exons)	FMNL2	FMNL2, KIAA1902	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMNL1 geni (Tüm Gen Dizi Analizi)	FMNL1 gene (Sequence Analysis-All Coding Exons)	FMNL1	FMNL, C17orf1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FMN1 geni (Tüm Gen Dizi Analizi)	FMN1 gene (Sequence Analysis-All Coding Exons)	FMN1	FMN, LD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLVCR1-AS1 geni (Tüm Gen Dizi Analizi)	FLVCR1-AS1 gene (Sequence Analysis-All Coding Exons)	FLVCR1-AS1	LQK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLT1 geni (Tüm Gen Dizi Analizi)	FLT1 gene (Sequence Analysis-All Coding Exons)	FLT1	FLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLRT2 geni (Tüm Gen Dizi Analizi)	FLRT2 gene (Sequence Analysis-All Coding Exons)	FLRT2	FLRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLRT1 geni (Tüm Gen Dizi Analizi)	FLRT1 gene (Sequence Analysis-All Coding Exons)	FLRT1	FLRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPZL1 geni (Tüm Gen Dizi Analizi)	MPZL1 gene (Sequence Analysis-All Coding Exons)	MPZL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLOT2 geni (Tüm Gen Dizi Analizi)	FLOT2 gene (Sequence Analysis-All Coding Exons)	FLOT2	FLOT2, M17S1, ECS1, ESA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLOT1 geni (Tüm Gen Dizi Analizi)	FLOT1 gene (Sequence Analysis-All Coding Exons)	FLOT1	FLOT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLII geni (Tüm Gen Dizi Analizi)	FLII gene (Sequence Analysis-All Coding Exons)	FLII	FLII	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLI1 geni (Tüm Gen Dizi Analizi)	FLI1 gene (Sequence Analysis-All Coding Exons)	FLI1	FLI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLI1 geni (Tüm Gen Dizi Analizi)	FLI1 gene (Sequence Analysis-All Coding Exons)	FLI1	FLI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLI1 geni (Tüm Gen Dizi Analizi)	FLI1 gene (Sequence Analysis-All Coding Exons)	FLI1	FLI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLI1 geni (Tüm Gen Dizi Analizi)	FLI1 gene (Sequence Analysis-All Coding Exons)	FLI1	FLI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLI1 geni (Tüm Gen Dizi Analizi)	FLI1 gene (Sequence Analysis-All Coding Exons)	FLI1	FLI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLG2 geni (Tüm Gen Dizi Analizi)	FLG2 gene (Sequence Analysis-All Coding Exons)	FLG2	FLG2, IFPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBPL geni (Tüm Gen Dizi Analizi)	FKBPL gene (Sequence Analysis-All Coding Exons)	FKBPL	FKBPL, DIR1, WISP39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP9 geni (Tüm Gen Dizi Analizi)	FKBP9 gene (Sequence Analysis-All Coding Exons)	FKBP9	FKBP9, FKBP60, FKBP63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP8 geni (Tüm Gen Dizi Analizi)	FKBP8 gene (Sequence Analysis-All Coding Exons)	FKBP8	FKBP8, FKBP38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP7 geni (Tüm Gen Dizi Analizi)	FKBP7 gene (Sequence Analysis-All Coding Exons)	FKBP7	FKBP7, FKBP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP6 geni (Tüm Gen Dizi Analizi)	FKBP6 gene (Sequence Analysis-All Coding Exons)	FKBP6	FKBP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP4 geni (Tüm Gen Dizi Analizi)	FKBP4 gene (Sequence Analysis-All Coding Exons)	FKBP4	FKBP4, FKBP52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP3 geni (Tüm Gen Dizi Analizi)	FKBP3 gene (Sequence Analysis-All Coding Exons)	FKBP3	FKBP3, FKBP25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP2 geni (Tüm Gen Dizi Analizi)	FKBP2 gene (Sequence Analysis-All Coding Exons)	FKBP2	FKBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP1B geni (Tüm Gen Dizi Analizi)	FKBP1B gene (Sequence Analysis-All Coding Exons)	FKBP1B	FKBP1B, PKBP1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP1A geni (Tüm Gen Dizi Analizi)	FKBP1A gene (Sequence Analysis-All Coding Exons)	FKBP1A	FKBP1A, FKBP12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FKBP11 geni (Tüm Gen Dizi Analizi)	FKBP11 gene (Sequence Analysis-All Coding Exons)	FKBP11	FKBP11, FKBP19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FJX1 geni (Tüm Gen Dizi Analizi)	FJX1 gene (Sequence Analysis-All Coding Exons)	FJX1	FJX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIZ1 geni (Tüm Gen Dizi Analizi)	FIZ1 gene (Sequence Analysis-All Coding Exons)	FIZ1	FIZ1, FLJ14768	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FITM2 geni (Tüm Gen Dizi Analizi)	FITM2 gene (Sequence Analysis-All Coding Exons)	FITM2	FITM2, C20orf142	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FITM1 geni (Tüm Gen Dizi Analizi)	FITM1 gene (Sequence Analysis-All Coding Exons)	FITM1	FIT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIS1 geni (Tüm Gen Dizi Analizi)	FIS1 gene (Sequence Analysis-All Coding Exons)	FIS1	TTC11, FIS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIP1L1 geni (Tüm Gen Dizi Analizi)	FIP1L1 gene (Sequence Analysis-All Coding Exons)	FIP1L1	FIP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIP1L1 geni (Tüm Gen Dizi Analizi)	FIP1L1 gene (Sequence Analysis-All Coding Exons)	FIP1L1	FIP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

FIP1L1 geni (Tüm Gen Dizi Analizi)	FIP1L1 gene (Sequence Analysis-All Coding Exons)	FIP1L1	FIP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPIG6B geni (Tüm Gen Dizi Analizi)	MPIG6B gene (Sequence Analysis-All Coding Exons)	MPIG6B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FILIP1L geni (Tüm Gen Dizi Analizi)	FILIP1L gene (Sequence Analysis-All Coding Exons)	FILIP1L	FILIP1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FILIP1 geni (Tüm Gen Dizi Analizi)	FILIP1 gene (Sequence Analysis-All Coding Exons)	FILIP1	FILIP, KIAA1275	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIGNL1 geni (Tüm Gen Dizi Analizi)	FIGNL1 gene (Sequence Analysis-All Coding Exons)	FIGNL1	FIGNL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIGN geni (Tüm Gen Dizi Analizi)	FIGN gene (Sequence Analysis-All Coding Exons)	FIGN	FIGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MPHOSPH10 geni (Tüm Gen Dizi Analizi)	MPHOSPH10 gene (Sequence Analysis-All Coding Exons)	MPHOSPH10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIBP geni (Tüm Gen Dizi Analizi)	FIBP gene (Sequence Analysis-All Coding Exons)	FIBP	FIBP, TROFAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIBIN geni (Tüm Gen Dizi Analizi)	FIBIN gene (Sequence Analysis-All Coding Exons)	FIBIN	FIBIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FIBCD1 geni (Tüm Gen Dizi Analizi)	FIBCD1 gene (Sequence Analysis-All Coding Exons)	FIBCD1	FIBCD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHOD3 geni (Tüm Gen Dizi Analizi)	FHOD3 gene (Sequence Analysis-All Coding Exons)	FHOD3	FHOD3, FHOS2, KIAA1695	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHOD1 geni (Tüm Gen Dizi Analizi)	FHOD1 gene (Sequence Analysis-All Coding Exons)	FHOD1	FHOD1, FHOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHL5 geni (Tüm Gen Dizi Analizi)	FHL5 gene (Sequence Analysis-All Coding Exons)	FHL5	FHL5, ACT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHL3 geni (Tüm Gen Dizi Analizi)	FHL3 gene (Sequence Analysis-All Coding Exons)	FHL3	FHL3, SLIM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHL2 geni (Tüm Gen Dizi Analizi)	FHL2 gene (Sequence Analysis-All Coding Exons)	FHL2	FHL2, DRAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FHIT geni (Tüm Gen Dizi Analizi)	FHIT gene (Sequence Analysis-All Coding Exons)	FHIT	FRA3B, FHIT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOSPD3 geni (Tüm Gen Dizi Analizi)	MOSPD3 gene (Sequence Analysis-All Coding Exons)	MOSPD3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGR geni (Tüm Gen Dizi Analizi)	FGR gene (Sequence Analysis-All Coding Exons)	FGR	FGR, SRC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGL2 geni (Tüm Gen Dizi Analizi)	FGL2 gene (Sequence Analysis-All Coding Exons)	FGL2	FGL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGL1 geni (Tüm Gen Dizi Analizi)	FGL1 gene (Sequence Analysis-All Coding Exons)	FGL1	FGL1, HFREP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGGY geni (Tüm Gen Dizi Analizi)	FGGY gene (Sequence Analysis-All Coding Exons)	FGGY	FGGY, FLJ10986	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGFRL1 geni (Tüm Gen Dizi Analizi)	FGFRL1 gene (Sequence Analysis-All Coding Exons)	FGFRL1	FGFRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGFBP2 geni (Tüm Gen Dizi Analizi)	FGFBP2 gene (Sequence Analysis-All Coding Exons)	FGFBP2	KSP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGFBP1 geni (Tüm Gen Dizi Analizi)	FGFBP1 gene (Sequence Analysis-All Coding Exons)	FGFBP1	FGFBP1, FGFBP, HBP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF7 geni (Tüm Gen Dizi Analizi)	FGF7 gene (Sequence Analysis-All Coding Exons)	FGF7	FGF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF6 geni (Tüm Gen Dizi Analizi)	FGF6 gene (Sequence Analysis-All Coding Exons)	FGF6	FGF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF4 geni (Tüm Gen Dizi Analizi)	FGF4 gene (Sequence Analysis-All Coding Exons)	FGF4	FGF4, HSTF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF22 geni (Tüm Gen Dizi Analizi)	FGF22 gene (Sequence Analysis-All Coding Exons)	FGF22	FGF22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF21 geni (Tüm Gen Dizi Analizi)	FGF21 gene (Sequence Analysis-All Coding Exons)	FGF21	FGF21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF2 geni (Tüm Gen Dizi Analizi)	FGF2 gene (Sequence Analysis-All Coding Exons)	FGF2	FGF2, FGFB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF19 geni (Tüm Gen Dizi Analizi)	FGF19 gene (Sequence Analysis-All Coding Exons)	FGF19	FGF19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF18 geni (Tüm Gen Dizi Analizi)	FGF18 gene (Sequence Analysis-All Coding Exons)	FGF18	FGF18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF13 geni (Tüm Gen Dizi Analizi)	FGF13 gene (Sequence Analysis-All Coding Exons)	FGF13	FGF13, FGF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF11 geni (Tüm Gen Dizi Analizi)	FGF11 gene (Sequence Analysis-All Coding Exons)	FGF11	FGF11, FHF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOCS3 geni (Tüm Gen Dizi Analizi)	MOCS3 gene (Sequence Analysis-All Coding Exons)	MOCS3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGF1 geni (Tüm Gen Dizi Analizi)	FGF1 gene (Sequence Analysis-All Coding Exons)	FGF1	FGF1, FGFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGD6 geni (Tüm Gen Dizi Analizi)	FGD6 gene (Sequence Analysis-All Coding Exons)	FGD6	FGD6, ZFYVE24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGD5 geni (Tüm Gen Dizi Analizi)	FGD5 gene (Sequence Analysis-All Coding Exons)	FGD5	FGD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGD2 geni (Tüm Gen Dizi Analizi)	FGD2 gene (Sequence Analysis-All Coding Exons)	FGD2	FGD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FFAR3 geni (Tüm Gen Dizi Analizi)	FFAR3 gene (Sequence Analysis-All Coding Exons)	FFAR3	FFAR3, GPR41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FFAR2 geni (Tüm Gen Dizi Analizi)	FFAR2 gene (Sequence Analysis-All Coding Exons)	FFAR2	FFAR2, GPR43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOB1B geni (Tüm Gen Dizi Analizi)	MOB1B gene (Sequence Analysis-All Coding Exons)	MOB1B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MOB1A geni (Tüm Gen Dizi Analizi)	MOB1A gene (Sequence Analysis-All Coding Exons)	MOB1A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FFAR1 geni (Tüm Gen Dizi Analizi)	FFAR1 gene (Sequence Analysis-All Coding Exons)	FFAR1	FFAR1, GPR40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEZF2 geni (Tüm Gen Dizi Analizi)	FEZF2 gene (Sequence Analysis-All Coding Exons)	FEZF2	FEZF2, ZNF312, FEZL, TOF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEZ2 geni (Tüm Gen Dizi Analizi)	FEZ2 gene (Sequence Analysis-All Coding Exons)	FEZ2	FEZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEZ1 geni (Tüm Gen Dizi Analizi)	FEZ1 gene (Sequence Analysis-All Coding Exons)	FEZ1	FEZ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEV geni (Tüm Gen Dizi Analizi)	FEV gene (Sequence Analysis-All Coding Exons)	FEV	FEV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FETUB geni (Tüm Gen Dizi Analizi)	FETUB gene (Sequence Analysis-All Coding Exons)	FETUB	FETUB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FES geni (Tüm Gen Dizi Analizi)	FES gene (Sequence Analysis-All Coding Exons)	FES	FES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FERMT2 geni (Tüm Gen Dizi Analizi)	FERMT2 gene (Sequence Analysis-All Coding Exons)	FERMT2	FERMT2, PLEKHC1, MIG2, UNC112, KIND2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FER geni (Tüm Gen Dizi Analizi)	FER gene (Sequence Analysis-All Coding Exons)	FER	FER, TYK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FENDRR geni (Tüm Gen Dizi Analizi)	FENDRR gene (Sequence Analysis-All Coding Exons)	FENDRR	FOXF1AS1, TCONS_00024240	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMRN2 geni (Tüm Gen Dizi Analizi)	MMRN2 gene (Sequence Analysis-All Coding Exons)	MMRN2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MMRN1 geni (Tüm Gen Dizi Analizi)	MMRN1 gene (Sequence Analysis-All Coding Exons)	MMRN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEN1 geni (Tüm Gen Dizi Analizi)	FEN1 gene (Sequence Analysis-All Coding Exons)	FEN1	FEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEM1B geni (Tüm Gen Dizi Analizi)	FEM1B gene (Sequence Analysis-All Coding Exons)	FEM1B	FEM1B, KIAA0396	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEM1A geni (Tüm Gen Dizi Analizi)	FEM1A gene (Sequence Analysis-All Coding Exons)	FEM1A	FEM1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FDXR geni (Tüm Gen Dizi Analizi)	FDXR gene (Sequence Analysis-All Coding Exons)	FDXR	FDXR, ADXR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FDX1L geni (Tüm Gen Dizi Analizi)	FDX1L gene (Sequence Analysis-All Coding Exons)	FDX1L	FDX1L, FDX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FDX1 geni (Tüm Gen Dizi Analizi)	FDX1 gene (Sequence Analysis-All Coding Exons)	FDX1	FDX1, ADX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FDFT1 geni (Tüm Gen Dizi Analizi)	FDFT1 gene (Sequence Analysis-All Coding Exons)	FDFT1	FDFT1, DGPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRLB geni (Tüm Gen Dizi Analizi)	FCRLB gene (Sequence Analysis-All Coding Exons)	FCRLB	FCRLB, FCRL2, FREB2, FCRY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRLA geni (Tüm Gen Dizi Analizi)	FCRLA gene (Sequence Analysis-All Coding Exons)	FCRLA	FREB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL5 geni (Tüm Gen Dizi Analizi)	FCRL5 gene (Sequence Analysis-All Coding Exons)	FCRL5	IRTA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL4 geni (Tüm Gen Dizi Analizi)	FCRL4 gene (Sequence Analysis-All Coding Exons)	FCRL4	IRTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL3 geni (Tüm Gen Dizi Analizi)	FCRL3 gene (Sequence Analysis-All Coding Exons)	FCRL3	FCRH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL2 geni (Tüm Gen Dizi Analizi)	FCRL2 gene (Sequence Analysis-All Coding Exons)	FCRL2	SPAP1, SPAP1A, SPAP1B, SPAP1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL1 geni (Tüm Gen Dizi Analizi)	FCRL1 gene (Sequence Analysis-All Coding Exons)	FCRL1	FCRL1, FCRH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCN2 geni (Tüm Gen Dizi Analizi)	FCN2 gene (Sequence Analysis-All Coding Exons)	FCN2	FCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCN1 geni (Tüm Gen Dizi Analizi)	FCN1 gene (Sequence Analysis-All Coding Exons)	FCN1	FCN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCMR geni (Tüm Gen Dizi Analizi)	FCMR gene (Sequence Analysis-All Coding Exons)	FCMR	FAIM3, TOSO, FCMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCHO2 geni (Tüm Gen Dizi Analizi)	FCHO2 gene (Sequence Analysis-All Coding Exons)	FCHO2	FCHO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCHO1 geni (Tüm Gen Dizi Analizi)	FCHO1 gene (Sequence Analysis-All Coding Exons)	FCHO1	FCHO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCGRT geni (Tüm Gen Dizi Analizi)	FCGRT gene (Sequence Analysis-All Coding Exons)	FCGRT	FCGRT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCGR1B geni (Tüm Gen Dizi Analizi)	FCGR1B gene (Sequence Analysis-All Coding Exons)	FCGR1B	FCGR1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCER2 geni (Tüm Gen Dizi Analizi)	FCER2 gene (Sequence Analysis-All Coding Exons)	FCER2	FCER2, FCE2, CD23, CLEC4J	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCER1G geni (Tüm Gen Dizi Analizi)	FCER1G gene (Sequence Analysis-All Coding Exons)	FCER1G	FCER1G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCER1A geni (Tüm Gen Dizi Analizi)	FCER1A gene (Sequence Analysis-All Coding Exons)	FCER1A	FCER1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCAR geni (Tüm Gen Dizi Analizi)	FCAR gene (Sequence Analysis-All Coding Exons)	FCAR	FCAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCAMR geni (Tüm Gen Dizi Analizi)	FCAMR gene (Sequence Analysis-All Coding Exons)	FCAMR	FCAMR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW9 geni (Tüm Gen Dizi Analizi)	FBXW9 gene (Sequence Analysis-All Coding Exons)	FBXW9	FBXW9, FBW9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW8 geni (Tüm Gen Dizi Analizi)	FBXW8 gene (Sequence Analysis-All Coding Exons)	FBXW8	FBXW8, FBW8, FBXW6, FBW6, FBXO29, FBX29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW7 geni (Tüm Gen Dizi Analizi)	FBXW7 gene (Sequence Analysis-All Coding Exons)	FBXW7	FBXW7, AGO, CDC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW5 geni (Tüm Gen Dizi Analizi)	FBXW5 gene (Sequence Analysis-All Coding Exons)	FBXW5	FBXW5, FBW5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW4 geni (Tüm Gen Dizi Analizi)	FBXW4 gene (Sequence Analysis-All Coding Exons)	FBXW4	FBXW4, DAC, FBW4, FBWD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW2 geni (Tüm Gen Dizi Analizi)	FBXW2 gene (Sequence Analysis-All Coding Exons)	FBXW2	FBXW2, FBW2, FWD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW12 geni (Tüm Gen Dizi Analizi)	FBXW12 gene (Sequence Analysis-All Coding Exons)	FBXW12	FBXW12, FBW12, FBXO35, FBWD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW11 geni (Tüm Gen Dizi Analizi)	FBXW11 gene (Sequence Analysis-All Coding Exons)	FBXW11	FBXW11, FBXW1B, BTRC2, BTRCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXW10 geni (Tüm Gen Dizi Analizi)	FBXW10 gene (Sequence Analysis-All Coding Exons)	FBXW10	FBXW10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO9 geni (Tüm Gen Dizi Analizi)	FBXO9 gene (Sequence Analysis-All Coding Exons)	FBXO9	FBXO9, FBX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO8 geni (Tüm Gen Dizi Analizi)	FBXO8 gene (Sequence Analysis-All Coding Exons)	FBXO8	FBXO8, FBX8, FBS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO6 geni (Tüm Gen Dizi Analizi)	FBXO6 gene (Sequence Analysis-All Coding Exons)	FBXO6	FBXO6, FBX6, FBG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

FBXO5 geni (Tüm Gen Dizi Analizi)	FBXO5 gene (Sequence Analysis-All Coding Exons)	FBXO5	FBXO5, FBX5, EMI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO47 geni (Tüm Gen Dizi Analizi)	FBXO47 gene (Sequence Analysis-All Coding Exons)	FBXO47	FBXO47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO46 geni (Tüm Gen Dizi Analizi)	FBXO46 gene (Sequence Analysis-All Coding Exons)	FBXO46	FBXO46, FBX46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO45 geni (Tüm Gen Dizi Analizi)	FBXO45 gene (Sequence Analysis-All Coding Exons)	FBXO45	FBXO45, FBX45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO44 geni (Tüm Gen Dizi Analizi)	FBXO44 gene (Sequence Analysis-All Coding Exons)	FBXO44	FBXO44, FBX44, FBX6A, FBX9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO43 geni (Tüm Gen Dizi Analizi)	FBXO43 gene (Sequence Analysis-All Coding Exons)	FBXO43	FBXO43, EMI2, ERP1, FBX43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO42 geni (Tüm Gen Dizi Analizi)	FBXO42 gene (Sequence Analysis-All Coding Exons)	FBXO42	FBXO42, FBX42, KIAA1332	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO41 geni (Tüm Gen Dizi Analizi)	FBXO41 gene (Sequence Analysis-All Coding Exons)	FBXO41	FBXO41, FBX41, KIAA1940	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO40 geni (Tüm Gen Dizi Analizi)	FBXO40 gene (Sequence Analysis-All Coding Exons)	FBXO40	FBXO40, FBX40, KIAA1195	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO4 geni (Tüm Gen Dizi Analizi)	FBXO4 gene (Sequence Analysis-All Coding Exons)	FBXO4	FBXO4, FBX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO39 geni (Tüm Gen Dizi Analizi)	FBXO39 gene (Sequence Analysis-All Coding Exons)	FBXO39	FBXO39, FBX39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO36 geni (Tüm Gen Dizi Analizi)	FBXO36 gene (Sequence Analysis-All Coding Exons)	FBXO36	FBXO36, FBX36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO34 geni (Tüm Gen Dizi Analizi)	FBXO34 gene (Sequence Analysis-All Coding Exons)	FBXO34	FBXO34, FBX34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO33 geni (Tüm Gen Dizi Analizi)	FBXO33 gene (Sequence Analysis-All Coding Exons)	FBXO33	FBXO33, FBX33, BMND12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO32 geni (Tüm Gen Dizi Analizi)	FBXO32 gene (Sequence Analysis-All Coding Exons)	FBXO32	FBXO32, MAFBX, FLJ32424, ATROGIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO30 geni (Tüm Gen Dizi Analizi)	FBXO30 gene (Sequence Analysis-All Coding Exons)	FBXO30	FBXO30, FBX30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO3 geni (Tüm Gen Dizi Analizi)	FBXO3 gene (Sequence Analysis-All Coding Exons)	FBXO3	FBXO3, FBX3, FBA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO28 geni (Tüm Gen Dizi Analizi)	FBXO28 gene (Sequence Analysis-All Coding Exons)	FBXO28	FBXO28, FBX28, KIAA0483	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO27 geni (Tüm Gen Dizi Analizi)	FBXO27 gene (Sequence Analysis-All Coding Exons)	FBXO27	FBXO27, FBX27, FBG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO25 geni (Tüm Gen Dizi Analizi)	FBXO25 gene (Sequence Analysis-All Coding Exons)	FBXO25	FBXO25, FBX25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO24 geni (Tüm Gen Dizi Analizi)	FBXO24 gene (Sequence Analysis-All Coding Exons)	FBXO24	FBXO24, FBX24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO22 geni (Tüm Gen Dizi Analizi)	FBXO22 gene (Sequence Analysis-All Coding Exons)	FBXO22	FBXO22, FBX22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO21 geni (Tüm Gen Dizi Analizi)	FBXO21 gene (Sequence Analysis-All Coding Exons)	FBXO21	FBXO21, FBX21, KIAA0875	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MKRN1 geni (Tüm Gen Dizi Analizi)	MKRN1 gene (Sequence Analysis-All Coding Exons)	MKRN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO2 geni (Tüm Gen Dizi Analizi)	FBXO2 gene (Sequence Analysis-All Coding Exons)	FBXO2	FBXO2, FBX2, FBG1, NFB42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO18 geni (Tüm Gen Dizi Analizi)	FBXO18 gene (Sequence Analysis-All Coding Exons)	FBXO18	FBXO18, FBH1, FBX18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO17 geni (Tüm Gen Dizi Analizi)	FBXO17 gene (Sequence Analysis-All Coding Exons)	FBXO17	FBXO17, FBX17, FBX26, FBG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO16 geni (Tüm Gen Dizi Analizi)	FBXO16 gene (Sequence Analysis-All Coding Exons)	FBXO16	FBXO16, FBX16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO15 geni (Tüm Gen Dizi Analizi)	FBXO15 gene (Sequence Analysis-All Coding Exons)	FBXO15	FBXO15, FBX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO11 geni (Tüm Gen Dizi Analizi)	FBXO11 gene (Sequence Analysis-All Coding Exons)	FBXO11	FBXO11, FBX11, VIT1, PRMT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXO10 geni (Tüm Gen Dizi Analizi)	FBXO10 gene (Sequence Analysis-All Coding Exons)	FBXO10	FBXO10, FBX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL8 geni (Tüm Gen Dizi Analizi)	FBXL8 gene (Sequence Analysis-All Coding Exons)	FBXL8	FBXL8, FBL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL7 geni (Tüm Gen Dizi Analizi)	FBXL7 gene (Sequence Analysis-All Coding Exons)	FBXL7	FBXL7, FBL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL6 geni (Tüm Gen Dizi Analizi)	FBXL6 gene (Sequence Analysis-All Coding Exons)	FBXL6	FBXL6, FBL6, FBL6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL5 geni (Tüm Gen Dizi Analizi)	FBXL5 gene (Sequence Analysis-All Coding Exons)	FBXL5	FBXL5, FBL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL3 geni (Tüm Gen Dizi Analizi)	FBXL3 gene (Sequence Analysis-All Coding Exons)	FBXL3	FBXL3A, FBL3A, FBL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL22 geni (Tüm Gen Dizi Analizi)	FBXL22 gene (Sequence Analysis-All Coding Exons)	FBXL22	FBXL22, FBL22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL20 geni (Tüm Gen Dizi Analizi)	FBXL20 gene (Sequence Analysis-All Coding Exons)	FBXL20	FBXL20, FBL20, FBL2, SCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL2 geni (Tüm Gen Dizi Analizi)	FBXL2 gene (Sequence Analysis-All Coding Exons)	FBXL2	FBXL2, FBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL19 geni (Tüm Gen Dizi Analizi)	FBXL19 gene (Sequence Analysis-All Coding Exons)	FBXL19	FBXL19, FBL19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL18 geni (Tüm Gen Dizi Analizi)	FBXL18 gene (Sequence Analysis-All Coding Exons)	FBXL18	FBXL18, FBL18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL17 geni (Tüm Gen Dizi Analizi)	FBXL17 gene (Sequence Analysis-All Coding Exons)	FBXL17	FBXL17, FBL17, FBX13, FBXO13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL16 geni (Tüm Gen Dizi Analizi)	FBXL16 gene (Sequence Analysis-All Coding Exons)	FBXL16	FBXL16, FBL16, C16orf22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL15 geni (Tüm Gen Dizi Analizi)	FBXL15 gene (Sequence Analysis-All Coding Exons)	FBXL15	FBXL15, JET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL14 geni (Tüm Gen Dizi Analizi)	FBXL14 gene (Sequence Analysis-All Coding Exons)	FBXL14	FBXL14, FBL14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL13 geni (Tüm Gen Dizi Analizi)	FBXL13 gene (Sequence Analysis-All Coding Exons)	FBXL13	FBXL13, FBL13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBXL12 geni (Tüm Gen Dizi Analizi)	FBXL12 gene (Sequence Analysis-All Coding Exons)	FBXL12	FBXL12, FBL12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBRS geni (Tüm Gen Dizi Analizi)	FBRS gene (Sequence Analysis-All Coding Exons)	FBRS	FBRS, FBS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBP2 geni (Tüm Gen Dizi Analizi)	FBP2 gene (Sequence Analysis-All Coding Exons)	FBP2	FBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBLN7 geni (Tüm Gen Dizi Analizi)	FBLN7 gene (Sequence Analysis-All Coding Exons)	FBLN7	FBLN7, TM14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBLN2 geni (Tüm Gen Dizi Analizi)	FBLN2 gene (Sequence Analysis-All Coding Exons)	FBLN2	FBLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBLIM1 geni (Tüm Gen Dizi Analizi)	FBLIM1 gene (Sequence Analysis-All Coding Exons)	FBLIM1	FBLP1, MIGFILIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBL geni (Tüm Gen Dizi Analizi)	FBL gene (Sequence Analysis-All Coding Exons)	FBL	FBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBF1 geni (Tüm Gen Dizi Analizi)	FBF1 gene (Sequence Analysis-All Coding Exons)	FBF1	FGF1, ALB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAU geni (Tüm Gen Dizi Analizi)	FAU gene (Sequence Analysis-All Coding Exons)	FAU	FAU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FATE1 geni (Tüm Gen Dizi Analizi)	FATE1 gene (Sequence Analysis-All Coding Exons)	FATE1	FATE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAT3 geni (Tüm Gen Dizi Analizi)	FAT3 gene (Sequence Analysis-All Coding Exons)	FAT3	FAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAT2 geni (Tüm Gen Dizi Analizi)	FAT2 gene (Sequence Analysis-All Coding Exons)	FAT2	FAT2, MEGF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAT1 geni (Tüm Gen Dizi Analizi)	FAT1 gene (Sequence Analysis-All Coding Exons)	FAT1	FAT1, FAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FASTKD5 geni (Tüm Gen Dizi Analizi)	FASTKD5 gene (Sequence Analysis-All Coding Exons)	FASTKD5	FASTKD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FASTK geni (Tüm Gen Dizi Analizi)	FASTK gene (Sequence Analysis-All Coding Exons)	FASTK	FASTK, FAST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FASN geni (Tüm Gen Dizi Analizi)	FASN gene (Sequence Analysis-All Coding Exons)	FASN	FASN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FARSB geni (Tüm Gen Dizi Analizi)	FARSB gene (Sequence Analysis-All Coding Exons)	FARSB	FARSLB, FRSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FARP1 geni (Tüm Gen Dizi Analizi)	FARP1 gene (Sequence Analysis-All Coding Exons)	FARP1	FARP1, CDEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAR2 geni (Tüm Gen Dizi Analizi)	FAR2 gene (Sequence Analysis-All Coding Exons)	FAR2	FAR2, MLSTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAP geni (Tüm Gen Dizi Analizi)	FAP gene (Sequence Analysis-All Coding Exons)	FAP	FAP, FAPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FANK1 geni (Tüm Gen Dizi Analizi)	FANK1 gene (Sequence Analysis-All Coding Exons)	FANK1	FANK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FANCM geni (Tüm Gen Dizi Analizi)	FANCM gene (Sequence Analysis-All Coding Exons)	FANCM	FANCM, KIAA1596	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM9C geni (Tüm Gen Dizi Analizi)	FAM9C gene (Sequence Analysis-All Coding Exons)	FAM9C	FAM9C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM9B geni (Tüm Gen Dizi Analizi)	FAM9B gene (Sequence Analysis-All Coding Exons)	FAM9B	FAM9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM9A geni (Tüm Gen Dizi Analizi)	FAM9A gene (Sequence Analysis-All Coding Exons)	FAM9A	FAM9A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM98B geni (Tüm Gen Dizi Analizi)	FAM98B gene (Sequence Analysis-All Coding Exons)	FAM98B	FAM98B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM96B geni (Tüm Gen Dizi Analizi)	FAM96B gene (Sequence Analysis-All Coding Exons)	FAM96B	FAM96B, MIP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM92B geni (Tüm Gen Dizi Analizi)	FAM92B gene (Sequence Analysis-All Coding Exons)	FAM92B	FAM92B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM92A geni (Tüm Gen Dizi Analizi)	FAM92A gene (Sequence Analysis-All Coding Exons)	FAM92A	FAM92A, FAM92A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM90A7P geni (Tüm Gen Dizi Analizi)	FAM90A7P gene (Sequence Analysis-All Coding Exons)	FAM90A7P	FAM90A7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM90A1 geni (Tüm Gen Dizi Analizi)	FAM90A1 gene (Sequence Analysis-All Coding Exons)	FAM90A1	FAM90A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM89B geni (Tüm Gen Dizi Analizi)	FAM89B gene (Sequence Analysis-All Coding Exons)	FAM89B	FAM89B, MTVR, LRAP25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM86C1 geni (Tüm Gen Dizi Analizi)	FAM86C1 gene (Sequence Analysis-All Coding Exons)	FAM86C1	FAM86C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM86B2 geni (Tüm Gen Dizi Analizi)	FAM86B2 gene (Sequence Analysis-All Coding Exons)	FAM86B2	FAM86B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM86B1 geni (Tüm Gen Dizi Analizi)	FAM86B1 gene (Sequence Analysis-All Coding Exons)	FAM86B1	FAM86B1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM84B geni (Tüm Gen Dizi Analizi)	FAM84B gene (Sequence Analysis-All Coding Exons)	FAM84B	FAM84B, BCMP101	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM84A geni (Tüm Gen Dizi Analizi)	FAM84A gene (Sequence Analysis-All Coding Exons)	FAM84A	FAM84A, NSE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM72D geni (Tüm Gen Dizi Analizi)	FAM72D gene (Sequence Analysis-All Coding Exons)	FAM72D	FAM72D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM72C geni (Tüm Gen Dizi Analizi)	FAM72C gene (Sequence Analysis-All Coding Exons)	FAM72C	FAM72C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM72B geni (Tüm Gen Dizi Analizi)	FAM72B gene (Sequence Analysis-All Coding Exons)	FAM72B	FAM72B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM72A geni (Tüm Gen Dizi Analizi)	FAM72A gene (Sequence Analysis-All Coding Exons)	FAM72A	FAM72A, LMPIP, UGENE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM69C geni (Tüm Gen Dizi Analizi)	FAM69C gene (Sequence Analysis-All Coding Exons)	FAM69C	FAM69C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM69B geni (Tüm Gen Dizi Analizi)	FAM69B gene (Sequence Analysis-All Coding Exons)	FAM69B	FAM69B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM69A geni (Tüm Gen Dizi Analizi)	FAM69A gene (Sequence Analysis-All Coding Exons)	FAM69A	FAM69A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM60A geni (Tüm Gen Dizi Analizi)	FAM60A gene (Sequence Analysis-All Coding Exons)	FAM60A	FAM60A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM57B geni (Tüm Gen Dizi Analizi)	FAM57B gene (Sequence Analysis-All Coding Exons)	FAM57B	FAM57B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM57A geni (Tüm Gen Dizi Analizi)	FAM57A gene (Sequence Analysis-All Coding Exons)	FAM57A	FAM57A, CT120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

FAM53C geni (Tüm Gen Dizi Analizi)	FAM53C gene (Sequence Analysis-All Coding Exons)	FAM53C	FAM53C, C5orf6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM53B geni (Tüm Gen Dizi Analizi)	FAM53B gene (Sequence Analysis-All Coding Exons)	FAM53B	FAM53B, KIAA0140	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM53A geni (Tüm Gen Dizi Analizi)	FAM53A gene (Sequence Analysis-All Coding Exons)	FAM53A	FAM53A, DNTNP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM50B geni (Tüm Gen Dizi Analizi)	FAM50B gene (Sequence Analysis-All Coding Exons)	FAM50B	FAM60B, XSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM50A geni (Tüm Gen Dizi Analizi)	FAM50A gene (Sequence Analysis-All Coding Exons)	FAM50A	FAM50A, DXS9928E, HXC26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM46D geni (Tüm Gen Dizi Analizi)	FAM46D gene (Sequence Analysis-All Coding Exons)	FAM46D	FAM46D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM46C geni (Tüm Gen Dizi Analizi)	FAM46C gene (Sequence Analysis-All Coding Exons)	FAM46C	FAM46C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM3D geni (Tüm Gen Dizi Analizi)	FAM3D gene (Sequence Analysis-All Coding Exons)	FAM3D	FAM3D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM3C geni (Tüm Gen Dizi Analizi)	FAM3C gene (Sequence Analysis-All Coding Exons)	FAM3C	FAM3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM3B geni (Tüm Gen Dizi Analizi)	FAM3B gene (Sequence Analysis-All Coding Exons)	FAM3B	FAM3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM3A geni (Tüm Gen Dizi Analizi)	FAM3A gene (Sequence Analysis-All Coding Exons)	FAM3A	FAM3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM32A geni (Tüm Gen Dizi Analizi)	FAM32A gene (Sequence Analysis-All Coding Exons)	FAM32A	FAM32A, OTAG12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM30A geni (Tüm Gen Dizi Analizi)	FAM30A gene (Sequence Analysis-All Coding Exons)	FAM30A	KIAA0125, FAM30A, C14orf110	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM26F geni (Tüm Gen Dizi Analizi)	FAM26F gene (Sequence Analysis-All Coding Exons)	FAM26F	FAM26F, INAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM220A geni (Tüm Gen Dizi Analizi)	FAM220A gene (Sequence Analysis-All Coding Exons)	FAM220A	FAM220A, SIPAR, ACPIN1, C7orf70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM213A geni (Tüm Gen Dizi Analizi)	FAM213A gene (Sequence Analysis-All Coding Exons)	FAM213A	FAM213A, PAMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM20B geni (Tüm Gen Dizi Analizi)	FAM20B gene (Sequence Analysis-All Coding Exons)	FAM20B	FAM20B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM208A geni (Tüm Gen Dizi Analizi)	FAM208A gene (Sequence Analysis-All Coding Exons)	FAM208A	FAM208A, RAP140, C3orf63, KIAA1105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM196A geni (Tüm Gen Dizi Analizi)	FAM196A gene (Sequence Analysis-All Coding Exons)	FAM196A	FAM196A, C10orf141, INSYN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM193B geni (Tüm Gen Dizi Analizi)	FAM193B gene (Sequence Analysis-All Coding Exons)	FAM193B	FAM193B, IRIZIO, KIAA1931	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM189A2 geni (Tüm Gen Dizi Analizi)	FAM189A2 gene (Sequence Analysis-All Coding Exons)	FAM189A2	X123	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM175B geni (Tüm Gen Dizi Analizi)	FAM175B gene (Sequence Analysis-All Coding Exons)	FAM175B	FAM175B, ABRO1, KIAA0157	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM175A geni (Tüm Gen Dizi Analizi)	FAM175A gene (Sequence Analysis-All Coding Exons)	FAM175A	FAM175A, CCDC98, ABRAXAS, ABRA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM169A geni (Tüm Gen Dizi Analizi)	FAM169A gene (Sequence Analysis-All Coding Exons)	FAM169A	FAM169A, SLAP75, KIAA0888	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM168A geni (Tüm Gen Dizi Analizi)	FAM168A gene (Sequence Analysis-All Coding Exons)	FAM168A	RAM168A, TCRP1, KIAA0280	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM167A geni (Tüm Gen Dizi Analizi)	FAM167A gene (Sequence Analysis-All Coding Exons)	FAM167A	FAM167A, C8orf13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM163A geni (Tüm Gen Dizi Analizi)	FAM163A gene (Sequence Analysis-All Coding Exons)	FAM163A	C1orf76, NDSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM162A geni (Tüm Gen Dizi Analizi)	FAM162A gene (Sequence Analysis-All Coding Exons)	FAM162A	C3orf28, EZIG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM160B1 geni (Tüm Gen Dizi Analizi)	FAM160B1 gene (Sequence Analysis-All Coding Exons)	FAM160B1	FAM160B1, KIAA1600	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM13B geni (Tüm Gen Dizi Analizi)	FAM13B gene (Sequence Analysis-All Coding Exons)	FAM13B	FAM13B, C5orf5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM13A-AS1 geni (Tüm Gen Dizi Analizi)	FAM13A-AS1 gene (Sequence Analysis-All Coding Exons)	FAM13A-AS1	FAM13AOS, FAM13A1OS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM13A geni (Tüm Gen Dizi Analizi)	FAM13A gene (Sequence Analysis-All Coding Exons)	FAM13A	FAM13A, FAM13A1, KIAA0914	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM136A geni (Tüm Gen Dizi Analizi)	FAM136A gene (Sequence Analysis-All Coding Exons)	FAM136A	FAM136A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM129C geni (Tüm Gen Dizi Analizi)	FAM129C gene (Sequence Analysis-All Coding Exons)	FAM129C	BCNP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM129B geni (Tüm Gen Dizi Analizi)	FAM129B gene (Sequence Analysis-All Coding Exons)	FAM129B	FAM129B, MINERVA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM127A geni (Tüm Gen Dizi Analizi)	FAM127A gene (Sequence Analysis-All Coding Exons)	FAM127A	CXX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM120C geni (Tüm Gen Dizi Analizi)	FAM120C gene (Sequence Analysis-All Coding Exons)	FAM120C	FAM120C, CXorf17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM120B geni (Tüm Gen Dizi Analizi)	FAM120B gene (Sequence Analysis-All Coding Exons)	FAM120B	FAM120B, PGCC1, CCPG, KIAA1838	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM120A geni (Tüm Gen Dizi Analizi)	FAM120A gene (Sequence Analysis-All Coding Exons)	FAM120A	FAM120A, C9orf10, KIAA0183	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM118B geni (Tüm Gen Dizi Analizi)	FAM118B gene (Sequence Analysis-All Coding Exons)	FAM118B	FAM118B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM110C geni (Tüm Gen Dizi Analizi)	FAM110C gene (Sequence Analysis-All Coding Exons)	FAM110C	FAM110C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM110B geni (Tüm Gen Dizi Analizi)	FAM110B gene (Sequence Analysis-All Coding Exons)	FAM110B	FAM110B, C8orf72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM110A geni (Tüm Gen Dizi Analizi)	FAM110A gene (Sequence Analysis-All Coding Exons)	FAM110A	FAM110A, C20orf55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM109B geni (Tüm Gen Dizi Analizi)	FAM109B gene (Sequence Analysis-All Coding Exons)	FAM109B	FAM109B, SES2, tPIP27B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM109A geni (Tüm Gen Dizi Analizi)	FAM109A gene (Sequence Analysis-All Coding Exons)	FAM109A	FAM109A, SES1, tPIP27A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM107A geni (Tüm Gen Dizi Analizi)	FAM107A gene (Sequence Analysis-All Coding Exons)	FAM107A	FAM107A, DRR1, TU3A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM103A1 geni (Tüm Gen Dizi Analizi)	FAM103A1 gene (Sequence Analysis-All Coding Exons)	FAM103A1	FAM103A1, RAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM102A geni (Tüm Gen Dizi Analizi)	FAM102A gene (Sequence Analysis-All Coding Exons)	FAM102A	FAM102A, EEIG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FALEC geni (Tüm Gen Dizi Analizi)	FALEC gene (Sequence Analysis-All Coding Exons)	FALEC	FALEC, FAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAIM2 geni (Tüm Gen Dizi Analizi)	FAIM2 gene (Sequence Analysis-All Coding Exons)	FAIM2	FAIM2, LFG, NMP35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAHD1 geni (Tüm Gen Dizi Analizi)	FAHD1 gene (Sequence Analysis-All Coding Exons)	FAHD1	FAHD1, FLJ36880	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAF2 geni (Tüm Gen Dizi Analizi)	FAF2 gene (Sequence Analysis-All Coding Exons)	FAF2	FAF2, ETEA, UBXD8, KIAA0867	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAF1 geni (Tüm Gen Dizi Analizi)	FAF1 gene (Sequence Analysis-All Coding Exons)	FAF1	FAF1, HFAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FADS3 geni (Tüm Gen Dizi Analizi)	FADS3 gene (Sequence Analysis-All Coding Exons)	FADS3	FADS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FADS2 geni (Tüm Gen Dizi Analizi)	FADS2 gene (Sequence Analysis-All Coding Exons)	FADS2	FADS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FADS1 geni (Tüm Gen Dizi Analizi)	FADS1 gene (Sequence Analysis-All Coding Exons)	FADS1	FADS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP7 geni (Tüm Gen Dizi Analizi)	FABP7 gene (Sequence Analysis-All Coding Exons)	FABP7	FABP7, FABP8, BLBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP6 geni (Tüm Gen Dizi Analizi)	FABP6 gene (Sequence Analysis-All Coding Exons)	FABP6	FABP6, ILLBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP5 geni (Tüm Gen Dizi Analizi)	FABP5 gene (Sequence Analysis-All Coding Exons)	FABP5	FABP5, PAFABP, EFABP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP4 geni (Tüm Gen Dizi Analizi)	FABP4 gene (Sequence Analysis-All Coding Exons)	FABP4	FABP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP3 geni (Tüm Gen Dizi Analizi)	FABP3 gene (Sequence Analysis-All Coding Exons)	FABP3	FABP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP2 geni (Tüm Gen Dizi Analizi)	FABP2 gene (Sequence Analysis-All Coding Exons)	FABP2	FABP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FABP1 geni (Tüm Gen Dizi Analizi)	FABP1 gene (Sequence Analysis-All Coding Exons)	FABP1	FABP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAAP24 geni (Tüm Gen Dizi Analizi)	FAAP24 gene (Sequence Analysis-All Coding Exons)	FAAP24	C19orf40, FAAP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAAP20 geni (Tüm Gen Dizi Analizi)	FAAP20 gene (Sequence Analysis-All Coding Exons)	FAAP20	C1orf86, FAAP20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAAP100 geni (Tüm Gen Dizi Analizi)	FAAP100 gene (Sequence Analysis-All Coding Exons)	FAAP100	C17orf70, FAAP100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAAH2 geni (Tüm Gen Dizi Analizi)	FAAH2 gene (Sequence Analysis-All Coding Exons)	FAAH2	FAAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F8A1 geni (Tüm Gen Dizi Analizi)	F8A1 gene (Sequence Analysis-All Coding Exons)	F8A1	F8A, DXS522E, HAP40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F3 geni (Tüm Gen Dizi Analizi)	F3 gene (Sequence Analysis-All Coding Exons)	F3	F3, TFA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F2RL3 geni (Tüm Gen Dizi Analizi)	F2RL3 gene (Sequence Analysis-All Coding Exons)	F2RL3	F2RL3, PAR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F2RL2 geni (Tüm Gen Dizi Analizi)	F2RL2 gene (Sequence Analysis-All Coding Exons)	F2RL2	F2RL2, PAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F2RL1 geni (Tüm Gen Dizi Analizi)	F2RL1 gene (Sequence Analysis-All Coding Exons)	F2RL1	F2RL1, GPR11, PAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F2R geni (Tüm Gen Dizi Analizi)	F2R gene (Sequence Analysis-All Coding Exons)	F2R	F2R, CF2R, PAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
F11R geni (Tüm Gen Dizi Analizi)	F11R gene (Sequence Analysis-All Coding Exons)	F11R	F11R, JAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EZR geni (Tüm Gen Dizi Analizi)	EZR gene (Sequence Analysis-All Coding Exons)	EZR	EZR, VIL2, CVL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EZH1 geni (Tüm Gen Dizi Analizi)	EZH1 gene (Sequence Analysis-All Coding Exons)	EZH1	EZH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EYA3 geni (Tüm Gen Dizi Analizi)	EYA3 gene (Sequence Analysis-All Coding Exons)	EYA3	EYA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EYA2 geni (Tüm Gen Dizi Analizi)	EYA2 gene (Sequence Analysis-All Coding Exons)	EYA2	EYA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXTL3 geni (Tüm Gen Dizi Analizi)	EXTL3 gene (Sequence Analysis-All Coding Exons)	EXTL3	EXTL3, EXTR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXTL2 geni (Tüm Gen Dizi Analizi)	EXTL2 gene (Sequence Analysis-All Coding Exons)	EXTL2	EXTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXTL1 geni (Tüm Gen Dizi Analizi)	EXTL1 gene (Sequence Analysis-All Coding Exons)	EXTL1	EXTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC9 geni (Tüm Gen Dizi Analizi)	EXOSC9 gene (Sequence Analysis-All Coding Exons)	EXOSC9	EXOSC9, PMSCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC7 geni (Tüm Gen Dizi Analizi)	EXOSC7 gene (Sequence Analysis-All Coding Exons)	EXOSC7	EXOSC7, RRP42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC6 geni (Tüm Gen Dizi Analizi)	EXOSC6 gene (Sequence Analysis-All Coding Exons)	EXOSC6	EXOSC6, MTR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC5 geni (Tüm Gen Dizi Analizi)	EXOSC5 gene (Sequence Analysis-All Coding Exons)	EXOSC5	EXOSC5, RRP46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC4 geni (Tüm Gen Dizi Analizi)	EXOSC4 gene (Sequence Analysis-All Coding Exons)	EXOSC4	EXOSC4, RRP41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC2 geni (Tüm Gen Dizi Analizi)	EXOSC2 gene (Sequence Analysis-All Coding Exons)	EXOSC2	EXOSC2, RRP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC10 geni (Tüm Gen Dizi Analizi)	EXOSC10 gene (Sequence Analysis-All Coding Exons)	EXOSC10	EXOSC10, PMSCL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOSC1 geni (Tüm Gen Dizi Analizi)	EXOSC1 gene (Sequence Analysis-All Coding Exons)	EXOSC1	EXOSC1, CSL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOG geni (Tüm Gen Dizi Analizi)	EXOG gene (Sequence Analysis-All Coding Exons)	EXOG	EXOG, ENDOGL1, ENGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC8 geni (Tüm Gen Dizi Analizi)	EXOC8 gene (Sequence Analysis-All Coding Exons)	EXOC8	EXOC8, EXO84, SEC84	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC7 geni (Tüm Gen Dizi Analizi)	EXOC7 gene (Sequence Analysis-All Coding Exons)	EXOC7	EXOC1, EX070, KIAA1067	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC6B geni (Tüm Gen Dizi Analizi)	EXOC6B gene (Sequence Analysis-All Coding Exons)	EXOC6B	EXOC6B, SEC15B, KIAA0919	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

EXOC6 geni (Tüm Gen Dizi Analizi)	EXOC6 gene (Sequence Analysis-All Coding Exons)	EXOC6	EXOC6, SEC15L1, SEC15L, SEC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC5 geni (Tüm Gen Dizi Analizi)	EXOC5 gene (Sequence Analysis-All Coding Exons)	EXOC5	EXOC5, SEC10L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC4 geni (Tüm Gen Dizi Analizi)	EXOC4 gene (Sequence Analysis-All Coding Exons)	EXOC4	EXOC4, SEC8, KIAA1699	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC3L1 geni (Tüm Gen Dizi Analizi)	EXOC3L1 gene (Sequence Analysis-All Coding Exons)	EXOC3L1	EXOC3L1, EXOC3L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC3 geni (Tüm Gen Dizi Analizi)	EXOC3 gene (Sequence Analysis-All Coding Exons)	EXOC3	EXOC3, SEC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC2 geni (Tüm Gen Dizi Analizi)	EXOC2 gene (Sequence Analysis-All Coding Exons)	EXOC2	EXOC2, SEC5L1, SEC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC1 geni (Tüm Gen Dizi Analizi)	EXOC1 gene (Sequence Analysis-All Coding Exons)	EXOC1	EXOC1, SEC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXO1 geni (Tüm Gen Dizi Analizi)	EXO1 gene (Sequence Analysis-All Coding Exons)	EXO1	EXO1, HEX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXD2 geni (Tüm Gen Dizi Analizi)	EXD2 gene (Sequence Analysis-All Coding Exons)	EXD2	EXD2, EXDL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EWSAT1 geni (Tüm Gen Dizi Analizi)	EWSAT1 gene (Sequence Analysis-All Coding Exons)	EWSAT1	EWSAT1, LINC00277	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVX2 geni (Tüm Gen Dizi Analizi)	EVX2 gene (Sequence Analysis-All Coding Exons)	EVX2	EVX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVX1 geni (Tüm Gen Dizi Analizi)	EVX1 gene (Sequence Analysis-All Coding Exons)	EVX1	EVX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVPL geni (Tüm Gen Dizi Analizi)	EVPL gene (Sequence Analysis-All Coding Exons)	EVPL	EVPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVL geni (Tüm Gen Dizi Analizi)	EVL gene (Sequence Analysis-All Coding Exons)	EVL	EVL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVI5 geni (Tüm Gen Dizi Analizi)	EVI5 gene (Sequence Analysis-All Coding Exons)	EVI5	EVI5, NB4S	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVI2B geni (Tüm Gen Dizi Analizi)	EVI2B gene (Sequence Analysis-All Coding Exons)	EVI2B	EVI2B, EVDB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EVI2A geni (Tüm Gen Dizi Analizi)	EVI2A gene (Sequence Analysis-All Coding Exons)	EVI2A	EVI2A, EVI2, EVDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV7 geni (Tüm Gen Dizi Analizi)	ETV7 gene (Sequence Analysis-All Coding Exons)	ETV7	ETV7, TEL2, TELB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV5 geni (Tüm Gen Dizi Analizi)	ETV5 gene (Sequence Analysis-All Coding Exons)	ETV5	ETV5, ERM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV4 geni (Tüm Gen Dizi Analizi)	ETV4 gene (Sequence Analysis-All Coding Exons)	ETV4	ETV4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV3 geni (Tüm Gen Dizi Analizi)	ETV3 gene (Sequence Analysis-All Coding Exons)	ETV3	ETV3, PE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV2 geni (Tüm Gen Dizi Analizi)	ETV2 gene (Sequence Analysis-All Coding Exons)	ETV2	ETV2, ETSRP71	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETV1 geni (Tüm Gen Dizi Analizi)	ETV1 gene (Sequence Analysis-All Coding Exons)	ETV1	ETV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETS2 geni (Tüm Gen Dizi Analizi)	ETS2 gene (Sequence Analysis-All Coding Exons)	ETS2	ETS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETS1 geni (Tüm Gen Dizi Analizi)	ETS1 gene (Sequence Analysis-All Coding Exons)	ETS1	ETS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETNPPL geni (Tüm Gen Dizi Analizi)	ETNPPL gene (Sequence Analysis-All Coding Exons)	ETNPPL	AGXT2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETNK2 geni (Tüm Gen Dizi Analizi)	ETNK2 gene (Sequence Analysis-All Coding Exons)	ETNK2	ETNK2, EKI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETNK1 geni (Tüm Gen Dizi Analizi)	ETNK1 gene (Sequence Analysis-All Coding Exons)	ETNK1	ETNK1, EKI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETFBKMT geni (Tüm Gen Dizi Analizi)	ETFBKMT gene (Sequence Analysis-All Coding Exons)	ETFBKMT	METTL20, C12orf72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETF1 geni (Tüm Gen Dizi Analizi)	ETF1 gene (Sequence Analysis-All Coding Exons)	ETF1	ETF1, RF1, ERF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ETAA1 geni (Tüm Gen Dizi Analizi)	ETAA1 gene (Sequence Analysis-All Coding Exons)	ETAA1	ETAA1, ETAA16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESYT3 geni (Tüm Gen Dizi Analizi)	ESYT3 gene (Sequence Analysis-All Coding Exons)	ESYT3	ESYT3, FAM62C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESYT2 geni (Tüm Gen Dizi Analizi)	ESYT2 gene (Sequence Analysis-All Coding Exons)	ESYT2	ESYT2, FAM62B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESYT1 geni (Tüm Gen Dizi Analizi)	ESYT1 gene (Sequence Analysis-All Coding Exons)	ESYT1	ESYT1, MBC2, FAM62A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESX1 geni (Tüm Gen Dizi Analizi)	ESX1 gene (Sequence Analysis-All Coding Exons)	ESX1	ESX1L, ESXR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESRRG geni (Tüm Gen Dizi Analizi)	ESRRG gene (Sequence Analysis-All Coding Exons)	ESRRG	ESRRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESRRA geni (Tüm Gen Dizi Analizi)	ESRRA gene (Sequence Analysis-All Coding Exons)	ESRRA	ESRRA, ESRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESRP2 geni (Tüm Gen Dizi Analizi)	ESRP2 gene (Sequence Analysis-All Coding Exons)	ESRP2	ESRP2, RBM35B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESRP1 geni (Tüm Gen Dizi Analizi)	ESRP1 gene (Sequence Analysis-All Coding Exons)	ESRP1	ESRP1, RMB35A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESRG geni (Tüm Gen Dizi Analizi)	ESRG gene (Sequence Analysis-All Coding Exons)	ESRG	HESRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESR2 geni (Tüm Gen Dizi Analizi)	ESR2 gene (Sequence Analysis-All Coding Exons)	ESR2	ESR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESPL1 geni (Tüm Gen Dizi Analizi)	ESPL1 gene (Sequence Analysis-All Coding Exons)	ESPL1	ESPL1, ESP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESM1 geni (Tüm Gen Dizi Analizi)	ESM1 gene (Sequence Analysis-All Coding Exons)	ESM1	ESM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESD geni (Tüm Gen Dizi Analizi)	ESD gene (Sequence Analysis-All Coding Exons)	ESD	ESD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESCO1 geni (Tüm Gen Dizi Analizi)	ESCO1 gene (Sequence Analysis-All Coding Exons)	ESCO1	ESCO1, ECO1, CTF, ESO1, EFO1, KIAA1911	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ESAM geni (Tüm Gen Dizi Analizi)	ESAM gene (Sequence Analysis-All Coding Exons)	ESAM	ESAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERVW-1 geni (Tüm Gen Dizi Analizi)	ERVW-1 gene (Sequence Analysis-All Coding Exons)	ERVW-1	ERVW1, ERVWE1, HERVW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERVFRD-1 geni (Tüm Gen Dizi Analizi)	ERVFRD-1 gene (Sequence Analysis-All Coding Exons)	ERVFRD-1	ERVFRD-1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERVS-1 geni (Tüm Gen Dizi Analizi)	ERVS-1 gene (Sequence Analysis-All Coding Exons)	ERVS-1	ERV3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERRFI1 geni (Tüm Gen Dizi Analizi)	ERRFI1 gene (Sequence Analysis-All Coding Exons)	ERRFI1	MIG6, RALT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERP44 geni (Tüm Gen Dizi Analizi)	ERP44 gene (Sequence Analysis-All Coding Exons)	ERP44	TXNDC4, KIAA0573	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERP29 geni (Tüm Gen Dizi Analizi)	ERP29 gene (Sequence Analysis-All Coding Exons)	ERP29	C12orf8, ERP28, ERP29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERO1B geni (Tüm Gen Dizi Analizi)	ERO1B gene (Sequence Analysis-All Coding Exons)	ERO1B	ERO1LB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERO1A geni (Tüm Gen Dizi Analizi)	ERO1A gene (Sequence Analysis-All Coding Exons)	ERO1A	ERO1L, ERO1LA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERN2 geni (Tüm Gen Dizi Analizi)	ERN2 gene (Sequence Analysis-All Coding Exons)	ERN2	ERN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERN1 geni (Tüm Gen Dizi Analizi)	ERN1 gene (Sequence Analysis-All Coding Exons)	ERN1	ERNL, IRE1, IRE1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERMP1 geni (Tüm Gen Dizi Analizi)	ERMP1 gene (Sequence Analysis-All Coding Exons)	ERMP1	ERMP1, KIAA1815, FXNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERMN geni (Tüm Gen Dizi Analizi)	ERMN gene (Sequence Analysis-All Coding Exons)	ERMN	ERMN, JN, KIAA1189	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERLEC1 geni (Tüm Gen Dizi Analizi)	ERLEC1 gene (Sequence Analysis-All Coding Exons)	ERLEC1	ERLEC1, C2orf30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERI3 geni (Tüm Gen Dizi Analizi)	ERI3 gene (Sequence Analysis-All Coding Exons)	ERI3	PRNPIP, PINT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERH geni (Tüm Gen Dizi Analizi)	ERH gene (Sequence Analysis-All Coding Exons)	ERH	ERH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERGIC3 geni (Tüm Gen Dizi Analizi)	ERGIC3 gene (Sequence Analysis-All Coding Exons)	ERGIC3	ERGIC3, C2orf47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERGIC2 geni (Tüm Gen Dizi Analizi)	ERGIC2 gene (Sequence Analysis-All Coding Exons)	ERGIC2	ERGIC2, CDA14, PTX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERG geni (Tüm Gen Dizi Analizi)	ERG gene (Sequence Analysis-All Coding Exons)	ERG	ERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERG geni (Tüm Gen Dizi Analizi)	ERG gene (Sequence Analysis-All Coding Exons)	ERG	ERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERG geni (Tüm Gen Dizi Analizi)	ERG gene (Sequence Analysis-All Coding Exons)	ERG	ERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERFE geni (Tüm Gen Dizi Analizi)	ERFE gene (Sequence Analysis-All Coding Exons)	ERFE	FAM132B, CTRP15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERCC6L geni (Tüm Gen Dizi Analizi)	ERCC6L gene (Sequence Analysis-All Coding Exons)	ERCC6L	ERCC6L, PICH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERC2 geni (Tüm Gen Dizi Analizi)	ERC2 gene (Sequence Analysis-All Coding Exons)	ERC2	ERC2, CAST, KIAA0378	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERBIN geni (Tüm Gen Dizi Analizi)	ERBIN gene (Sequence Analysis-All Coding Exons)	ERBIN	ERBB2IP, ERBIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERAS geni (Tüm Gen Dizi Analizi)	ERAS gene (Sequence Analysis-All Coding Exons)	ERAS	HRAS2, HRASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERAP2 geni (Tüm Gen Dizi Analizi)	ERAP2 gene (Sequence Analysis-All Coding Exons)	ERAP2	ERAP2, LRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERAP1 geni (Tüm Gen Dizi Analizi)	ERAP1 gene (Sequence Analysis-All Coding Exons)	ERAP1	ERAP1, ALAP, ARTS1, PILSAP, KIAA0525	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPYC geni (Tüm Gen Dizi Analizi)	EPYC gene (Sequence Analysis-All Coding Exons)	EPYC	EPYC, DSPG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPSTI1 geni (Tüm Gen Dizi Analizi)	EPSTI1 gene (Sequence Analysis-All Coding Exons)	EPSTI1	EPSTI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPS8L3 geni (Tüm Gen Dizi Analizi)	EPS8L3 gene (Sequence Analysis-All Coding Exons)	EPS8L3	EPS8L3, EPS8R3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPS8L2 geni (Tüm Gen Dizi Analizi)	EPS8L2 gene (Sequence Analysis-All Coding Exons)	EPS8L2	EPS8L2, EPS8R2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPS8L1 geni (Tüm Gen Dizi Analizi)	EPS8L1 gene (Sequence Analysis-All Coding Exons)	EPS8L1	EPS8L1, EPS8R1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPS15L1 geni (Tüm Gen Dizi Analizi)	EPS15L1 gene (Sequence Analysis-All Coding Exons)	EPS15L1	EPS15L1, EPS15R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPS15 geni (Tüm Gen Dizi Analizi)	EPS15 gene (Sequence Analysis-All Coding Exons)	EPS15	EPS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPRS geni (Tüm Gen Dizi Analizi)	EPRS gene (Sequence Analysis-All Coding Exons)	EPRS	EPRS, PARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPPK1 geni (Tüm Gen Dizi Analizi)	EPPK1 gene (Sequence Analysis-All Coding Exons)	EPPK1	EPPK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPPIN geni (Tüm Gen Dizi Analizi)	EPPIN gene (Sequence Analysis-All Coding Exons)	EPPIN	SPINLW1, EPPIN, WAP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPN3 geni (Tüm Gen Dizi Analizi)	EPN3 gene (Sequence Analysis-All Coding Exons)	EPN3	EPN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPN2 geni (Tüm Gen Dizi Analizi)	EPN2 gene (Sequence Analysis-All Coding Exons)	EPN2	EPN2, KIAA1065	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPN1 geni (Tüm Gen Dizi Analizi)	EPN1 gene (Sequence Analysis-All Coding Exons)	EPN1	EPN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPM2AIP1 geni (Tüm Gen Dizi Analizi)	EPM2AIP1 gene (Sequence Analysis-All Coding Exons)	EPM2AIP1	EPM2AIP1, KIAA0766	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHB6 geni (Tüm Gen Dizi Analizi)	EPHB6 gene (Sequence Analysis-All Coding Exons)	EPHB6	EPHB6, HEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHB3 geni (Tüm Gen Dizi Analizi)	EPHB3 gene (Sequence Analysis-All Coding Exons)	EPHB3	EPHB3, ETK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHB1 geni (Tüm Gen Dizi Analizi)	EPHB1 gene (Sequence Analysis-All Coding Exons)	EPHB1	EPHB1, EPHT2, NET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA8 geni (Tüm Gen Dizi Analizi)	EPHA8 gene (Sequence Analysis-All Coding Exons)	EPHA8	EPHA8, EEK, HEK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA7 geni (Tüm Gen Dizi Analizi)	EPHA7 gene (Sequence Analysis-All Coding Exons)	EPHA7	EPHA7, HEK11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA6 geni (Tüm Gen Dizi Analizi)	EPHA6 gene (Sequence Analysis-All Coding Exons)	EPHA6	EPHA6, HEK12, EHK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

EPHA5 geni (Tüm Gen Dizi Analizi)	EPHA5 gene (Sequence Analysis-All Coding Exons)	EPHA5	EPHA5, TYRO4, HEK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA3 geni (Tüm Gen Dizi Analizi)	EPHA3 gene (Sequence Analysis-All Coding Exons)	EPHA3	EPHA3, ETK1, HEK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA10 geni (Tüm Gen Dizi Analizi)	EPHA10 gene (Sequence Analysis-All Coding Exons)	EPHA10	EPHA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA1 geni (Tüm Gen Dizi Analizi)	EPHA1 gene (Sequence Analysis-All Coding Exons)	EPHA1	EPHA1, EPHT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPC2 geni (Tüm Gen Dizi Analizi)	EPC2 gene (Sequence Analysis-All Coding Exons)	EPC2	EPC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPC1 geni (Tüm Gen Dizi Analizi)	EPC1 gene (Sequence Analysis-All Coding Exons)	EPC1	EPC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPB41L5 geni (Tüm Gen Dizi Analizi)	EPB41L5 gene (Sequence Analysis-All Coding Exons)	EPB41L5	EPB41L5, KIAA1548	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPB41L4B geni (Tüm Gen Dizi Analizi)	EPB41L4B gene (Sequence Analysis-All Coding Exons)	EPB41L4B	EPB41L4B, EHM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIOX geni (Tüm Gen Dizi Analizi)	MIOX gene (Sequence Analysis-All Coding Exons)	MIOX		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPB41L4A geni (Tüm Gen Dizi Analizi)	EPB41L4A gene (Sequence Analysis-All Coding Exons)	EPB41L4A	EPB41L4A, EPB41L4, NBL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPB41L3 geni (Tüm Gen Dizi Analizi)	EPB41L3 gene (Sequence Analysis-All Coding Exons)	EPB41L3	EPB41L3, DAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPB41L2 geni (Tüm Gen Dizi Analizi)	EPB41L2 gene (Sequence Analysis-All Coding Exons)	EPB41L2	EPB41L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EP400 geni (Tüm Gen Dizi Analizi)	EP400 gene (Sequence Analysis-All Coding Exons)	EP400	EP400, TNRC12, KIAA1498,	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EOMES geni (Tüm Gen Dizi Analizi)	EOMES gene (Sequence Analysis-All Coding Exons)	EOMES	EOMES, TBR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIIP geni (Tüm Gen Dizi Analizi)	MIIP gene (Sequence Analysis-All Coding Exons)	MIIP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MIGA2 geni (Tüm Gen Dizi Analizi)	MIGA2 gene (Sequence Analysis-All Coding Exons)	MIGA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD8 geni (Tüm Gen Dizi Analizi)	ENTPD8 gene (Sequence Analysis-All Coding Exons)	ENTPD8	ENTPD8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD7 geni (Tüm Gen Dizi Analizi)	ENTPD7 gene (Sequence Analysis-All Coding Exons)	ENTPD7	ENTPD7, LALP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD6 geni (Tüm Gen Dizi Analizi)	ENTPD6 gene (Sequence Analysis-All Coding Exons)	ENTPD6	ENTPD6, CD39L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD5 geni (Tüm Gen Dizi Analizi)	ENTPD5 gene (Sequence Analysis-All Coding Exons)	ENTPD5	ENTPD5, CD39L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD4 geni (Tüm Gen Dizi Analizi)	ENTPD4 gene (Sequence Analysis-All Coding Exons)	ENTPD4	ENTPD4, LYSAL1, LAP70, KIAA0392	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD3 geni (Tüm Gen Dizi Analizi)	ENTPD3 gene (Sequence Analysis-All Coding Exons)	ENTPD3	ENTPD3, CD39L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENTPD2 geni (Tüm Gen Dizi Analizi)	ENTPD2 gene (Sequence Analysis-All Coding Exons)	ENTPD2	ENTPD2, CD39L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENSA geni (Tüm Gen Dizi Analizi)	ENSA gene (Sequence Analysis-All Coding Exons)	ENSA	ENSA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP7 geni (Tüm Gen Dizi Analizi)	ENPP7 gene (Sequence Analysis-All Coding Exons)	ENPP7	ENPP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP6 geni (Tüm Gen Dizi Analizi)	ENPP6 gene (Sequence Analysis-All Coding Exons)	ENPP6	ENPP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP5 geni (Tüm Gen Dizi Analizi)	ENPP5 gene (Sequence Analysis-All Coding Exons)	ENPP5	ENPP5, NPP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP4 geni (Tüm Gen Dizi Analizi)	ENPP4 gene (Sequence Analysis-All Coding Exons)	ENPP4	ENPP4, NPP4, KIAA0879	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP3 geni (Tüm Gen Dizi Analizi)	ENPP3 gene (Sequence Analysis-All Coding Exons)	ENPP3	ENPP3, PDNP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPP2 geni (Tüm Gen Dizi Analizi)	ENPP2 gene (Sequence Analysis-All Coding Exons)	ENPP2	ENPP2, PDNP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENPEP geni (Tüm Gen Dizi Analizi)	ENPEP gene (Sequence Analysis-All Coding Exons)	ENPEP	ENPEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENOX2 geni (Tüm Gen Dizi Analizi)	ENOX2 gene (Sequence Analysis-All Coding Exons)	ENOX2	ENOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENOX1 geni (Tüm Gen Dizi Analizi)	ENOX1 gene (Sequence Analysis-All Coding Exons)	ENOX1	ENOX1, CNOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENOSF1 geni (Tüm Gen Dizi Analizi)	ENOSF1 gene (Sequence Analysis-All Coding Exons)	ENOSF1	ENOSF1, RTS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENO2 geni (Tüm Gen Dizi Analizi)	ENO2 gene (Sequence Analysis-All Coding Exons)	ENO2	ENO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENKUR geni (Tüm Gen Dizi Analizi)	ENKUR gene (Sequence Analysis-All Coding Exons)	ENKUR	ENKUR, C10orf63	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENGASE geni (Tüm Gen Dizi Analizi)	ENGASE gene (Sequence Analysis-All Coding Exons)	ENGASE	NAGLUEB, FLJ21865	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENDOU geni (Tüm Gen Dizi Analizi)	ENDOU gene (Sequence Analysis-All Coding Exons)	ENDOU	ENDOU, PP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENDOG geni (Tüm Gen Dizi Analizi)	ENDOG gene (Sequence Analysis-All Coding Exons)	ENDOG	ENDOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENC1 geni (Tüm Gen Dizi Analizi)	ENC1 gene (Sequence Analysis-All Coding Exons)	ENC1	ENC1, PIG10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ENAH geni (Tüm Gen Dizi Analizi)	ENAH gene (Sequence Analysis-All Coding Exons)	ENAH	ENAH, ENA, MENA, NDPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EN2 geni (Tüm Gen Dizi Analizi)	EN2 gene (Sequence Analysis-All Coding Exons)	EN2	EN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EN1 geni (Tüm Gen Dizi Analizi)	EN1 gene (Sequence Analysis-All Coding Exons)	EN1	EN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMX2OS geni (Tüm Gen Dizi Analizi)	EMX2OS gene (Sequence Analysis-All Coding Exons)	EMX2OS	EMX2OS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMX1 geni (Tüm Gen Dizi Analizi)	EMX1 gene (Sequence Analysis-All Coding Exons)	EMX1	EMX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMSY geni (Tüm Gen Dizi Analizi)	EMSY gene (Sequence Analysis-All Coding Exons)	EMSY	EMSY, C11orf30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMP3 geni (Tüm Gen Dizi Analizi)	EMP3 gene (Sequence Analysis-All Coding Exons)	EMP3	EMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMP1 geni (Tüm Gen Dizi Analizi)	EMP1 gene (Sequence Analysis-All Coding Exons)	EMP1	EMP1, TMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EML4 geni (Tüm Gen Dizi Analizi)	EML4 gene (Sequence Analysis-All Coding Exons)	EML4	EML4, ROPP120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMILIN1 geni (Tüm Gen Dizi Analizi)	EMILIN1 gene (Sequence Analysis-All Coding Exons)	EMILIN1	EMILIN1, EMILIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EME2 geni (Tüm Gen Dizi Analizi)	EME2 gene (Sequence Analysis-All Coding Exons)	EME2	EME2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EME1 geni (Tüm Gen Dizi Analizi)	EME1 gene (Sequence Analysis-All Coding Exons)	EME1	EME1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMC8 geni (Tüm Gen Dizi Analizi)	EMC8 gene (Sequence Analysis-All Coding Exons)	EMC8	EMC8, NOC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMC4 geni (Tüm Gen Dizi Analizi)	EMC4 gene (Sequence Analysis-All Coding Exons)	EMC4	EMC4, TMEM85, PIG17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMC2 geni (Tüm Gen Dizi Analizi)	EMC2 gene (Sequence Analysis-All Coding Exons)	EMC2	EMC2, KIAA0103	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMC10 geni (Tüm Gen Dizi Analizi)	EMC10 gene (Sequence Analysis-All Coding Exons)	EMC10	EMC10, C15orf63, HSS1, HSM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMB geni (Tüm Gen Dizi Analizi)	EMB gene (Sequence Analysis-All Coding Exons)	EMB	EMB, GP70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELSPBP1 geni (Tüm Gen Dizi Analizi)	ELSPBP1 gene (Sequence Analysis-All Coding Exons)	ELSPBP1	ELSPBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELP6 geni (Tüm Gen Dizi Analizi)	ELP6 gene (Sequence Analysis-All Coding Exons)	ELP6	ELP6, TMEM103, C3orf75	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELP5 geni (Tüm Gen Dizi Analizi)	ELP5 gene (Sequence Analysis-All Coding Exons)	ELP5	ELP5, DERP6, C17orf81	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELP3 geni (Tüm Gen Dizi Analizi)	ELP3 gene (Sequence Analysis-All Coding Exons)	ELP3	ELP3, KAT9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOVL7 geni (Tüm Gen Dizi Analizi)	ELOVL7 gene (Sequence Analysis-All Coding Exons)	ELOVL7	ELOVL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOVL6 geni (Tüm Gen Dizi Analizi)	ELOVL6 gene (Sequence Analysis-All Coding Exons)	ELOVL6	ELOVL6, LCE, FACE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOVL3 geni (Tüm Gen Dizi Analizi)	ELOVL3 gene (Sequence Analysis-All Coding Exons)	ELOVL3	ELOVL3, CIG30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOVL2 geni (Tüm Gen Dizi Analizi)	ELOVL2 gene (Sequence Analysis-All Coding Exons)	ELOVL2	ELOVL2, SSC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOVL1 geni (Tüm Gen Dizi Analizi)	ELOVL1 gene (Sequence Analysis-All Coding Exons)	ELOVL1	ELOVL1, SSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOC geni (Tüm Gen Dizi Analizi)	ELOC gene (Sequence Analysis-All Coding Exons)	ELOC	TCEB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOA geni (Tüm Gen Dizi Analizi)	ELOA gene (Sequence Analysis-All Coding Exons)	ELOA	TCEB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELMOD2 geni (Tüm Gen Dizi Analizi)	ELMOD2 gene (Sequence Analysis-All Coding Exons)	ELMOD2	ELMOD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELMOD1 geni (Tüm Gen Dizi Analizi)	ELMOD1 gene (Sequence Analysis-All Coding Exons)	ELMOD1	ELMOD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELMO3 geni (Tüm Gen Dizi Analizi)	ELMO3 gene (Sequence Analysis-All Coding Exons)	ELMO3	ELMO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELMO1 geni (Tüm Gen Dizi Analizi)	ELMO1 gene (Sequence Analysis-All Coding Exons)	ELMO1	ELMO1, CED12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELL3 geni (Tüm Gen Dizi Analizi)	ELL3 gene (Sequence Analysis-All Coding Exons)	ELL3	ELL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MFN1 geni (Tüm Gen Dizi Analizi)	MFN1 gene (Sequence Analysis-All Coding Exons)	MFN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELL2 geni (Tüm Gen Dizi Analizi)	ELL2 gene (Sequence Analysis-All Coding Exons)	ELL2	ELL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELL geni (Tüm Gen Dizi Analizi)	ELL gene (Sequence Analysis-All Coding Exons)	ELL	ELL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELK4 geni (Tüm Gen Dizi Analizi)	ELK4 gene (Sequence Analysis-All Coding Exons)	ELK4	ELK4, SAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELK3 geni (Tüm Gen Dizi Analizi)	ELK3 gene (Sequence Analysis-All Coding Exons)	ELK3	ELK3, SAP2, ERP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELK1 geni (Tüm Gen Dizi Analizi)	ELK1 gene (Sequence Analysis-All Coding Exons)	ELK1	ELK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELFN1 geni (Tüm Gen Dizi Analizi)	ELFN1 gene (Sequence Analysis-All Coding Exons)	ELFN1	ELFN1, PPP1R28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELF5 geni (Tüm Gen Dizi Analizi)	ELF5 gene (Sequence Analysis-All Coding Exons)	ELF5	ELF5, ESE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELF4 geni (Tüm Gen Dizi Analizi)	ELF4 gene (Sequence Analysis-All Coding Exons)	ELF4	ELF4, MEF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELF3 geni (Tüm Gen Dizi Analizi)	ELF3 gene (Sequence Analysis-All Coding Exons)	ELF3	ELF3, ESX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELF1 geni (Tüm Gen Dizi Analizi)	ELF1 gene (Sequence Analysis-All Coding Exons)	ELF1	ELF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELAVL3 geni (Tüm Gen Dizi Analizi)	ELAVL3 gene (Sequence Analysis-All Coding Exons)	ELAVL3	ELAVL3, HUC, PLE21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELAVL2 geni (Tüm Gen Dizi Analizi)	ELAVL2 gene (Sequence Analysis-All Coding Exons)	ELAVL2	ELAVL2, HELN1, HUB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELAVL1 geni (Tüm Gen Dizi Analizi)	ELAVL1 gene (Sequence Analysis-All Coding Exons)	ELAVL1	ELAVL1, HUR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELAC1 geni (Tüm Gen Dizi Analizi)	ELAC1 gene (Sequence Analysis-All Coding Exons)	ELAC1	ELAC1, D29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL8 geni (Tüm Gen Dizi Analizi)	METTL8 gene (Sequence Analysis-All Coding Exons)	METTL8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF6 geni (Tüm Gen Dizi Analizi)	EIF6 gene (Sequence Analysis-All Coding Exons)	EIF6	EIF6, EIF3A, ITGB4BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
METTL2B geni (Tüm Gen Dizi Analizi)	METTL2B gene (Sequence Analysis-All Coding Exons)	METTL2B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF5A2 geni (Tüm Gen Dizi Analizi)	EIF5A2 gene (Sequence Analysis-All Coding Exons)	EIF5A2	EIF5A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF5A geni (Tüm Gen Dizi Analizi)	EIF5A gene (Sequence Analysis-All Coding Exons)	EIF5A	EIF5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

EIF5 geni (Tüm Gen Dizi Analizi)	EIF5 gene (Sequence Analysis-All Coding Exons)	EIF5	EIF5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4H geni (Tüm Gen Dizi Analizi)	EIF4H gene (Sequence Analysis-All Coding Exons)	EIF4H	EIF4H, WBSCR1, WSCR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4G3 geni (Tüm Gen Dizi Analizi)	EIF4G3 gene (Sequence Analysis-All Coding Exons)	EIF4G3	EIF4G3, EIF4GII	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4G2 geni (Tüm Gen Dizi Analizi)	EIF4G2 gene (Sequence Analysis-All Coding Exons)	EIF4G2	EIF4G2, DAP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4ENIF1 geni (Tüm Gen Dizi Analizi)	EIF4ENIF1 gene (Sequence Analysis-All Coding Exons)	EIF4ENIF1	EIF4ENIF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4EBP2 geni (Tüm Gen Dizi Analizi)	EIF4EBP2 gene (Sequence Analysis-All Coding Exons)	EIF4EBP2	EIF4EBP2, 4EBP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4EBP1 geni (Tüm Gen Dizi Analizi)	EIF4EBP1 gene (Sequence Analysis-All Coding Exons)	EIF4EBP1	EIF4EBP1, 4EBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4E3 geni (Tüm Gen Dizi Analizi)	EIF4E3 gene (Sequence Analysis-All Coding Exons)	EIF4E3	EIF4E3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4E2 geni (Tüm Gen Dizi Analizi)	EIF4E2 gene (Sequence Analysis-All Coding Exons)	EIF4E2	EIF4E2, EIF4EL3, 4EHP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4B geni (Tüm Gen Dizi Analizi)	EIF4B gene (Sequence Analysis-All Coding Exons)	EIF4B	EIF4B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4A2 geni (Tüm Gen Dizi Analizi)	EIF4A2 gene (Sequence Analysis-All Coding Exons)	EIF4A2	EIF4A2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4A1 geni (Tüm Gen Dizi Analizi)	EIF4A1 gene (Sequence Analysis-All Coding Exons)	EIF4A1	EIF4A1, DDX2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3M geni (Tüm Gen Dizi Analizi)	EIF3M gene (Sequence Analysis-All Coding Exons)	EIF3M	EIF3M, PCID1, B5, GA17	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3K geni (Tüm Gen Dizi Analizi)	EIF3K gene (Sequence Analysis-All Coding Exons)	EIF3K	EIF3K, PLAC24, EIF3S12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3J geni (Tüm Gen Dizi Analizi)	EIF3J gene (Sequence Analysis-All Coding Exons)	EIF3J	EIF3J, EIF3S1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3I geni (Tüm Gen Dizi Analizi)	EIF3I gene (Sequence Analysis-All Coding Exons)	EIF3I	EIF3I, EIF3S2, TRIP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3H geni (Tüm Gen Dizi Analizi)	EIF3H gene (Sequence Analysis-All Coding Exons)	EIF3H	EIF3H, EIF3S3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3G geni (Tüm Gen Dizi Analizi)	EIF3G gene (Sequence Analysis-All Coding Exons)	EIF3G	EIF3G, EIF3S4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3F geni (Tüm Gen Dizi Analizi)	EIF3F gene (Sequence Analysis-All Coding Exons)	EIF3F	EIF3F, EIF3S5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3E geni (Tüm Gen Dizi Analizi)	EIF3E gene (Sequence Analysis-All Coding Exons)	EIF3E	EIF3E, EIF3S6, INT6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3D geni (Tüm Gen Dizi Analizi)	EIF3D gene (Sequence Analysis-All Coding Exons)	EIF3D	EIF3D, EIF3S7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3C geni (Tüm Gen Dizi Analizi)	EIF3C gene (Sequence Analysis-All Coding Exons)	EIF3C	EIF3C, EIF3S8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3B geni (Tüm Gen Dizi Analizi)	EIF3B gene (Sequence Analysis-All Coding Exons)	EIF3B	EIF3B, EIF3S9, PRT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF3A geni (Tüm Gen Dizi Analizi)	EIF3A gene (Sequence Analysis-All Coding Exons)	EIF3A	EIF3A, EIF3S10, P167	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2D geni (Tüm Gen Dizi Analizi)	EIF2D gene (Sequence Analysis-All Coding Exons)	EIF2D	EIF2D, HCA56	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2AK2 geni (Tüm Gen Dizi Analizi)	EIF2AK2 gene (Sequence Analysis-All Coding Exons)	EIF2AK2	PRKR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2AK1 geni (Tüm Gen Dizi Analizi)	EIF2AK1 gene (Sequence Analysis-All Coding Exons)	EIF2AK1	EIF2AK1, HRI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2A geni (Tüm Gen Dizi Analizi)	EIF2A gene (Sequence Analysis-All Coding Exons)	EIF2A	EIF2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF1AY geni (Tüm Gen Dizi Analizi)	EIF1AY gene (Sequence Analysis-All Coding Exons)	EIF1AY	EIF1AY	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF1AX geni (Tüm Gen Dizi Analizi)	EIF1AX gene (Sequence Analysis-All Coding Exons)	EIF1AX	EIF1A, EIF4C, EIF1AX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EID3 geni (Tüm Gen Dizi Analizi)	EID3 gene (Sequence Analysis-All Coding Exons)	EID3	EID3, NSMCE4B, NSE4B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EID2 geni (Tüm Gen Dizi Analizi)	EID2 gene (Sequence Analysis-All Coding Exons)	EID2	EID2, CRI2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EID1 geni (Tüm Gen Dizi Analizi)	EID1 gene (Sequence Analysis-All Coding Exons)	EID1	EID1, CRI1, C15orf3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EI24 geni (Tüm Gen Dizi Analizi)	EI24 gene (Sequence Analysis-All Coding Exons)	EI24	EI24, PIG8, EPG4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHMT2 geni (Tüm Gen Dizi Analizi)	EHMT2 gene (Sequence Analysis-All Coding Exons)	EHMT2	EHMT2, GAT8, G9A, NG36	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHF geni (Tüm Gen Dizi Analizi)	EHF gene (Sequence Analysis-All Coding Exons)	EHF	EHF, ESE3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHD4 geni (Tüm Gen Dizi Analizi)	EHD4 gene (Sequence Analysis-All Coding Exons)	EHD4	EHD4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHD3 geni (Tüm Gen Dizi Analizi)	EHD3 gene (Sequence Analysis-All Coding Exons)	EHD3	EHD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHD2 geni (Tüm Gen Dizi Analizi)	EHD2 gene (Sequence Analysis-All Coding Exons)	EHD2	EHD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EHD1 geni (Tüm Gen Dizi Analizi)	EHD1 gene (Sequence Analysis-All Coding Exons)	EHD1	EHD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGR4 geni (Tüm Gen Dizi Analizi)	EGR4 gene (Sequence Analysis-All Coding Exons)	EGR4	EGR4, NGFIC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ME1 geni (Tüm Gen Dizi Analizi)	ME1 gene (Sequence Analysis-All Coding Exons)	ME1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGR3 geni (Tüm Gen Dizi Analizi)	EGR3 gene (Sequence Analysis-All Coding Exons)	EGR3	EGR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGR1 geni (Tüm Gen Dizi Analizi)	EGR1 gene (Sequence Analysis-All Coding Exons)	EGR1	EGR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGOT geni (Tüm Gen Dizi Analizi)	EGOT gene (Sequence Analysis-All Coding Exons)	EGOT	EGO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGLN3 geni (Tüm Gen Dizi Analizi)	EGLN3 gene (Sequence Analysis-All Coding Exons)	EGLN3	EGLN3, PHD3, HIFP4H3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGLN2 geni (Tüm Gen Dizi Analizi)	EGLN2 gene (Sequence Analysis-All Coding Exons)	EGLN2	EGLN2, PHD1, HFPH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGFL8 geni (Tüm Gen Dizi Analizi)	EGFL8 gene (Sequence Analysis-All Coding Exons)	EGFL8	EGFL8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGFL7 geni (Tüm Gen Dizi Analizi)	EGFL7 gene (Sequence Analysis-All Coding Exons)	EGFL7	EGFL7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EGFL6 geni (Tüm Gen Dizi Analizi)	EGFL6 gene (Sequence Analysis-All Coding Exons)	EGFL6	EGFL6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFS geni (Tüm Gen Dizi Analizi)	EFS gene (Sequence Analysis-All Coding Exons)	EFS	EFS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFR3B geni (Tüm Gen Dizi Analizi)	EFR3B gene (Sequence Analysis-All Coding Exons)	EFR3B	EFR3B, KIAA0953	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFR3A geni (Tüm Gen Dizi Analizi)	EFR3A gene (Sequence Analysis-All Coding Exons)	EFR3A	EFR3A, KIAA0143	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNB3 geni (Tüm Gen Dizi Analizi)	EFNB3 gene (Sequence Analysis-All Coding Exons)	EFNB3	EFNB3, EPLG8, LERK8, EFL6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNB2 geni (Tüm Gen Dizi Analizi)	EFNB2 gene (Sequence Analysis-All Coding Exons)	EFNB2	EFNB2, EPLG5, LERK5, HTK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNA4 geni (Tüm Gen Dizi Analizi)	EFNA4 gene (Sequence Analysis-All Coding Exons)	EFNA4	EFNA4, EPLG4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNA3 geni (Tüm Gen Dizi Analizi)	EFNA3 gene (Sequence Analysis-All Coding Exons)	EFNA3	EFNA3, EPLG3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNA2 geni (Tüm Gen Dizi Analizi)	EFNA2 gene (Sequence Analysis-All Coding Exons)	EFNA2	EFNA2, EPLG6, LERK6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNA1 geni (Tüm Gen Dizi Analizi)	EFNA1 gene (Sequence Analysis-All Coding Exons)	EFNA1	EFNA1, EPLG1, TNFAIP4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFHD2 geni (Tüm Gen Dizi Analizi)	EFHD2 gene (Sequence Analysis-All Coding Exons)	EFHD2	EFHD2, SWS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFHD1 geni (Tüm Gen Dizi Analizi)	EFHD1 gene (Sequence Analysis-All Coding Exons)	EFHD1	EFHD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFHC2 geni (Tüm Gen Dizi Analizi)	EFHC2 gene (Sequence Analysis-All Coding Exons)	EFHC2	EFHC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED6 geni (Tüm Gen Dizi Analizi)	MED6 gene (Sequence Analysis-All Coding Exons)	MED6		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEPD1 geni (Tüm Gen Dizi Analizi)	EEPD1 gene (Sequence Analysis-All Coding Exons)	EEPD1	EEPD1, KIAA1706	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEFSEC geni (Tüm Gen Dizi Analizi)	EEFSEC gene (Sequence Analysis-All Coding Exons)	EEFSEC	EEFSEC, SELB, EFSEC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF2KMT geni (Tüm Gen Dizi Analizi)	EEF2KMT gene (Sequence Analysis-All Coding Exons)	EEF2KMT	EEF2KMT, FAM86A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF2K geni (Tüm Gen Dizi Analizi)	EEF2K gene (Sequence Analysis-All Coding Exons)	EEF2K	EEF2K	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1G geni (Tüm Gen Dizi Analizi)	EEF1G gene (Sequence Analysis-All Coding Exons)	EEF1G	EEF1G, EF1G	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1E1 geni (Tüm Gen Dizi Analizi)	EEF1E1 gene (Sequence Analysis-All Coding Exons)	EEF1E1	EEF1E1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1D geni (Tüm Gen Dizi Analizi)	EEF1D gene (Sequence Analysis-All Coding Exons)	EEF1D	EEF1D, EF1D	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1B2 geni (Tüm Gen Dizi Analizi)	EEF1B2 gene (Sequence Analysis-All Coding Exons)	EEF1B2	EEF1B2, EEF1B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1AKMT3 geni (Tüm Gen Dizi Analizi)	EEF1AKMT3 gene (Sequence Analysis-All Coding Exons)	EEF1AKMT3	METTL21B, FAM119B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEF1A1 geni (Tüm Gen Dizi Analizi)	EEF1A1 gene (Sequence Analysis-All Coding Exons)	EEF1A1	EEF1A1, EF1A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EED geni (Tüm Gen Dizi Analizi)	EED gene (Sequence Analysis-All Coding Exons)	EED	EED, WAIT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EEA1 geni (Tüm Gen Dizi Analizi)	EEA1 gene (Sequence Analysis-All Coding Exons)	EEA1	EEA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDN2 geni (Tüm Gen Dizi Analizi)	EDN2 gene (Sequence Analysis-All Coding Exons)	EDN2	EDN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDEM2 geni (Tüm Gen Dizi Analizi)	EDEM2 gene (Sequence Analysis-All Coding Exons)	EDEM2	EDEM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDEM1 geni (Tüm Gen Dizi Analizi)	EDEM1 gene (Sequence Analysis-All Coding Exons)	EDEM1	EDEM, KIAA0212	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDDM3B geni (Tüm Gen Dizi Analizi)	EDDM3B gene (Sequence Analysis-All Coding Exons)	EDDM3B	FAM12B, HE3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MED16 geni (Tüm Gen Dizi Analizi)	MED16 gene (Sequence Analysis-All Coding Exons)	MED16		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDDM3A geni (Tüm Gen Dizi Analizi)	EDDM3A gene (Sequence Analysis-All Coding Exons)	EDDM3A	FAM12A, HE3A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDC4 geni (Tüm Gen Dizi Analizi)	EDC4 gene (Sequence Analysis-All Coding Exons)	EDC4	EDC4, RCD8, GE1, HEDLS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDA2R geni (Tüm Gen Dizi Analizi)	EDA2R gene (Sequence Analysis-All Coding Exons)	EDA2R	EDAR2, XEDAR, EDA4R	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECT2 geni (Tüm Gen Dizi Analizi)	ECT2 gene (Sequence Analysis-All Coding Exons)	ECT2	ECT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECSIT geni (Tüm Gen Dizi Analizi)	ECSIT gene (Sequence Analysis-All Coding Exons)	ECSIT	SITPEC, ECSIT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECSCR geni (Tüm Gen Dizi Analizi)	ECSCR gene (Sequence Analysis-All Coding Exons)	ECSCR	ECSCR, ECSM2, ARIA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECM2 geni (Tüm Gen Dizi Analizi)	ECM2 gene (Sequence Analysis-All Coding Exons)	ECM2	ECM2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECI2 geni (Tüm Gen Dizi Analizi)	ECI2 gene (Sequence Analysis-All Coding Exons)	ECI2	ECI2, PECI, DRS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECI1 geni (Tüm Gen Dizi Analizi)	ECI1 gene (Sequence Analysis-All Coding Exons)	ECI1	ECI1, DCI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECHDC1 geni (Tüm Gen Dizi Analizi)	ECHDC1 gene (Sequence Analysis-All Coding Exons)	ECHDC1	ECHDC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECH1 geni (Tüm Gen Dizi Analizi)	ECH1 gene (Sequence Analysis-All Coding Exons)	ECH1	ECH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECE2 geni (Tüm Gen Dizi Analizi)	ECE2 gene (Sequence Analysis-All Coding Exons)	ECE2	ECE2, KIAA0604	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ECD geni (Tüm Gen Dizi Analizi)	ECD gene (Sequence Analysis-All Coding Exons)	ECD	ECD, SGT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

EBPL geni (Tüm Gen Dizi Analizi)	EBPL gene (Sequence Analysis-All Coding Exons)	EBPL	EBPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBNA1BP2 geni (Tüm Gen Dizi Analizi)	EBNA1BP2 gene (Sequence Analysis-All Coding Exons)	EBNA1BP2	EBNA1BP2, EBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBLN2 geni (Tüm Gen Dizi Analizi)	EBLN2 gene (Sequence Analysis-All Coding Exons)	EBLN2	EBLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBLN1 geni (Tüm Gen Dizi Analizi)	EBLN1 gene (Sequence Analysis-All Coding Exons)	EBLN1	EBLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBI3 geni (Tüm Gen Dizi Analizi)	EBI3 gene (Sequence Analysis-All Coding Exons)	EBI3	EBI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBF4 geni (Tüm Gen Dizi Analizi)	EBF4 gene (Sequence Analysis-All Coding Exons)	EBF4	EBF4, KIAA1442	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBF2 geni (Tüm Gen Dizi Analizi)	EBF2 gene (Sequence Analysis-All Coding Exons)	EBF2	EBF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBF1 geni (Tüm Gen Dizi Analizi)	EBF1 gene (Sequence Analysis-All Coding Exons)	EBF1	EBF, OLF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EBAG9 geni (Tüm Gen Dizi Analizi)	EBAG9 gene (Sequence Analysis-All Coding Exons)	EBAG9	EBAG9, RCAS1, EB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EAF2 geni (Tüm Gen Dizi Analizi)	EAF2 gene (Sequence Analysis-All Coding Exons)	EAF2	EAF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EAF1 geni (Tüm Gen Dizi Analizi)	EAF1 gene (Sequence Analysis-All Coding Exons)	EAF1	EAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E4F1 geni (Tüm Gen Dizi Analizi)	E4F1 gene (Sequence Analysis-All Coding Exons)	E4F1	E4F1, E4F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F8 geni (Tüm Gen Dizi Analizi)	E2F8 gene (Sequence Analysis-All Coding Exons)	E2F8	E2F8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MDS2 geni (Tüm Gen Dizi Analizi)	MDS2 gene (Sequence Analysis-All Coding Exons)	MDS2	MDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F7 geni (Tüm Gen Dizi Analizi)	E2F7 gene (Sequence Analysis-All Coding Exons)	E2F7	E2F7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F5 geni (Tüm Gen Dizi Analizi)	E2F5 gene (Sequence Analysis-All Coding Exons)	E2F5	E2F5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F4 geni (Tüm Gen Dizi Analizi)	E2F4 gene (Sequence Analysis-All Coding Exons)	E2F4	E2F4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F3 geni (Tüm Gen Dizi Analizi)	E2F3 gene (Sequence Analysis-All Coding Exons)	E2F3	E2F3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F2 geni (Tüm Gen Dizi Analizi)	E2F2 gene (Sequence Analysis-All Coding Exons)	E2F2	E2F2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F1 geni (Tüm Gen Dizi Analizi)	E2F1 gene (Sequence Analysis-All Coding Exons)	E2F1	E2F1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYRK4 geni (Tüm Gen Dizi Analizi)	DYRK4 gene (Sequence Analysis-All Coding Exons)	DYRK4	DYRK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYRK3 geni (Tüm Gen Dizi Analizi)	DYRK3 gene (Sequence Analysis-All Coding Exons)	DYRK3	DYRK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYRK2 geni (Tüm Gen Dizi Analizi)	DYRK2 gene (Sequence Analysis-All Coding Exons)	DYRK2	DYRK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLT3 geni (Tüm Gen Dizi Analizi)	DYNLT3 gene (Sequence Analysis-All Coding Exons)	DYNLT3	TCTEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLT1 geni (Tüm Gen Dizi Analizi)	DYNLT1 gene (Sequence Analysis-All Coding Exons)	DYNLT1	TCTEL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLRB2 geni (Tüm Gen Dizi Analizi)	DYNLRB2 gene (Sequence Analysis-All Coding Exons)	DYNLRB2	DYNLRB2, DNCL2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLRB1 geni (Tüm Gen Dizi Analizi)	DYNLRB1 gene (Sequence Analysis-All Coding Exons)	DYNLRB1	DYNLRB1, DNCL2A, DNLC2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLL2 geni (Tüm Gen Dizi Analizi)	DYNLL2 gene (Sequence Analysis-All Coding Exons)	DYNLL2	DYNLL2, DLC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNLL1 geni (Tüm Gen Dizi Analizi)	DYNLL1 gene (Sequence Analysis-All Coding Exons)	DYNLL1	DYNLL1, DNCL1, DLC1, PIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNC1LI1 geni (Tüm Gen Dizi Analizi)	DYNC1LI1 gene (Sequence Analysis-All Coding Exons)	DYNC1LI1	LIC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNC1I2 geni (Tüm Gen Dizi Analizi)	DYNC1I2 gene (Sequence Analysis-All Coding Exons)	DYNC1I2	DYNC1I2, DNC2, IC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNC1I1 geni (Tüm Gen Dizi Analizi)	DYNC1I1 gene (Sequence Analysis-All Coding Exons)	DYNC1I1	DNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYDC1 geni (Tüm Gen Dizi Analizi)	DYDC1 gene (Sequence Analysis-All Coding Exons)	DYDC1	DYDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DXO geni (Tüm Gen Dizi Analizi)	DXO gene (Sequence Analysis-All Coding Exons)	DXO	DOM3Z	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUXA geni (Tüm Gen Dizi Analizi)	DUXA gene (Sequence Analysis-All Coding Exons)	DUXA	DUXA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUT geni (Tüm Gen Dizi Analizi)	DUT gene (Sequence Analysis-All Coding Exons)	DUT	DUT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP9 geni (Tüm Gen Dizi Analizi)	DUSP9 gene (Sequence Analysis-All Coding Exons)	DUSP9	DUSP9, MKP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP8 geni (Tüm Gen Dizi Analizi)	DUSP8 gene (Sequence Analysis-All Coding Exons)	DUSP8	DUSP8, HVH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP7 geni (Tüm Gen Dizi Analizi)	DUSP7 gene (Sequence Analysis-All Coding Exons)	DUSP7	DUSP7, MKPX, PYST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP5 geni (Tüm Gen Dizi Analizi)	DUSP5 gene (Sequence Analysis-All Coding Exons)	DUSP5	DUSP5, HVH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP4 geni (Tüm Gen Dizi Analizi)	DUSP4 gene (Sequence Analysis-All Coding Exons)	DUSP4	DUSP4, MKP2, HVH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP3 geni (Tüm Gen Dizi Analizi)	DUSP3 gene (Sequence Analysis-All Coding Exons)	DUSP3	DUSP3, VHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP22 geni (Tüm Gen Dizi Analizi)	DUSP22 gene (Sequence Analysis-All Coding Exons)	DUSP22	DUSP22, LMWDSP2, MKPX, VHX, IKAP, PTP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP21 geni (Tüm Gen Dizi Analizi)	DUSP21 gene (Sequence Analysis-All Coding Exons)	DUSP21	DUSP21, LMWDSP21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP2 geni (Tüm Gen Dizi Analizi)	DUSP2 gene (Sequence Analysis-All Coding Exons)	DUSP2	DUSP2, PAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP16 geni (Tüm Gen Dizi Analizi)	DUSP16 gene (Sequence Analysis-All Coding Exons)	DUSP16	DUSP16, MKP7, KIAA1700	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP15 geni (Tüm Gen Dizi Analizi)	DUSP15 gene (Sequence Analysis-All Coding Exons)	DUSP15	DUSP15, VHY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP14 geni (Tüm Gen Dizi Analizi)	DUSP14 gene (Sequence Analysis-All Coding Exons)	DUSP14	DUSP14, MKP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP13 geni (Tüm Gen Dizi Analizi)	DUSP13 gene (Sequence Analysis-All Coding Exons)	DUSP13	DUSP13, TMDP, MDSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP12 geni (Tüm Gen Dizi Analizi)	DUSP12 gene (Sequence Analysis-All Coding Exons)	DUSP12	DUSP1, YVH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP11 geni (Tüm Gen Dizi Analizi)	DUSP11 gene (Sequence Analysis-All Coding Exons)	DUSP11	DUSP11, PIR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP10 geni (Tüm Gen Dizi Analizi)	DUSP10 gene (Sequence Analysis-All Coding Exons)	DUSP10	DUSP10, MKP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP1 geni (Tüm Gen Dizi Analizi)	DUSP1 gene (Sequence Analysis-All Coding Exons)	DUSP1	DUSP1, CL100, PTPN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUS2 geni (Tüm Gen Dizi Analizi)	DUS2 gene (Sequence Analysis-All Coding Exons)	DUS2	DUS2L, DUS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUOXA1 geni (Tüm Gen Dizi Analizi)	DUOXA1 gene (Sequence Analysis-All Coding Exons)	DUOXA1	DUOXA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUOX1 geni (Tüm Gen Dizi Analizi)	DUOX1 gene (Sequence Analysis-All Coding Exons)	DUOX1	DUOX1, THOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUBR geni (Tüm Gen Dizi Analizi)	DUBR gene (Sequence Analysis-All Coding Exons)	DUBR	DUBR, DUM, LINC00883	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTYMK geni (Tüm Gen Dizi Analizi)	DTYMK gene (Sequence Analysis-All Coding Exons)	DTYMK	DTYMK, TYMK, CDC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTX4 geni (Tüm Gen Dizi Analizi)	DTX4 gene (Sequence Analysis-All Coding Exons)	DTX4	DTX4, RNF155, KIAA0937	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTX3L geni (Tüm Gen Dizi Analizi)	DTX3L gene (Sequence Analysis-All Coding Exons)	DTX3L	DTX3L, BBAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTX3 geni (Tüm Gen Dizi Analizi)	DTX3 gene (Sequence Analysis-All Coding Exons)	DTX3	DTX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTX2 geni (Tüm Gen Dizi Analizi)	DTX2 gene (Sequence Analysis-All Coding Exons)	DTX2	DTX2, KIAA1528	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTX1 geni (Tüm Gen Dizi Analizi)	DTX1 gene (Sequence Analysis-All Coding Exons)	DTX1	DTX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTNB geni (Tüm Gen Dizi Analizi)	DTNB gene (Sequence Analysis-All Coding Exons)	DTNB	DTNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTL geni (Tüm Gen Dizi Analizi)	DTL gene (Sequence Analysis-All Coding Exons)	DTL	DTL, RAMP, DCAF2, CDT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTHD1 geni (Tüm Gen Dizi Analizi)	DTHD1 gene (Sequence Analysis-All Coding Exons)	DTHD1	DTHD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DTD1 geni (Tüm Gen Dizi Analizi)	DTD1 gene (Sequence Analysis-All Coding Exons)	DTD1	DTD1, C20orf88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSTN geni (Tüm Gen Dizi Analizi)	DSTN gene (Sequence Analysis-All Coding Exons)	DSTN	DSTN, ADF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSN1 geni (Tüm Gen Dizi Analizi)	DSN1 gene (Sequence Analysis-All Coding Exons)	DSN1	DSN1, C20orf172	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSG3 geni (Tüm Gen Dizi Analizi)	DSG3 gene (Sequence Analysis-All Coding Exons)	DSG3	DSG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSEL geni (Tüm Gen Dizi Analizi)	DSEL gene (Sequence Analysis-All Coding Exons)	DSEL	DSEL, C18orf4, NCAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCR8 geni (Tüm Gen Dizi Analizi)	DSCR8 gene (Sequence Analysis-All Coding Exons)	DSCR8	DSCR8, MMA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCR4 geni (Tüm Gen Dizi Analizi)	DSCR4 gene (Sequence Analysis-All Coding Exons)	DSCR4	DSCR4, DSCR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCR3 geni (Tüm Gen Dizi Analizi)	DSCR3 gene (Sequence Analysis-All Coding Exons)	DSCR3	DSCR3, DSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCC1 geni (Tüm Gen Dizi Analizi)	DSCC1 gene (Sequence Analysis-All Coding Exons)	DSCC1	DSCC1, DCC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCAML1 geni (Tüm Gen Dizi Analizi)	DSCAML1 gene (Sequence Analysis-All Coding Exons)	DSCAML1	DSCAML1, KIAA1132	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSCAM geni (Tüm Gen Dizi Analizi)	DSCAM gene (Sequence Analysis-All Coding Exons)	DSCAM	DSCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DSC1 geni (Tüm Gen Dizi Analizi)	DSC1 gene (Sequence Analysis-All Coding Exons)	DSC1	DSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRP2 geni (Tüm Gen Dizi Analizi)	DRP2 gene (Sequence Analysis-All Coding Exons)	DRP2	DRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DROSHA geni (Tüm Gen Dizi Analizi)	DROSHA gene (Sequence Analysis-All Coding Exons)	DROSHA	RNASEN, DROSHA, RANSE3L, RN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRGX geni (Tüm Gen Dizi Analizi)	DRGX gene (Sequence Analysis-All Coding Exons)	DRGX	DRGX, DRG11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRG2 geni (Tüm Gen Dizi Analizi)	DRG2 gene (Sequence Analysis-All Coding Exons)	DRG2	DRG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MBNL2 geni (Tüm Gen Dizi Analizi)	MBNL2 gene (Sequence Analysis-All Coding Exons)	MBNL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRD2 geni (Tüm Gen Dizi Analizi)	DRD2 gene (Sequence Analysis-All Coding Exons)	DRD2	DRD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRD1 geni (Tüm Gen Dizi Analizi)	DRD1 gene (Sequence Analysis-All Coding Exons)	DRD1	DRD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRAXIN geni (Tüm Gen Dizi Analizi)	DRAXIN gene (Sequence Analysis-All Coding Exons)	DRAXIN	DRAXIN, C1orf187	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRAP1 geni (Tüm Gen Dizi Analizi)	DRAP1 gene (Sequence Analysis-All Coding Exons)	DRAP1	DRAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRAM1 geni (Tüm Gen Dizi Analizi)	DRAM1 gene (Sequence Analysis-All Coding Exons)	DRAM1	DRAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DR1 geni (Tüm Gen Dizi Analizi)	DR1 gene (Sequence Analysis-All Coding Exons)	DR1	DR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPYSL5 geni (Tüm Gen Dizi Analizi)	DPYSL5 gene (Sequence Analysis-All Coding Exons)	DPYSL5	DPYSL5, CRMP5, CRAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPYSL4 geni (Tüm Gen Dizi Analizi)	DPYSL4 gene (Sequence Analysis-All Coding Exons)	DPYSL4	DPYSL4, CRMP3, ULIP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPYSL3 geni (Tüm Gen Dizi Analizi)	DPYSL3 gene (Sequence Analysis-All Coding Exons)	DPYSL3	DPYSL3, ULIP, DRP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPYSL2 geni (Tüm Gen Dizi Analizi)	DPYSL2 gene (Sequence Analysis-All Coding Exons)	DPYSL2	DPYSL2, DRP2, CRMP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

DPY30 geni (Tüm Gen Dizi Analizi)	DPY30 gene (Sequence Analysis-All Coding Exons)	DPY30	DPY30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPY19L4 geni (Tüm Gen Dizi Analizi)	DPY19L4 gene (Sequence Analysis-All Coding Exons)	DPY19L4	DPY19L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPY19L3 geni (Tüm Gen Dizi Analizi)	DPY19L3 gene (Sequence Analysis-All Coding Exons)	DPY19L3	DPY19L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPY19L1 geni (Tüm Gen Dizi Analizi)	DPY19L1 gene (Sequence Analysis-All Coding Exons)	DPY19L1	DPY19L1, KIAA0877	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPT geni (Tüm Gen Dizi Analizi)	DPT gene (Sequence Analysis-All Coding Exons)	DPT	DPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPRX geni (Tüm Gen Dizi Analizi)	DPRX gene (Sequence Analysis-All Coding Exons)	DPRX	DPRX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPPA5 geni (Tüm Gen Dizi Analizi)	DPPA5 gene (Sequence Analysis-All Coding Exons)	DPPA5	DPPA5, ESG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPPA4 geni (Tüm Gen Dizi Analizi)	DPPA4 gene (Sequence Analysis-All Coding Exons)	DPPA4	DPPA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPPA3 geni (Tüm Gen Dizi Analizi)	DPPA3 gene (Sequence Analysis-All Coding Exons)	DPPA3	DPPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPPA2 geni (Tüm Gen Dizi Analizi)	DPPA2 gene (Sequence Analysis-All Coding Exons)	DPPA2	DPPA2, PESCRG1, ECAT15-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP8 geni (Tüm Gen Dizi Analizi)	DPP8 gene (Sequence Analysis-All Coding Exons)	DPP8	DPP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP7 geni (Tüm Gen Dizi Analizi)	DPP7 gene (Sequence Analysis-All Coding Exons)	DPP7	DPP7, QPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP4 geni (Tüm Gen Dizi Analizi)	DPP4 gene (Sequence Analysis-All Coding Exons)	DPP4	DPP4, CD26, ADCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP3 geni (Tüm Gen Dizi Analizi)	DPP3 gene (Sequence Analysis-All Coding Exons)	DPP3	DPP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAT2B geni (Tüm Gen Dizi Analizi)	MAT2B gene (Sequence Analysis-All Coding Exons)	MAT2B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP10 geni (Tüm Gen Dizi Analizi)	DPP10 gene (Sequence Analysis-All Coding Exons)	DPP10	DPP10, DPRP3, KIAA1492	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPH7 geni (Tüm Gen Dizi Analizi)	DPH7 gene (Sequence Analysis-All Coding Exons)	DPH7	WDR85	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPH5 geni (Tüm Gen Dizi Analizi)	DPH5 gene (Sequence Analysis-All Coding Exons)	DPH5	DPH5, HSPC143	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPH3 geni (Tüm Gen Dizi Analizi)	DPH3 gene (Sequence Analysis-All Coding Exons)	DPH3	DPH3, KTI11, ZCSL2, DELGIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPH2 geni (Tüm Gen Dizi Analizi)	DPH2 gene (Sequence Analysis-All Coding Exons)	DPH2	DPH2, DPH2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPF3 geni (Tüm Gen Dizi Analizi)	DPF3 gene (Sequence Analysis-All Coding Exons)	DPF3	DPF3, CERD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPF2 geni (Tüm Gen Dizi Analizi)	DPF2 gene (Sequence Analysis-All Coding Exons)	DPF2	DPF2, REQ, UBID4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPF1 geni (Tüm Gen Dizi Analizi)	DPF1 gene (Sequence Analysis-All Coding Exons)	DPF1	DPF1, NEUD4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPEP3 geni (Tüm Gen Dizi Analizi)	DPEP3 gene (Sequence Analysis-All Coding Exons)	DPEP3	DPEP3, MBD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPEP2 geni (Tüm Gen Dizi Analizi)	DPEP2 gene (Sequence Analysis-All Coding Exons)	DPEP2	DPEP2, MBD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPEP1 geni (Tüm Gen Dizi Analizi)	DPEP1 gene (Sequence Analysis-All Coding Exons)	DPEP1	DPEP1, RDP, MDP, MBD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPCR1 geni (Tüm Gen Dizi Analizi)	DPCR1 gene (Sequence Analysis-All Coding Exons)	DPCR1	DPCR1, C6orf37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPCD geni (Tüm Gen Dizi Analizi)	DPCD gene (Sequence Analysis-All Coding Exons)	DPCD	DPCD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOT1L geni (Tüm Gen Dizi Analizi)	DOT1L gene (Sequence Analysis-All Coding Exons)	DOT1L	DOT1L, DOT1, KIAA1814	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOPEY2 geni (Tüm Gen Dizi Analizi)	DOPEY2 gene (Sequence Analysis-All Coding Exons)	DOPEY2	DOPEY2, 21orf5, KIAA0933	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOPEY1 geni (Tüm Gen Dizi Analizi)	DOPEY1 gene (Sequence Analysis-All Coding Exons)	DOPEY1	DOPEY1, DOP1, KIAA1117	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOLPP1 geni (Tüm Gen Dizi Analizi)	DOLPP1 gene (Sequence Analysis-All Coding Exons)	DOLPP1	DOLPP1, LSFR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK5 geni (Tüm Gen Dizi Analizi)	DOK5 gene (Sequence Analysis-All Coding Exons)	DOK5	DOK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK4 geni (Tüm Gen Dizi Analizi)	DOK4 gene (Sequence Analysis-All Coding Exons)	DOK4	DOK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK3 geni (Tüm Gen Dizi Analizi)	DOK3 gene (Sequence Analysis-All Coding Exons)	DOK3	DOK3, DOKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK2 geni (Tüm Gen Dizi Analizi)	DOK2 gene (Sequence Analysis-All Coding Exons)	DOK2	DOK2, P56DOK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK1 geni (Tüm Gen Dizi Analizi)	DOK1 gene (Sequence Analysis-All Coding Exons)	DOK1	DOK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOHH geni (Tüm Gen Dizi Analizi)	DOHH gene (Sequence Analysis-All Coding Exons)	DOHH	DOHH, HLRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK9 geni (Tüm Gen Dizi Analizi)	DOCK9 gene (Sequence Analysis-All Coding Exons)	DOCK9	ZIZ1, KIAA1058	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK5 geni (Tüm Gen Dizi Analizi)	DOCK5 gene (Sequence Analysis-All Coding Exons)	DOCK5	DOCK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK4 geni (Tüm Gen Dizi Analizi)	DOCK4 gene (Sequence Analysis-All Coding Exons)	DOCK4	DOCK4, KIAA0716	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK3 geni (Tüm Gen Dizi Analizi)	DOCK3 gene (Sequence Analysis-All Coding Exons)	DOCK3	DOCK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK11 geni (Tüm Gen Dizi Analizi)	DOCK11 gene (Sequence Analysis-All Coding Exons)	DOCK11	DOCK11, ACG, ZIZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK10 geni (Tüm Gen Dizi Analizi)	DOCK10 gene (Sequence Analysis-All Coding Exons)	DOCK10	DOCK10, KIAA0694, ZIZ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOCK1 geni (Tüm Gen Dizi Analizi)	DOCK1 gene (Sequence Analysis-All Coding Exons)	DOCK1	DOCK1, DOCK180	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOC2B geni (Tüm Gen Dizi Analizi)	DOC2B gene (Sequence Analysis-All Coding Exons)	DOC2B	DOC2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOC2A geni (Tüm Gen Dizi Analizi)	DOC2A gene (Sequence Analysis-All Coding Exons)	DOC2A	DOC2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNTTIP2 geni (Tüm Gen Dizi Analizi)	DNTTIP2 gene (Sequence Analysis-All Coding Exons)	DNTTIP2	DNTTIP2, ERBP, FCF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNTTIP1 geni (Tüm Gen Dizi Analizi)	DNTTIP1 gene (Sequence Analysis-All Coding Exons)	DNTTIP1	DNTTIP1, TDIF1, C20orf167	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNTT geni (Tüm Gen Dizi Analizi)	DNTT gene (Sequence Analysis-All Coding Exons)	DNTT	DNTT, TDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNPEP geni (Tüm Gen Dizi Analizi)	DNPEP gene (Sequence Analysis-All Coding Exons)	DNPEP	DNPEP, ASPEP, DAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNMT3L geni (Tüm Gen Dizi Analizi)	DNMT3L gene (Sequence Analysis-All Coding Exons)	DNMT3L	DNMT3L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNMBP geni (Tüm Gen Dizi Analizi)	DNMBP gene (Sequence Analysis-All Coding Exons)	DNMBP	DNMBP, TUBA, KIAA1010	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNM3 geni (Tüm Gen Dizi Analizi)	DNM3 gene (Sequence Analysis-All Coding Exons)	DNM3	DNM3, KIAA0820	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNHD1 geni (Tüm Gen Dizi Analizi)	DNHD1 gene (Sequence Analysis-All Coding Exons)	DNHD1	DNHD1, FLJ00251	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNER geni (Tüm Gen Dizi Analizi)	DNER gene (Sequence Analysis-All Coding Exons)	DNER	DNER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DND1 geni (Tüm Gen Dizi Analizi)	DND1 gene (Sequence Analysis-All Coding Exons)	DND1	DND1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNASE2B geni (Tüm Gen Dizi Analizi)	DNASE2B gene (Sequence Analysis-All Coding Exons)	DNASE2B	DNASE2B, DLAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNASE2 geni (Tüm Gen Dizi Analizi)	DNASE2 gene (Sequence Analysis-All Coding Exons)	DNASE2	DNASE2, DNL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNASE1L1 geni (Tüm Gen Dizi Analizi)	DNASE1L1 gene (Sequence Analysis-All Coding Exons)	DNASE1L1	DNASE1L1, DNL1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK6 geni (Tüm Gen Dizi Analizi)	MAPK6 gene (Sequence Analysis-All Coding Exons)	MAPK6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNALI1 geni (Tüm Gen Dizi Analizi)	DNALI1 gene (Sequence Analysis-All Coding Exons)	DNALI1	DNALI1, P28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC9 geni (Tüm Gen Dizi Analizi)	DNAJC9 gene (Sequence Analysis-All Coding Exons)	DNAJC9	DNAJC9, JDD1, KIAA0974	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC7 geni (Tüm Gen Dizi Analizi)	DNAJC7 gene (Sequence Analysis-All Coding Exons)	DNAJC7	DANJC7, TTC2, TPR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC5G geni (Tüm Gen Dizi Analizi)	DNAJC5G gene (Sequence Analysis-All Coding Exons)	DNAJC5G	DNAJC5G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK13 geni (Tüm Gen Dizi Analizi)	MAPK13 gene (Sequence Analysis-All Coding Exons)	MAPK13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC5B geni (Tüm Gen Dizi Analizi)	DNAJC5B gene (Sequence Analysis-All Coding Exons)	DNAJC5B	DNAJC5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAPK11 geni (Tüm Gen Dizi Analizi)	MAPK11 gene (Sequence Analysis-All Coding Exons)	MAPK11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC4 geni (Tüm Gen Dizi Analizi)	DNAJC4 gene (Sequence Analysis-All Coding Exons)	DNAJC4	DANJC4, HSPF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC27 geni (Tüm Gen Dizi Analizi)	DNAJC27 gene (Sequence Analysis-All Coding Exons)	DNAJC27	DNAJC27, RBJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC24 geni (Tüm Gen Dizi Analizi)	DNAJC24 gene (Sequence Analysis-All Coding Exons)	DNAJC24	DPH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC2 geni (Tüm Gen Dizi Analizi)	DNAJC2 gene (Sequence Analysis-All Coding Exons)	DNAJC2	DNAJC2, ZRF1, MPP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC17 geni (Tüm Gen Dizi Analizi)	DNAJC17 gene (Sequence Analysis-All Coding Exons)	DNAJC17	DNAJC17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC15 geni (Tüm Gen Dizi Analizi)	DNAJC15 gene (Sequence Analysis-All Coding Exons)	DNAJC15	DNAJC15, MCJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP6 geni (Tüm Gen Dizi Analizi)	MAP6 gene (Sequence Analysis-All Coding Exons)	MAP6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC14 geni (Tüm Gen Dizi Analizi)	DNAJC14 gene (Sequence Analysis-All Coding Exons)	DNAJC14	DRIP78, HDJ3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC13 geni (Tüm Gen Dizi Analizi)	DNAJC13 gene (Sequence Analysis-All Coding Exons)	DNAJC13	DNAJC13, RME8, KIAA0678	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC11 geni (Tüm Gen Dizi Analizi)	DNAJC11 gene (Sequence Analysis-All Coding Exons)	DNAJC11	DNAJC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC10 geni (Tüm Gen Dizi Analizi)	DNAJC10 gene (Sequence Analysis-All Coding Exons)	DNAJC10	DNAJC10, ERDJ5, JPDI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJC1 geni (Tüm Gen Dizi Analizi)	DNAJC1 gene (Sequence Analysis-All Coding Exons)	DNAJC1	DNAJC1, HTJ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB9 geni (Tüm Gen Dizi Analizi)	DNAJB9 gene (Sequence Analysis-All Coding Exons)	DNAJB9	DNAJB9, MDG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB8 geni (Tüm Gen Dizi Analizi)	DNAJB8 gene (Sequence Analysis-All Coding Exons)	DNAJB8	DNAJB8, DJ6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB7 geni (Tüm Gen Dizi Analizi)	DNAJB7 gene (Sequence Analysis-All Coding Exons)	DNAJB7	DNAJB7, DJ5, HSC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB5 geni (Tüm Gen Dizi Analizi)	DNAJB5 gene (Sequence Analysis-All Coding Exons)	DNAJB5	DNAJB5, KIAA1045, HSC40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB4 geni (Tüm Gen Dizi Analizi)	DNAJB4 gene (Sequence Analysis-All Coding Exons)	DNAJB4	DNAJB4, HLJ1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB12 geni (Tüm Gen Dizi Analizi)	DNAJB12 gene (Sequence Analysis-All Coding Exons)	DNAJB12	DNAJB12, DJ10, FLJ00027	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB11 geni (Tüm Gen Dizi Analizi)	DNAJB11 gene (Sequence Analysis-All Coding Exons)	DNAJB11	DNAJB11, HEDJ, DJ9, ABBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJB1 geni (Tüm Gen Dizi Analizi)	DNAJB1 gene (Sequence Analysis-All Coding Exons)	DNAJB1	DNAJB1, HSPF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJA3 geni (Tüm Gen Dizi Analizi)	DNAJA3 gene (Sequence Analysis-All Coding Exons)	DNAJA3	DNAJA3, TID1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJA2 geni (Tüm Gen Dizi Analizi)	DNAJA2 gene (Sequence Analysis-All Coding Exons)	DNAJA2	DNAJA2, DJ3, CPR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH9 geni (Tüm Gen Dizi Analizi)	DNAH9 gene (Sequence Analysis-All Coding Exons)	DNAH9	DNAH9, DNAH17L, DNEL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH8 geni (Tüm Gen Dizi Analizi)	DNAH8 gene (Sequence Analysis-All Coding Exons)	DNAH8	DNAH8, HDHC9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH7 geni (Tüm Gen Dizi Analizi)	DNAH7 gene (Sequence Analysis-All Coding Exons)	DNAH7	DNAH7, KIAA0944	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

DNAH6 geni (Tüm Gen Dizi Analizi)	DNAH6 gene (Sequence Analysis-All Coding Exons)	DNAH6	DNAH6, HL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH3 geni (Tüm Gen Dizi Analizi)	DNAH3 gene (Sequence Analysis-All Coding Exons)	DNAH3	DNAH3, DNAHC3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH2 geni (Tüm Gen Dizi Analizi)	DNAH2 gene (Sequence Analysis-All Coding Exons)	DNAH2	DNAH2, DNAHC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH17 geni (Tüm Gen Dizi Analizi)	DNAH17 gene (Sequence Analysis-All Coding Exons)	DNAH17	DNAH17, DNEL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH14 geni (Tüm Gen Dizi Analizi)	DNAH14 gene (Sequence Analysis-All Coding Exons)	DNAH14	DNAH14, HL18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH12 geni (Tüm Gen Dizi Analizi)	DNAH12 gene (Sequence Analysis-All Coding Exons)	DNAH12	DNAH12, DNAHC3, HL19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH10 geni (Tüm Gen Dizi Analizi)	DNAH10 gene (Sequence Analysis-All Coding Exons)	DNAH10	DNAH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAH1 geni (Tüm Gen Dizi Analizi)	DNAH1 gene (Sequence Analysis-All Coding Exons)	DNAH1	DNAH1, HL11, DNAHC1, HDHC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMXL1 geni (Tüm Gen Dizi Analizi)	DMXL1 gene (Sequence Analysis-All Coding Exons)	DMXL1	DMXL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMWD geni (Tüm Gen Dizi Analizi)	DMWD gene (Sequence Analysis-All Coding Exons)	DMWD	DMWD, DMRN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMTN geni (Tüm Gen Dizi Analizi)	DMTN gene (Sequence Analysis-All Coding Exons)	DMTN	EPB49, DMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMTF1 geni (Tüm Gen Dizi Analizi)	DMTF1 gene (Sequence Analysis-All Coding Exons)	DMTF1	DMTF1, DMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRTC2 geni (Tüm Gen Dizi Analizi)	DMRTC2 gene (Sequence Analysis-All Coding Exons)	DMRTC2	DMRTC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRTC1 geni (Tüm Gen Dizi Analizi)	DMRTC1 gene (Sequence Analysis-All Coding Exons)	DMRTC1	DMRTC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRTB1 geni (Tüm Gen Dizi Analizi)	DMRTB1 gene (Sequence Analysis-All Coding Exons)	DMRTB1	DMRTB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRTA2 geni (Tüm Gen Dizi Analizi)	DMRTA2 gene (Sequence Analysis-All Coding Exons)	DMRTA2	DMRTA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAP1LC3C geni (Tüm Gen Dizi Analizi)	MAP1LC3C gene (Sequence Analysis-All Coding Exons)	MAP1LC3C		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRTA1 geni (Tüm Gen Dizi Analizi)	DMRTA1 gene (Sequence Analysis-All Coding Exons)	DMRTA1	DMRTA1, DMRT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRT3 geni (Tüm Gen Dizi Analizi)	DMRT3 gene (Sequence Analysis-All Coding Exons)	DMRT3	DMRT3, DMRTA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRT2 geni (Tüm Gen Dizi Analizi)	DMRT2 gene (Sequence Analysis-All Coding Exons)	DMRT2	DMRT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMRT1 geni (Tüm Gen Dizi Analizi)	DMRT1 gene (Sequence Analysis-All Coding Exons)	DMRT1	DMRT1, DMT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMKN geni (Tüm Gen Dizi Analizi)	DMKN gene (Sequence Analysis-All Coding Exons)	DMKN	DMKN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMC1 geni (Tüm Gen Dizi Analizi)	DMC1 gene (Sequence Analysis-All Coding Exons)	DMC1	DMC1, LIM15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMBX1 geni (Tüm Gen Dizi Analizi)	DMBX1 gene (Sequence Analysis-All Coding Exons)	DMBX1	DMBX1, OTX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMBT1 geni (Tüm Gen Dizi Analizi)	DMBT1 gene (Sequence Analysis-All Coding Exons)	DMBT1	DMBT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DMAP1 geni (Tüm Gen Dizi Analizi)	DMAP1 gene (Sequence Analysis-All Coding Exons)	DMAP1	DNMAP1, DMAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLX6 geni (Tüm Gen Dizi Analizi)	DLX6 gene (Sequence Analysis-All Coding Exons)	DLX6	DLX6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLX2 geni (Tüm Gen Dizi Analizi)	DLX2 gene (Sequence Analysis-All Coding Exons)	DLX2	DLX2, TES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLX1 geni (Tüm Gen Dizi Analizi)	DLX1 gene (Sequence Analysis-All Coding Exons)	DLX1	DLX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLST geni (Tüm Gen Dizi Analizi)	DLST gene (Sequence Analysis-All Coding Exons)	DLST	DLST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLK1 geni (Tüm Gen Dizi Analizi)	DLK1 gene (Sequence Analysis-All Coding Exons)	DLK1	DLK1, PREF1, FA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLK1 geni (Tüm Gen Dizi Analizi)	DLK1 gene (Sequence Analysis-All Coding Exons)	DLK1	DLK1, PREF1, FA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLK1 geni (Tüm Gen Dizi Analizi)	DLK1 gene (Sequence Analysis-All Coding Exons)	DLK1	DLK1, PREF1, FA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLK1 geni (Tüm Gen Dizi Analizi)	DLK1 gene (Sequence Analysis-All Coding Exons)	DLK1	DLK1, PREF1, FA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLK1 geni (Tüm Gen Dizi Analizi)	DLK1 gene (Sequence Analysis-All Coding Exons)	DLK1	DLK1, PREF1, FA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLGAP4 geni (Tüm Gen Dizi Analizi)	DLGAP4 gene (Sequence Analysis-All Coding Exons)	DLGAP4	DLGAP4, SAPAP4, KIAA0964	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLGAP2 geni (Tüm Gen Dizi Analizi)	DLGAP2 gene (Sequence Analysis-All Coding Exons)	DLGAP2	DLGAP2, DAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLGAP1 geni (Tüm Gen Dizi Analizi)	DLGAP1 gene (Sequence Analysis-All Coding Exons)	DLGAP1	DLGAP1, DAP1, DLGAP1A, DLGAP1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLG5 geni (Tüm Gen Dizi Analizi)	DLG5 gene (Sequence Analysis-All Coding Exons)	DLG5	DLG5, PDLG, KIAA0583	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLG4 geni (Tüm Gen Dizi Analizi)	DLG4 gene (Sequence Analysis-All Coding Exons)	DLG4	DLG4, PSD95, SAP90	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLG2 geni (Tüm Gen Dizi Analizi)	DLG2 gene (Sequence Analysis-All Coding Exons)	DLG2	DLG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLG1 geni (Tüm Gen Dizi Analizi)	DLG1 gene (Sequence Analysis-All Coding Exons)	DLG1	DLG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DKKL1 geni (Tüm Gen Dizi Analizi)	DKKL1 gene (Sequence Analysis-All Coding Exons)	DKKL1	DKKL1, SGY1, SGY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DKK4 geni (Tüm Gen Dizi Analizi)	DKK4 gene (Sequence Analysis-All Coding Exons)	DKK4	DKK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DKK3 geni (Tüm Gen Dizi Analizi)	DKK3 gene (Sequence Analysis-All Coding Exons)	DKK3	DKK3, RIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
MAL2 geni (Tüm Gen Dizi Analizi)	MAL2 gene (Sequence Analysis-All Coding Exons)	MAL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DKK1 geni (Tüm Gen Dizi Analizi)	DKK1 gene (Sequence Analysis-All Coding Exons)	DKK1	DKK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIXDC1 geni (Tüm Gen Dizi Analizi)	DIXDC1 gene (Sequence Analysis-All Coding Exons)	DIXDC1	DIXDC1, CCD1, KIAA1735	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP3 geni (Tüm Gen Dizi Analizi)	DISP3 gene (Sequence Analysis-All Coding Exons)	DISP3	PTCHD2, DISP3, KIAA1337	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP2 geni (Tüm Gen Dizi Analizi)	DISP2 gene (Sequence Analysis-All Coding Exons)	DISP2	DISPB, KIAA1742	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DISP1 geni (Tüm Gen Dizi Analizi)	DISP1 gene (Sequence Analysis-All Coding Exons)	DISP1	DISPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIS3L geni (Tüm Gen Dizi Analizi)	DIS3L gene (Sequence Analysis-All Coding Exons)	DIS3L	DIS3L, DIS3L1, KIAA1955	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIS3 geni (Tüm Gen Dizi Analizi)	DIS3 gene (Sequence Analysis-All Coding Exons)	DIS3	DIS3, KIAA1008	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIRC1 geni (Tüm Gen Dizi Analizi)	DIRC1 gene (Sequence Analysis-All Coding Exons)	DIRC1	DIRC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIRAS3 geni (Tüm Gen Dizi Analizi)	DIRAS3 gene (Sequence Analysis-All Coding Exons)	DIRAS3	DIRAS3, ARHI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIRAS2 geni (Tüm Gen Dizi Analizi)	DIRAS2 gene (Sequence Analysis-All Coding Exons)	DIRAS2	DIRAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIRAS1 geni (Tüm Gen Dizi Analizi)	DIRAS1 gene (Sequence Analysis-All Coding Exons)	DIRAS1	DIRAS1, RIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIP2C geni (Tüm Gen Dizi Analizi)	DIP2C gene (Sequence Analysis-All Coding Exons)	DIP2C	DIP2C, KIAA0934	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIP2A geni (Tüm Gen Dizi Analizi)	DIP2A gene (Sequence Analysis-All Coding Exons)	DIP2A	DIP2, KIAA0184	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIO3 geni (Tüm Gen Dizi Analizi)	DIO3 gene (Sequence Analysis-All Coding Exons)	DIO3	DIO3, TXDI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIO2 geni (Tüm Gen Dizi Analizi)	DIO2 gene (Sequence Analysis-All Coding Exons)	DIO2	DIO2, TXDI2, D2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIMT1 geni (Tüm Gen Dizi Analizi)	DIMT1 gene (Sequence Analysis-All Coding Exons)	DIMT1	DIMT1L, DIM1, HUSSY5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIDO1 geni (Tüm Gen Dizi Analizi)	DIDO1 gene (Sequence Analysis-All Coding Exons)	DIDO1	DATF1, DIO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIAPH2-AS1 geni (Tüm Gen Dizi Analizi)	DIAPH2-AS1 gene (Sequence Analysis-All Coding Exons)	DIAPH2-AS1	EPAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX9 geni (Tüm Gen Dizi Analizi)	DHX9 gene (Sequence Analysis-All Coding Exons)	DHX9	DHX9, DDX9, NDHII	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX8 geni (Tüm Gen Dizi Analizi)	DHX8 gene (Sequence Analysis-All Coding Exons)	DHX8	DHX8, DDX8, HRH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX58 geni (Tüm Gen Dizi Analizi)	DHX58 gene (Sequence Analysis-All Coding Exons)	DHX58	DHX58, LGP2, D111GP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX40 geni (Tüm Gen Dizi Analizi)	DHX40 gene (Sequence Analysis-All Coding Exons)	DHX40	DHX40, DDX40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX38 geni (Tüm Gen Dizi Analizi)	DHX38 gene (Sequence Analysis-All Coding Exons)	DHX38	DHX38, DDX38, PRP16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX37 geni (Tüm Gen Dizi Analizi)	DHX37 gene (Sequence Analysis-All Coding Exons)	DHX37	DHX37, KIAA1517	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX36 geni (Tüm Gen Dizi Analizi)	DHX36 gene (Sequence Analysis-All Coding Exons)	DHX36	DHX36, G4R1, RHAU, KIAA1488	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX34 geni (Tüm Gen Dizi Analizi)	DHX34 gene (Sequence Analysis-All Coding Exons)	DHX34	DHX34, KIAA0134	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX33 geni (Tüm Gen Dizi Analizi)	DHX33 gene (Sequence Analysis-All Coding Exons)	DHX33	DHX33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX32 geni (Tüm Gen Dizi Analizi)	DHX32 gene (Sequence Analysis-All Coding Exons)	DHX32	DHX32, DDX32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX30 geni (Tüm Gen Dizi Analizi)	DHX30 gene (Sequence Analysis-All Coding Exons)	DHX30	DHX30, DDX30, RETCOR, KIAA0990	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX29 geni (Tüm Gen Dizi Analizi)	DHX29 gene (Sequence Analysis-All Coding Exons)	DHX29	DHX29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX16 geni (Tüm Gen Dizi Analizi)	DHX16 gene (Sequence Analysis-All Coding Exons)	DHX16	DHX16, DDX16, DBP2, PRP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHX15 geni (Tüm Gen Dizi Analizi)	DHX15 gene (Sequence Analysis-All Coding Exons)	DHX15	DHX15, DDX15, DBP1, HRH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS9 geni (Tüm Gen Dizi Analizi)	DHRS9 gene (Sequence Analysis-All Coding Exons)	DHRS9	DHRS9, RDHTBE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS7C geni (Tüm Gen Dizi Analizi)	DHRS7C gene (Sequence Analysis-All Coding Exons)	DHRS7C	DHRS7C, SDR32C2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS7B geni (Tüm Gen Dizi Analizi)	DHRS7B gene (Sequence Analysis-All Coding Exons)	DHRS7B	DHRS7B, SDR32C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS7 geni (Tüm Gen Dizi Analizi)	DHRS7 gene (Sequence Analysis-All Coding Exons)	DHRS7	DHRS7, RETSDR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS4L2 geni (Tüm Gen Dizi Analizi)	DHRS4L2 gene (Sequence Analysis-All Coding Exons)	DHRS4L2	DHRS4L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS4L1 geni (Tüm Gen Dizi Analizi)	DHRS4L1 gene (Sequence Analysis-All Coding Exons)	DHRS4L1	DHRS4L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS4-AS1 geni (Tüm Gen Dizi Analizi)	DHRS4-AS1 gene (Sequence Analysis-All Coding Exons)	DHRS4-AS1	DHRS4AS1, DHRS4AS1, AS1DHRS4, C14orf67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS4 geni (Tüm Gen Dizi Analizi)	DHRS4 gene (Sequence Analysis-All Coding Exons)	DHRS4	DHRS4, NRDR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS3 geni (Tüm Gen Dizi Analizi)	DHRS3 gene (Sequence Analysis-All Coding Exons)	DHRS3	DHRS3, RETSDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

DHRS2 geni (Tüm Gen Dizi Analizi)	DHRS2 gene (Sequence Analysis-All Coding Exons)	DHRS2	DHRS2, HEP27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS13 geni (Tüm Gen Dizi Analizi)	DHRS13 gene (Sequence Analysis-All Coding Exons)	DHRS13	DHRS13, SDR7C5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS12 geni (Tüm Gen Dizi Analizi)	DHRS12 gene (Sequence Analysis-All Coding Exons)	DHRS12	DHRS12, SDR40C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS11 geni (Tüm Gen Dizi Analizi)	DHRS11 gene (Sequence Analysis-All Coding Exons)	DHRS11	DHRS11, SDR24C1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHRS1 geni (Tüm Gen Dizi Analizi)	DHRS1 gene (Sequence Analysis-All Coding Exons)	DHRS1	DHRS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHPS geni (Tüm Gen Dizi Analizi)	DHPS gene (Sequence Analysis-All Coding Exons)	DHPS	DHPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHFR2 geni (Tüm Gen Dizi Analizi)	DHFR2 gene (Sequence Analysis-All Coding Exons)	DHFR2	DHFRL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKZ geni (Tüm Gen Dizi Analizi)	DGKZ gene (Sequence Analysis-All Coding Exons)	DGKZ	DGKZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKQ geni (Tüm Gen Dizi Analizi)	DGKQ gene (Sequence Analysis-All Coding Exons)	DGKQ	DGKQ, DAGK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKK geni (Tüm Gen Dizi Analizi)	DGKK gene (Sequence Analysis-All Coding Exons)	DGKK	DGKK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKI geni (Tüm Gen Dizi Analizi)	DGKI gene (Sequence Analysis-All Coding Exons)	DGKI	DGKI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKH geni (Tüm Gen Dizi Analizi)	DGKH gene (Sequence Analysis-All Coding Exons)	DGKH	DGKH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKG geni (Tüm Gen Dizi Analizi)	DGKG gene (Sequence Analysis-All Coding Exons)	DGKG	DGKG, DAGK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKD geni (Tüm Gen Dizi Analizi)	DGKD gene (Sequence Analysis-All Coding Exons)	DGKD	DGKD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKB geni (Tüm Gen Dizi Analizi)	DGKB gene (Sequence Analysis-All Coding Exons)	DGKB	DGKB, DGK, KIAA0718	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGKA geni (Tüm Gen Dizi Analizi)	DGKA gene (Sequence Analysis-All Coding Exons)	DGKA	DGKA, DAGK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGCR8 geni (Tüm Gen Dizi Analizi)	DGCR8 gene (Sequence Analysis-All Coding Exons)	DGCR8	DGCR8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGCR6L geni (Tüm Gen Dizi Analizi)	DGCR6L gene (Sequence Analysis-All Coding Exons)	DGCR6L	DGCR6L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGCR6 geni (Tüm Gen Dizi Analizi)	DGCR6 gene (Sequence Analysis-All Coding Exons)	DGCR6	DGCR6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGCR2 geni (Tüm Gen Dizi Analizi)	DGCR2 gene (Sequence Analysis-All Coding Exons)	DGCR2	DGCR2, IDD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGCR14 geni (Tüm Gen Dizi Analizi)	DGCR14 gene (Sequence Analysis-All Coding Exons)	DGCR14	DGSI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGAT2L6 geni (Tüm Gen Dizi Analizi)	DGAT2L6 gene (Sequence Analysis-All Coding Exons)	DGAT2L6	DGAT2L6, DC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DGAT2 geni (Tüm Gen Dizi Analizi)	DGAT2 gene (Sequence Analysis-All Coding Exons)	DGAT2	DGAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DFFB geni (Tüm Gen Dizi Analizi)	DFFB gene (Sequence Analysis-All Coding Exons)	DFFB	DFFB, CAD, DFF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DFFA geni (Tüm Gen Dizi Analizi)	DFFA gene (Sequence Analysis-All Coding Exons)	DFFA	DFFA, DFF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEUP1 geni (Tüm Gen Dizi Analizi)	DEUP1 gene (Sequence Analysis-All Coding Exons)	DEUP1	DEUP1, CCDC67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DET1 geni (Tüm Gen Dizi Analizi)	DET1 gene (Sequence Analysis-All Coding Exons)	DET1	DET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DESI2 geni (Tüm Gen Dizi Analizi)	DESI2 gene (Sequence Analysis-All Coding Exons)	DESI2	DESI2, PPPDE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DESI1 geni (Tüm Gen Dizi Analizi)	DESI1 gene (Sequence Analysis-All Coding Exons)	DESI1	DESI1, PPPDE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DERL3 geni (Tüm Gen Dizi Analizi)	DERL3 gene (Sequence Analysis-All Coding Exons)	DERL3	DERL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DERL2 geni (Tüm Gen Dizi Analizi)	DERL2 gene (Sequence Analysis-All Coding Exons)	DERL2	DERL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DERL1 geni (Tüm Gen Dizi Analizi)	DERL1 gene (Sequence Analysis-All Coding Exons)	DERL1	DERL1, DER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEPTOR geni (Tüm Gen Dizi Analizi)	DEPTOR gene (Sequence Analysis-All Coding Exons)	DEPTOR	DEPDC6, DEPTOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEPDC7 geni (Tüm Gen Dizi Analizi)	DEPDC7 gene (Sequence Analysis-All Coding Exons)	DEPDC7	DEPDC7, TR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEPDC1B geni (Tüm Gen Dizi Analizi)	DEPDC1B gene (Sequence Analysis-All Coding Exons)	DEPDC1B	DEPDC1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEPDC1 geni (Tüm Gen Dizi Analizi)	DEPDC1 gene (Sequence Analysis-All Coding Exons)	DEPDC1	DEPDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENR geni (Tüm Gen Dizi Analizi)	DENR gene (Sequence Analysis-All Coding Exons)	DENR	DENR, DRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND5B geni (Tüm Gen Dizi Analizi)	DENND5B gene (Sequence Analysis-All Coding Exons)	DENND5B	DENND5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND4A geni (Tüm Gen Dizi Analizi)	DENND4A gene (Sequence Analysis-All Coding Exons)	DENND4A	DENND4A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND2D geni (Tüm Gen Dizi Analizi)	DENND2D gene (Sequence Analysis-All Coding Exons)	DENND2D	DENND2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND1C geni (Tüm Gen Dizi Analizi)	DENND1C gene (Sequence Analysis-All Coding Exons)	DENND1C	DENND1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND1B geni (Tüm Gen Dizi Analizi)	DENND1B gene (Sequence Analysis-All Coding Exons)	DENND1B	DENND1B, FAM31B, C1orf18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DENND1A geni (Tüm Gen Dizi Analizi)	DENND1A gene (Sequence Analysis-All Coding Exons)	DENND1A	DENND1A, KIAA1608	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEGS2 geni (Tüm Gen Dizi Analizi)	DEGS2 gene (Sequence Analysis-All Coding Exons)	DEGS2	DEGS2, DES2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEGS1 geni (Tüm Gen Dizi Analizi)	DEGS1 gene (Sequence Analysis-All Coding Exons)	DEGS1	DEGS1, DES1, MLD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB4A geni (Tüm Gen Dizi Analizi)	DEFB4A gene (Sequence Analysis-All Coding Exons)	DEFB4A	DEFB4A, DEFB4, DEFB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB123 geni (Tüm Gen Dizi Analizi)	DEFB123 gene (Sequence Analysis-All Coding Exons)	DEFB123	DEFB123, DEFB23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB122 geni (Tüm Gen Dizi Analizi)	DEFB122 gene (Sequence Analysis-All Coding Exons)	DEFB122	DEFB122, DEFB22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB121 geni (Tüm Gen Dizi Analizi)	DEFB121 gene (Sequence Analysis-All Coding Exons)	DEFB121	DEFB121, DEFB21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB119 geni (Tüm Gen Dizi Analizi)	DEFB119 gene (Sequence Analysis-All Coding Exons)	DEFB119	DEFB119, DEFB120, DEFB20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB118 geni (Tüm Gen Dizi Analizi)	DEFB118 gene (Sequence Analysis-All Coding Exons)	DEFB118	DEFB118, C20orf63, ESC42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB114 geni (Tüm Gen Dizi Analizi)	DEFB114 gene (Sequence Analysis-All Coding Exons)	DEFB114	DEFB114, DEFB14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB103B geni (Tüm Gen Dizi Analizi)	DEFB103B gene (Sequence Analysis-All Coding Exons)	DEFB103B	DEFB103A, DEFB3, HBD3, HBP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY75 geni (Tüm Gen Dizi Analizi)	LY75 gene (Sequence Analysis-All Coding Exons)	LY75		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFB1 geni (Tüm Gen Dizi Analizi)	DEFB1 gene (Sequence Analysis-All Coding Exons)	DEFB1	DEFB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFA6 geni (Tüm Gen Dizi Analizi)	DEFA6 gene (Sequence Analysis-All Coding Exons)	DEFA6	DEFA6, DEF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G6F geni (Tüm Gen Dizi Analizi)	LY6G6F gene (Sequence Analysis-All Coding Exons)	LY6G6F		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LY6G6E geni (Tüm Gen Dizi Analizi)	LY6G6E gene (Sequence Analysis-All Coding Exons)	LY6G6E	LY6G6E, G6E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFA5 geni (Tüm Gen Dizi Analizi)	DEFA5 gene (Sequence Analysis-All Coding Exons)	DEFA5	DEFA5, DEF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFA4 geni (Tüm Gen Dizi Analizi)	DEFA4 gene (Sequence Analysis-All Coding Exons)	DEFA4	DEFA4, DEF4, HP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFA1 geni (Tüm Gen Dizi Analizi)	DEFA1 gene (Sequence Analysis-All Coding Exons)	DEFA1	DEFA1, DEF1, MRS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEF6 geni (Tüm Gen Dizi Analizi)	DEF6 gene (Sequence Analysis-All Coding Exons)	DEF6	DEF6, IBP, SLAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEDD2 geni (Tüm Gen Dizi Analizi)	DEDD2 gene (Sequence Analysis-All Coding Exons)	DEDD2	DEDD2, FLAME3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEDD geni (Tüm Gen Dizi Analizi)	DEDD gene (Sequence Analysis-All Coding Exons)	DEDD	DEDD, DEFT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DECR2 geni (Tüm Gen Dizi Analizi)	DECR2 gene (Sequence Analysis-All Coding Exons)	DECR2	DECR2, PDCR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DECR1 geni (Tüm Gen Dizi Analizi)	DECR1 gene (Sequence Analysis-All Coding Exons)	DECR1	DECR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX60L geni (Tüm Gen Dizi Analizi)	DDX60L gene (Sequence Analysis-All Coding Exons)	DDX60L	DDX60L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX60 geni (Tüm Gen Dizi Analizi)	DDX60 gene (Sequence Analysis-All Coding Exons)	DDX60	DDX60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX6 geni (Tüm Gen Dizi Analizi)	DDX6 gene (Sequence Analysis-All Coding Exons)	DDX6	DDX6, HLR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX56 geni (Tüm Gen Dizi Analizi)	DDX56 gene (Sequence Analysis-All Coding Exons)	DDX56	DDX56, NOH61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX54 geni (Tüm Gen Dizi Analizi)	DDX54 gene (Sequence Analysis-All Coding Exons)	DDX54	DDX54, DP97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX52 geni (Tüm Gen Dizi Analizi)	DDX52 gene (Sequence Analysis-All Coding Exons)	DDX52	DDX52, ROK1, HUSSY19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX50 geni (Tüm Gen Dizi Analizi)	DDX50 gene (Sequence Analysis-All Coding Exons)	DDX50	DDX50, GUB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX5 geni (Tüm Gen Dizi Analizi)	DDX5 gene (Sequence Analysis-All Coding Exons)	DDX5	DDX5, HLR1, G17P1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX47 geni (Tüm Gen Dizi Analizi)	DDX47 gene (Sequence Analysis-All Coding Exons)	DDX47	DDX47	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX43 geni (Tüm Gen Dizi Analizi)	DDX43 gene (Sequence Analysis-All Coding Exons)	DDX43	DDX43, HAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX42 geni (Tüm Gen Dizi Analizi)	DDX42 gene (Sequence Analysis-All Coding Exons)	DDX42	DDX42, RHELP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX4 geni (Tüm Gen Dizi Analizi)	DDX4 gene (Sequence Analysis-All Coding Exons)	DDX4	DDX4, VASA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX3Y geni (Tüm Gen Dizi Analizi)	DDX3Y gene (Sequence Analysis-All Coding Exons)	DDX3Y	DDX3Y, DBY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX39B geni (Tüm Gen Dizi Analizi)	DDX39B gene (Sequence Analysis-All Coding Exons)	DDX39B	DDX39B, BAT1, D6S81E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX31 geni (Tüm Gen Dizi Analizi)	DDX31 gene (Sequence Analysis-All Coding Exons)	DDX31	DDX31, PPP1R25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX28 geni (Tüm Gen Dizi Analizi)	DDX28 gene (Sequence Analysis-All Coding Exons)	DDX28	DDX28, MDDX28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX27 geni (Tüm Gen Dizi Analizi)	DDX27 gene (Sequence Analysis-All Coding Exons)	DDX27	DDX27, RHLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX25 geni (Tüm Gen Dizi Analizi)	DDX25 gene (Sequence Analysis-All Coding Exons)	DDX25	GRTH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX24 geni (Tüm Gen Dizi Analizi)	DDX24 gene (Sequence Analysis-All Coding Exons)	DDX24	DDX24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX23 geni (Tüm Gen Dizi Analizi)	DDX23 gene (Sequence Analysis-All Coding Exons)	DDX23	DDX23, PRP28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX21 geni (Tüm Gen Dizi Analizi)	DDX21 gene (Sequence Analysis-All Coding Exons)	DDX21	DDX21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX20 geni (Tüm Gen Dizi Analizi)	DDX20 gene (Sequence Analysis-All Coding Exons)	DDX20	DDX20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX19B geni (Tüm Gen Dizi Analizi)	DDX19B gene (Sequence Analysis-All Coding Exons)	DDX19B	DDX19, DBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX18 geni (Tüm Gen Dizi Analizi)	DDX18 gene (Sequence Analysis-All Coding Exons)	DDX18	DDX18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX17 geni (Tüm Gen Dizi Analizi)	DDX17 gene (Sequence Analysis-All Coding Exons)	DDX17	DDX17, RH70, P72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX10 geni (Tüm Gen Dizi Analizi)	DDX10 gene (Sequence Analysis-All Coding Exons)	DDX10	DDX10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDX1 geni (Tüm Gen Dizi Analizi)	DDX1 gene (Sequence Analysis-All Coding Exons)	DDX1	DDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

DDT geni (Tüm Gen Dizi Analizi)	DDT gene (Sequence Analysis-All Coding Exons)	DDT	DDT	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDRGK1 geni (Tüm Gen Dizi Analizi)	DDRGK1 gene (Sequence Analysis-All Coding Exons)	DDRGK1	DDRGK1, UFBP1, C20orf116	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDR1 geni (Tüm Gen Dizi Analizi)	DDR1 gene (Sequence Analysis-All Coding Exons)	DDR1	NEP, EDDR1, NTRK4, TRKE	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDO geni (Tüm Gen Dizi Analizi)	DDO gene (Sequence Analysis-All Coding Exons)	DDO	DDO, DASOX	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDN geni (Tüm Gen Dizi Analizi)	DDN gene (Sequence Analysis-All Coding Exons)	DDN	DDN, KIAA0749	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDIT4L geni (Tüm Gen Dizi Analizi)	DDIT4L gene (Sequence Analysis-All Coding Exons)	DDIT4L	DDIT4L, REDD2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDIT4 geni (Tüm Gen Dizi Analizi)	DDIT4 gene (Sequence Analysis-All Coding Exons)	DDIT4	DDIT4, REDD1, RTP801	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDB1 geni (Tüm Gen Dizi Analizi)	DDB1 gene (Sequence Analysis-All Coding Exons)	DDB1	DDB1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDAH2 geni (Tüm Gen Dizi Analizi)	DDAH2 gene (Sequence Analysis-All Coding Exons)	DDAH2	DDAH2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DDAH1 geni (Tüm Gen Dizi Analizi)	DDAH1 gene (Sequence Analysis-All Coding Exons)	DDAH1	DDAH1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCUN1D5 geni (Tüm Gen Dizi Analizi)	DCUN1D5 gene (Sequence Analysis-All Coding Exons)	DCUN1D5	DCUN1D5, SCCRO5	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCUN1D4 geni (Tüm Gen Dizi Analizi)	DCUN1D4 gene (Sequence Analysis-All Coding Exons)	DCUN1D4	DCUN1D4, KIAA0276	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCUN1D3 geni (Tüm Gen Dizi Analizi)	DCUN1D3 gene (Sequence Analysis-All Coding Exons)	DCUN1D3	DCUN1D3, SCCRO3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCUN1D1 geni (Tüm Gen Dizi Analizi)	DCUN1D1 gene (Sequence Analysis-All Coding Exons)	DCUN1D1	DCUN1D1, RP42	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTPP1 geni (Tüm Gen Dizi Analizi)	DCTPP1 gene (Sequence Analysis-All Coding Exons)	DCTPP1	DCTPP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTN6 geni (Tüm Gen Dizi Analizi)	DCTN6 gene (Sequence Analysis-All Coding Exons)	DCTN6	DCTN6, WS3, P27	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTN5 geni (Tüm Gen Dizi Analizi)	DCTN5 gene (Sequence Analysis-All Coding Exons)	DCTN5	DCTN5, p25	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTN4 geni (Tüm Gen Dizi Analizi)	DCTN4 gene (Sequence Analysis-All Coding Exons)	DCTN4	DCTN4, P62, DYNI4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTN3 geni (Tüm Gen Dizi Analizi)	DCTN3 gene (Sequence Analysis-All Coding Exons)	DCTN3	DCTN3, DCTN22	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTN2 geni (Tüm Gen Dizi Analizi)	DCTN2 gene (Sequence Analysis-All Coding Exons)	DCTN2	DCTN2, DCTN50	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCTD geni (Tüm Gen Dizi Analizi)	DCTD gene (Sequence Analysis-All Coding Exons)	DCTD	DCTD	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCT geni (Tüm Gen Dizi Analizi)	DCT gene (Sequence Analysis-All Coding Exons)	DCT	DCT, TYRP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCSTAMP geni (Tüm Gen Dizi Analizi)	DCSTAMP gene (Sequence Analysis-All Coding Exons)	DCSTAMP	TM7SF4, DCSTAMP, FIND	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCP2 geni (Tüm Gen Dizi Analizi)	DCP2 gene (Sequence Analysis-All Coding Exons)	DCP2	DCP2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCP1B geni (Tüm Gen Dizi Analizi)	DCP1B gene (Sequence Analysis-All Coding Exons)	DCP1B	DCP1B, DCP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCP1A geni (Tüm Gen Dizi Analizi)	DCP1A gene (Sequence Analysis-All Coding Exons)	DCP1A	DCP1A, SMIF	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCLRE1B geni (Tüm Gen Dizi Analizi)	DCLRE1B gene (Sequence Analysis-All Coding Exons)	DCLRE1B	DCLRE1B, SNM1B, APOLLO	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCLRE1A geni (Tüm Gen Dizi Analizi)	DCLRE1A gene (Sequence Analysis-All Coding Exons)	DCLRE1A	DCLRE1A, SNM1, SNM1A, KIAA0086	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCLK3 geni (Tüm Gen Dizi Analizi)	DCLK3 gene (Sequence Analysis-All Coding Exons)	DCLK3	DCLK3, DCK3, CLR	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCLK2 geni (Tüm Gen Dizi Analizi)	DCLK2 gene (Sequence Analysis-All Coding Exons)	DCLK2	DCLK2, DCK2, CLIK2, CL2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCLK1 geni (Tüm Gen Dizi Analizi)	DCLK1 gene (Sequence Analysis-All Coding Exons)	DCLK1	DCLK1, DCAMKL1, CLICK1, CL1, KIAA0369	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCK geni (Tüm Gen Dizi Analizi)	DCK gene (Sequence Analysis-All Coding Exons)	DCK	DCK	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCHS2 geni (Tüm Gen Dizi Analizi)	DCHS2 gene (Sequence Analysis-All Coding Exons)	DCHS2	DCHS2, CDHJ, PCDHJ	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCDC5 geni (Tüm Gen Dizi Analizi)	DCDC5 gene (Sequence Analysis-All Coding Exons)	DCDC5	DCDC5, KIAA1493	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCDC1 geni (Tüm Gen Dizi Analizi)	DCDC1 gene (Sequence Analysis-All Coding Exons)	DCDC1	DCDC1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCD geni (Tüm Gen Dizi Analizi)	DCD gene (Sequence Analysis-All Coding Exons)	DCD	DCD	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCBLD2 geni (Tüm Gen Dizi Analizi)	DCBLD2 gene (Sequence Analysis-All Coding Exons)	DCBLD2	DCBLD2, ESDN	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCANP1 geni (Tüm Gen Dizi Analizi)	DCANP1 gene (Sequence Analysis-All Coding Exons)	DCANP1	CSorf20, DCNP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF6 geni (Tüm Gen Dizi Analizi)	DCAF6 gene (Sequence Analysis-All Coding Exons)	DCAF6	DCAF6, IQWD1, NRIP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF5 geni (Tüm Gen Dizi Analizi)	DCAF5 gene (Sequence Analysis-All Coding Exons)	DCAF5	DCAF5, WDR22, BCRP2, 7B4S1461F	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF4 geni (Tüm Gen Dizi Analizi)	DCAF4 gene (Sequence Analysis-All Coding Exons)	DCAF4	DCAF4, WDR21, WDR21A	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF13 geni (Tüm Gen Dizi Analizi)	DCAF13 gene (Sequence Analysis-All Coding Exons)	DCAF13	DCAF13, WDSOF1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF11 geni (Tüm Gen Dizi Analizi)	DCAF11 gene (Sequence Analysis-All Coding Exons)	DCAF11	DCAF11, WDR23	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF1 geni (Tüm Gen Dizi Analizi)	DCAF1 gene (Sequence Analysis-All Coding Exons)	DCAF1	DCAF1, RIP, VPRBP, KIAA0800	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBR1 geni (Tüm Gen Dizi Analizi)	DBR1 gene (Sequence Analysis-All Coding Exons)	DBR1	DBR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBP geni (Tüm Gen Dizi Analizi)	DBP gene (Sequence Analysis-All Coding Exons)	DBP	DBP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBNL geni (Tüm Gen Dizi Analizi)	DBNL gene (Sequence Analysis-All Coding Exons)	DBNL	DBNL, HIP55	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBNDD2 geni (Tüm Gen Dizi Analizi)	DBNDD2 gene (Sequence Analysis-All Coding Exons)	DBNDD2	DBNDD2, HSMNP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBN1 geni (Tüm Gen Dizi Analizi)	DBN1 gene (Sequence Analysis-All Coding Exons)	DBN1	DBN1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBI geni (Tüm Gen Dizi Analizi)	DBI gene (Sequence Analysis-All Coding Exons)	DBI	DBI	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBF4B geni (Tüm Gen Dizi Analizi)	DBF4B gene (Sequence Analysis-All Coding Exons)	DBF4B	DBF4B, DRF1, ASKL1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBF4 geni (Tüm Gen Dizi Analizi)	DBF4 gene (Sequence Analysis-All Coding Exons)	DBF4	ASK, DBF4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRR1 geni (Tüm Gen Dizi Analizi)	LRR1 gene (Sequence Analysis-All Coding Exons)	LRR1		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DBET geni (Tüm Gen Dizi Analizi)	DBET gene (Sequence Analysis-All Coding Exons)	DBET	DBET	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAZ3 geni (Tüm Gen Dizi Analizi)	DAZ3 gene (Sequence Analysis-All Coding Exons)	DAZ3	DAZ3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAZ2 geni (Tüm Gen Dizi Analizi)	DAZ2 gene (Sequence Analysis-All Coding Exons)	DAZ2	DAZ2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAXX geni (Tüm Gen Dizi Analizi)	DAXX gene (Sequence Analysis-All Coding Exons)	DAXX	DAXX	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAPP1 geni (Tüm Gen Dizi Analizi)	DAPP1 gene (Sequence Analysis-All Coding Exons)	DAPP1	DAPP1, BAM32	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAPK3 geni (Tüm Gen Dizi Analizi)	DAPK3 gene (Sequence Analysis-All Coding Exons)	DAPK3	ZIPK	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAPK2 geni (Tüm Gen Dizi Analizi)	DAPK2 gene (Sequence Analysis-All Coding Exons)	DAPK2	DAPK2, DRP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAPK1 geni (Tüm Gen Dizi Analizi)	DAPK1 gene (Sequence Analysis-All Coding Exons)	DAPK1	DAPK1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAP3 geni (Tüm Gen Dizi Analizi)	DAP3 gene (Sequence Analysis-All Coding Exons)	DAP3	DAP3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAP geni (Tüm Gen Dizi Analizi)	DAP gene (Sequence Analysis-All Coding Exons)	DAP	DAP	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAOA-AS1 geni (Tüm Gen Dizi Analizi)	DAOA-AS1 gene (Sequence Analysis-All Coding Exons)	DAOA-AS1	DAOAAS, G30	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAND5 geni (Tüm Gen Dizi Analizi)	DAND5 gene (Sequence Analysis-All Coding Exons)	DAND5	DAND5, CER2, CERL2, DANTE, COCO	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DANCR geni (Tüm Gen Dizi Analizi)	DANCR gene (Sequence Analysis-All Coding Exons)	DANCR	DANCR, ANCR, KIAA0114	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAGLB geni (Tüm Gen Dizi Analizi)	DAGLB gene (Sequence Analysis-All Coding Exons)	DAGLB	DAGLB	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAGLA geni (Tüm Gen Dizi Analizi)	DAGLA gene (Sequence Analysis-All Coding Exons)	DAGLA	DAGLA, KIA00659	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAD1 geni (Tüm Gen Dizi Analizi)	DAD1 gene (Sequence Analysis-All Coding Exons)	DAD1	DAD1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACT3 geni (Tüm Gen Dizi Analizi)	DACT3 gene (Sequence Analysis-All Coding Exons)	DACT3	DACT3	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACT2 geni (Tüm Gen Dizi Analizi)	DACT2 gene (Sequence Analysis-All Coding Exons)	DACT2	DACT2, DPR2, DAPPER2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LRP1B geni (Tüm Gen Dizi Analizi)	LRP1B gene (Sequence Analysis-All Coding Exons)	LRP1B		Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACT1 geni (Tüm Gen Dizi Analizi)	DACT1 gene (Sequence Analysis-All Coding Exons)	DACT1	DACT1, DPR1, DAPPER1, FRODO	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACT1 geni (Tüm Gen Dizi Analizi)	DACT1 gene (Sequence Analysis-All Coding Exons)	DACT1	DACT1, DPR1, DAPPER1, FRODO	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACH2 geni (Tüm Gen Dizi Analizi)	DACH2 gene (Sequence Analysis-All Coding Exons)	DACH2	DACH2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DACH1 geni (Tüm Gen Dizi Analizi)	DACH1 gene (Sequence Analysis-All Coding Exons)	DACH1	DACH	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAB2IP geni (Tüm Gen Dizi Analizi)	DAB2IP gene (Sequence Analysis-All Coding Exons)	DAB2IP	DAB2IP, AIP1, KIAA1743	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAB2 geni (Tüm Gen Dizi Analizi)	DAB2 gene (Sequence Analysis-All Coding Exons)	DAB2	DAB2, DOC2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAB1 geni (Tüm Gen Dizi Analizi)	DAB1 gene (Sequence Analysis-All Coding Exons)	DAB1	DAB1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAAM2 geni (Tüm Gen Dizi Analizi)	DAAM2 gene (Sequence Analysis-All Coding Exons)	DAAM2	DAAM2, KIAA0381	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAAM1 geni (Tüm Gen Dizi Analizi)	DAAM1 gene (Sequence Analysis-All Coding Exons)	DAAM1	DAAM1, KIAA0666	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYYR1-AS1 geni (Tüm Gen Dizi Analizi)	CYYR1-AS1 gene (Sequence Analysis-All Coding Exons)	CYYR1-AS1	CYYR1AS1, CYYR1-AS1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYYR1 geni (Tüm Gen Dizi Analizi)	CYYR1 gene (Sequence Analysis-All Coding Exons)	CYYR1	CYYR1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTL1 geni (Tüm Gen Dizi Analizi)	CYTL1 gene (Sequence Analysis-All Coding Exons)	CYTL1	CYTL1, C17	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTIP geni (Tüm Gen Dizi Analizi)	CYTIP gene (Sequence Analysis-All Coding Exons)	CYTIP	CYTIP, PSCDBP, HE	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTH4 geni (Tüm Gen Dizi Analizi)	CYTH4 gene (Sequence Analysis-All Coding Exons)	CYTH4	CYTH4, PSCD4, CYT4	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTH3 geni (Tüm Gen Dizi Analizi)	CYTH3 gene (Sequence Analysis-All Coding Exons)	CYTH3	CYTH3, PSCD3, ARNO3, GRP1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTH2 geni (Tüm Gen Dizi Analizi)	CYTH2 gene (Sequence Analysis-All Coding Exons)	CYTH2	CYTH2, PSCD2, ARNO, PSCD9	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYTH1 geni (Tüm Gen Dizi Analizi)	CYTH1 gene (Sequence Analysis-All Coding Exons)	CYTH1	CYTH1, D17S811E, SEC7	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYSLTR2 geni (Tüm Gen Dizi Analizi)	CYSLTR2 gene (Sequence Analysis-All Coding Exons)	CYSLTR2	CYSLTR2, CYSLT2	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYSLTR1 geni (Tüm Gen Dizi Analizi)	CYSLTR1 gene (Sequence Analysis-All Coding Exons)	CYSLTR1	CYSLTR1, CYSLT1	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYR61 geni (Tüm Gen Dizi Analizi)	CYR61 gene (Sequence Analysis-All Coding Exons)	CYR61	CYR61, IGFBP10	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP8B1 geni (Tüm Gen Dizi Analizi)	CYP8B1 gene (Sequence Analysis-All Coding Exons)	CYP8B1	CYP8B1, CYP12	Tüm Gen Dizi Analizi Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CYP7A1 geni (Tüm Gen Dizi Analizi)	CYP7A1 gene (Sequence Analysis-All Coding Exons)	CYP7A1	CYP7A1, CYP7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP51A1 geni (Tüm Gen Dizi Analizi)	CYP51A1 gene (Sequence Analysis-All Coding Exons)	CYP51A1	CYP51A1, CYP51	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4X1 geni (Tüm Gen Dizi Analizi)	CYP4X1 gene (Sequence Analysis-All Coding Exons)	CYP4X1	CYP4X1, CYPIVX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4F8 geni (Tüm Gen Dizi Analizi)	CYP4F8 gene (Sequence Analysis-All Coding Exons)	CYP4F8	CYP4F8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4F3 geni (Tüm Gen Dizi Analizi)	CYP4F3 gene (Sequence Analysis-All Coding Exons)	CYP4F3	CYP4F3, LTB4H	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4F2 geni (Tüm Gen Dizi Analizi)	CYP4F2 gene (Sequence Analysis-All Coding Exons)	CYP4F2	CYP4F2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4F12 geni (Tüm Gen Dizi Analizi)	CYP4F12 gene (Sequence Analysis-All Coding Exons)	CYP4F12	CYP4F12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4F11 geni (Tüm Gen Dizi Analizi)	CYP4F11 gene (Sequence Analysis-All Coding Exons)	CYP4F11	CYP4F11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4B1 geni (Tüm Gen Dizi Analizi)	CYP4B1 gene (Sequence Analysis-All Coding Exons)	CYP4B1	CYP4B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4A22 geni (Tüm Gen Dizi Analizi)	CYP4A22 gene (Sequence Analysis-All Coding Exons)	CYP4A22	CYP4A22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LPGAT1 geni (Tüm Gen Dizi Analizi)	LPGAT1 gene (Sequence Analysis-All Coding Exons)	LPGAT1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP4A11 geni (Tüm Gen Dizi Analizi)	CYP4A11 gene (Sequence Analysis-All Coding Exons)	CYP4A11	CYP4A11	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP46A1 geni (Tüm Gen Dizi Analizi)	CYP46A1 gene (Sequence Analysis-All Coding Exons)	CYP46A1	CYP46A1, CYP46	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP3A7 geni (Tüm Gen Dizi Analizi)	CYP3A7 gene (Sequence Analysis-All Coding Exons)	CYP3A7	CYP3A7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP3A43 geni (Tüm Gen Dizi Analizi)	CYP3A43 gene (Sequence Analysis-All Coding Exons)	CYP3A43	CYP3A43	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP3A4 geni (Tüm Gen Dizi Analizi)	CYP3A4 gene (Sequence Analysis-All Coding Exons)	CYP3A4	CYP3A4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP39A1 geni (Tüm Gen Dizi Analizi)	CYP39A1 gene (Sequence Analysis-All Coding Exons)	CYP39A1	CYP39A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2W1 geni (Tüm Gen Dizi Analizi)	CYP2W1 gene (Sequence Analysis-All Coding Exons)	CYP2W1	CYP2W1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2S1 geni (Tüm Gen Dizi Analizi)	CYP2S1 gene (Sequence Analysis-All Coding Exons)	CYP2S1	CYP2S1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2J2 geni (Tüm Gen Dizi Analizi)	CYP2J2 gene (Sequence Analysis-All Coding Exons)	CYP2J2	CYP2J2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2F1 geni (Tüm Gen Dizi Analizi)	CYP2F1 gene (Sequence Analysis-All Coding Exons)	CYP2F1	CYP2F1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2E1 geni (Tüm Gen Dizi Analizi)	CYP2E1 gene (Sequence Analysis-All Coding Exons)	CYP2E1	CYP2E, CYP2E1, P450C2E	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2A7 geni (Tüm Gen Dizi Analizi)	CYP2A7 gene (Sequence Analysis-All Coding Exons)	CYP2A7	CYP2A7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2A13 geni (Tüm Gen Dizi Analizi)	CYP2A13 gene (Sequence Analysis-All Coding Exons)	CYP2A13	CYP2A13	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP26A1 geni (Tüm Gen Dizi Analizi)	CYP26A1 gene (Sequence Analysis-All Coding Exons)	CYP26A1	CYP26A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP1A1 geni (Tüm Gen Dizi Analizi)	CYP1A1 gene (Sequence Analysis-All Coding Exons)	CYP1A1	CYP1A1, CYP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYLC2 geni (Tüm Gen Dizi Analizi)	CYLC2 gene (Sequence Analysis-All Coding Exons)	CYLC2	CYLC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYLC1 geni (Tüm Gen Dizi Analizi)	CYLC1 gene (Sequence Analysis-All Coding Exons)	CYLC1	CYLC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYHR1 geni (Tüm Gen Dizi Analizi)	CYHR1 gene (Sequence Analysis-All Coding Exons)	CYHR1	CYHR1, KIAA0496	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYGB geni (Tüm Gen Dizi Analizi)	CYGB gene (Sequence Analysis-All Coding Exons)	CYGB	CYGB, HGB, STAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYFIP2 geni (Tüm Gen Dizi Analizi)	CYFIP2 gene (Sequence Analysis-All Coding Exons)	CYFIP2	CYFIP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYFIP1 geni (Tüm Gen Dizi Analizi)	CYFIP1 gene (Sequence Analysis-All Coding Exons)	CYFIP1	CYFIP1, KIAA0068	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB5R4 geni (Tüm Gen Dizi Analizi)	CYB5R4 gene (Sequence Analysis-All Coding Exons)	CYB5R4	CYB5R4, NCB5OR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB5R2 geni (Tüm Gen Dizi Analizi)	CYB5R2 gene (Sequence Analysis-All Coding Exons)	CYB5R2	CYB5R2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB5R1 geni (Tüm Gen Dizi Analizi)	CYB5R1 gene (Sequence Analysis-All Coding Exons)	CYB5R1	CYB5BR1, NQO3A2, BSR.1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB5B geni (Tüm Gen Dizi Analizi)	CYB5B gene (Sequence Analysis-All Coding Exons)	CYB5B	CYB5B, CYPB5M	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB561D2 geni (Tüm Gen Dizi Analizi)	CYB561D2 gene (Sequence Analysis-All Coding Exons)	CYB561D2	CYB561D2, 101F6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYB561 geni (Tüm Gen Dizi Analizi)	CYB561 gene (Sequence Analysis-All Coding Exons)	CYB561	CYB561	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXXC5 geni (Tüm Gen Dizi Analizi)	CXXC5 gene (Sequence Analysis-All Coding Exons)	CXXC5	CXXC5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXXC4 geni (Tüm Gen Dizi Analizi)	CXXC4 gene (Sequence Analysis-All Coding Exons)	CXXC4	CXXC4, IDAX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXorf40A geni (Tüm Gen Dizi Analizi)	CXorf40A gene (Sequence Analysis-All Coding Exons)	CXorf40A	CXorf40A, EOLA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXorf36 geni (Tüm Gen Dizi Analizi)	CXorf36 gene (Sequence Analysis-All Coding Exons)	CXorf36	CXorf36, DIA1R	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCR6 geni (Tüm Gen Dizi Analizi)	CXCR6 gene (Sequence Analysis-All Coding Exons)	CXCR6	CXCR6, STRL33, BONZO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCR5 geni (Tüm Gen Dizi Analizi)	CXCR5 gene (Sequence Analysis-All Coding Exons)	CXCR5	CXCR5, BLR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCR3 geni (Tüm Gen Dizi Analizi)	CXCR3 gene (Sequence Analysis-All Coding Exons)	CXCR3	CXCR3, GPR9, CD182	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCR2 geni (Tüm Gen Dizi Analizi)	CXCR2 gene (Sequence Analysis-All Coding Exons)	CXCR2	CXCR2, IL8RB, IL8R2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL9 geni (Tüm Gen Dizi Analizi)	CXCL9 gene (Sequence Analysis-All Coding Exons)	CXCL9	CXCL9, MIG, SCYB9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL8 geni (Tüm Gen Dizi Analizi)	CXCL8 gene (Sequence Analysis-All Coding Exons)	CXCL8	IL8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL6 geni (Tüm Gen Dizi Analizi)	CXCL6 gene (Sequence Analysis-All Coding Exons)	CXCL6	CXCL6, SCYB6, GCP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL5 geni (Tüm Gen Dizi Analizi)	CXCL5 gene (Sequence Analysis-All Coding Exons)	CXCL5	CXCL5, SCYB5, ENA78	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL3 geni (Tüm Gen Dizi Analizi)	CXCL3 gene (Sequence Analysis-All Coding Exons)	CXCL3	CXCL3, GRO3, MIP2B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL2 geni (Tüm Gen Dizi Analizi)	CXCL2 gene (Sequence Analysis-All Coding Exons)	CXCL2	CXCL2, GRO2, MIP2A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL17 geni (Tüm Gen Dizi Analizi)	CXCL17 gene (Sequence Analysis-All Coding Exons)	CXCL17	CXCL17, VCC1, DMC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL16 geni (Tüm Gen Dizi Analizi)	CXCL16 gene (Sequence Analysis-All Coding Exons)	CXCL16	CXCL16	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL14 geni (Tüm Gen Dizi Analizi)	CXCL14 gene (Sequence Analysis-All Coding Exons)	CXCL14	CXCL14, SCYB14, BRAK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL13 geni (Tüm Gen Dizi Analizi)	CXCL13 gene (Sequence Analysis-All Coding Exons)	CXCL13	CXCL13, SCYB13, BCA1, BLC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL11 geni (Tüm Gen Dizi Analizi)	CXCL11 gene (Sequence Analysis-All Coding Exons)	CXCL11	CXCL11, SCYB11, IP9, CXCL11_SCYB9B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL10 geni (Tüm Gen Dizi Analizi)	CXCL10 gene (Sequence Analysis-All Coding Exons)	CXCL10	CXCL10, INP10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXCL1 geni (Tüm Gen Dizi Analizi)	CXCL1 gene (Sequence Analysis-All Coding Exons)	CXCL1	CXCL1, GRO1, MGSA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXADR geni (Tüm Gen Dizi Analizi)	CXADR gene (Sequence Analysis-All Coding Exons)	CXADR	CXADR, CAR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CX3CL1 geni (Tüm Gen Dizi Analizi)	CX3CL1 gene (Sequence Analysis-All Coding Exons)	CX3CL1	CX3CL1, SCYD1, NTT, NTN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CWC27 geni (Tüm Gen Dizi Analizi)	CWC27 gene (Sequence Analysis-All Coding Exons)	CWC27	CWC27, SDCCAG10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CWC22 geni (Tüm Gen Dizi Analizi)	CWC22 gene (Sequence Analysis-All Coding Exons)	CWC22	CWC2, KIAA1604, NCM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUZD1 geni (Tüm Gen Dizi Analizi)	CUZD1 gene (Sequence Analysis-All Coding Exons)	CUZD1	CUZD1, ERG1, ITMAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUX1 geni (Tüm Gen Dizi Analizi)	CUX1 gene (Sequence Analysis-All Coding Exons)	CUX1	CUTL1, CDP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUTC geni (Tüm Gen Dizi Analizi)	CUTC gene (Sequence Analysis-All Coding Exons)	CUTC	CUTC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUTA geni (Tüm Gen Dizi Analizi)	CUTA gene (Sequence Analysis-All Coding Exons)	CUTA	CUTA, ACHAP, C6orf82	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUL9 geni (Tüm Gen Dizi Analizi)	CUL9 gene (Sequence Analysis-All Coding Exons)	CUL9	CUL9, PARC, KIAA0708	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUL5 geni (Tüm Gen Dizi Analizi)	CUL5 gene (Sequence Analysis-All Coding Exons)	CUL5	CUL5, VACM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUL4A geni (Tüm Gen Dizi Analizi)	CUL4A gene (Sequence Analysis-All Coding Exons)	CUL4A	CUL4A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUL2 geni (Tüm Gen Dizi Analizi)	CUL2 gene (Sequence Analysis-All Coding Exons)	CUL2	CUL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUL1 geni (Tüm Gen Dizi Analizi)	CUL1 gene (Sequence Analysis-All Coding Exons)	CUL1	CUL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUEDC2 geni (Tüm Gen Dizi Analizi)	CUEDC2 gene (Sequence Analysis-All Coding Exons)	CUEDC2	CUEDC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMAN2 geni (Tüm Gen Dizi Analizi)	LMAN2 gene (Sequence Analysis-All Coding Exons)	LMAN2		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LMAN1L geni (Tüm Gen Dizi Analizi)	LMAN1L gene (Sequence Analysis-All Coding Exons)	LMAN1L		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTXN1 geni (Tüm Gen Dizi Analizi)	CTXN1 gene (Sequence Analysis-All Coding Exons)	CTXN1	CTXN1, CTXN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTU1 geni (Tüm Gen Dizi Analizi)	CTU1 gene (Sequence Analysis-All Coding Exons)	CTU1	CTU1, ATPBD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTTNBP2NL geni (Tüm Gen Dizi Analizi)	CTTNBP2NL gene (Sequence Analysis-All Coding Exons)	CTTNBP2NL	CTTNBP2NL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTTNBP2 geni (Tüm Gen Dizi Analizi)	CTTNBP2 gene (Sequence Analysis-All Coding Exons)	CTTNBP2	CTTNBP2, CORTBP2, KIAA1758	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTTN geni (Tüm Gen Dizi Analizi)	CTTN gene (Sequence Analysis-All Coding Exons)	CTTN	CTTN, EMS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSZ geni (Tüm Gen Dizi Analizi)	CTSZ gene (Sequence Analysis-All Coding Exons)	CTSZ	CTSZ	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSW geni (Tüm Gen Dizi Analizi)	CTSW gene (Sequence Analysis-All Coding Exons)	CTSW	CTSW	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSV geni (Tüm Gen Dizi Analizi)	CTSV gene (Sequence Analysis-All Coding Exons)	CTSV	CTSL2, CTSV	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSS geni (Tüm Gen Dizi Analizi)	CTSS gene (Sequence Analysis-All Coding Exons)	CTSS	CTSS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSO geni (Tüm Gen Dizi Analizi)	CTSO gene (Sequence Analysis-All Coding Exons)	CTSO	CTSO	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSL geni (Tüm Gen Dizi Analizi)	CTSL gene (Sequence Analysis-All Coding Exons)	CTSL	CTSL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSH geni (Tüm Gen Dizi Analizi)	CTSH gene (Sequence Analysis-All Coding Exons)	CTSH	CTSH	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSG geni (Tüm Gen Dizi Analizi)	CTSG gene (Sequence Analysis-All Coding Exons)	CTSG	CTSG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSE geni (Tüm Gen Dizi Analizi)	CTSE gene (Sequence Analysis-All Coding Exons)	CTSE	CTSE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIPF geni (Tüm Gen Dizi Analizi)	LIPF gene (Sequence Analysis-All Coding Exons)	LIPF		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTSB geni (Tüm Gen Dizi Analizi)	CTSB gene (Sequence Analysis-All Coding Exons)	CTSB	CTSB, CPSB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTRL geni (Tüm Gen Dizi Analizi)	CTRL gene (Sequence Analysis-All Coding Exons)	CTRL	CTRL	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTRB1 geni (Tüm Gen Dizi Analizi)	CTRB1 gene (Sequence Analysis-All Coding Exons)	CTRB1	CTRB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CTR9 geni (Tüm Gen Dizi Analizi)	CTR9 gene (Sequence Analysis-All Coding Exons)	CTR9	SH2BP1, KIAA0155, p150	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTPS2 geni (Tüm Gen Dizi Analizi)	CTPS2 gene (Sequence Analysis-All Coding Exons)	CTPS2	CTPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNND2 geni (Tüm Gen Dizi Analizi)	CTNND2 gene (Sequence Analysis-All Coding Exons)	CTNND2	CTNND2, NPRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNND1 geni (Tüm Gen Dizi Analizi)	CTNND1 gene (Sequence Analysis-All Coding Exons)	CTNND1	CTNND1, CTNND	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNNBL1 geni (Tüm Gen Dizi Analizi)	CTNNBL1 gene (Sequence Analysis-All Coding Exons)	CTNNBL1	CTNNBL1, NAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNNBIP1 geni (Tüm Gen Dizi Analizi)	CTNNBIP1 gene (Sequence Analysis-All Coding Exons)	CTNNBIP1	CTNNBIP1, ICAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNNAL1 geni (Tüm Gen Dizi Analizi)	CTNNAL1 gene (Sequence Analysis-All Coding Exons)	CTNNAL1	CTNNAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTNNA2 geni (Tüm Gen Dizi Analizi)	CTNNA2 gene (Sequence Analysis-All Coding Exons)	CTNNA2	CTNNA2, CAPR, CTNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTIF geni (Tüm Gen Dizi Analizi)	CTIF gene (Sequence Analysis-All Coding Exons)	CTIF	KIAA0427, CTIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTGF geni (Tüm Gen Dizi Analizi)	CTGF gene (Sequence Analysis-All Coding Exons)	CTGF	CTGF, NOV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTGF geni (Tüm Gen Dizi Analizi)	CTGF gene (Sequence Analysis-All Coding Exons)	CTGF	CTGF, NOV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTF1 geni (Tüm Gen Dizi Analizi)	CTF1 gene (Sequence Analysis-All Coding Exons)	CTF1	CTF1, CT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTDSP1 geni (Tüm Gen Dizi Analizi)	CTDSP1 gene (Sequence Analysis-All Coding Exons)	CTDSP1	CTDSP1, NLIIF, SCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTDNEP1 geni (Tüm Gen Dizi Analizi)	CTDNEP1 gene (Sequence Analysis-All Coding Exons)	CTDNEP1	CTDNEP1, DULLARD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTCFL geni (Tüm Gen Dizi Analizi)	CTCFL gene (Sequence Analysis-All Coding Exons)	CTCFL	CTCFL, BORIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTBS geni (Tüm Gen Dizi Analizi)	CTBS gene (Sequence Analysis-All Coding Exons)	CTBS	CTBS, CTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTBP2 geni (Tüm Gen Dizi Analizi)	CTBP2 gene (Sequence Analysis-All Coding Exons)	CTBP2	CTBP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTBP1 geni (Tüm Gen Dizi Analizi)	CTBP1 gene (Sequence Analysis-All Coding Exons)	CTBP1	CTBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTAG2 geni (Tüm Gen Dizi Analizi)	CTAG2 gene (Sequence Analysis-All Coding Exons)	CTAG2	CTAG2, LAGE1, CAMEL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTAG1B geni (Tüm Gen Dizi Analizi)	CTAG1B gene (Sequence Analysis-All Coding Exons)	CTAG1B	CTAG1B, CTAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTAG1A geni (Tüm Gen Dizi Analizi)	CTAG1A gene (Sequence Analysis-All Coding Exons)	CTAG1A	CTAG1A, LAGE2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT83 geni (Tüm Gen Dizi Analizi)	CT83 gene (Sequence Analysis-All Coding Exons)	CT83	KKLC1, CXorf61	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIN7A geni (Tüm Gen Dizi Analizi)	LIN7A gene (Sequence Analysis-All Coding Exons)	LIN7A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47B1 geni (Tüm Gen Dizi Analizi)	CT47B1 gene (Sequence Analysis-All Coding Exons)	CT47B1	CT47B1, CT47A13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A9 geni (Tüm Gen Dizi Analizi)	CT47A9 gene (Sequence Analysis-All Coding Exons)	CT47A9	CT47A9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A8 geni (Tüm Gen Dizi Analizi)	CT47A8 gene (Sequence Analysis-All Coding Exons)	CT47A8	CT47A8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A7 geni (Tüm Gen Dizi Analizi)	CT47A7 gene (Sequence Analysis-All Coding Exons)	CT47A7	CT47A7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LIMS1 geni (Tüm Gen Dizi Analizi)	LIMS1 gene (Sequence Analysis-All Coding Exons)	LIMS1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A6 geni (Tüm Gen Dizi Analizi)	CT47A6 gene (Sequence Analysis-All Coding Exons)	CT47A6	CT47A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A5 geni (Tüm Gen Dizi Analizi)	CT47A5 gene (Sequence Analysis-All Coding Exons)	CT47A5	CT47A5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A4 geni (Tüm Gen Dizi Analizi)	CT47A4 gene (Sequence Analysis-All Coding Exons)	CT47A4	CT47A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A3 geni (Tüm Gen Dizi Analizi)	CT47A3 gene (Sequence Analysis-All Coding Exons)	CT47A3	CT47A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A2 geni (Tüm Gen Dizi Analizi)	CT47A2 gene (Sequence Analysis-All Coding Exons)	CT47A2	CT47A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A11 geni (Tüm Gen Dizi Analizi)	CT47A11 gene (Sequence Analysis-All Coding Exons)	CT47A11	CT47A11, LOC255313	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A10 geni (Tüm Gen Dizi Analizi)	CT47A10 gene (Sequence Analysis-All Coding Exons)	CT47A10	CT47A10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT47A1 geni (Tüm Gen Dizi Analizi)	CT47A1 gene (Sequence Analysis-All Coding Exons)	CT47A1	CT47A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT45A6 geni (Tüm Gen Dizi Analizi)	CT45A6 gene (Sequence Analysis-All Coding Exons)	CT45A6	CT45A6, CT45.6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT45A5 geni (Tüm Gen Dizi Analizi)	CT45A5 gene (Sequence Analysis-All Coding Exons)	CT45A5	CT45A5, CT45.5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT45A3 geni (Tüm Gen Dizi Analizi)	CT45A3 gene (Sequence Analysis-All Coding Exons)	CT45A3	CT45A3, CT45.3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT45A2 geni (Tüm Gen Dizi Analizi)	CT45A2 gene (Sequence Analysis-All Coding Exons)	CT45A2	CT45A2, CT45.2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CT45A1 geni (Tüm Gen Dizi Analizi)	CT45A1 gene (Sequence Analysis-All Coding Exons)	CT45A1	CT45A1, CT45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LILRA3 geni (Tüm Gen Dizi Analizi)	LILRA3 gene (Sequence Analysis-All Coding Exons)	LILRA3	LILRA3, LIR4, ILT6, HM43	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSTF3 geni (Tüm Gen Dizi Analizi)	CSTF3 gene (Sequence Analysis-All Coding Exons)	CSTF3	CSTF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSTF2T geni (Tüm Gen Dizi Analizi)	CSTF2T gene (Sequence Analysis-All Coding Exons)	CSTF2T	CSTF2T, KIAA0689	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSTF2 geni (Tüm Gen Dizi Analizi)	CSTF2 gene (Sequence Analysis-All Coding Exons)	CSTF2	CSTF64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSTF1 geni (Tüm Gen Dizi Analizi)	CSTF1 gene (Sequence Analysis-All Coding Exons)	CSTF1	CSTF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST9L geni (Tüm Gen Dizi Analizi)	CST9L gene (Sequence Analysis-All Coding Exons)	CST9L	CST9L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST9 geni (Tüm Gen Dizi Analizi)	CST9 gene (Sequence Analysis-All Coding Exons)	CST9	CST9, CLM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST7 geni (Tüm Gen Dizi Analizi)	CST7 gene (Sequence Analysis-All Coding Exons)	CST7	CST7, CMAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST6 geni (Tüm Gen Dizi Analizi)	CST6 gene (Sequence Analysis-All Coding Exons)	CST6	CST6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST5 geni (Tüm Gen Dizi Analizi)	CST5 gene (Sequence Analysis-All Coding Exons)	CST5	CST5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST4 geni (Tüm Gen Dizi Analizi)	CST4 gene (Sequence Analysis-All Coding Exons)	CST4	CST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST2 geni (Tüm Gen Dizi Analizi)	CST2 gene (Sequence Analysis-All Coding Exons)	CST2	CST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST11 geni (Tüm Gen Dizi Analizi)	CST11 gene (Sequence Analysis-All Coding Exons)	CST11	CST11, CST8L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST1 geni (Tüm Gen Dizi Analizi)	CST1 gene (Sequence Analysis-All Coding Exons)	CST1	CST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSRP2 geni (Tüm Gen Dizi Analizi)	CSRP2 gene (Sequence Analysis-All Coding Exons)	CSRP2	CSRP2, LMO5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSRP1 geni (Tüm Gen Dizi Analizi)	CSRP1 gene (Sequence Analysis-All Coding Exons)	CSRP1	CSRP1, CSRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSRNP1 geni (Tüm Gen Dizi Analizi)	CSRNP1 gene (Sequence Analysis-All Coding Exons)	CSRNP1	AXUD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSPG4P1Y geni (Tüm Gen Dizi Analizi)	CSPG4P1Y gene (Sequence Analysis-All Coding Exons)	CSPG4P1Y	CSPG4LY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSPG4 geni (Tüm Gen Dizi Analizi)	CSPG4 gene (Sequence Analysis-All Coding Exons)	CSPG4	CSPG4, MCSPG, MSK16, NG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK2B geni (Tüm Gen Dizi Analizi)	CSNK2B gene (Sequence Analysis-All Coding Exons)	CSNK2B	CSNK2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK2A2 geni (Tüm Gen Dizi Analizi)	CSNK2A2 gene (Sequence Analysis-All Coding Exons)	CSNK2A2	CSNK2A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK1G3 geni (Tüm Gen Dizi Analizi)	CSNK1G3 gene (Sequence Analysis-All Coding Exons)	CSNK1G3	CSNK1G3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK1G2 geni (Tüm Gen Dizi Analizi)	CSNK1G2 gene (Sequence Analysis-All Coding Exons)	CSNK1G2	CSNK1G2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK1G1 geni (Tüm Gen Dizi Analizi)	CSNK1G1 gene (Sequence Analysis-All Coding Exons)	CSNK1G1	CSNK1G1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK1E geni (Tüm Gen Dizi Analizi)	CSNK1E gene (Sequence Analysis-All Coding Exons)	CSNK1E	CSNK1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSNK1A1 geni (Tüm Gen Dizi Analizi)	CSNK1A1 gene (Sequence Analysis-All Coding Exons)	CSNK1A1	CSNK1A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSN3 geni (Tüm Gen Dizi Analizi)	CSN3 gene (Sequence Analysis-All Coding Exons)	CSN3	CSN3, CNS10, CSNK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSN2 geni (Tüm Gen Dizi Analizi)	CSN2 gene (Sequence Analysis-All Coding Exons)	CSN2	CSN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSN1S1 geni (Tüm Gen Dizi Analizi)	CSN1S1 gene (Sequence Analysis-All Coding Exons)	CSN1S1	CSN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSMD3 geni (Tüm Gen Dizi Analizi)	CSMD3 gene (Sequence Analysis-All Coding Exons)	CSMD3	CSMD3, KIAA1894	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LGSN geni (Tüm Gen Dizi Analizi)	LGSN gene (Sequence Analysis-All Coding Exons)	LGSN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSMD2 geni (Tüm Gen Dizi Analizi)	CSMD2 gene (Sequence Analysis-All Coding Exons)	CSMD2	CSMD2, KIAA1884	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSMD1 geni (Tüm Gen Dizi Analizi)	CSMD1 gene (Sequence Analysis-All Coding Exons)	CSMD1	CSMD1, KIAA1890	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSK geni (Tüm Gen Dizi Analizi)	CSK gene (Sequence Analysis-All Coding Exons)	CSK	CSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSHL1 geni (Tüm Gen Dizi Analizi)	CSHL1 gene (Sequence Analysis-All Coding Exons)	CSHL1	CSHL1, CSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSH2 geni (Tüm Gen Dizi Analizi)	CSH2 gene (Sequence Analysis-All Coding Exons)	CSH2	CSH2, CSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSGALNACT2 geni (Tüm Gen Dizi Analizi)	CSGALNACT2 gene (Sequence Analysis-All Coding Exons)	CSGALNACT2	CSGALNACT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSGALNACT1 geni (Tüm Gen Dizi Analizi)	CSGALNACT1 gene (Sequence Analysis-All Coding Exons)	CSGALNACT1	CSGALNACT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSF3 geni (Tüm Gen Dizi Analizi)	CSF3 gene (Sequence Analysis-All Coding Exons)	CSF3	CSF3, GCSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSF2RA geni (Tüm Gen Dizi Analizi)	CSF2RA gene (Sequence Analysis-All Coding Exons)	CSF2RA	CSF2RY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Pulmonary surfactant metabolism dysfunction 4 (SMDP4) [MIM:300770]: A rare lung disorder due to impaired surfactant homeostasis. It is characterized by alveolar filling with flocular material that stains positive using the periodic acid-Schiff method and is derived from surfactant phospholipids and protein components. Excessive lipoproteins accumulation in the alveoli results in severe respiratory distress. {ECO:0000269|PubMed:18955567, ECO:0000269|PubMed:18955570}. Note=The disease is caused by mutations affecting the gene represented in this entry..	EDTA Blood Sample
CSF2 geni (Tüm Gen Dizi Analizi)	CSF2 gene (Sequence Analysis-All Coding Exons)	CSF2	CSF2, GMCSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSF1 geni (Tüm Gen Dizi Analizi)	CSF1 gene (Sequence Analysis-All Coding Exons)	CSF1	CSF1, MCSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSE1L geni (Tüm Gen Dizi Analizi)	CSE1L gene (Sequence Analysis-All Coding Exons)	CSE1L	CSE1L, CAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSDE1 geni (Tüm Gen Dizi Analizi)	CSDE1 gene (Sequence Analysis-All Coding Exons)	CSDE1	CSDE1, D1S155E, UNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSAG1 geni (Tüm Gen Dizi Analizi)	CSAG1 gene (Sequence Analysis-All Coding Exons)	CSAG1	CSAG1, CSAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSAD geni (Tüm Gen Dizi Analizi)	CSAD gene (Sequence Analysis-All Coding Exons)	CSAD	CSAD, CSD, PCAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CS geni (Tüm Gen Dizi Analizi)	CS gene (Sequence Analysis-All Coding Exons)	CS	CS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CRYZL1 geni (Tüm Gen Dizi Analizi)	CRYZL1 gene (Sequence Analysis-All Coding Exons)	CRYZL1	CRYZL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRYZ geni (Tüm Gen Dizi Analizi)	CRYZ gene (Sequence Analysis-All Coding Exons)	CRYZ	CRYZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRYL1 geni (Tüm Gen Dizi Analizi)	CRYL1 gene (Sequence Analysis-All Coding Exons)	CRYL1	CRYL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRYGA geni (Tüm Gen Dizi Analizi)	CRYGA gene (Sequence Analysis-All Coding Exons)	CRYGA	CRYGA, CRYG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRY2 geni (Tüm Gen Dizi Analizi)	CRY2 gene (Sequence Analysis-All Coding Exons)	CRY2	CRY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRY1 geni (Tüm Gen Dizi Analizi)	CRY1 gene (Sequence Analysis-All Coding Exons)	CRY1	CRY1, PHLL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRTC3 geni (Tüm Gen Dizi Analizi)	CRTC3 gene (Sequence Analysis-All Coding Exons)	CRTC3	CRTC3, TORC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRTC2 geni (Tüm Gen Dizi Analizi)	CRTC2 gene (Sequence Analysis-All Coding Exons)	CRTC2	CRTC2, TORC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRTAM geni (Tüm Gen Dizi Analizi)	CRTAM gene (Sequence Analysis-All Coding Exons)	CRTAM	CRTAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRP geni (Tüm Gen Dizi Analizi)	CRP gene (Sequence Analysis-All Coding Exons)	CRP	CRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CROT geni (Tüm Gen Dizi Analizi)	CROT gene (Sequence Analysis-All Coding Exons)	CROT	CROT, COT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CROCC geni (Tüm Gen Dizi Analizi)	CROCC gene (Sequence Analysis-All Coding Exons)	CROCC	CROCC, ROOTLETIN, KIAA0445	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRNKL1 geni (Tüm Gen Dizi Analizi)	CRNKL1 gene (Sequence Analysis-All Coding Exons)	CRNKL1	CRNKL1, CRN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRMP1 geni (Tüm Gen Dizi Analizi)	CRMP1 gene (Sequence Analysis-All Coding Exons)	CRMP1	CRMP1, DPYSL1, DRP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRLS1 geni (Tüm Gen Dizi Analizi)	CRLS1 gene (Sequence Analysis-All Coding Exons)	CRLS1	CRLS1, GCD10, C20orf155	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRLF3 geni (Tüm Gen Dizi Analizi)	CRLF3 gene (Sequence Analysis-All Coding Exons)	CRLF3	CRLF3, CYTOR4, CRLM9, CSRSMER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRLF2 geni (Tüm Gen Dizi Analizi)	CRLF2 gene (Sequence Analysis-All Coding Exons)	CRLF2	CRLF2, CRL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRLF2 geni (Tüm Gen Dizi Analizi)	CRLF2 gene (Sequence Analysis-All Coding Exons)	CRLF2	CRLF2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRKL geni (Tüm Gen Dizi Analizi)	CRKL gene (Sequence Analysis-All Coding Exons)	CRKL	CRKL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRK geni (Tüm Gen Dizi Analizi)	CRK gene (Sequence Analysis-All Coding Exons)	CRK	CRK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRISPLD2 geni (Tüm Gen Dizi Analizi)	CRISPLD2 gene (Sequence Analysis-All Coding Exons)	CRISPLD2	CRISPLD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRISP2 geni (Tüm Gen Dizi Analizi)	CRISP2 gene (Sequence Analysis-All Coding Exons)	CRISP2	CRISP2, TPX1, TSP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRISP1 geni (Tüm Gen Dizi Analizi)	CRISP1 gene (Sequence Analysis-All Coding Exons)	CRISP1	CRISP1, AEGL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRIPAK geni (Tüm Gen Dizi Analizi)	CRIPAK gene (Sequence Analysis-All Coding Exons)	CRIPAK	CRIPAK, FLJ33443	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRIP2 geni (Tüm Gen Dizi Analizi)	CRIP2 gene (Sequence Analysis-All Coding Exons)	CRIP2	CRIP2, CRP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRIP1 geni (Tüm Gen Dizi Analizi)	CRIP1 gene (Sequence Analysis-All Coding Exons)	CRIP1	CRIP1, CRIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRIM1 geni (Tüm Gen Dizi Analizi)	CRIM1 gene (Sequence Analysis-All Coding Exons)	CRIM1	CRIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRHR2 geni (Tüm Gen Dizi Analizi)	CRHR2 gene (Sequence Analysis-All Coding Exons)	CRHR2	CRHR2, CRFR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRHR1 geni (Tüm Gen Dizi Analizi)	CRHR1 gene (Sequence Analysis-All Coding Exons)	CRHR1	CRHR1, CRHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRHBP geni (Tüm Gen Dizi Analizi)	CRHBP gene (Sequence Analysis-All Coding Exons)	CRHBP	CRHBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRH geni (Tüm Gen Dizi Analizi)	CRH gene (Sequence Analysis-All Coding Exons)	CRH	CRH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREM geni (Tüm Gen Dizi Analizi)	CREM gene (Sequence Analysis-All Coding Exons)	CREM	CREM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREBRF geni (Tüm Gen Dizi Analizi)	CREBRF gene (Sequence Analysis-All Coding Exons)	CREBRF	CREBRF, LRF, C5orf41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREBL2 geni (Tüm Gen Dizi Analizi)	CREBL2 gene (Sequence Analysis-All Coding Exons)	CREBL2	CREBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3L4 geni (Tüm Gen Dizi Analizi)	CREB3L4 gene (Sequence Analysis-All Coding Exons)	CREB3L4	CREB3L4, CREB4, AIBZIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3L3 geni (Tüm Gen Dizi Analizi)	CREB3L3 gene (Sequence Analysis-All Coding Exons)	CREB3L3	CREB3L3, CREBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3L2 geni (Tüm Gen Dizi Analizi)	CREB3L2 gene (Sequence Analysis-All Coding Exons)	CREB3L2	CREB3L2, BBF2H7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3L1 geni (Tüm Gen Dizi Analizi)	CREB3L1 gene (Sequence Analysis-All Coding Exons)	CREB3L1	CREB3L1, OASIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3L1 geni (Tüm Gen Dizi Analizi)	CREB3L1 gene (Sequence Analysis-All Coding Exons)	CREB3L1	CREB3L1, OASIS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREB3 geni (Tüm Gen Dizi Analizi)	CREB3 gene (Sequence Analysis-All Coding Exons)	CREB3	CREB3, LZIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRCP geni (Tüm Gen Dizi Analizi)	CRCP gene (Sequence Analysis-All Coding Exons)	CRCP	RCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRB3 geni (Tüm Gen Dizi Analizi)	CRB3 gene (Sequence Analysis-All Coding Exons)	CRB3	CRB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
LDAH geni (Tüm Gen Dizi Analizi)	LDAH gene (Sequence Analysis-All Coding Exons)	LDAH	LDAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRACR2B geni (Tüm Gen Dizi Analizi)	CRACR2B gene (Sequence Analysis-All Coding Exons)	CRACR2B	EFCAB4A, CRACR2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRACR2A geni (Tüm Gen Dizi Analizi)	CRACR2A gene (Sequence Analysis-All Coding Exons)	CRACR2A	EFCAB4B, CRACR2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRABP2 geni (Tüm Gen Dizi Analizi)	CRABP2 gene (Sequence Analysis-All Coding Exons)	CRABP2	CRABP2, RBP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRABP1 geni (Tüm Gen Dizi Analizi)	CRABP1 gene (Sequence Analysis-All Coding Exons)	CRABP1	CRABP1, RBP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CR1L geni (Tüm Gen Dizi Analizi)	CR1L gene (Sequence Analysis-All Coding Exons)	CR1L	CR1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPZ geni (Tüm Gen Dizi Analizi)	CPZ gene (Sequence Analysis-All Coding Exons)	CPZ	CPZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPXM2 geni (Tüm Gen Dizi Analizi)	CPXM2 gene (Sequence Analysis-All Coding Exons)	CPXM2	CPXM2, CPX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPVL geni (Tüm Gen Dizi Analizi)	CPVL gene (Sequence Analysis-All Coding Exons)	CPVL	CPVL, HVLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPTP geni (Tüm Gen Dizi Analizi)	CPTP gene (Sequence Analysis-All Coding Exons)	CPTP	GLTPD1, CPTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPT1B geni (Tüm Gen Dizi Analizi)	CPT1B gene (Sequence Analysis-All Coding Exons)	CPT1B	CPT1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPSF6 geni (Tüm Gen Dizi Analizi)	CPSF6 gene (Sequence Analysis-All Coding Exons)	CPSF6	CPSF6, CFIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPSF4 geni (Tüm Gen Dizi Analizi)	CPSF4 gene (Sequence Analysis-All Coding Exons)	CPSF4	CPSF4, CPSF30, NEB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPPED1 geni (Tüm Gen Dizi Analizi)	CPPED1 gene (Sequence Analysis-All Coding Exons)	CPPED1	CPPED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE7 geni (Tüm Gen Dizi Analizi)	CPNE7 gene (Sequence Analysis-All Coding Exons)	CPNE7	CPNE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE6 geni (Tüm Gen Dizi Analizi)	CPNE6 gene (Sequence Analysis-All Coding Exons)	CPNE6	CPNE6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE5 geni (Tüm Gen Dizi Analizi)	CPNE5 gene (Sequence Analysis-All Coding Exons)	CPNE5	CPNE5, CPN5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE4 geni (Tüm Gen Dizi Analizi)	CPNE4 gene (Sequence Analysis-All Coding Exons)	CPNE4	CPNE4, CPN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE3 geni (Tüm Gen Dizi Analizi)	CPNE3 gene (Sequence Analysis-All Coding Exons)	CPNE3	CPNE3, CPN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE2 geni (Tüm Gen Dizi Analizi)	CPNE2 gene (Sequence Analysis-All Coding Exons)	CPNE2	CPNE2, CPN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPNE1 geni (Tüm Gen Dizi Analizi)	CPNE1 gene (Sequence Analysis-All Coding Exons)	CPNE1	CPNE1, CPN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPN2 geni (Tüm Gen Dizi Analizi)	CPN2 gene (Sequence Analysis-All Coding Exons)	CPN2	CPN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPM geni (Tüm Gen Dizi Analizi)	CPM gene (Sequence Analysis-All Coding Exons)	CPM	CPM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPLX2 geni (Tüm Gen Dizi Analizi)	CPLX2 gene (Sequence Analysis-All Coding Exons)	CPLX2	CPLX2, CPX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPLX1 geni (Tüm Gen Dizi Analizi)	CPLX1 gene (Sequence Analysis-All Coding Exons)	CPLX1	CPLX1, CPX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPEB4 geni (Tüm Gen Dizi Analizi)	CPEB4 gene (Sequence Analysis-All Coding Exons)	CPEB4	CPEB4, KIAA1673	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPEB3 geni (Tüm Gen Dizi Analizi)	CPEB3 gene (Sequence Analysis-All Coding Exons)	CPEB3	CPEB3, KIAA0940	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPEB2 geni (Tüm Gen Dizi Analizi)	CPEB2 gene (Sequence Analysis-All Coding Exons)	CPEB2	CPEB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPEB1 geni (Tüm Gen Dizi Analizi)	CPEB1 gene (Sequence Analysis-All Coding Exons)	CPEB1	CPEB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPE geni (Tüm Gen Dizi Analizi)	CPE gene (Sequence Analysis-All Coding Exons)	CPE	CPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPD geni (Tüm Gen Dizi Analizi)	CPD gene (Sequence Analysis-All Coding Exons)	CPD	CPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPB2 geni (Tüm Gen Dizi Analizi)	CPB2 gene (Sequence Analysis-All Coding Exons)	CPB2	CPB2, CPU, TAFI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPB1 geni (Tüm Gen Dizi Analizi)	CPB1 gene (Sequence Analysis-All Coding Exons)	CPB1	CPB1, PCPB, PASP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPA4 geni (Tüm Gen Dizi Analizi)	CPA4 gene (Sequence Analysis-All Coding Exons)	CPA4	CPA4, CPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPA3 geni (Tüm Gen Dizi Analizi)	CPA3 gene (Sequence Analysis-All Coding Exons)	CPA3	CPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPA2 geni (Tüm Gen Dizi Analizi)	CPA2 gene (Sequence Analysis-All Coding Exons)	CPA2	CPA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPA1 geni (Tüm Gen Dizi Analizi)	CPA1 gene (Sequence Analysis-All Coding Exons)	CPA1	CPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX8C geni (Tüm Gen Dizi Analizi)	COX8C gene (Sequence Analysis-All Coding Exons)	COX8C	COX8C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX7C geni (Tüm Gen Dizi Analizi)	COX7C gene (Sequence Analysis-All Coding Exons)	COX7C	COX7C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX7B2 geni (Tüm Gen Dizi Analizi)	COX7B2 gene (Sequence Analysis-All Coding Exons)	COX7B2	COX7B2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX7A2L geni (Tüm Gen Dizi Analizi)	COX7A2L gene (Sequence Analysis-All Coding Exons)	COX7A2L	COX7A2L, COX7RP, EB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX7A2 geni (Tüm Gen Dizi Analizi)	COX7A2 gene (Sequence Analysis-All Coding Exons)	COX7A2	COX7A2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX7A1 geni (Tüm Gen Dizi Analizi)	COX7A1 gene (Sequence Analysis-All Coding Exons)	COX7A1	COX7A1, COX7AM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX6C geni (Tüm Gen Dizi Analizi)	COX6C gene (Sequence Analysis-All Coding Exons)	COX6C	COX6C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX5B geni (Tüm Gen Dizi Analizi)	COX5B gene (Sequence Analysis-All Coding Exons)	COX5B	COX5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX5A geni (Tüm Gen Dizi Analizi)	COX5A gene (Sequence Analysis-All Coding Exons)	COX5A	COX5A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX4I1 geni (Tüm Gen Dizi Analizi)	COX4I1 gene (Sequence Analysis-All Coding Exons)	COX4I1	COX4I1, COX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX19 geni (Tüm Gen Dizi Analizi)	COX19 gene (Sequence Analysis-All Coding Exons)	COX19	COX19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX18 geni (Tüm Gen Dizi Analizi)	COX18 gene (Sequence Analysis-All Coding Exons)	COX18	COX18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX17 geni (Tüm Gen Dizi Analizi)	COX17 gene (Sequence Analysis-All Coding Exons)	COX17	COX17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX11 geni (Tüm Gen Dizi Analizi)	COX11 gene (Sequence Analysis-All Coding Exons)	COX11	COX11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

COTL1 geni (Tüm Gen Dizi Analizi)	COTL1 gene (Sequence Analysis-All Coding Exons)	COTL1	COTL1, CLP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORT geni (Tüm Gen Dizi Analizi)	CORT gene (Sequence Analysis-All Coding Exons)	CORT	CORT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORO7 geni (Tüm Gen Dizi Analizi)	CORO7 gene (Sequence Analysis-All Coding Exons)	CORO7	CORO7, PQD1, CRN7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORO2B geni (Tüm Gen Dizi Analizi)	CORO2B gene (Sequence Analysis-All Coding Exons)	CORO2B	CORO2B, CLIPINC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORO2A geni (Tüm Gen Dizi Analizi)	CORO2A gene (Sequence Analysis-All Coding Exons)	CORO2A	CORO2A, WDR2, IR10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORO1C geni (Tüm Gen Dizi Analizi)	CORO1C gene (Sequence Analysis-All Coding Exons)	CORO1C	CORO1C	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CORO1B geni (Tüm Gen Dizi Analizi)	CORO1B gene (Sequence Analysis-All Coding Exons)	CORO1B	CORO1B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COQ5 geni (Tüm Gen Dizi Analizi)	COQ5 gene (Sequence Analysis-All Coding Exons)	COQ5	COQ5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPZ2 geni (Tüm Gen Dizi Analizi)	COPZ2 gene (Sequence Analysis-All Coding Exons)	COPZ2	COPZ2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPZ1 geni (Tüm Gen Dizi Analizi)	COPZ1 gene (Sequence Analysis-All Coding Exons)	COPZ1	COPZ1, COPZ	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS8 geni (Tüm Gen Dizi Analizi)	COPS8 gene (Sequence Analysis-All Coding Exons)	COPS8	COPS8, CSN8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS7B geni (Tüm Gen Dizi Analizi)	COPS7B gene (Sequence Analysis-All Coding Exons)	COPS7B	COPS7B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS7A geni (Tüm Gen Dizi Analizi)	COPS7A gene (Sequence Analysis-All Coding Exons)	COPS7A	COPS7A, CSN7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS6 geni (Tüm Gen Dizi Analizi)	COPS6 gene (Sequence Analysis-All Coding Exons)	COPS6	COPS6, CSN6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS5 geni (Tüm Gen Dizi Analizi)	COPS5 gene (Sequence Analysis-All Coding Exons)	COPS5	COPS5, JAB1, SGN5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS4 geni (Tüm Gen Dizi Analizi)	COPS4 gene (Sequence Analysis-All Coding Exons)	COPS4	COPS4, CSN4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS3 geni (Tüm Gen Dizi Analizi)	COPS3 gene (Sequence Analysis-All Coding Exons)	COPS3	COPS3, SGN3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPS2 geni (Tüm Gen Dizi Analizi)	COPS2 gene (Sequence Analysis-All Coding Exons)	COPS2	TRIP15, SGN2, COPS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPG2 geni (Tüm Gen Dizi Analizi)	COPG2 gene (Sequence Analysis-All Coding Exons)	COPG2	COPG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPG1 geni (Tüm Gen Dizi Analizi)	COPG1 gene (Sequence Analysis-All Coding Exons)	COPG1	COPG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPE geni (Tüm Gen Dizi Analizi)	COPE gene (Sequence Analysis-All Coding Exons)	COPE	COPE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPB2 geni (Tüm Gen Dizi Analizi)	COPB2 gene (Sequence Analysis-All Coding Exons)	COPB2	COPB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COPB1 geni (Tüm Gen Dizi Analizi)	COPB1 gene (Sequence Analysis-All Coding Exons)	COPB1	COPB1, COPB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD9 geni (Tüm Gen Dizi Analizi)	COMMD9 gene (Sequence Analysis-All Coding Exons)	COMMD9	COMM9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD8 geni (Tüm Gen Dizi Analizi)	COMMD8 gene (Sequence Analysis-All Coding Exons)	COMMD8	COMMD8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD7 geni (Tüm Gen Dizi Analizi)	COMMD7 gene (Sequence Analysis-All Coding Exons)	COMMD7	COMMD7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD6 geni (Tüm Gen Dizi Analizi)	COMMD6 gene (Sequence Analysis-All Coding Exons)	COMMD6	COMMD6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD5 geni (Tüm Gen Dizi Analizi)	COMMD5 gene (Sequence Analysis-All Coding Exons)	COMMD5	COMMD5, HCARG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD4 geni (Tüm Gen Dizi Analizi)	COMMD4 gene (Sequence Analysis-All Coding Exons)	COMMD4	COMMD4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD3 geni (Tüm Gen Dizi Analizi)	COMMD3 gene (Sequence Analysis-All Coding Exons)	COMMD3	COMMD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD2 geni (Tüm Gen Dizi Analizi)	COMMD2 gene (Sequence Analysis-All Coding Exons)	COMMD2	COMMD2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD10 geni (Tüm Gen Dizi Analizi)	COMMD10 gene (Sequence Analysis-All Coding Exons)	COMMD10	COMMD10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COMMD1 geni (Tüm Gen Dizi Analizi)	COMMD1 gene (Sequence Analysis-All Coding Exons)	COMMD1	COMMD1, MURR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COLEC12 geni (Tüm Gen Dizi Analizi)	COLEC12 gene (Sequence Analysis-All Coding Exons)	COLEC12	COLEC12, SRCL, CLP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COLEC10 geni (Tüm Gen Dizi Analizi)	COLEC10 gene (Sequence Analysis-All Coding Exons)	COLEC10	COLEC10, CLL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COLCA2 geni (Tüm Gen Dizi Analizi)	COLCA2 gene (Sequence Analysis-All Coding Exons)	COLCA2	COLCA2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL8A1 geni (Tüm Gen Dizi Analizi)	COL8A1 gene (Sequence Analysis-All Coding Exons)	COL8A1	COL8A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL6A6 geni (Tüm Gen Dizi Analizi)	COL6A6 gene (Sequence Analysis-All Coding Exons)	COL6A6	COL6A6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL6A5 geni (Tüm Gen Dizi Analizi)	COL6A5 gene (Sequence Analysis-All Coding Exons)	COL6A5	COL6A5, COL29A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL64AP1 geni (Tüm Gen Dizi Analizi)	COL64AP1 gene (Sequence Analysis-All Coding Exons)	COL64AP1	COL64AP1, DVWA, DIVA, LOC144875	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL5A3 geni (Tüm Gen Dizi Analizi)	COL5A3 gene (Sequence Analysis-All Coding Exons)	COL5A3	COL5A3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL28A1 geni (Tüm Gen Dizi Analizi)	COL28A1 gene (Sequence Analysis-All Coding Exons)	COL28A1	COL28A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL24A1 geni (Tüm Gen Dizi Analizi)	COL24A1 gene (Sequence Analysis-All Coding Exons)	COL24A1	COL24A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL23A1 geni (Tüm Gen Dizi Analizi)	COL23A1 gene (Sequence Analysis-All Coding Exons)	COL23A1	COL23A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL22A1 geni (Tüm Gen Dizi Analizi)	COL22A1 gene (Sequence Analysis-All Coding Exons)	COL22A1	COL22A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL21A1 geni (Tüm Gen Dizi Analizi)	COL21A1 gene (Sequence Analysis-All Coding Exons)	COL21A1	COL21A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL19A1 geni (Tüm Gen Dizi Analizi)	COL19A1 gene (Sequence Analysis-All Coding Exons)	COL19A1	COL19A1, D6S228E, COL9A1L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL16A1 geni (Tüm Gen Dizi Analizi)	COL16A1 gene (Sequence Analysis-All Coding Exons)	COL16A1	COL16A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL15A1 geni (Tüm Gen Dizi Analizi)	COL15A1 gene (Sequence Analysis-All Coding Exons)	COL15A1	COL15A1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL14A1 geni (Tüm Gen Dizi Analizi)	COL14A1 gene (Sequence Analysis-All Coding Exons)	COL14A1	COL14A1, UND	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COIL geni (Tüm Gen Dizi Analizi)	COIL gene (Sequence Analysis-All Coding Exons)	COIL	COIL, CLN80	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COG3 geni (Tüm Gen Dizi Analizi)	COG3 gene (Sequence Analysis-All Coding Exons)	COG3	COG3, SEC34	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COG2 geni (Tüm Gen Dizi Analizi)	COG2 gene (Sequence Analysis-All Coding Exons)	COG2	COG2, LDLC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COBLL1 geni (Tüm Gen Dizi Analizi)	COBLL1 gene (Sequence Analysis-All Coding Exons)	COBLL1	COBLL1, COBLR1, KIAA0977	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COBL geni (Tüm Gen Dizi Analizi)	COBL gene (Sequence Analysis-All Coding Exons)	COBL	COBL, KIAA0833	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COA7 geni (Tüm Gen Dizi Analizi)	COA7 gene (Sequence Analysis-All Coding Exons)	COA7	COA7, RESA1, SELRC1, C1orf163	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COA4 geni (Tüm Gen Dizi Analizi)	COA4 gene (Sequence Analysis-All Coding Exons)	COA4	CHCHD8, E2IG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COA3 geni (Tüm Gen Dizi Analizi)	COA3 gene (Sequence Analysis-All Coding Exons)	COA3	COA3, CCDC56	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COA1 geni (Tüm Gen Dizi Analizi)	COA1 gene (Sequence Analysis-All Coding Exons)	COA1	COA1, C7orf44	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTROB geni (Tüm Gen Dizi Analizi)	CNTROB gene (Sequence Analysis-All Coding Exons)	CNTROB	CNTROB, LIP8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTRL geni (Tüm Gen Dizi Analizi)	CNTRL gene (Sequence Analysis-All Coding Exons)	CNTRL	CEP1, CEP110, FAN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTNAP5 geni (Tüm Gen Dizi Analizi)	CNTNAP5 gene (Sequence Analysis-All Coding Exons)	CNTNAP5	CNTNAP5, CASPR5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTNAP4 geni (Tüm Gen Dizi Analizi)	CNTNAP4 gene (Sequence Analysis-All Coding Exons)	CNTNAP4	CNTNAP4, CASPR4, KIAA1763	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTNAP3 geni (Tüm Gen Dizi Analizi)	CNTNAP3 gene (Sequence Analysis-All Coding Exons)	CNTNAP3	CNTNAP3, KIAA1714	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTN6 geni (Tüm Gen Dizi Analizi)	CNTN6 gene (Sequence Analysis-All Coding Exons)	CNTN6	CNTN6, NB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTN5 geni (Tüm Gen Dizi Analizi)	CNTN5 gene (Sequence Analysis-All Coding Exons)	CNTN5	CNTN5, NB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTN4 geni (Tüm Gen Dizi Analizi)	CNTN4 gene (Sequence Analysis-All Coding Exons)	CNTN4	CNTN4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTN3 geni (Tüm Gen Dizi Analizi)	CNTN3 gene (Sequence Analysis-All Coding Exons)	CNTN3	CNTN3, PANG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTLN geni (Tüm Gen Dizi Analizi)	CNTLN gene (Sequence Analysis-All Coding Exons)	CNTLN	CNTLN, C9orf101, C9orf39	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTFR geni (Tüm Gen Dizi Analizi)	CNTFR gene (Sequence Analysis-All Coding Exons)	CNTFR	CNTFR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNTF geni (Tüm Gen Dizi Analizi)	CNTF gene (Sequence Analysis-All Coding Exons)	CNTF	CNTF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNST geni (Tüm Gen Dizi Analizi)	CNST gene (Sequence Analysis-All Coding Exons)	CNST	CNST	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNR2 geni (Tüm Gen Dizi Analizi)	CNR2 gene (Sequence Analysis-All Coding Exons)	CNR2	CNR2, CB2, CX5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNR1 geni (Tüm Gen Dizi Analizi)	CNR1 gene (Sequence Analysis-All Coding Exons)	CNR1	CNR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNPY4 geni (Tüm Gen Dizi Analizi)	CNPY4 gene (Sequence Analysis-All Coding Exons)	CNPY4	CNPY4, PRAT4B, MGC40499	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNPY3 geni (Tüm Gen Dizi Analizi)	CNPY3 gene (Sequence Analysis-All Coding Exons)	CNPY3	CNPY3, TNRC5, PRAT4A, PRAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNPY2 geni (Tüm Gen Dizi Analizi)	CNPY2 gene (Sequence Analysis-All Coding Exons)	CNPY2	CNPY4, TMEM4, MSAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNPY1 geni (Tüm Gen Dizi Analizi)	CNPY1 gene (Sequence Analysis-All Coding Exons)	CNPY1	CNPY1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNP geni (Tüm Gen Dizi Analizi)	CNP gene (Sequence Analysis-All Coding Exons)	CNP	CNP, CNP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT9 geni (Tüm Gen Dizi Analizi)	CNOT9 gene (Sequence Analysis-All Coding Exons)	CNOT9	RQCD1, RCD1, CNOT9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT8 geni (Tüm Gen Dizi Analizi)	CNOT8 gene (Sequence Analysis-All Coding Exons)	CNOT8	CNOT8, CALIF, POP2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT7 geni (Tüm Gen Dizi Analizi)	CNOT7 gene (Sequence Analysis-All Coding Exons)	CNOT7	CNOT7, CAF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT6 geni (Tüm Gen Dizi Analizi)	CNOT6 gene (Sequence Analysis-All Coding Exons)	CNOT6	CNOT6, CCR4, KIAA1194	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT4 geni (Tüm Gen Dizi Analizi)	CNOT4 gene (Sequence Analysis-All Coding Exons)	CNOT4	CNOT4, NOT4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT3 geni (Tüm Gen Dizi Analizi)	CNOT3 gene (Sequence Analysis-All Coding Exons)	CNOT3	CNOT3, NOT3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT2 geni (Tüm Gen Dizi Analizi)	CNOT2 gene (Sequence Analysis-All Coding Exons)	CNOT2	CNOT2, NOT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNOT1 geni (Tüm Gen Dizi Analizi)	CNOT1 gene (Sequence Analysis-All Coding Exons)	CNOT1	CNOT1, NOT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNNM3 geni (Tüm Gen Dizi Analizi)	CNNM3 gene (Sequence Analysis-All Coding Exons)	CNNM3	CNNM3, ACDP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNNM1 geni (Tüm Gen Dizi Analizi)	CNNM1 gene (Sequence Analysis-All Coding Exons)	CNNM1	CNNM1, ACDP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNN3 geni (Tüm Gen Dizi Analizi)	CNN3 gene (Sequence Analysis-All Coding Exons)	CNN3	CNN3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNN2 geni (Tüm Gen Dizi Analizi)	CNN2 gene (Sequence Analysis-All Coding Exons)	CNN2	CNN2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNN1 geni (Tüm Gen Dizi Analizi)	CNN1 gene (Sequence Analysis-All Coding Exons)	CNN1	CNN1, SMCC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNMD geni (Tüm Gen Dizi Analizi)	CNMD gene (Sequence Analysis-All Coding Exons)	CNMD	LECT1, CHM1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CNKSR2 geni (Tüm Gen Dizi Analizi)	CNKSR2 gene (Sequence Analysis-All Coding Exons)	CNKSR2	CNKSR2, CNK2, KIAA0902	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNKSR1 geni (Tüm Gen Dizi Analizi)	CNKSR1 gene (Sequence Analysis-All Coding Exons)	CNKSR1	CNKSR1, CNK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNIH2 geni (Tüm Gen Dizi Analizi)	CNIH2 gene (Sequence Analysis-All Coding Exons)	CNIH2	CNIH2, CNIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNIH1 geni (Tüm Gen Dizi Analizi)	CNIH1 gene (Sequence Analysis-All Coding Exons)	CNIH1	CNIH, TGAM77	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNGA2 geni (Tüm Gen Dizi Analizi)	CNGA2 gene (Sequence Analysis-All Coding Exons)	CNGA2	CNGA2, CNG2, OCNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNFN geni (Tüm Gen Dizi Analizi)	CNFN gene (Sequence Analysis-All Coding Exons)	CNFN	CNFN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNEP1R1 geni (Tüm Gen Dizi Analizi)	CNEP1R1 gene (Sequence Analysis-All Coding Exons)	CNEP1R1	CNEP1R1, NEP1R1, TMEM188	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNDP2 geni (Tüm Gen Dizi Analizi)	CNDP2 gene (Sequence Analysis-All Coding Exons)	CNDP2	PEPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNDP1 geni (Tüm Gen Dizi Analizi)	CNDP1 gene (Sequence Analysis-All Coding Exons)	CNDP1	CNDP1, CN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMYA5 geni (Tüm Gen Dizi Analizi)	CMYA5 gene (Sequence Analysis-All Coding Exons)	CMYA5	CMYA5, MYOSPRYN, TRIM76	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTR2 geni (Tüm Gen Dizi Analizi)	CMTR2 gene (Sequence Analysis-All Coding Exons)	CMTR2	CMTR2, MTR2, FTSJD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTR1 geni (Tüm Gen Dizi Analizi)	CMTR1 gene (Sequence Analysis-All Coding Exons)	CMTR1	CMTR1, MTR1, ISG95, FTSJD2, KIAA0082	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM8 geni (Tüm Gen Dizi Analizi)	CMTM8 gene (Sequence Analysis-All Coding Exons)	CMTM8	CMTM8, CKLFSF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM7 geni (Tüm Gen Dizi Analizi)	CMTM7 gene (Sequence Analysis-All Coding Exons)	CMTM7	CMTM7, CKLFSF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM6 geni (Tüm Gen Dizi Analizi)	CMTM6 gene (Sequence Analysis-All Coding Exons)	CMTM6	CMTM6, CKLFSF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM5 geni (Tüm Gen Dizi Analizi)	CMTM5 gene (Sequence Analysis-All Coding Exons)	CMTM5	CMTM5, CKLFSF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM4 geni (Tüm Gen Dizi Analizi)	CMTM4 gene (Sequence Analysis-All Coding Exons)	CMTM4	CMTM4, CKLFSF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM3 geni (Tüm Gen Dizi Analizi)	CMTM3 gene (Sequence Analysis-All Coding Exons)	CMTM3	CMTM3, CKLFSF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM2 geni (Tüm Gen Dizi Analizi)	CMTM2 gene (Sequence Analysis-All Coding Exons)	CMTM2	CMTM2, CKLFSF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMTM1 geni (Tüm Gen Dizi Analizi)	CMTM1 gene (Sequence Analysis-All Coding Exons)	CMTM1	CMTM1, CKLFSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMPK2 geni (Tüm Gen Dizi Analizi)	CMPK2 gene (Sequence Analysis-All Coding Exons)	CMPK2	CMPK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMPK1 geni (Tüm Gen Dizi Analizi)	CMPK1 gene (Sequence Analysis-All Coding Exons)	CMPK1	CMPK1, CMPK, UMPK, UMK, CMK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMKLR1 geni (Tüm Gen Dizi Analizi)	CMKLR1 gene (Sequence Analysis-All Coding Exons)	CMKLR1	CMKLR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMIP geni (Tüm Gen Dizi Analizi)	CMIP gene (Sequence Analysis-All Coding Exons)	CMIP	CMIP, KIAA1694, TCMIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMC1 geni (Tüm Gen Dizi Analizi)	CMC1 gene (Sequence Analysis-All Coding Exons)	CMC1	CMC1, C3orf68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMBL geni (Tüm Gen Dizi Analizi)	CMBL gene (Sequence Analysis-All Coding Exons)	CMBL	CMBL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMAS geni (Tüm Gen Dizi Analizi)	CMAS gene (Sequence Analysis-All Coding Exons)	CMAS	CMAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CMA1 geni (Tüm Gen Dizi Analizi)	CMA1 gene (Sequence Analysis-All Coding Exons)	CMA1	CMA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLYBL geni (Tüm Gen Dizi Analizi)	CLYBL gene (Sequence Analysis-All Coding Exons)	CLYBL	CLYBL, CLB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLVS2 geni (Tüm Gen Dizi Analizi)	CLVS2 gene (Sequence Analysis-All Coding Exons)	CLVS2	CLVS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLVS1 geni (Tüm Gen Dizi Analizi)	CLVS1 gene (Sequence Analysis-All Coding Exons)	CLVS1	CLVS1, RLBP1L1, CRALBPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLUL1 geni (Tüm Gen Dizi Analizi)	CLUL1 gene (Sequence Analysis-All Coding Exons)	CLUL1	CLUL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLUH geni (Tüm Gen Dizi Analizi)	CLUH gene (Sequence Analysis-All Coding Exons)	CLUH	CLUH, CLU1, KIAA0664	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLUAP1 geni (Tüm Gen Dizi Analizi)	CLUAP1 gene (Sequence Analysis-All Coding Exons)	CLUAP1	CLUAP1, QILIN, FAP22, KIAA0645	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLU geni (Tüm Gen Dizi Analizi)	CLU gene (Sequence Analysis-All Coding Exons)	CLU	CLU, CL1, SGP2, TRPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLTCL1 geni (Tüm Gen Dizi Analizi)	CLTCL1 gene (Sequence Analysis-All Coding Exons)	CLTCL1	CLTCL1, CLTD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLTC geni (Tüm Gen Dizi Analizi)	CLTC gene (Sequence Analysis-All Coding Exons)	CLTC	CLTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLTC geni (Tüm Gen Dizi Analizi)	CLTC gene (Sequence Analysis-All Coding Exons)	CLTC	CLTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLTB geni (Tüm Gen Dizi Analizi)	CLTB gene (Sequence Analysis-All Coding Exons)	CLTB	CLTB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLTA geni (Tüm Gen Dizi Analizi)	CLTA gene (Sequence Analysis-All Coding Exons)	CLTA	CLTA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLSTN3 geni (Tüm Gen Dizi Analizi)	CLSTN3 gene (Sequence Analysis-All Coding Exons)	CLSTN3	CLSTN3, KIAA0726	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLSTN2 geni (Tüm Gen Dizi Analizi)	CLSTN2 gene (Sequence Analysis-All Coding Exons)	CLSTN2	CLSTN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLSTN1 geni (Tüm Gen Dizi Analizi)	CLSTN1 gene (Sequence Analysis-All Coding Exons)	CLSTN1	CLSTN1, KIAA0911	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLSPN geni (Tüm Gen Dizi Analizi)	CLSPN gene (Sequence Analysis-All Coding Exons)	CLSPN	CLASPIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLPX geni (Tüm Gen Dizi Analizi)	CLPX gene (Sequence Analysis-All Coding Exons)	CLPX	CLPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLPTM1L geni (Tüm Gen Dizi Analizi)	CLPTM1L gene (Sequence Analysis-All Coding Exons)	CLPTM1L	CLPTM1L, CRR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLPTM1 geni (Tüm Gen Dizi Analizi)	CLPTM1 gene (Sequence Analysis-All Coding Exons)	CLPTM1	CLPTM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLPS geni (Tüm Gen Dizi Analizi)	CLPS gene (Sequence Analysis-All Coding Exons)	CLPS	CLPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLOCK geni (Tüm Gen Dizi Analizi)	CLOCK gene (Sequence Analysis-All Coding Exons)	CLOCK	CLOCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLNS1A geni (Tüm Gen Dizi Analizi)	CLNS1A gene (Sequence Analysis-All Coding Exons)	CLNS1A	CLNS1A, CLNS1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLNK geni (Tüm Gen Dizi Analizi)	CLNK gene (Sequence Analysis-All Coding Exons)	CLNK	CLNK, MIST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLMN geni (Tüm Gen Dizi Analizi)	CLMN gene (Sequence Analysis-All Coding Exons)	CLMN	CLMN, KIAA1188	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLLU1OS geni (Tüm Gen Dizi Analizi)	CLLU1OS gene (Sequence Analysis-All Coding Exons)	CLLU1OS	CLLU1OS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLLU1 geni (Tüm Gen Dizi Analizi)	CLLU1 gene (Sequence Analysis-All Coding Exons)	CLLU1	CLLU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLK4 geni (Tüm Gen Dizi Analizi)	CLK4 gene (Sequence Analysis-All Coding Exons)	CLK4	CLK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLK3 geni (Tüm Gen Dizi Analizi)	CLK3 gene (Sequence Analysis-All Coding Exons)	CLK3	CLK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLK2 geni (Tüm Gen Dizi Analizi)	CLK2 gene (Sequence Analysis-All Coding Exons)	CLK2	CLK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLK1 geni (Tüm Gen Dizi Analizi)	CLK1 gene (Sequence Analysis-All Coding Exons)	CLK1	CLK1, CLK, STY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIP3 geni (Tüm Gen Dizi Analizi)	CLIP3 gene (Sequence Analysis-All Coding Exons)	CLIP3	CLIP3, CLIPR59	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIP2 geni (Tüm Gen Dizi Analizi)	CLIP2 gene (Sequence Analysis-All Coding Exons)	CLIP2	CLIP2, CYLN2, WBSCR4, WSCR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIP1 geni (Tüm Gen Dizi Analizi)	CLIP1 gene (Sequence Analysis-All Coding Exons)	CLIP1	CLIP1, RSN, CYLN1, CLIP170	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLINT1 geni (Tüm Gen Dizi Analizi)	CLINT1 gene (Sequence Analysis-All Coding Exons)	CLINT1	CLINT1, EPN4, EPNR, KIAA0171	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIC6 geni (Tüm Gen Dizi Analizi)	CLIC6 gene (Sequence Analysis-All Coding Exons)	CLIC6	CLIC6, CLIC1L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIC3 geni (Tüm Gen Dizi Analizi)	CLIC3 gene (Sequence Analysis-All Coding Exons)	CLIC3	CLIC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLGN geni (Tüm Gen Dizi Analizi)	CLGN gene (Sequence Analysis-All Coding Exons)	CLGN	CLGN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLECL1 geni (Tüm Gen Dizi Analizi)	CLECL1 gene (Sequence Analysis-All Coding Exons)	CLECL1	CLECL1, DCAL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC9A geni (Tüm Gen Dizi Analizi)	CLEC9A gene (Sequence Analysis-All Coding Exons)	CLEC9A	CLEC9A, DNGR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC6A geni (Tüm Gen Dizi Analizi)	CLEC6A gene (Sequence Analysis-All Coding Exons)	CLEC6A	CLEC6A, CLEC4N	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC5A geni (Tüm Gen Dizi Analizi)	CLEC5A gene (Sequence Analysis-All Coding Exons)	CLEC5A	CLEC5A, CLECSF5, MDL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4M geni (Tüm Gen Dizi Analizi)	CLEC4M gene (Sequence Analysis-All Coding Exons)	CLEC4M	CLEC4M, CD209L, L-SIGN, DCSIGNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4G geni (Tüm Gen Dizi Analizi)	CLEC4G gene (Sequence Analysis-All Coding Exons)	CLEC4G	CLEC4G, LSECTIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4E geni (Tüm Gen Dizi Analizi)	CLEC4E gene (Sequence Analysis-All Coding Exons)	CLEC4E	CLEC4E, MINCLE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4D geni (Tüm Gen Dizi Analizi)	CLEC4D gene (Sequence Analysis-All Coding Exons)	CLEC4D	CLEC4D, CLECSF8, CLEC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4C geni (Tüm Gen Dizi Analizi)	CLEC4C gene (Sequence Analysis-All Coding Exons)	CLEC4C	CLEC4C, CLECSF11, DLEC, BDCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC4A geni (Tüm Gen Dizi Analizi)	CLEC4A gene (Sequence Analysis-All Coding Exons)	CLEC4A	CLEC4A, CLECSF6, DCIR, DDB27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC3B geni (Tüm Gen Dizi Analizi)	CLEC3B gene (Sequence Analysis-All Coding Exons)	CLEC3B	TNA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC3A geni (Tüm Gen Dizi Analizi)	CLEC3A gene (Sequence Analysis-All Coding Exons)	CLEC3A	CLEC3A, CLECSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC2D geni (Tüm Gen Dizi Analizi)	CLEC2D gene (Sequence Analysis-All Coding Exons)	CLEC2D	CLEC2D, LLT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC2B geni (Tüm Gen Dizi Analizi)	CLEC2B gene (Sequence Analysis-All Coding Exons)	CLEC2B	CLEC2B, CLECSF2, AICL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC2A geni (Tüm Gen Dizi Analizi)	CLEC2A gene (Sequence Analysis-All Coding Exons)	CLEC2A	CLEC2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC1B geni (Tüm Gen Dizi Analizi)	CLEC1B gene (Sequence Analysis-All Coding Exons)	CLEC1B	CLEC2B, CLEC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC1A geni (Tüm Gen Dizi Analizi)	CLEC1A gene (Sequence Analysis-All Coding Exons)	CLEC1A	CLEC1A, CLEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC18C geni (Tüm Gen Dizi Analizi)	CLEC18C gene (Sequence Analysis-All Coding Exons)	CLEC18C	CLEC18C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC18B geni (Tüm Gen Dizi Analizi)	CLEC18B gene (Sequence Analysis-All Coding Exons)	CLEC18B	CLEC18B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC18A geni (Tüm Gen Dizi Analizi)	CLEC18A gene (Sequence Analysis-All Coding Exons)	CLEC18A	CLEC18A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC17A geni (Tüm Gen Dizi Analizi)	CLEC17A gene (Sequence Analysis-All Coding Exons)	CLEC17A	CLEC17A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC16A geni (Tüm Gen Dizi Analizi)	CLEC16A gene (Sequence Analysis-All Coding Exons)	CLEC16A	CLEC16A, KIAA0350	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC14A geni (Tüm Gen Dizi Analizi)	CLEC14A gene (Sequence Analysis-All Coding Exons)	CLEC14A	CLEC14A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC12A geni (Tüm Gen Dizi Analizi)	CLEC12A gene (Sequence Analysis-All Coding Exons)	CLEC12A	CLEC12A, MICL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC11A geni (Tüm Gen Dizi Analizi)	CLEC11A gene (Sequence Analysis-All Coding Exons)	CLEC11A	SCGF, LSLCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLEC10A geni (Tüm Gen Dizi Analizi)	CLEC10A gene (Sequence Analysis-All Coding Exons)	CLEC10A	CLEC10A, CLECSF14, MGL, HML, CD301	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN9 geni (Tüm Gen Dizi Analizi)	CLDN9 gene (Sequence Analysis-All Coding Exons)	CLDN9	CLDN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN8 geni (Tüm Gen Dizi Analizi)	CLDN8 gene (Sequence Analysis-All Coding Exons)	CLDN8	CLDN8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CLDN6 geni (Tüm Gen Dizi Analizi)	CLDN6 gene (Sequence Analysis-All Coding Exons)	CLDN6	CLDN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN5 geni (Tüm Gen Dizi Analizi)	CLDN5 gene (Sequence Analysis-All Coding Exons)	CLDN5	CLDN5, TMVCF, AWAL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN4 geni (Tüm Gen Dizi Analizi)	CLDN4 gene (Sequence Analysis-All Coding Exons)	CLDN4	CLDN4, CPETR1, CPER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN3 geni (Tüm Gen Dizi Analizi)	CLDN3 gene (Sequence Analysis-All Coding Exons)	CLDN3	CLDN3, CPETR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN23 geni (Tüm Gen Dizi Analizi)	CLDN23 gene (Sequence Analysis-All Coding Exons)	CLDN23	CLDN23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN2 geni (Tüm Gen Dizi Analizi)	CLDN2 gene (Sequence Analysis-All Coding Exons)	CLDN2	CLDN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN17 geni (Tüm Gen Dizi Analizi)	CLDN17 gene (Sequence Analysis-All Coding Exons)	CLDN17	CLDN17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KMT5A geni (Tüm Gen Dizi Analizi)	KMT5A gene (Sequence Analysis-All Coding Exons)	KMT5A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN15 geni (Tüm Gen Dizi Analizi)	CLDN15 gene (Sequence Analysis-All Coding Exons)	CLDN15	CLDN15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN12 geni (Tüm Gen Dizi Analizi)	CLDN12 gene (Sequence Analysis-All Coding Exons)	CLDN12	CLKDN12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN11 geni (Tüm Gen Dizi Analizi)	CLDN11 gene (Sequence Analysis-All Coding Exons)	CLDN11	CLDN11, OTM, OSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCN6 geni (Tüm Gen Dizi Analizi)	CLCN6 gene (Sequence Analysis-All Coding Exons)	CLCN6	CLCN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCN3 geni (Tüm Gen Dizi Analizi)	CLCN3 gene (Sequence Analysis-All Coding Exons)	CLCN3	CLCN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCA4 geni (Tüm Gen Dizi Analizi)	CLCA4 gene (Sequence Analysis-All Coding Exons)	CLCA4	CLCA4, CACC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCA3P geni (Tüm Gen Dizi Analizi)	CLCA3P gene (Sequence Analysis-All Coding Exons)	CLCA3P	CLCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCA2 geni (Tüm Gen Dizi Analizi)	CLCA2 gene (Sequence Analysis-All Coding Exons)	CLCA2	CLCA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLCA1 geni (Tüm Gen Dizi Analizi)	CLCA1 gene (Sequence Analysis-All Coding Exons)	CLCA1	CLCA1, CACC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLC geni (Tüm Gen Dizi Analizi)	CLC gene (Sequence Analysis-All Coding Exons)	CLC	CLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLASP2 geni (Tüm Gen Dizi Analizi)	CLASP2 gene (Sequence Analysis-All Coding Exons)	CLASP2	CLASP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLASP1 geni (Tüm Gen Dizi Analizi)	CLASP1 gene (Sequence Analysis-All Coding Exons)	CLASP1	CLASP1, KIAA0622	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKS2 geni (Tüm Gen Dizi Analizi)	CKS2 gene (Sequence Analysis-All Coding Exons)	CKS2	CKS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKS1B geni (Tüm Gen Dizi Analizi)	CKS1B gene (Sequence Analysis-All Coding Exons)	CKS1B	CKS1B, CKS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKMT2 geni (Tüm Gen Dizi Analizi)	CKMT2 gene (Sequence Analysis-All Coding Exons)	CKMT2	CKMT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKMT1B geni (Tüm Gen Dizi Analizi)	CKMT1B gene (Sequence Analysis-All Coding Exons)	CKMT1B	CKMT1B, CKMT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKMT1A geni (Tüm Gen Dizi Analizi)	CKMT1A gene (Sequence Analysis-All Coding Exons)	CKMT1A	CKMT1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKM geni (Tüm Gen Dizi Analizi)	CKM gene (Sequence Analysis-All Coding Exons)	CKM	CKM, CKMM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKLF geni (Tüm Gen Dizi Analizi)	CKLF gene (Sequence Analysis-All Coding Exons)	CKLF	CKLF, HSPC224	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKB geni (Tüm Gen Dizi Analizi)	CKB gene (Sequence Analysis-All Coding Exons)	CKB	CKB, CKBB, BCK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKAP5 geni (Tüm Gen Dizi Analizi)	CKAP5 gene (Sequence Analysis-All Coding Exons)	CKAP5	CKAP5, CHTOG, MSPS, KIAA0097	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CKAP2 geni (Tüm Gen Dizi Analizi)	CKAP2 gene (Sequence Analysis-All Coding Exons)	CKAP2	CKAP2, TMAP, LB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIZ1 geni (Tüm Gen Dizi Analizi)	CIZ1 gene (Sequence Analysis-All Coding Exons)	CIZ1	CIZ1, ZNF356, LSFR1, NP94	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CITED4 geni (Tüm Gen Dizi Analizi)	CITED4 gene (Sequence Analysis-All Coding Exons)	CITED4	CITED4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CITED1 geni (Tüm Gen Dizi Analizi)	CITED1 gene (Sequence Analysis-All Coding Exons)	CITED1	MSG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CISD3 geni (Tüm Gen Dizi Analizi)	CISD3 gene (Sequence Analysis-All Coding Exons)	CISD3	CISD3, MINER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CISD1 geni (Tüm Gen Dizi Analizi)	CISD1 gene (Sequence Analysis-All Coding Exons)	CISD1	CISD1, MITONEET	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIRBP geni (Tüm Gen Dizi Analizi)	CIRBP gene (Sequence Analysis-All Coding Exons)	CIRBP	CIRBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIR1 geni (Tüm Gen Dizi Analizi)	CIR1 gene (Sequence Analysis-All Coding Exons)	CIR1	CIR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIPC geni (Tüm Gen Dizi Analizi)	CIPC gene (Sequence Analysis-All Coding Exons)	CIPC	CIPC, KIAA1737	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CINP geni (Tüm Gen Dizi Analizi)	CINP gene (Sequence Analysis-All Coding Exons)	CINP	CINP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CILP2 geni (Tüm Gen Dizi Analizi)	CILP2 gene (Sequence Analysis-All Coding Exons)	CILP2	CILP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIDEA geni (Tüm Gen Dizi Analizi)	CIDEA gene (Sequence Analysis-All Coding Exons)	CIDEA	CIDEA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIC geni (Tüm Gen Dizi Analizi)	CIC gene (Sequence Analysis-All Coding Exons)	CIC	CIC, KIAA0306	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIB1 geni (Tüm Gen Dizi Analizi)	CIB1 gene (Sequence Analysis-All Coding Exons)	CIB1	CIB, KIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIART geni (Tüm Gen Dizi Analizi)	CIART gene (Sequence Analysis-All Coding Exons)	CIART	CIART, CHRONO, GM129	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIAPIN1 geni (Tüm Gen Dizi Analizi)	CIAPIN1 gene (Sequence Analysis-All Coding Exons)	CIAPIN1	CIAPIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIAO1 geni (Tüm Gen Dizi Analizi)	CIAO1 gene (Sequence Analysis-All Coding Exons)	CIAO1	CIAO1, WDR39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHURC1 geni (Tüm Gen Dizi Analizi)	CHURC1 gene (Sequence Analysis-All Coding Exons)	CHURC1	CHURC1, CHCH, C14orf52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHTOP geni (Tüm Gen Dizi Analizi)	CHTOP gene (Sequence Analysis-All Coding Exons)	CHTOP	CHTOP, C10orf77, SRAG, FOP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHTF8 geni (Tüm Gen Dizi Analizi)	CHTF8 gene (Sequence Analysis-All Coding Exons)	CHTF8	CHTF8, CTF8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHTF18 geni (Tüm Gen Dizi Analizi)	CHTF18 gene (Sequence Analysis-All Coding Exons)	CHTF18	CHTF18, CHL12, CTF18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHSY3 geni (Tüm Gen Dizi Analizi)	CHSY3 gene (Sequence Analysis-All Coding Exons)	CHSY3	CHSY3, CSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST9 geni (Tüm Gen Dizi Analizi)	CHST9 gene (Sequence Analysis-All Coding Exons)	CHST9	CHST9, GalNAc4ST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST7 geni (Tüm Gen Dizi Analizi)	CHST7 gene (Sequence Analysis-All Coding Exons)	CHST7	CHST7, C6ST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST5 geni (Tüm Gen Dizi Analizi)	CHST5 gene (Sequence Analysis-All Coding Exons)	CHST5	CHST5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST2 geni (Tüm Gen Dizi Analizi)	CHST2 gene (Sequence Analysis-All Coding Exons)	CHST2	CHST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST15 geni (Tüm Gen Dizi Analizi)	CHST15 gene (Sequence Analysis-All Coding Exons)	CHST15	CHST15, BRAG, KIAA0598	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST13 geni (Tüm Gen Dizi Analizi)	CHST13 gene (Sequence Analysis-All Coding Exons)	CHST13	CHST13, C4ST3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST12 geni (Tüm Gen Dizi Analizi)	CHST12 gene (Sequence Analysis-All Coding Exons)	CHST12	CHST12, C4ST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST11 geni (Tüm Gen Dizi Analizi)	CHST11 gene (Sequence Analysis-All Coding Exons)	CHST11	CHST11, C4ST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST10 geni (Tüm Gen Dizi Analizi)	CHST10 gene (Sequence Analysis-All Coding Exons)	CHST10	CHST10, HNK1ST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHST1 geni (Tüm Gen Dizi Analizi)	CHST1 gene (Sequence Analysis-All Coding Exons)	CHST1	CHST1, KSGAL6ST	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNB4 geni (Tüm Gen Dizi Analizi)	CHRNB4 gene (Sequence Analysis-All Coding Exons)	CHRNB4	CHRNB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNB3 geni (Tüm Gen Dizi Analizi)	CHRNB3 gene (Sequence Analysis-All Coding Exons)	CHRNB3	CHRNB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNA9 geni (Tüm Gen Dizi Analizi)	CHRNA9 gene (Sequence Analysis-All Coding Exons)	CHRNA9	CHRNA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNA7 geni (Tüm Gen Dizi Analizi)	CHRNA7 gene (Sequence Analysis-All Coding Exons)	CHRNA7	CHRNA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNA6 geni (Tüm Gen Dizi Analizi)	CHRNA6 gene (Sequence Analysis-All Coding Exons)	CHRNA6	CHRNA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRNA10 geni (Tüm Gen Dizi Analizi)	CHRNA10 gene (Sequence Analysis-All Coding Exons)	CHRNA10	CHRNA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRM5 geni (Tüm Gen Dizi Analizi)	CHRM5 gene (Sequence Analysis-All Coding Exons)	CHRM5	CHRM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRM4 geni (Tüm Gen Dizi Analizi)	CHRM4 gene (Sequence Analysis-All Coding Exons)	CHRM4	CHRM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRM2 geni (Tüm Gen Dizi Analizi)	CHRM2 gene (Sequence Analysis-All Coding Exons)	CHRM2	CHRM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRM1 geni (Tüm Gen Dizi Analizi)	CHRM1 gene (Sequence Analysis-All Coding Exons)	CHRM1	CHRM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRDL2 geni (Tüm Gen Dizi Analizi)	CHRDL2 gene (Sequence Analysis-All Coding Exons)	CHRDL2	CHRDL2, CHL2, BNF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRD geni (Tüm Gen Dizi Analizi)	CHRD gene (Sequence Analysis-All Coding Exons)	CHRD	CHRD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KLF5 geni (Tüm Gen Dizi Analizi)	KLF5 gene (Sequence Analysis-All Coding Exons)	KLF5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRAC1 geni (Tüm Gen Dizi Analizi)	CHRAC1 gene (Sequence Analysis-All Coding Exons)	CHRAC1	CHARC1, CHARC15, YCL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHPT1 geni (Tüm Gen Dizi Analizi)	CHPT1 gene (Sequence Analysis-All Coding Exons)	CHPT1	CHPT1, CPT, CPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHPF2 geni (Tüm Gen Dizi Analizi)	CHPF2 gene (Sequence Analysis-All Coding Exons)	CHPF2	CHPF2, KIAA1402, CSGlcAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHPF geni (Tüm Gen Dizi Analizi)	CHPF gene (Sequence Analysis-All Coding Exons)	CHPF	CHPF, CSS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHP1 geni (Tüm Gen Dizi Analizi)	CHP1 gene (Sequence Analysis-All Coding Exons)	CHP1	CHP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHODL geni (Tüm Gen Dizi Analizi)	CHODL gene (Sequence Analysis-All Coding Exons)	CHODL	CHODL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHN2 geni (Tüm Gen Dizi Analizi)	CHN2 gene (Sequence Analysis-All Coding Exons)	CHN2	CHN2, ARHGAP3, RHOGAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP7 geni (Tüm Gen Dizi Analizi)	CHMP7 gene (Sequence Analysis-All Coding Exons)	CHMP7	CHMP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP6 geni (Tüm Gen Dizi Analizi)	CHMP6 gene (Sequence Analysis-All Coding Exons)	CHMP6	CHMP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP5 geni (Tüm Gen Dizi Analizi)	CHMP5 gene (Sequence Analysis-All Coding Exons)	CHMP5	CHMP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP4C geni (Tüm Gen Dizi Analizi)	CHMP4C gene (Sequence Analysis-All Coding Exons)	CHMP4C	CHMP4C, SNF7-3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP4A geni (Tüm Gen Dizi Analizi)	CHMP4A gene (Sequence Analysis-All Coding Exons)	CHMP4A	CHMP4A, SNF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP3 geni (Tüm Gen Dizi Analizi)	CHMP3 gene (Sequence Analysis-All Coding Exons)	CHMP3	VPS24, CHMP3, NEDF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP2A geni (Tüm Gen Dizi Analizi)	CHMP2A gene (Sequence Analysis-All Coding Exons)	CHMP2A	CHMP2A, VPS2A, VPS2, BC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHMP1B geni (Tüm Gen Dizi Analizi)	CHMP1B gene (Sequence Analysis-All Coding Exons)	CHMP1B	CHMP1B, C10orf2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHML geni (Tüm Gen Dizi Analizi)	CHML gene (Sequence Analysis-All Coding Exons)	CHML	CHML, REP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHL1 geni (Tüm Gen Dizi Analizi)	CHL1 gene (Sequence Analysis-All Coding Exons)	CHL1	CHL1, CALL, L1CAM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHKA geni (Tüm Gen Dizi Analizi)	CHKA gene (Sequence Analysis-All Coding Exons)	CHKA	CHKA, CHK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHID1 geni (Tüm Gen Dizi Analizi)	CHID1 gene (Sequence Analysis-All Coding Exons)	CHID1	CHID1, SICLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CHIC1 geni (Tüm Gen Dizi Analizi)	CHIC1 gene (Sequence Analysis-All Coding Exons)	CHIC1	CHIC1, BRX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHIA geni (Tüm Gen Dizi Analizi)	CHIA gene (Sequence Analysis-All Coding Exons)	CHIA	CHIA, TSA1902, CHT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHI3L2 geni (Tüm Gen Dizi Analizi)	CHI3L2 gene (Sequence Analysis-All Coding Exons)	CHI3L2	CHI3L2, YKL39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHGB geni (Tüm Gen Dizi Analizi)	CHGB gene (Sequence Analysis-All Coding Exons)	CHGB	CHGB, SCG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHGA geni (Tüm Gen Dizi Analizi)	CHGA gene (Sequence Analysis-All Coding Exons)	CHGA	CHGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHFR geni (Tüm Gen Dizi Analizi)	CHFR gene (Sequence Analysis-All Coding Exons)	CHFR	CHFR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHEK1 geni (Tüm Gen Dizi Analizi)	CHEK1 gene (Sequence Analysis-All Coding Exons)	CHEK1	CHEK1, CHK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD9 geni (Tüm Gen Dizi Analizi)	CHD9 gene (Sequence Analysis-All Coding Exons)	CHD9	CHD9, CREMM, PRIC320, KIAA0308	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD6 geni (Tüm Gen Dizi Analizi)	CHD6 gene (Sequence Analysis-All Coding Exons)	CHD6	CHD6, CHD5, RIGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD5 geni (Tüm Gen Dizi Analizi)	CHD5 gene (Sequence Analysis-All Coding Exons)	CHD5	CHD5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD3 geni (Tüm Gen Dizi Analizi)	CHD3 gene (Sequence Analysis-All Coding Exons)	CHD3	CHD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD1L geni (Tüm Gen Dizi Analizi)	CHD1L gene (Sequence Analysis-All Coding Exons)	CHD1L	CHD1L, ALC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHD1 geni (Tüm Gen Dizi Analizi)	CHD1 gene (Sequence Analysis-All Coding Exons)	CHD1	CHD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD7 geni (Tüm Gen Dizi Analizi)	CHCHD7 gene (Sequence Analysis-All Coding Exons)	CHCHD7	CHCHD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD6 geni (Tüm Gen Dizi Analizi)	CHCHD6 gene (Sequence Analysis-All Coding Exons)	CHCHD6	CHCHD6, CHCM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD5 geni (Tüm Gen Dizi Analizi)	CHCHD5 gene (Sequence Analysis-All Coding Exons)	CHCHD5	CHCHD5, MIC14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD4 geni (Tüm Gen Dizi Analizi)	CHCHD4 gene (Sequence Analysis-All Coding Exons)	CHCHD4	CHDH4, MIA40	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD3 geni (Tüm Gen Dizi Analizi)	CHCHD3 gene (Sequence Analysis-All Coding Exons)	CHCHD3	CHCHD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHAF1B geni (Tüm Gen Dizi Analizi)	CHAF1B gene (Sequence Analysis-All Coding Exons)	CHAF1B	CAF1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DS5 geni (Tüm Gen Dizi Analizi)	KIR2DS5 gene (Sequence Analysis-All Coding Exons)	KIR2DS5	KIR2DS5, NKAT9, CD158G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHADL geni (Tüm Gen Dizi Analizi)	CHADL gene (Sequence Analysis-All Coding Exons)	CHADL	CHADL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DS3 geni (Tüm Gen Dizi Analizi)	KIR2DS3 gene (Sequence Analysis-All Coding Exons)	KIR2DS3	KIR2DS3, NKAT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DS2 geni (Tüm Gen Dizi Analizi)	KIR2DS2 gene (Sequence Analysis-All Coding Exons)	KIR2DS2	KIR2DS2, NKAT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DS1 geni (Tüm Gen Dizi Analizi)	KIR2DS1 gene (Sequence Analysis-All Coding Exons)	KIR2DS1	KIR2DS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL5B geni (Tüm Gen Dizi Analizi)	KIR2DL5B gene (Sequence Analysis-All Coding Exons)	KIR2DL5B	KIR2DL5B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL5A geni (Tüm Gen Dizi Analizi)	KIR2DL5A gene (Sequence Analysis-All Coding Exons)	KIR2DL5A	KIR2DL5A, KIR2DL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHAD geni (Tüm Gen Dizi Analizi)	CHAD gene (Sequence Analysis-All Coding Exons)	CHAD	CHAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHAC1 geni (Tüm Gen Dizi Analizi)	CHAC1 gene (Sequence Analysis-All Coding Exons)	CHAC1	CHAC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIR2DL2 geni (Tüm Gen Dizi Analizi)	KIR2DL2 gene (Sequence Analysis-All Coding Exons)	KIR2DL2	KIR2DL2, NKAT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CH2SH geni (Tüm Gen Dizi Analizi)	CH2SH gene (Sequence Analysis-All Coding Exons)	CH2SH	CH2SH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGRRF1 geni (Tüm Gen Dizi Analizi)	CGRRF1 gene (Sequence Analysis-All Coding Exons)	CGRRF1	GCRRF1, CGR19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGREF1 geni (Tüm Gen Dizi Analizi)	CGREF1 gene (Sequence Analysis-All Coding Exons)	CGREF1	CGREF1, CGR11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGNL1 geni (Tüm Gen Dizi Analizi)	CGNL1 gene (Sequence Analysis-All Coding Exons)	CGNL1	CGNL1, FLJ14957, KIAA1749	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGGBP1 geni (Tüm Gen Dizi Analizi)	CGGBP1 gene (Sequence Analysis-All Coding Exons)	CGGBP1	CGGBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB7 geni (Tüm Gen Dizi Analizi)	CGB7 gene (Sequence Analysis-All Coding Exons)	CGB7	CGB7, CGB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB3 geni (Tüm Gen Dizi Analizi)	CGB3 gene (Sequence Analysis-All Coding Exons)	CGB3	CGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB1 geni (Tüm Gen Dizi Analizi)	CGB1 gene (Sequence Analysis-All Coding Exons)	CGB1	CGB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGA geni (Tüm Gen Dizi Analizi)	CGA gene (Sequence Analysis-All Coding Exons)	CGA	CGA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFLAR geni (Tüm Gen Dizi Analizi)	CFLAR gene (Sequence Analysis-All Coding Exons)	CFLAR	CFLAR, FLIP, CASPER, FLAME1, CASH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFL1 geni (Tüm Gen Dizi Analizi)	CFL1 gene (Sequence Analysis-All Coding Exons)	CFL1	CFL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFHR4 geni (Tüm Gen Dizi Analizi)	CFHR4 gene (Sequence Analysis-All Coding Exons)	CFHR4	CFHR4, FHR4, CFHL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFHR2 geni (Tüm Gen Dizi Analizi)	CFHR2 gene (Sequence Analysis-All Coding Exons)	CFHR2	CFHR2, FHR2, HFL3, CFHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFDP1 geni (Tüm Gen Dizi Analizi)	CFDP1 gene (Sequence Analysis-All Coding Exons)	CFDP1	CFDP1, CP27, BCNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP97 geni (Tüm Gen Dizi Analizi)	CFAP97 gene (Sequence Analysis-All Coding Exons)	CFAP97	CFAP97, HMW, KIAA1430	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP65 geni (Tüm Gen Dizi Analizi)	CFAP65 gene (Sequence Analysis-All Coding Exons)	CFAP65	CCDC108	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP57 geni (Tüm Gen Dizi Analizi)	CFAP57 gene (Sequence Analysis-All Coding Exons)	CFAP57	WDR65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP52 geni (Tüm Gen Dizi Analizi)	CFAP52 gene (Sequence Analysis-All Coding Exons)	CFAP52	WDR16, WDRPLH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP45 geni (Tüm Gen Dizi Analizi)	CFAP45 gene (Sequence Analysis-All Coding Exons)	CFAP45	CCDC19, NESG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CFAP126 geni (Tüm Gen Dizi Analizi)	CFAP126 gene (Sequence Analysis-All Coding Exons)	CFAP126	CFAP126, FLTP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CETN3 geni (Tüm Gen Dizi Analizi)	CETN3 gene (Sequence Analysis-All Coding Exons)	CETN3	CETN3, CDC31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CETN2 geni (Tüm Gen Dizi Analizi)	CETN2 gene (Sequence Analysis-All Coding Exons)	CETN2	CETN2, CALT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CETN1 geni (Tüm Gen Dizi Analizi)	CETN1 gene (Sequence Analysis-All Coding Exons)	CETN1	CETN1, CEN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CES3 geni (Tüm Gen Dizi Analizi)	CES3 gene (Sequence Analysis-All Coding Exons)	CES3	CES3, ES31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CES2 geni (Tüm Gen Dizi Analizi)	CES2 gene (Sequence Analysis-All Coding Exons)	CES2	CES2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERS6 geni (Tüm Gen Dizi Analizi)	CERS6 gene (Sequence Analysis-All Coding Exons)	CERS6	CERS6, LASS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERS5 geni (Tüm Gen Dizi Analizi)	CERS5 gene (Sequence Analysis-All Coding Exons)	CERS5	CERS5, LASS5, TRH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERS4 geni (Tüm Gen Dizi Analizi)	CERS4 gene (Sequence Analysis-All Coding Exons)	CERS4	CERS4, LASS4, TRH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERS2 geni (Tüm Gen Dizi Analizi)	CERS2 gene (Sequence Analysis-All Coding Exons)	CERS2	CERS2, LASS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERK geni (Tüm Gen Dizi Analizi)	CERK gene (Sequence Analysis-All Coding Exons)	CERK	CERK, KIAA1646	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CERCAM geni (Tüm Gen Dizi Analizi)	CERCAM gene (Sequence Analysis-All Coding Exons)	CERCAM	CERCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CER1 geni (Tüm Gen Dizi Analizi)	CER1 gene (Sequence Analysis-All Coding Exons)	CER1	CER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEPT1 geni (Tüm Gen Dizi Analizi)	CEPT1 gene (Sequence Analysis-All Coding Exons)	CEPT1	CEPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP97 geni (Tüm Gen Dizi Analizi)	CEP97 gene (Sequence Analysis-All Coding Exons)	CEP97	CEP97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP89 geni (Tüm Gen Dizi Analizi)	CEP89 gene (Sequence Analysis-All Coding Exons)	CEP89	CEP89, CCDC123, CEP123, FLJ14640	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP72 geni (Tüm Gen Dizi Analizi)	CEP72 gene (Sequence Analysis-All Coding Exons)	CEP72	CEP72, KIAA1519	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP70 geni (Tüm Gen Dizi Analizi)	CEP70 gene (Sequence Analysis-All Coding Exons)	CEP70	CEP70	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP68 geni (Tüm Gen Dizi Analizi)	CEP68 gene (Sequence Analysis-All Coding Exons)	CEP68	CEP68, KIAA0582	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP55 geni (Tüm Gen Dizi Analizi)	CEP55 gene (Sequence Analysis-All Coding Exons)	CEP55	CEP55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP250 geni (Tüm Gen Dizi Analizi)	CEP250 gene (Sequence Analysis-All Coding Exons)	CEP250	CEP2, CEP250, CNAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP192 geni (Tüm Gen Dizi Analizi)	CEP192 gene (Sequence Analysis-All Coding Exons)	CEP192	CEP192, PPP1R62, KIAA1569	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP170 geni (Tüm Gen Dizi Analizi)	CEP170 gene (Sequence Analysis-All Coding Exons)	CEP170	CEP170, KIAA0470	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP162 geni (Tüm Gen Dizi Analizi)	CEP162 gene (Sequence Analysis-All Coding Exons)	CEP162	CEP162, KIAA1009	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP131 geni (Tüm Gen Dizi Analizi)	CEP131 gene (Sequence Analysis-All Coding Exons)	CEP131	AZI1, ZA1, KIAA1118	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEP126 geni (Tüm Gen Dizi Analizi)	CEP126 gene (Sequence Analysis-All Coding Exons)	CEP126	KIAA1377	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPX geni (Tüm Gen Dizi Analizi)	CENPX gene (Sequence Analysis-All Coding Exons)	CENPX	STRAI3, CENPX, FAAP10, MHF2_09	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPW geni (Tüm Gen Dizi Analizi)	CENPW gene (Sequence Analysis-All Coding Exons)	CENPW	CEMPW, C6orf173, CUG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPV geni (Tüm Gen Dizi Analizi)	CENPV gene (Sequence Analysis-All Coding Exons)	CENPV	CENPV, P30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPU geni (Tüm Gen Dizi Analizi)	CENPU gene (Sequence Analysis-All Coding Exons)	CENPU	MLF1IP, CENPU, CENP50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPT geni (Tüm Gen Dizi Analizi)	CENPT gene (Sequence Analysis-All Coding Exons)	CENPT	CENPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPS geni (Tüm Gen Dizi Analizi)	CENPS gene (Sequence Analysis-All Coding Exons)	CENPS	APITD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPQ geni (Tüm Gen Dizi Analizi)	CENPQ gene (Sequence Analysis-All Coding Exons)	CENPQ	CENPQ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPP geni (Tüm Gen Dizi Analizi)	CENPP gene (Sequence Analysis-All Coding Exons)	CENPP	CENPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPO geni (Tüm Gen Dizi Analizi)	CENPO gene (Sequence Analysis-All Coding Exons)	CENPO	CENPO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPN geni (Tüm Gen Dizi Analizi)	CENPN gene (Sequence Analysis-All Coding Exons)	CENPN	CENPN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPM geni (Tüm Gen Dizi Analizi)	CENPM gene (Sequence Analysis-All Coding Exons)	CENPM	PANE1, C22orf18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPL geni (Tüm Gen Dizi Analizi)	CENPL gene (Sequence Analysis-All Coding Exons)	CENPL	CENPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPK geni (Tüm Gen Dizi Analizi)	CENPK gene (Sequence Analysis-All Coding Exons)	CENPK	CEMPK, SOLT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPI geni (Tüm Gen Dizi Analizi)	CENPI gene (Sequence Analysis-All Coding Exons)	CENPI		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPH geni (Tüm Gen Dizi Analizi)	CENPH gene (Sequence Analysis-All Coding Exons)	CENPH	CENPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPC geni (Tüm Gen Dizi Analizi)	CENPC gene (Sequence Analysis-All Coding Exons)	CENPC	CENPC1, CENPC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPB geni (Tüm Gen Dizi Analizi)	CENPB gene (Sequence Analysis-All Coding Exons)	CENPB	CENPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CENPA geni (Tüm Gen Dizi Analizi)	CENPA gene (Sequence Analysis-All Coding Exons)	CENPA	CENPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEND1 geni (Tüm Gen Dizi Analizi)	CEND1 gene (Sequence Analysis-All Coding Exons)	CEND1	CEND1, BM88	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CEMP1 geni (Tüm Gen Dizi Analizi)	CEMP1 gene (Sequence Analysis-All Coding Exons)	CEMP1	CEMP1, CP23	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEMIP geni (Tüm Gen Dizi Analizi)	CEMIP gene (Sequence Analysis-All Coding Exons)	CEMIP	KIAA1199	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELSR3 geni (Tüm Gen Dizi Analizi)	CELSR3 gene (Sequence Analysis-All Coding Exons)	CELSR3	CELSR3, EGFL1, MEGF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELSR2 geni (Tüm Gen Dizi Analizi)	CELSR2 gene (Sequence Analysis-All Coding Exons)	CELSR2	CELSR2, EGFL2, MEGF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELSR1 geni (Tüm Gen Dizi Analizi)	CELSR1 gene (Sequence Analysis-All Coding Exons)	CELSR1	CELSR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF6 geni (Tüm Gen Dizi Analizi)	CELF6 gene (Sequence Analysis-All Coding Exons)	CELF6	CELF6, BRUNOL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF5 geni (Tüm Gen Dizi Analizi)	CELF5 gene (Sequence Analysis-All Coding Exons)	CELF5	CELF5, BRUNOL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF4 geni (Tüm Gen Dizi Analizi)	CELF4 gene (Sequence Analysis-All Coding Exons)	CELF4	CELF4, BRUNOL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF3 geni (Tüm Gen Dizi Analizi)	CELF3 gene (Sequence Analysis-All Coding Exons)	CELF3	CELF3, TNRC4, BRUNOL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF2 geni (Tüm Gen Dizi Analizi)	CELF2 gene (Sequence Analysis-All Coding Exons)	CELF2	CELF2, CUGBP2, ETR3, BRUNOL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELF1 geni (Tüm Gen Dizi Analizi)	CELF1 gene (Sequence Analysis-All Coding Exons)	CELF1	CELF1, CUGBP1, NAB50, BRUNOL2, CUGBP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELA2B geni (Tüm Gen Dizi Analizi)	CELA2B gene (Sequence Analysis-All Coding Exons)	CELA2B	CELA2B, ELA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELA2A geni (Tüm Gen Dizi Analizi)	CELA2A gene (Sequence Analysis-All Coding Exons)	CELA2A	CELA2A, ELA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CELA1 geni (Tüm Gen Dizi Analizi)	CELA1 gene (Sequence Analysis-All Coding Exons)	CELA1	CELA1, ELA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KIAA0100 geni (Tüm Gen Dizi Analizi)	KIAA0100 gene (Sequence Analysis-All Coding Exons)	KIAA0100		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CECR2 geni (Tüm Gen Dizi Analizi)	CECR2 gene (Sequence Analysis-All Coding Exons)	CECR2	CECR2, KIAA1740	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEBPZ geni (Tüm Gen Dizi Analizi)	CEBPZ gene (Sequence Analysis-All Coding Exons)	CEBPZ	CEBPZ, CBF, NOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEBPG geni (Tüm Gen Dizi Analizi)	CEBPG gene (Sequence Analysis-All Coding Exons)	CEBPG	CEBPG, GPE1BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEBPD geni (Tüm Gen Dizi Analizi)	CEBPD gene (Sequence Analysis-All Coding Exons)	CEBPD	CEBPD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEBPB geni (Tüm Gen Dizi Analizi)	CEBPB gene (Sequence Analysis-All Coding Exons)	CEBPB	CEBPB, TCF5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM8 geni (Tüm Gen Dizi Analizi)	CEACAM8 gene (Sequence Analysis-All Coding Exons)	CEACAM8	CEACAM8, CD66B, CGM6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM6 geni (Tüm Gen Dizi Analizi)	CEACAM6 gene (Sequence Analysis-All Coding Exons)	CEACAM6	CEACAM6, NCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM5 geni (Tüm Gen Dizi Analizi)	CEACAM5 gene (Sequence Analysis-All Coding Exons)	CEACAM5	CEACAM5, CEA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM1 geni (Tüm Gen Dizi Analizi)	CEACAM1 gene (Sequence Analysis-All Coding Exons)	CEACAM1	CEACAM1, BGP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDYL geni (Tüm Gen Dizi Analizi)	CDYL gene (Sequence Analysis-All Coding Exons)	CDYL	CDYL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDY2A geni (Tüm Gen Dizi Analizi)	CDY2A gene (Sequence Analysis-All Coding Exons)	CDY2A	CDY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDY1 geni (Tüm Gen Dizi Analizi)	CDY1 gene (Sequence Analysis-All Coding Exons)	CDY1	CDY1, CDY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDX4 geni (Tüm Gen Dizi Analizi)	CDX4 gene (Sequence Analysis-All Coding Exons)	CDX4	CDX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDX2 geni (Tüm Gen Dizi Analizi)	CDX2 gene (Sequence Analysis-All Coding Exons)	CDX2	CDX2, CDX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDX1 geni (Tüm Gen Dizi Analizi)	CDX1 gene (Sequence Analysis-All Coding Exons)	CDX1	CDX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDS2 geni (Tüm Gen Dizi Analizi)	CDS2 gene (Sequence Analysis-All Coding Exons)	CDS2	CDS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDS1 geni (Tüm Gen Dizi Analizi)	CDS1 gene (Sequence Analysis-All Coding Exons)	CDS1	CDS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDR2 geni (Tüm Gen Dizi Analizi)	CDR2 gene (Sequence Analysis-All Coding Exons)	CDR2	CDR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDR1 geni (Tüm Gen Dizi Analizi)	CDR1 gene (Sequence Analysis-All Coding Exons)	CDR1	CDR1, CDR62A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDO1 geni (Tüm Gen Dizi Analizi)	CDO1 gene (Sequence Analysis-All Coding Exons)	CDO1	CDO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDNF geni (Tüm Gen Dizi Analizi)	CDNF gene (Sequence Analysis-All Coding Exons)	CDNF	ARMETL1, CDNF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN3 geni (Tüm Gen Dizi Analizi)	CDKN3 gene (Sequence Analysis-All Coding Exons)	CDKN3	CDKN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2D geni (Tüm Gen Dizi Analizi)	CDKN2D gene (Sequence Analysis-All Coding Exons)	CDKN2D	CDKN2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2C geni (Tüm Gen Dizi Analizi)	CDKN2C gene (Sequence Analysis-All Coding Exons)	CDKN2C	CDKN2C, INK4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2B-AS1 geni (Tüm Gen Dizi Analizi)	CDKN2B-AS1 gene (Sequence Analysis-All Coding Exons)	CDKN2B-AS1	CDKN2BAS, ANRIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2B geni (Tüm Gen Dizi Analizi)	CDKN2B gene (Sequence Analysis-All Coding Exons)	CDKN2B	CDKN2B, MTS2, P15, INK4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2B geni (Tüm Gen Dizi Analizi)	CDKN2B gene (Sequence Analysis-All Coding Exons)	CDKN2B	CDKN2B, MTS2, P15, INK4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN2AIP geni (Tüm Gen Dizi Analizi)	CDKN2AIP gene (Sequence Analysis-All Coding Exons)	CDKN2AIP	CDKN2AIP, CARF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKN1A geni (Tüm Gen Dizi Analizi)	CDKN1A gene (Sequence Analysis-All Coding Exons)	CDKN1A	CDKN1A, WAF1, CIP1, CDKN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKL3 geni (Tüm Gen Dizi Analizi)	CDKL3 gene (Sequence Analysis-All Coding Exons)	CDKL3	CDKL3, NKIAMRE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKL2 geni (Tüm Gen Dizi Analizi)	CDKL2 gene (Sequence Analysis-All Coding Exons)	CDKL2	CDKL2, KKIAMRE, P56	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDKL1 geni (Tüm Gen Dizi Analizi)	CDKL1 gene (Sequence Analysis-All Coding Exons)	CDKL1	CDKL1, KKIALRE, p42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK9 geni (Tüm Gen Dizi Analizi)	CDK9 gene (Sequence Analysis-All Coding Exons)	CDK9	CDK9, CDC2L4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KDELR2 geni (Tüm Gen Dizi Analizi)	KDELR2 gene (Sequence Analysis-All Coding Exons)	KDELR2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK8 geni (Tüm Gen Dizi Analizi)	CDK8 gene (Sequence Analysis-All Coding Exons)	CDK8	CDK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK7 geni (Tüm Gen Dizi Analizi)	CDK7 gene (Sequence Analysis-All Coding Exons)	CDK7	CDK7, STK1, CAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK5RAP3 geni (Tüm Gen Dizi Analizi)	CDK5RAP3 gene (Sequence Analysis-All Coding Exons)	CDK5RAP3	CDK5RAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK5RAP1 geni (Tüm Gen Dizi Analizi)	CDK5RAP1 gene (Sequence Analysis-All Coding Exons)	CDK5RAP1	CDK5RAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK5R2 geni (Tüm Gen Dizi Analizi)	CDK5R2 gene (Sequence Analysis-All Coding Exons)	CDK5R2	CDK5R2, NCK5AI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK5R1 geni (Tüm Gen Dizi Analizi)	CDK5R1 gene (Sequence Analysis-All Coding Exons)	CDK5R1	CDK5R1, p35	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK3 geni (Tüm Gen Dizi Analizi)	CDK3 gene (Sequence Analysis-All Coding Exons)	CDK3	CDK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK2AP1 geni (Tüm Gen Dizi Analizi)	CDK2AP1 gene (Sequence Analysis-All Coding Exons)	CDK2AP1	CDK2AP1, DOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK20 geni (Tüm Gen Dizi Analizi)	CDK20 gene (Sequence Analysis-All Coding Exons)	CDK20	CDK20, CCRK, CDCH, P42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK2 geni (Tüm Gen Dizi Analizi)	CDK2 gene (Sequence Analysis-All Coding Exons)	CDK2	CDK2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK19 geni (Tüm Gen Dizi Analizi)	CDK19 gene (Sequence Analysis-All Coding Exons)	CDK19	CDK19, KIAA1028	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK18 geni (Tüm Gen Dizi Analizi)	CDK18 gene (Sequence Analysis-All Coding Exons)	CDK18	CDK18, PCTK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK17 geni (Tüm Gen Dizi Analizi)	CDK17 gene (Sequence Analysis-All Coding Exons)	CDK17	CDK17, PCTK2, PCTAIRE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK16 geni (Tüm Gen Dizi Analizi)	CDK16 gene (Sequence Analysis-All Coding Exons)	CDK16	CDK16, PCTK1, PCTAIRE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK15 geni (Tüm Gen Dizi Analizi)	CDK15 gene (Sequence Analysis-All Coding Exons)	CDK15	CDK15, PFTAIRE2, ALS2CR7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK14 geni (Tüm Gen Dizi Analizi)	CDK14 gene (Sequence Analysis-All Coding Exons)	CDK14	CDK14, PFTK1, PFTAIRE1, KIAA0834	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK12 geni (Tüm Gen Dizi Analizi)	CDK12 gene (Sequence Analysis-All Coding Exons)	CDK12	CDK12, CRKRS, CRK7, KIAA0904	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK11B geni (Tüm Gen Dizi Analizi)	CDK11B gene (Sequence Analysis-All Coding Exons)	CDK11B	CDK11B, CDC2L1, P58, CDK11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK11A geni (Tüm Gen Dizi Analizi)	CDK11A gene (Sequence Analysis-All Coding Exons)	CDK11A	CDK11A, CDC2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK10 geni (Tüm Gen Dizi Analizi)	CDK10 gene (Sequence Analysis-All Coding Exons)	CDK10	CDK10, PISSLRE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDK1 geni (Tüm Gen Dizi Analizi)	CDK1 gene (Sequence Analysis-All Coding Exons)	CDK1	CDK1, CDC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDIP1 geni (Tüm Gen Dizi Analizi)	CDIP1 gene (Sequence Analysis-All Coding Exons)	CDIP1	C16orf5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDHR5 geni (Tüm Gen Dizi Analizi)	CDHR5 gene (Sequence Analysis-All Coding Exons)	CDHR5	CDHR5, MUCDHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDHR3 geni (Tüm Gen Dizi Analizi)	CDHR3 gene (Sequence Analysis-All Coding Exons)	CDHR3	CDHR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH9 geni (Tüm Gen Dizi Analizi)	CDH9 gene (Sequence Analysis-All Coding Exons)	CDH9	CDH9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH8 geni (Tüm Gen Dizi Analizi)	CDH8 gene (Sequence Analysis-All Coding Exons)	CDH8	CDH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH7 geni (Tüm Gen Dizi Analizi)	CDH7 gene (Sequence Analysis-All Coding Exons)	CDH7	CDH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH6 geni (Tüm Gen Dizi Analizi)	CDH6 gene (Sequence Analysis-All Coding Exons)	CDH6	CDH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH5 geni (Tüm Gen Dizi Analizi)	CDH5 gene (Sequence Analysis-All Coding Exons)	CDH5	CDH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH4 geni (Tüm Gen Dizi Analizi)	CDH4 gene (Sequence Analysis-All Coding Exons)	CDH4	CDH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH22 geni (Tüm Gen Dizi Analizi)	CDH22 gene (Sequence Analysis-All Coding Exons)	CDH22	CDH22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH20 geni (Tüm Gen Dizi Analizi)	CDH20 gene (Sequence Analysis-All Coding Exons)	CDH20	CDH20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH2 geni (Tüm Gen Dizi Analizi)	CDH2 gene (Sequence Analysis-All Coding Exons)	CDH2	CDH2, NCAD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH19 geni (Tüm Gen Dizi Analizi)	CDH19 gene (Sequence Analysis-All Coding Exons)	CDH19	CDH19, CDH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH18 geni (Tüm Gen Dizi Analizi)	CDH18 gene (Sequence Analysis-All Coding Exons)	CDH18	CDH18, CDH14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH17 geni (Tüm Gen Dizi Analizi)	CDH17 gene (Sequence Analysis-All Coding Exons)	CDH17	CDH17, CDH16, HPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH16 geni (Tüm Gen Dizi Analizi)	CDH16 gene (Sequence Analysis-All Coding Exons)	CDH16	CDH16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH13 geni (Tüm Gen Dizi Analizi)	CDH13 gene (Sequence Analysis-All Coding Exons)	CDH13	CDH13, CDHH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH12 geni (Tüm Gen Dizi Analizi)	CDH12 gene (Sequence Analysis-All Coding Exons)	CDH12	CDH12, CDHB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH11 geni (Tüm Gen Dizi Analizi)	CDH11 gene (Sequence Analysis-All Coding Exons)	CDH11	CDH11, CAD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDH10 geni (Tüm Gen Dizi Analizi)	CDH10 gene (Sequence Analysis-All Coding Exons)	CDH10	CDH10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCP2 geni (Tüm Gen Dizi Analizi)	CDCP2 gene (Sequence Analysis-All Coding Exons)	CDCP2	CDCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCP1 geni (Tüm Gen Dizi Analizi)	CDCP1 gene (Sequence Analysis-All Coding Exons)	CDCP1	CDCP1, SIMA135	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNN1 geni (Tüm Gen Dizi Analizi)	KCNN1 gene (Sequence Analysis-All Coding Exons)	KCNN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CDCA8 geni (Tüm Gen Dizi Analizi)	CDCA8 gene (Sequence Analysis-All Coding Exons)	CDCA8	CDCA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCA7L geni (Tüm Gen Dizi Analizi)	CDCA7L gene (Sequence Analysis-All Coding Exons)	CDCA7L	CDCA7L, R1, JPO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCA5 geni (Tüm Gen Dizi Analizi)	CDCA5 gene (Sequence Analysis-All Coding Exons)	CDCA5	CDCA5, SORORIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCA4 geni (Tüm Gen Dizi Analizi)	CDCA4 gene (Sequence Analysis-All Coding Exons)	CDCA4	CDCA4, HEPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC7 geni (Tüm Gen Dizi Analizi)	CDC7 gene (Sequence Analysis-All Coding Exons)	CDC7	CDC7L1, CDC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC5L geni (Tüm Gen Dizi Analizi)	CDC5L gene (Sequence Analysis-All Coding Exons)	CDC5L	CDC5L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42EP5 geni (Tüm Gen Dizi Analizi)	CDC42EP5 gene (Sequence Analysis-All Coding Exons)	CDC42EP5	CDC42EP5, CEP5, BORG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42EP4 geni (Tüm Gen Dizi Analizi)	CDC42EP4 gene (Sequence Analysis-All Coding Exons)	CDC42EP4	CDC42EP4, BORG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42EP3 geni (Tüm Gen Dizi Analizi)	CDC42EP3 gene (Sequence Analysis-All Coding Exons)	CDC42EP3	CDC42EP3, CEP3, BORG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42EP2 geni (Tüm Gen Dizi Analizi)	CDC42EP2 gene (Sequence Analysis-All Coding Exons)	CDC42EP2	CDC42EP2, CEP2, BORG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42EP1 geni (Tüm Gen Dizi Analizi)	CDC42EP1 gene (Sequence Analysis-All Coding Exons)	CDC42EP1	CDC42EP1, MSE55, BORG5, CEP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42BPG geni (Tüm Gen Dizi Analizi)	CDC42BPG gene (Sequence Analysis-All Coding Exons)	CDC42BPG	CDC42BPG, MRCKG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42BPB geni (Tüm Gen Dizi Analizi)	CDC42BPB gene (Sequence Analysis-All Coding Exons)	CDC42BPB	CDC42BPB, MRCKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC42BPA geni (Tüm Gen Dizi Analizi)	CDC42BPA gene (Sequence Analysis-All Coding Exons)	CDC42BPA	CDC42BPA, PK428, MRCKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC40 geni (Tüm Gen Dizi Analizi)	CDC40 gene (Sequence Analysis-All Coding Exons)	CDC40	CDC40, PRP17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC37L1 geni (Tüm Gen Dizi Analizi)	CDC37L1 gene (Sequence Analysis-All Coding Exons)	CDC37L1	CDC37L1, HARC, FLJ20639	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC37 geni (Tüm Gen Dizi Analizi)	CDC37 gene (Sequence Analysis-All Coding Exons)	CDC37	CDC37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC34 geni (Tüm Gen Dizi Analizi)	CDC34 gene (Sequence Analysis-All Coding Exons)	CDC34	CDC34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC27 geni (Tüm Gen Dizi Analizi)	CDC27 gene (Sequence Analysis-All Coding Exons)	CDC27	CDC27	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC26 geni (Tüm Gen Dizi Analizi)	CDC26 gene (Sequence Analysis-All Coding Exons)	CDC26	CDC26, APC12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC25C geni (Tüm Gen Dizi Analizi)	CDC25C gene (Sequence Analysis-All Coding Exons)	CDC25C	CDC25C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC25B geni (Tüm Gen Dizi Analizi)	CDC25B gene (Sequence Analysis-All Coding Exons)	CDC25B	CDC25B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC25A geni (Tüm Gen Dizi Analizi)	CDC25A gene (Sequence Analysis-All Coding Exons)	CDC25A	CDC25A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC23 geni (Tüm Gen Dizi Analizi)	CDC23 gene (Sequence Analysis-All Coding Exons)	CDC23	CDC23, APC8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC20 geni (Tüm Gen Dizi Analizi)	CDC20 gene (Sequence Analysis-All Coding Exons)	CDC20	CDC20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC16 geni (Tüm Gen Dizi Analizi)	CDC16 gene (Sequence Analysis-All Coding Exons)	CDC16	CDC16, APC6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDC14B geni (Tüm Gen Dizi Analizi)	CDC14B gene (Sequence Analysis-All Coding Exons)	CDC14B	CDC14B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDA geni (Tüm Gen Dizi Analizi)	CDA gene (Sequence Analysis-All Coding Exons)	CDA	CDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD99L2 geni (Tüm Gen Dizi Analizi)	CD99L2 gene (Sequence Analysis-All Coding Exons)	CD99L2	CD99L2, CD99B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD99 geni (Tüm Gen Dizi Analizi)	CD99 gene (Sequence Analysis-All Coding Exons)	CD99	CD99, MIC2, MIC2X	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD99 geni (Tüm Gen Dizi Analizi)	CD99 gene (Sequence Analysis-All Coding Exons)	CD99	MIC2Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD93 geni (Tüm Gen Dizi Analizi)	CD93 gene (Sequence Analysis-All Coding Exons)	CD93	C1QR1, CD93, C1QR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD9 geni (Tüm Gen Dizi Analizi)	CD9 gene (Sequence Analysis-All Coding Exons)	CD9	CD9, MIC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD8B geni (Tüm Gen Dizi Analizi)	CD8B gene (Sequence Analysis-All Coding Exons)	CD8B	CD8B1, CD8B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNJ14 geni (Tüm Gen Dizi Analizi)	KCNJ14 gene (Sequence Analysis-All Coding Exons)	KCNJ14		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD86 geni (Tüm Gen Dizi Analizi)	CD86 gene (Sequence Analysis-All Coding Exons)	CD86	CD86, CD28LG2, LAB72	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD83 geni (Tüm Gen Dizi Analizi)	CD83 gene (Sequence Analysis-All Coding Exons)	CD83	CD83, HB15, BL11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD80 geni (Tüm Gen Dizi Analizi)	CD80 gene (Sequence Analysis-All Coding Exons)	CD80	CD80, CD28LG, LAB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD74 geni (Tüm Gen Dizi Analizi)	CD74 gene (Sequence Analysis-All Coding Exons)	CD74	CD74, DHLAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD72 geni (Tüm Gen Dizi Analizi)	CD72 gene (Sequence Analysis-All Coding Exons)	CD72	CD72, LYB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD70 geni (Tüm Gen Dizi Analizi)	CD70 gene (Sequence Analysis-All Coding Exons)	CD70	TNFSF7, CD70, CD27L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD7 geni (Tüm Gen Dizi Analizi)	CD7 gene (Sequence Analysis-All Coding Exons)	CD7	CD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD69 geni (Tüm Gen Dizi Analizi)	CD69 gene (Sequence Analysis-All Coding Exons)	CD69	CD69	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD68 geni (Tüm Gen Dizi Analizi)	CD68 gene (Sequence Analysis-All Coding Exons)	CD68	CD68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD63 geni (Tüm Gen Dizi Analizi)	CD63 gene (Sequence Analysis-All Coding Exons)	CD63	CD63, MLA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD6 geni (Tüm Gen Dizi Analizi)	CD6 gene (Sequence Analysis-All Coding Exons)	CD6	CD6, TP120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDSL geni (Tüm Gen Dizi Analizi)	CDSL gene (Sequence Analysis-All Coding Exons)	CDSL	CDSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDS8 geni (Tüm Gen Dizi Analizi)	CDS8 gene (Sequence Analysis-All Coding Exons)	CDS8	CDS8, LFA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNIP2 geni (Tüm Gen Dizi Analizi)	KCNIP2 gene (Sequence Analysis-All Coding Exons)	KCNIP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNIP1 geni (Tüm Gen Dizi Analizi)	KCNIP1 gene (Sequence Analysis-All Coding Exons)	KCNIP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH8 geni (Tüm Gen Dizi Analizi)	KCNH8 gene (Sequence Analysis-All Coding Exons)	KCNH8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDS3 geni (Tüm Gen Dizi Analizi)	CDS3 gene (Sequence Analysis-All Coding Exons)	CDS3	CDS3, MOX44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDS2 geni (Tüm Gen Dizi Analizi)	CDS2 gene (Sequence Analysis-All Coding Exons)	CDS2	CDS2, CDW52	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD5 geni (Tüm Gen Dizi Analizi)	CD5 gene (Sequence Analysis-All Coding Exons)	CD5	CD5, LEU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNH4 geni (Tüm Gen Dizi Analizi)	KCNH4 gene (Sequence Analysis-All Coding Exons)	KCNH4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD48 geni (Tüm Gen Dizi Analizi)	CD48 gene (Sequence Analysis-All Coding Exons)	CD48	CD48, BCM1, BLAST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD47 geni (Tüm Gen Dizi Analizi)	CD47 gene (Sequence Analysis-All Coding Exons)	CD47	CD47, MER6, IAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD3EAP geni (Tüm Gen Dizi Analizi)	CD3EAP gene (Sequence Analysis-All Coding Exons)	CD3EAP	CD3EAP, ASE1, PAF49	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD38 geni (Tüm Gen Dizi Analizi)	CD38 gene (Sequence Analysis-All Coding Exons)	CD38	CD38	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD37 geni (Tüm Gen Dizi Analizi)	CD37 gene (Sequence Analysis-All Coding Exons)	CD37	CD37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD34 geni (Tüm Gen Dizi Analizi)	CD34 gene (Sequence Analysis-All Coding Exons)	CD34	CD34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD33 geni (Tüm Gen Dizi Analizi)	CD33 gene (Sequence Analysis-All Coding Exons)	CD33	CD33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD302 geni (Tüm Gen Dizi Analizi)	CD302 gene (Sequence Analysis-All Coding Exons)	CD302	CD302, DCL1, KIAA0022	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KCNG2 geni (Tüm Gen Dizi Analizi)	KCNG2 gene (Sequence Analysis-All Coding Exons)	KCNG2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300LG geni (Tüm Gen Dizi Analizi)	CD300LG gene (Sequence Analysis-All Coding Exons)	CD300LG	CD300LG, TREM4, CLM9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300LF geni (Tüm Gen Dizi Analizi)	CD300LF gene (Sequence Analysis-All Coding Exons)	CD300LF	CD300LF, IGSF13, IREM1, CLM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300LD geni (Tüm Gen Dizi Analizi)	CD300LD gene (Sequence Analysis-All Coding Exons)	CD300LD	CD300LD, CD300D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300LB geni (Tüm Gen Dizi Analizi)	CD300LB gene (Sequence Analysis-All Coding Exons)	CD300LB	CD300LB, TREM5, IREM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300E geni (Tüm Gen Dizi Analizi)	CD300E gene (Sequence Analysis-All Coding Exons)	CD300E	CD300E, CD300LF, IREM2, CLM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300C geni (Tüm Gen Dizi Analizi)	CD300C gene (Sequence Analysis-All Coding Exons)	CD300C	CMRF35, CMRF35A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD300A geni (Tüm Gen Dizi Analizi)	CD300A gene (Sequence Analysis-All Coding Exons)	CD300A	CMRF35H, CMRF35H9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD2BP2 geni (Tüm Gen Dizi Analizi)	CD2BP2 gene (Sequence Analysis-All Coding Exons)	CD2BP2	CD2BP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD28 geni (Tüm Gen Dizi Analizi)	CD28 gene (Sequence Analysis-All Coding Exons)	CD28	CD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD28 geni (Tüm Gen Dizi Analizi)	CD28 gene (Sequence Analysis-All Coding Exons)	CD28	CD28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD276 geni (Tüm Gen Dizi Analizi)	CD276 gene (Sequence Analysis-All Coding Exons)	CD276	CD276, B7H3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD274 geni (Tüm Gen Dizi Analizi)	CD274 gene (Sequence Analysis-All Coding Exons)	CD274	PDCD1LG1, B7H1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD248 geni (Tüm Gen Dizi Analizi)	CD248 gene (Sequence Analysis-All Coding Exons)	CD248	CD248, TEM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD24 geni (Tüm Gen Dizi Analizi)	CD24 gene (Sequence Analysis-All Coding Exons)	CD24	CD24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD226 geni (Tüm Gen Dizi Analizi)	CD226 gene (Sequence Analysis-All Coding Exons)	CD226	DNAM1, CD226	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD22 geni (Tüm Gen Dizi Analizi)	CD22 gene (Sequence Analysis-All Coding Exons)	CD22	CD22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD200R1 geni (Tüm Gen Dizi Analizi)	CD200R1 gene (Sequence Analysis-All Coding Exons)	CD200R1	CD200R1, MOX2R, OX2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD200 geni (Tüm Gen Dizi Analizi)	CD200 gene (Sequence Analysis-All Coding Exons)	CD200	MOX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD2 geni (Tüm Gen Dizi Analizi)	CD2 gene (Sequence Analysis-All Coding Exons)	CD2	CD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD1E geni (Tüm Gen Dizi Analizi)	CD1E gene (Sequence Analysis-All Coding Exons)	CD1E	CD1E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD1D geni (Tüm Gen Dizi Analizi)	CD1D gene (Sequence Analysis-All Coding Exons)	CD1D	CD1D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD1C geni (Tüm Gen Dizi Analizi)	CD1C gene (Sequence Analysis-All Coding Exons)	CD1C	CD1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD1B geni (Tüm Gen Dizi Analizi)	CD1B gene (Sequence Analysis-All Coding Exons)	CD1B	CD1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD1A geni (Tüm Gen Dizi Analizi)	CD1A gene (Sequence Analysis-All Coding Exons)	CD1A	CD1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD180 geni (Tüm Gen Dizi Analizi)	CD180 gene (Sequence Analysis-All Coding Exons)	CD180	CD180, LY64, RP105	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD177 geni (Tüm Gen Dizi Analizi)	CD177 gene (Sequence Analysis-All Coding Exons)	CD177	NB1, PRV1, CD177, HNA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD163L1 geni (Tüm Gen Dizi Analizi)	CD163L1 gene (Sequence Analysis-All Coding Exons)	CD163L1	CD164L1, CD163B, M160	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD163 geni (Tüm Gen Dizi Analizi)	CD163 gene (Sequence Analysis-All Coding Exons)	CD163	CD163	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD160 geni (Tüm Gen Dizi Analizi)	CD160 gene (Sequence Analysis-All Coding Exons)	CD160	CD160, BY55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CD14 geni (Tüm Gen Dizi Analizi)	CD14 gene (Sequence Analysis-All Coding Exons)	CD14	CD14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD109 geni (Tüm Gen Dizi Analizi)	CD109 gene (Sequence Analysis-All Coding Exons)	CD109	CD109	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT7 geni (Tüm Gen Dizi Analizi)	CCT7 gene (Sequence Analysis-All Coding Exons)	CCT7	CCT7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT6B geni (Tüm Gen Dizi Analizi)	CCT6B gene (Sequence Analysis-All Coding Exons)	CCT6B	CCT6B, CCT22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
KAZALD1 geni (Tüm Gen Dizi Analizi)	KAZALD1 gene (Sequence Analysis-All Coding Exons)	KAZALD1		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT6A geni (Tüm Gen Dizi Analizi)	CCT6A gene (Sequence Analysis-All Coding Exons)	CCT6A	CCT6A, CCT6, HTR3, TCP20	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT4 geni (Tüm Gen Dizi Analizi)	CCT4 gene (Sequence Analysis-All Coding Exons)	CCT4	CCT4, CCTD, SRB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT3 geni (Tüm Gen Dizi Analizi)	CCT3 gene (Sequence Analysis-All Coding Exons)	CCT3	CCT3, TRIC5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCT2 geni (Tüm Gen Dizi Analizi)	CCT2 gene (Sequence Analysis-All Coding Exons)	CCT2	CCT2, CCTB	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCSAP geni (Tüm Gen Dizi Analizi)	CCSAP gene (Sequence Analysis-All Coding Exons)	CCSAP	CCSAP, CSAP, C1orf96	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCS geni (Tüm Gen Dizi Analizi)	CCS gene (Sequence Analysis-All Coding Exons)	CCS	CCS	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCRL2 geni (Tüm Gen Dizi Analizi)	CCRL2 gene (Sequence Analysis-All Coding Exons)	CCRL2	CCRL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR9 geni (Tüm Gen Dizi Analizi)	CCR9 gene (Sequence Analysis-All Coding Exons)	CCR9	CCR9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR8 geni (Tüm Gen Dizi Analizi)	CCR8 gene (Sequence Analysis-All Coding Exons)	CCR8	CCR8, CMKBR8, CMKBR2, CKR-L1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR7 geni (Tüm Gen Dizi Analizi)	CCR7 gene (Sequence Analysis-All Coding Exons)	CCR7	CCR7, CMKBR7, EBI1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR6 geni (Tüm Gen Dizi Analizi)	CCR6 gene (Sequence Analysis-All Coding Exons)	CCR6	CCR6, CMKBR6, CKRL3, GPR29, GPRCY4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR6 geni (Tüm Gen Dizi Analizi)	CCR6 gene (Sequence Analysis-All Coding Exons)	CCR6	CCR6, CMKBR6, CKRL3, GPR29, GPRCY4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR4 geni (Tüm Gen Dizi Analizi)	CCR4 gene (Sequence Analysis-All Coding Exons)	CCR4	CCR4, CKR4, CMKBR4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR3 geni (Tüm Gen Dizi Analizi)	CCR3 gene (Sequence Analysis-All Coding Exons)	CCR3	CCR3, CKR3, CMKBR3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR10 geni (Tüm Gen Dizi Analizi)	CCR10 gene (Sequence Analysis-All Coding Exons)	CCR10	CCR10, GPR2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCR1 geni (Tüm Gen Dizi Analizi)	CCR1 gene (Sequence Analysis-All Coding Exons)	CCR1	CCR1, CMKBR1, CKR1, HM145	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCPG1 geni (Tüm Gen Dizi Analizi)	CCPG1 gene (Sequence Analysis-All Coding Exons)	CCPG1	CCPG1, KIAA1254, CPR8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCP110 geni (Tüm Gen Dizi Analizi)	CCP110 gene (Sequence Analysis-All Coding Exons)	CCP110	CCP110, CP110, KIAA0419	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNY geni (Tüm Gen Dizi Analizi)	CCNY gene (Sequence Analysis-All Coding Exons)	CCNY	CCNY, CFP1, CCNX, C10orf9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNT2 geni (Tüm Gen Dizi Analizi)	CCNT2 gene (Sequence Analysis-All Coding Exons)	CCNT2	CCNT2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNT1 geni (Tüm Gen Dizi Analizi)	CCNT1 gene (Sequence Analysis-All Coding Exons)	CCNT1	CCNT1, CCNT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNL2 geni (Tüm Gen Dizi Analizi)	CCNL2 gene (Sequence Analysis-All Coding Exons)	CCNL2	CCNL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNL1 geni (Tüm Gen Dizi Analizi)	CCNL1 gene (Sequence Analysis-All Coding Exons)	CCNL1	CCNL1, ANA6A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNK geni (Tüm Gen Dizi Analizi)	CCNK gene (Sequence Analysis-All Coding Exons)	CCNK	CCNK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNH geni (Tüm Gen Dizi Analizi)	CCNH gene (Sequence Analysis-All Coding Exons)	CCNH	CCNH, CAK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNG2 geni (Tüm Gen Dizi Analizi)	CCNG2 gene (Sequence Analysis-All Coding Exons)	CCNG2	CCNG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNG1 geni (Tüm Gen Dizi Analizi)	CCNG1 gene (Sequence Analysis-All Coding Exons)	CCNG1	CCNG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNF geni (Tüm Gen Dizi Analizi)	CCNF gene (Sequence Analysis-All Coding Exons)	CCNF	CCNF, FBX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNE2 geni (Tüm Gen Dizi Analizi)	CCNE2 gene (Sequence Analysis-All Coding Exons)	CCNE2	CCNE2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNE1 geni (Tüm Gen Dizi Analizi)	CCNE1 gene (Sequence Analysis-All Coding Exons)	CCNE1	CCNE1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNDBP1 geni (Tüm Gen Dizi Analizi)	CCNDBP1 gene (Sequence Analysis-All Coding Exons)	CCNDBP1	CCNDBP1, GCIP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCND3 geni (Tüm Gen Dizi Analizi)	CCND3 gene (Sequence Analysis-All Coding Exons)	CCND3	CCND3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNC geni (Tüm Gen Dizi Analizi)	CCNC gene (Sequence Analysis-All Coding Exons)	CCNC	CCNC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNB3 geni (Tüm Gen Dizi Analizi)	CCNB3 gene (Sequence Analysis-All Coding Exons)	CCNB3	CCNB3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNB2 geni (Tüm Gen Dizi Analizi)	CCNB2 gene (Sequence Analysis-All Coding Exons)	CCNB2	CCNB2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNB1IP1 geni (Tüm Gen Dizi Analizi)	CCNB1IP1 gene (Sequence Analysis-All Coding Exons)	CCNB1IP1	C14orf18, HEI10	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNB1 geni (Tüm Gen Dizi Analizi)	CCNB1 gene (Sequence Analysis-All Coding Exons)	CCNB1	CCNB1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNA2 geni (Tüm Gen Dizi Analizi)	CCNA2 gene (Sequence Analysis-All Coding Exons)	CCNA2	CCNA, CCN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCNA1 geni (Tüm Gen Dizi Analizi)	CCNA1 gene (Sequence Analysis-All Coding Exons)	CCNA1	CCNA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL8 geni (Tüm Gen Dizi Analizi)	CCL8 gene (Sequence Analysis-All Coding Exons)	CCL8	CCL8, SCYA8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL7 geni (Tüm Gen Dizi Analizi)	CCL7 gene (Sequence Analysis-All Coding Exons)	CCL7	CCL7, SCYA7, MCP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL4L2 geni (Tüm Gen Dizi Analizi)	CCL4L2 gene (Sequence Analysis-All Coding Exons)	CCL4L2	CCL4L2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL4L1 geni (Tüm Gen Dizi Analizi)	CCL4L1 gene (Sequence Analysis-All Coding Exons)	CCL4L1	CCL4L, SCYA4L, LAG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL4 geni (Tüm Gen Dizi Analizi)	CCL4 gene (Sequence Analysis-All Coding Exons)	CCL4	CCL4, SCYA4, ACT2, MIP1B, 6T244.1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL28 geni (Tüm Gen Dizi Analizi)	CCL28 gene (Sequence Analysis-All Coding Exons)	CCL28	CCL28	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL27 geni (Tüm Gen Dizi Analizi)	CCL27 gene (Sequence Analysis-All Coding Exons)	CCL27	CCL27, SCYA27, CCL27, ILC, CTACK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL26 geni (Tüm Gen Dizi Analizi)	CCL26 gene (Sequence Analysis-All Coding Exons)	CCL26	CCL26, SCYA26	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL25 geni (Tüm Gen Dizi Analizi)	CCL25 gene (Sequence Analysis-All Coding Exons)	CCL25	CCL25, SCYA25, TECK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL24 geni (Tüm Gen Dizi Analizi)	CCL24 gene (Sequence Analysis-All Coding Exons)	CCL24	CCL24, SCYA24, MPIF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL23 geni (Tüm Gen Dizi Analizi)	CCL23 gene (Sequence Analysis-All Coding Exons)	CCL23	CCL23, SCYA23	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL22 geni (Tüm Gen Dizi Analizi)	CCL22 gene (Sequence Analysis-All Coding Exons)	CCL22	CCL22, SCYA22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL21 geni (Tüm Gen Dizi Analizi)	CCL21 gene (Sequence Analysis-All Coding Exons)	CCL21	CCL21, SCYA21, SLC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL20 geni (Tüm Gen Dizi Analizi)	CCL20 gene (Sequence Analysis-All Coding Exons)	CCL20	CCL20, SCYA20, MIP3A, LARC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL19 geni (Tüm Gen Dizi Analizi)	CCL19 gene (Sequence Analysis-All Coding Exons)	CCL19	CCL19, SCYA19, ELC, MIP3B	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL18 geni (Tüm Gen Dizi Analizi)	CCL18 gene (Sequence Analysis-All Coding Exons)	CCL18	CCL18, SCYA18, PARC, AMAC1, DCCK1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL17 geni (Tüm Gen Dizi Analizi)	CCL17 gene (Sequence Analysis-All Coding Exons)	CCL17	CCL17, SCYA17, TARC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL16 geni (Tüm Gen Dizi Analizi)	CCL16 gene (Sequence Analysis-All Coding Exons)	CCL16	CCL16, SCYA16, NCC4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL15 geni (Tüm Gen Dizi Analizi)	CCL15 gene (Sequence Analysis-All Coding Exons)	CCL15	CCL15, SCYA15, NCC3, LKN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL14 geni (Tüm Gen Dizi Analizi)	CCL14 gene (Sequence Analysis-All Coding Exons)	CCL14	CCL14, SCYA14, NCC2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL13 geni (Tüm Gen Dizi Analizi)	CCL13 gene (Sequence Analysis-All Coding Exons)	CCL13	CCL13, SCYA13, NCC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL1 geni (Tüm Gen Dizi Analizi)	CCL1 gene (Sequence Analysis-All Coding Exons)	CCL1	CCL1, SCYA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCKBR geni (Tüm Gen Dizi Analizi)	CCKBR gene (Sequence Analysis-All Coding Exons)	CCKBR	CCKBR, GASR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCKAR geni (Tüm Gen Dizi Analizi)	CCKAR gene (Sequence Analysis-All Coding Exons)	CCKAR	CCKAR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCK geni (Tüm Gen Dizi Analizi)	CCK gene (Sequence Analysis-All Coding Exons)	CCK	CCK	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCIN geni (Tüm Gen Dizi Analizi)	CCIN gene (Sequence Analysis-All Coding Exons)	CCIN	CCIN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCHCR1 geni (Tüm Gen Dizi Analizi)	CCHCR1 gene (Sequence Analysis-All Coding Exons)	CCHCR1	CCHCR1, HCR, C6ORF18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC91 geni (Tüm Gen Dizi Analizi)	CCDC91 gene (Sequence Analysis-All Coding Exons)	CCDC91	CCDC91	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC88B geni (Tüm Gen Dizi Analizi)	CCDC88B gene (Sequence Analysis-All Coding Exons)	CCDC88B	CCDC88B, GIPIE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC86 geni (Tüm Gen Dizi Analizi)	CCDC86 gene (Sequence Analysis-All Coding Exons)	CCDC86	CCDC86, CYCLON	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC85B geni (Tüm Gen Dizi Analizi)	CCDC85B gene (Sequence Analysis-All Coding Exons)	CCDC85B	DIPA	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC80 geni (Tüm Gen Dizi Analizi)	CCDC80 gene (Sequence Analysis-All Coding Exons)	CCDC80	URB, SSG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC73 geni (Tüm Gen Dizi Analizi)	CCDC73 gene (Sequence Analysis-All Coding Exons)	CCDC73	CCDC73	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC68 geni (Tüm Gen Dizi Analizi)	CCDC68 gene (Sequence Analysis-All Coding Exons)	CCDC68	CCDC68	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC62 geni (Tüm Gen Dizi Analizi)	CCDC62 gene (Sequence Analysis-All Coding Exons)	CCDC62	CCDC62, ERAP75	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC6 geni (Tüm Gen Dizi Analizi)	CCDC6 gene (Sequence Analysis-All Coding Exons)	CCDC6	CCDC6, D10S170, H4, TST1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC34 geni (Tüm Gen Dizi Analizi)	CCDC34 gene (Sequence Analysis-All Coding Exons)	CCDC34	CCDC34	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC28A geni (Tüm Gen Dizi Analizi)	CCDC28A gene (Sequence Analysis-All Coding Exons)	CCDC28A	CCDC28A, C6orf80	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC26 geni (Tüm Gen Dizi Analizi)	CCDC26 gene (Sequence Analysis-All Coding Exons)	CCDC26	CCD26, RAM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC183 geni (Tüm Gen Dizi Analizi)	CCDC183 gene (Sequence Analysis-All Coding Exons)	CCDC183	CCDC183, KIAA1984	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC141 geni (Tüm Gen Dizi Analizi)	CCDC141 gene (Sequence Analysis-All Coding Exons)	CCDC141	CCDC141, CAMDI	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC14 geni (Tüm Gen Dizi Analizi)	CCDC14 gene (Sequence Analysis-All Coding Exons)	CCDC14	CCDC14	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC137 geni (Tüm Gen Dizi Analizi)	CCDC137 gene (Sequence Analysis-All Coding Exons)	CCDC137	CCDC137	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC136 geni (Tüm Gen Dizi Analizi)	CCDC136 gene (Sequence Analysis-All Coding Exons)	CCDC136	CCDC136, NAG6, KIAA1793	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC125 geni (Tüm Gen Dizi Analizi)	CCDC125 gene (Sequence Analysis-All Coding Exons)	CCDC125	CCDC125, KENAE	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC122 geni (Tüm Gen Dizi Analizi)	CCDC122 gene (Sequence Analysis-All Coding Exons)	CCDC122	CCDC122	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC120 geni (Tüm Gen Dizi Analizi)	CCDC120 gene (Sequence Analysis-All Coding Exons)	CCDC120	CCDC120	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC113 geni (Tüm Gen Dizi Analizi)	CCDC113 gene (Sequence Analysis-All Coding Exons)	CCDC113	CCDC113	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCDC110 geni (Tüm Gen Dizi Analizi)	CCDC110 gene (Sequence Analysis-All Coding Exons)	CCDC110	KMHN1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CCDC106 geni (Tüm Gen Dizi Analizi)	CCDC106 gene (Sequence Analysis-All Coding Exons)	CCDC106	CCDC106	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCAR2 geni (Tüm Gen Dizi Analizi)	CCAR2 gene (Sequence Analysis-All Coding Exons)	CCAR2	DBC1, KIAA1967	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCAR1 geni (Tüm Gen Dizi Analizi)	CCAR1 gene (Sequence Analysis-All Coding Exons)	CCAR1	CCAR1, CARP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBY1 geni (Tüm Gen Dizi Analizi)	CBY1 gene (Sequence Analysis-All Coding Exons)	CBY1	CBY, C22orf2, HSSB15A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX8 geni (Tüm Gen Dizi Analizi)	CBX8 gene (Sequence Analysis-All Coding Exons)	CBX8	CBX8, PC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX7 geni (Tüm Gen Dizi Analizi)	CBX7 gene (Sequence Analysis-All Coding Exons)	CBX7	CBX7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX5 geni (Tüm Gen Dizi Analizi)	CBX5 gene (Sequence Analysis-All Coding Exons)	CBX5	CBX5, HP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX4 geni (Tüm Gen Dizi Analizi)	CBX4 gene (Sequence Analysis-All Coding Exons)	CBX4	CBX4, PC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX3 geni (Tüm Gen Dizi Analizi)	CBX3 gene (Sequence Analysis-All Coding Exons)	CBX3	CBX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBWD3 geni (Tüm Gen Dizi Analizi)	CBWD3 gene (Sequence Analysis-All Coding Exons)	CBWD3	CBWD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBWD2 geni (Tüm Gen Dizi Analizi)	CBWD2 gene (Sequence Analysis-All Coding Exons)	CBWD2	CBWD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBWD1 geni (Tüm Gen Dizi Analizi)	CBWD1 gene (Sequence Analysis-All Coding Exons)	CBWD1	CBWD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBR3 geni (Tüm Gen Dizi Analizi)	CBR3 gene (Sequence Analysis-All Coding Exons)	CBR3	CBR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ITGB8 geni (Tüm Gen Dizi Analizi)	ITGB8 gene (Sequence Analysis-All Coding Exons)	ITGB8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBR1 geni (Tüm Gen Dizi Analizi)	CBR1 gene (Sequence Analysis-All Coding Exons)	CBR1	CBR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLN4 geni (Tüm Gen Dizi Analizi)	CBLN4 gene (Sequence Analysis-All Coding Exons)	CBLN4	CBLN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLN3 geni (Tüm Gen Dizi Analizi)	CBLN3 gene (Sequence Analysis-All Coding Exons)	CBLN3	CBLN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLN2 geni (Tüm Gen Dizi Analizi)	CBLN2 gene (Sequence Analysis-All Coding Exons)	CBLN2	CBLN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLN1 geni (Tüm Gen Dizi Analizi)	CBLN1 gene (Sequence Analysis-All Coding Exons)	CBLN1	CLN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLL1 geni (Tüm Gen Dizi Analizi)	CBLL1 gene (Sequence Analysis-All Coding Exons)	CBLL1	CBLL1, HAKAI, RNF188	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLC geni (Tüm Gen Dizi Analizi)	CBLC gene (Sequence Analysis-All Coding Exons)	CBLC	CBLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBLB geni (Tüm Gen Dizi Analizi)	CBLB gene (Sequence Analysis-All Coding Exons)	CBLB	CBLB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBFA2T3 geni (Tüm Gen Dizi Analizi)	CBFA2T3 gene (Sequence Analysis-All Coding Exons)	CBFA2T3	CBFA2T3, MTGR2, MTG16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBFA2T2 geni (Tüm Gen Dizi Analizi)	CBFA2T2 gene (Sequence Analysis-All Coding Exons)	CBFA2T2	CBFA2T2, MTGR1, ETO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAV2 geni (Tüm Gen Dizi Analizi)	CAV2 gene (Sequence Analysis-All Coding Exons)	CAV2	CAV2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSPERG geni (Tüm Gen Dizi Analizi)	CATSPERG gene (Sequence Analysis-All Coding Exons)	CATSPERG	CATSPERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSPERB geni (Tüm Gen Dizi Analizi)	CATSPERB gene (Sequence Analysis-All Coding Exons)	CATSPERB	CATSPERB, C14orf161	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSPER4 geni (Tüm Gen Dizi Analizi)	CATSPER4 gene (Sequence Analysis-All Coding Exons)	CATSPER4	CATSPER4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSPER3 geni (Tüm Gen Dizi Analizi)	CATSPER3 gene (Sequence Analysis-All Coding Exons)	CATSPER3	CATSPER3, CACRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CATSPER2 geni (Tüm Gen Dizi Analizi)	CATSPER2 gene (Sequence Analysis-All Coding Exons)	CATSPER2	CATSPER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASZ1 geni (Tüm Gen Dizi Analizi)	CASZ1 gene (Sequence Analysis-All Coding Exons)	CASZ1	CASZ1, SRG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASTOR2 geni (Tüm Gen Dizi Analizi)	CASTOR2 gene (Sequence Analysis-All Coding Exons)	CASTOR2	GATSL2, CASTOR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASTOR1 geni (Tüm Gen Dizi Analizi)	CASTOR1 gene (Sequence Analysis-All Coding Exons)	CASTOR1	GATSL3, CASTOR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP9 geni (Tüm Gen Dizi Analizi)	CASP9 gene (Sequence Analysis-All Coding Exons)	CASP9	CASP9, APAF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP8AP2 geni (Tüm Gen Dizi Analizi)	CASP8AP2 gene (Sequence Analysis-All Coding Exons)	CASP8AP2	CASP8AP2, FLASH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP7 geni (Tüm Gen Dizi Analizi)	CASP7 gene (Sequence Analysis-All Coding Exons)	CASP7	CASP7, MCH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP6 geni (Tüm Gen Dizi Analizi)	CASP6 gene (Sequence Analysis-All Coding Exons)	CASP6	CASP6, MCH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP5 geni (Tüm Gen Dizi Analizi)	CASP5 gene (Sequence Analysis-All Coding Exons)	CASP5	CASP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP4 geni (Tüm Gen Dizi Analizi)	CASP4 gene (Sequence Analysis-All Coding Exons)	CASP4	CASP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP3 geni (Tüm Gen Dizi Analizi)	CASP3 gene (Sequence Analysis-All Coding Exons)	CASP3	CASP3, CPP32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP2 geni (Tüm Gen Dizi Analizi)	CASP2 gene (Sequence Analysis-All Coding Exons)	CASP2	CASP2, NEDD2, ICH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASP1 geni (Tüm Gen Dizi Analizi)	CASP1 gene (Sequence Analysis-All Coding Exons)	CASP1	CASP1, IL1BC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASKIN2 geni (Tüm Gen Dizi Analizi)	CASKIN2 gene (Sequence Analysis-All Coding Exons)	CASKIN2	CASKIN2, KIAA1139	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASKIN1 geni (Tüm Gen Dizi Analizi)	CASKIN1 gene (Sequence Analysis-All Coding Exons)	CASKIN1	CASKIN1, KIAA1306	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASD1 geni (Tüm Gen Dizi Analizi)	CASD1 gene (Sequence Analysis-All Coding Exons)	CASD1	CASD1, C7orf12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASC3 geni (Tüm Gen Dizi Analizi)	CASC3 gene (Sequence Analysis-All Coding Exons)	CASC3	MLN51, CASC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASC2 geni (Tüm Gen Dizi Analizi)	CASC2 gene (Sequence Analysis-All Coding Exons)	CASC2	CASC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASC15 geni (Tüm Gen Dizi Analizi)	CASC15 gene (Sequence Analysis-All Coding Exons)	CASC15	CASC15, LINC00340	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASC1 geni (Tüm Gen Dizi Analizi)	CASC1 gene (Sequence Analysis-All Coding Exons)	CASC1	CASC1, LAS1, PPP1R54	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARS geni (Tüm Gen Dizi Analizi)	CARS gene (Sequence Analysis-All Coding Exons)	CARS	CARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARNS1 geni (Tüm Gen Dizi Analizi)	CARNS1 gene (Sequence Analysis-All Coding Exons)	CARNS1	CARNS1, ATPGD1, KIAA1394	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARNMT1 geni (Tüm Gen Dizi Analizi)	CARNMT1 gene (Sequence Analysis-All Coding Exons)	CARNMT1	CARNMT1, C9orf41, UPF0586	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARMIL3 geni (Tüm Gen Dizi Analizi)	CARMIL3 gene (Sequence Analysis-All Coding Exons)	CARMIL3	LRRC16B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARMIL2 geni (Tüm Gen Dizi Analizi)	CARMIL2 gene (Sequence Analysis-All Coding Exons)	CARMIL2	CARMIL2, RLTPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARMIL1 geni (Tüm Gen Dizi Analizi)	CARMIL1 gene (Sequence Analysis-All Coding Exons)	CARMIL1	LRRC16A, LRRC16, CARML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARM1 geni (Tüm Gen Dizi Analizi)	CARM1 gene (Sequence Analysis-All Coding Exons)	CARM1	CARM1, PRMT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARHSP1 geni (Tüm Gen Dizi Analizi)	CARHSP1 gene (Sequence Analysis-All Coding Exons)	CARHSP1	CARHSP1, CRHSP24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARF geni (Tüm Gen Dizi Analizi)	CARF gene (Sequence Analysis-All Coding Exons)	CARF	ALS2CR8, CARF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD8 geni (Tüm Gen Dizi Analizi)	CARD8 gene (Sequence Analysis-All Coding Exons)	CARD8	CARD8, TUCAN, KIAA0955, NDPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD6 geni (Tüm Gen Dizi Analizi)	CARD6 gene (Sequence Analysis-All Coding Exons)	CARD6	CARD6, CINCIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD18 geni (Tüm Gen Dizi Analizi)	CARD18 gene (Sequence Analysis-All Coding Exons)	CARD18	CARD18, ICEBERG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD17 geni (Tüm Gen Dizi Analizi)	CARD17 gene (Sequence Analysis-All Coding Exons)	CARD17	INCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD16 geni (Tüm Gen Dizi Analizi)	CARD16 gene (Sequence Analysis-All Coding Exons)	CARD16	CARD16, COP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ISL2 geni (Tüm Gen Dizi Analizi)	ISL2 gene (Sequence Analysis-All Coding Exons)	ISL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CARD10 geni (Tüm Gen Dizi Analizi)	CARD10 gene (Sequence Analysis-All Coding Exons)	CARD10	CARD10, CARMA3, BIMP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPZB geni (Tüm Gen Dizi Analizi)	CAPZB gene (Sequence Analysis-All Coding Exons)	CAPZB	CAPZB, CAPPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPZA3 geni (Tüm Gen Dizi Analizi)	CAPZA3 gene (Sequence Analysis-All Coding Exons)	CAPZA3	CAPZA3, CAPPA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPZA2 geni (Tüm Gen Dizi Analizi)	CAPZA2 gene (Sequence Analysis-All Coding Exons)	CAPZA2	CAPZA2, CAPPA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPZA1 geni (Tüm Gen Dizi Analizi)	CAPZA1 gene (Sequence Analysis-All Coding Exons)	CAPZA1	CAPZA1, CAPPA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPS2 geni (Tüm Gen Dizi Analizi)	CAPS2 gene (Sequence Analysis-All Coding Exons)	CAPS2	CAPS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPS geni (Tüm Gen Dizi Analizi)	CAPS gene (Sequence Analysis-All Coding Exons)	CAPS	CAPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPRIN2 geni (Tüm Gen Dizi Analizi)	CAPRIN2 gene (Sequence Analysis-All Coding Exons)	CAPRIN2	CAPRIN2, C1QDC1, EEG1, KIAA1873	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPRIN1 geni (Tüm Gen Dizi Analizi)	CAPRIN1 gene (Sequence Analysis-All Coding Exons)	CAPRIN1	CAPRIN1, M11S1, GPIAP1, GPIP137	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPNS1 geni (Tüm Gen Dizi Analizi)	CAPNS1 gene (Sequence Analysis-All Coding Exons)	CAPNS1	CAPNS1, CAPN4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN9 geni (Tüm Gen Dizi Analizi)	CAPN9 gene (Sequence Analysis-All Coding Exons)	CAPN9	CAPN9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN7 geni (Tüm Gen Dizi Analizi)	CAPN7 gene (Sequence Analysis-All Coding Exons)	CAPN7	CAPN7, PALBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN6 geni (Tüm Gen Dizi Analizi)	CAPN6 gene (Sequence Analysis-All Coding Exons)	CAPN6	CAPN6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN2 geni (Tüm Gen Dizi Analizi)	CAPN2 gene (Sequence Analysis-All Coding Exons)	CAPN2	CAPN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN15 geni (Tüm Gen Dizi Analizi)	CAPN15 gene (Sequence Analysis-All Coding Exons)	CAPN15	SOLH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN12 geni (Tüm Gen Dizi Analizi)	CAPN12 gene (Sequence Analysis-All Coding Exons)	CAPN12	CAPN12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN11 geni (Tüm Gen Dizi Analizi)	CAPN11 gene (Sequence Analysis-All Coding Exons)	CAPN11	CAPN11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPG geni (Tüm Gen Dizi Analizi)	CAPG gene (Sequence Analysis-All Coding Exons)	CAPG	CAPG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CANX geni (Tüm Gen Dizi Analizi)	CANX gene (Sequence Analysis-All Coding Exons)	CANX	CANX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAND2 geni (Tüm Gen Dizi Analizi)	CAND2 gene (Sequence Analysis-All Coding Exons)	CAND2	CAND2, TIP120B, KIAA0667	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAND1 geni (Tüm Gen Dizi Analizi)	CAND1 gene (Sequence Analysis-All Coding Exons)	CAND1	CAND1, TIP120A, TIP120, KIAA0829	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMTA2 geni (Tüm Gen Dizi Analizi)	CAMTA2 gene (Sequence Analysis-All Coding Exons)	CAMTA2	CAMTA2, KIAA0909	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMSAP3 geni (Tüm Gen Dizi Analizi)	CAMSAP3 gene (Sequence Analysis-All Coding Exons)	CAMSAP3	NEZHA, KIAA1543	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMSAP2 geni (Tüm Gen Dizi Analizi)	CAMSAP2 gene (Sequence Analysis-All Coding Exons)	CAMSAP2	CAMSAP1L, CAMSAP2, KIAA1078	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMSAP1 geni (Tüm Gen Dizi Analizi)	CAMSAP1 gene (Sequence Analysis-All Coding Exons)	CAMSAP1	CAMSAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMP geni (Tüm Gen Dizi Analizi)	CAMP gene (Sequence Analysis-All Coding Exons)	CAMP	CAMP, FALL39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMLG geni (Tüm Gen Dizi Analizi)	CAMLG gene (Sequence Analysis-All Coding Exons)	CAMLG	CAMLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMKV geni (Tüm Gen Dizi Analizi)	CAMKV gene (Sequence Analysis-All Coding Exons)	CAMKV	CAMKV, 1G5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMKMT geni (Tüm Gen Dizi Analizi)	CAMKMT gene (Sequence Analysis-All Coding Exons)	CAMKMT	CAMKMT, C2orf34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CAMKMT geni (Tüm Gen Dizi Analizi)	CAMKMT gene (Sequence Analysis-All Coding Exons)	CAMKMT	CAMKMT, C2orf34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMKMT geni (Tüm Gen Dizi Analizi)	CAMKMT gene (Sequence Analysis-All Coding Exons)	CAMKMT	CAMKMT, C2orf34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMKK2 geni (Tüm Gen Dizi Analizi)	CAMKK2 gene (Sequence Analysis-All Coding Exons)	CAMKK2	CAMKK2, KIAA0787, CAMKKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK4 geni (Tüm Gen Dizi Analizi)	CAMK4 gene (Sequence Analysis-All Coding Exons)	CAMK4	CAMK4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2N2 geni (Tüm Gen Dizi Analizi)	CAMK2N2 gene (Sequence Analysis-All Coding Exons)	CAMK2N2	CAMK2N2, CAMKIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2N1 geni (Tüm Gen Dizi Analizi)	CAMK2N1 gene (Sequence Analysis-All Coding Exons)	CAMK2N1	CAMK2N1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2G geni (Tüm Gen Dizi Analizi)	CAMK2G gene (Sequence Analysis-All Coding Exons)	CAMK2G	CAMK2G, CAMKG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2D geni (Tüm Gen Dizi Analizi)	CAMK2D gene (Sequence Analysis-All Coding Exons)	CAMK2D	CAMK2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2B geni (Tüm Gen Dizi Analizi)	CAMK2B gene (Sequence Analysis-All Coding Exons)	CAMK2B	CAMK2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK2A geni (Tüm Gen Dizi Analizi)	CAMK2A gene (Sequence Analysis-All Coding Exons)	CAMK2A	CAMK2A, KIAA0968, CAMKA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK1G geni (Tüm Gen Dizi Analizi)	CAMK1G gene (Sequence Analysis-All Coding Exons)	CAMK1G	CAMK1G, CLICK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK1D geni (Tüm Gen Dizi Analizi)	CAMK1D gene (Sequence Analysis-All Coding Exons)	CAMK1D	CAMK1D, CKLIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMK1 geni (Tüm Gen Dizi Analizi)	CAMK1 gene (Sequence Analysis-All Coding Exons)	CAMK1	CAMK1, CAMKI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALY geni (Tüm Gen Dizi Analizi)	CALY gene (Sequence Analysis-All Coding Exons)	CALY	CALY, DRD1IP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALU geni (Tüm Gen Dizi Analizi)	CALU gene (Sequence Analysis-All Coding Exons)	CALU	CALU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALML6 geni (Tüm Gen Dizi Analizi)	CALML6 gene (Sequence Analysis-All Coding Exons)	CALML6	CALML6, CAGLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALML3 geni (Tüm Gen Dizi Analizi)	CALML3 gene (Sequence Analysis-All Coding Exons)	CALML3	CALML3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALM3 geni (Tüm Gen Dizi Analizi)	CALM3 gene (Sequence Analysis-All Coding Exons)	CALM3	CALM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALHM2 geni (Tüm Gen Dizi Analizi)	CALHM2 gene (Sequence Analysis-All Coding Exons)	CALHM2	CALHM2, FAM26B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALHM1 geni (Tüm Gen Dizi Analizi)	CALHM1 gene (Sequence Analysis-All Coding Exons)	CALHM1	CALHM1, FAM26C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALD1 geni (Tüm Gen Dizi Analizi)	CALD1 gene (Sequence Analysis-All Coding Exons)	CALD1	CALD1, CDM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALCRL geni (Tüm Gen Dizi Analizi)	CALCRL gene (Sequence Analysis-All Coding Exons)	CALCRL	CALCRL, CGRPR, CRLR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALCB geni (Tüm Gen Dizi Analizi)	CALCB gene (Sequence Analysis-All Coding Exons)	CALCB	CALCB, CALC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALCA geni (Tüm Gen Dizi Analizi)	CALCA gene (Sequence Analysis-All Coding Exons)	CALCA	CALCA, CALC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALB2 geni (Tüm Gen Dizi Analizi)	CALB2 gene (Sequence Analysis-All Coding Exons)	CALB2	CALB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALB1 geni (Tüm Gen Dizi Analizi)	CALB1 gene (Sequence Analysis-All Coding Exons)	CALB1	CALB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAHM geni (Tüm Gen Dizi Analizi)	CAHM gene (Sequence Analysis-All Coding Exons)	CAHM	CAHM, LINC00468	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAGE1 geni (Tüm Gen Dizi Analizi)	CAGE1 gene (Sequence Analysis-All Coding Exons)	CAGE1	CTAG3, CAGE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IPO5 geni (Tüm Gen Dizi Analizi)	IPO5 gene (Sequence Analysis-All Coding Exons)	IPO5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADPS2 geni (Tüm Gen Dizi Analizi)	CADPS2 gene (Sequence Analysis-All Coding Exons)	CADPS2	CADPS2, KIAA1591	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADPS geni (Tüm Gen Dizi Analizi)	CADPS gene (Sequence Analysis-All Coding Exons)	CADPS	CADPS, CAPS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADM4 geni (Tüm Gen Dizi Analizi)	CADM4 gene (Sequence Analysis-All Coding Exons)	CADM4	IGSF4C, TSLL2, NECL4, SYNCAM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADM3 geni (Tüm Gen Dizi Analizi)	CADM3 gene (Sequence Analysis-All Coding Exons)	CADM3	IGSF4B, TSLL1, NECL1, SYNCAM3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADM2 geni (Tüm Gen Dizi Analizi)	CADM2 gene (Sequence Analysis-All Coding Exons)	CADM2	IGSF4D, SYNCAM2, NECL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CADM1 geni (Tüm Gen Dizi Analizi)	CADM1 gene (Sequence Analysis-All Coding Exons)	CADM1	IGSF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACYBP geni (Tüm Gen Dizi Analizi)	CACYBP gene (Sequence Analysis-All Coding Exons)	CACYBP	SIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG8 geni (Tüm Gen Dizi Analizi)	CACNG8 gene (Sequence Analysis-All Coding Exons)	CACNG8	CACNG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG7 geni (Tüm Gen Dizi Analizi)	CACNG7 gene (Sequence Analysis-All Coding Exons)	CACNG7	CACNG7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG6 geni (Tüm Gen Dizi Analizi)	CACNG6 gene (Sequence Analysis-All Coding Exons)	CACNG6	CACNG6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG5 geni (Tüm Gen Dizi Analizi)	CACNG5 gene (Sequence Analysis-All Coding Exons)	CACNG5	CACNG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG3 geni (Tüm Gen Dizi Analizi)	CACNG3 gene (Sequence Analysis-All Coding Exons)	CACNG3	CACNG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG1 geni (Tüm Gen Dizi Analizi)	CACNG1 gene (Sequence Analysis-All Coding Exons)	CACNG1	CACNG1, CACNLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNB3 geni (Tüm Gen Dizi Analizi)	CACNB3 gene (Sequence Analysis-All Coding Exons)	CACNB3	CACNB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNB1 geni (Tüm Gen Dizi Analizi)	CACNB1 gene (Sequence Analysis-All Coding Exons)	CACNB1	CACNB1, CACNLB1, CCHLB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA2D3 geni (Tüm Gen Dizi Analizi)	CACNA2D3 gene (Sequence Analysis-All Coding Exons)	CACNA2D3	CACNA2D3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA2D2 geni (Tüm Gen Dizi Analizi)	CACNA2D2 gene (Sequence Analysis-All Coding Exons)	CACNA2D2	CACNA2D2, KIAA0558	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA2D1 geni (Tüm Gen Dizi Analizi)	CACNA2D1 gene (Sequence Analysis-All Coding Exons)	CACNA2D1	CACNA2, CACNL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA2D1 geni (Tüm Gen Dizi Analizi)	CACNA2D1 gene (Sequence Analysis-All Coding Exons)	CACNA2D1	CACNA2, CACNL2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA1I geni (Tüm Gen Dizi Analizi)	CACNA1I gene (Sequence Analysis-All Coding Exons)	CACNA1I	CACNA1I, KIAA1120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNA1E geni (Tüm Gen Dizi Analizi)	CACNA1E gene (Sequence Analysis-All Coding Exons)	CACNA1E	CACNA1E, CACNL1A6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACFD1 geni (Tüm Gen Dizi Analizi)	CACFD1 gene (Sequence Analysis-All Coding Exons)	CACFD1	C9orf7, FLOWER	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CABYR geni (Tüm Gen Dizi Analizi)	CABYR gene (Sequence Analysis-All Coding Exons)	CABYR	CABYR, FSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CABP5 geni (Tüm Gen Dizi Analizi)	CABP5 gene (Sequence Analysis-All Coding Exons)	CABP5	CABP5, CABP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CABIN1 geni (Tüm Gen Dizi Analizi)	CABIN1 gene (Sequence Analysis-All Coding Exons)	CABIN1	CABIN1, CAIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAB39L geni (Tüm Gen Dizi Analizi)	CAB39L gene (Sequence Analysis-All Coding Exons)	CAB39L	CAB39L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAB39 geni (Tüm Gen Dizi Analizi)	CAB39 gene (Sequence Analysis-All Coding Exons)	CAB39	CAB39	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA9 geni (Tüm Gen Dizi Analizi)	CA9 gene (Sequence Analysis-All Coding Exons)	CA9	CA9, MN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA7 geni (Tüm Gen Dizi Analizi)	CA7 gene (Sequence Analysis-All Coding Exons)	CA7	CA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA6 geni (Tüm Gen Dizi Analizi)	CA6 gene (Sequence Analysis-All Coding Exons)	CA6	CA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CASB geni (Tüm Gen Dizi Analizi)	CASB gene (Sequence Analysis-All Coding Exons)	CASB	CASB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA3 geni (Tüm Gen Dizi Analizi)	CA3 gene (Sequence Analysis-All Coding Exons)	CA3	CA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA14 geni (Tüm Gen Dizi Analizi)	CA14 gene (Sequence Analysis-All Coding Exons)	CA14	CA14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA11 geni (Tüm Gen Dizi Analizi)	CA11 gene (Sequence Analysis-All Coding Exons)	CA11	CA11, CARP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA10 geni (Tüm Gen Dizi Analizi)	CA10 gene (Sequence Analysis-All Coding Exons)	CA10	CA10, CARPX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA1 geni (Tüm Gen Dizi Analizi)	CA1 gene (Sequence Analysis-All Coding Exons)	CA1	CA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C9orf64 geni (Tüm Gen Dizi Analizi)	C9orf64 gene (Sequence Analysis-All Coding Exons)	C9orf64	C9orf64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C9orf142 geni (Tüm Gen Dizi Analizi)	C9orf142 gene (Sequence Analysis-All Coding Exons)	C9orf142	PAXX, C9orf142	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C9orf116 geni (Tüm Gen Dizi Analizi)	C9orf116 gene (Sequence Analysis-All Coding Exons)	C9orf116	C9orf116, PIERCE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C8orf4 geni (Tüm Gen Dizi Analizi)	C8orf4 gene (Sequence Analysis-All Coding Exons)	C8orf4	C8orf4, TC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C8G geni (Tüm Gen Dizi Analizi)	C8G gene (Sequence Analysis-All Coding Exons)	C8G	C8G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C7orf49 geni (Tüm Gen Dizi Analizi)	C7orf49 gene (Sequence Analysis-All Coding Exons)	C7orf49	C7orf49, MRI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C7orf31 geni (Tüm Gen Dizi Analizi)	C7orf31 gene (Sequence Analysis-All Coding Exons)	C7orf31	C7orf31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C7orf13 geni (Tüm Gen Dizi Analizi)	C7orf13 gene (Sequence Analysis-All Coding Exons)	C7orf13	C7orf13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf89 geni (Tüm Gen Dizi Analizi)	C6orf89 gene (Sequence Analysis-All Coding Exons)	C6orf89	C6orf89, BRAP, AMFION	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf48 geni (Tüm Gen Dizi Analizi)	C6orf48 gene (Sequence Analysis-All Coding Exons)	C6orf48	G8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf120 geni (Tüm Gen Dizi Analizi)	C6orf120 gene (Sequence Analysis-All Coding Exons)	C6orf120	C6orf120	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf106 geni (Tüm Gen Dizi Analizi)	C6orf106 gene (Sequence Analysis-All Coding Exons)	C6orf106	C6orf106	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
INPP4B geni (Tüm Gen Dizi Analizi)	INPP4B gene (Sequence Analysis-All Coding Exons)	INPP4B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf1 geni (Tüm Gen Dizi Analizi)	C6orf1 gene (Sequence Analysis-All Coding Exons)	C6orf1	C6orf1, LBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C5orf38 geni (Tüm Gen Dizi Analizi)	C5orf38 gene (Sequence Analysis-All Coding Exons)	C5orf38	CEI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C5orf30 geni (Tüm Gen Dizi Analizi)	C5orf30 gene (Sequence Analysis-All Coding Exons)	C5orf30	C5orf30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C5AR2 geni (Tüm Gen Dizi Analizi)	C5AR2 gene (Sequence Analysis-All Coding Exons)	C5AR2	C5AR2, GPF77, C5L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C5AR1 geni (Tüm Gen Dizi Analizi)	C5AR1 gene (Sequence Analysis-All Coding Exons)	C5AR1	C5AR1, CSR1, C5AR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C4orf48 geni (Tüm Gen Dizi Analizi)	C4orf48 gene (Sequence Analysis-All Coding Exons)	C4orf48	C4orf48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C4orf46 geni (Tüm Gen Dizi Analizi)	C4orf46 gene (Sequence Analysis-All Coding Exons)	C4orf46	C4orf46, RCDG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C4BPB geni (Tüm Gen Dizi Analizi)	C4BPB gene (Sequence Analysis-All Coding Exons)	C4BPB	C4BPB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C4BPA geni (Tüm Gen Dizi Analizi)	C4BPA gene (Sequence Analysis-All Coding Exons)	C4BPA	C4BPA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C3orf58 geni (Tüm Gen Dizi Analizi)	C3orf58 gene (Sequence Analysis-All Coding Exons)	C3orf58	C3orf58, DIA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C3orf52 geni (Tüm Gen Dizi Analizi)	C3orf52 gene (Sequence Analysis-All Coding Exons)	C3orf52	C3orf52, TTMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C3AR1 geni (Tüm Gen Dizi Analizi)	C3AR1 gene (Sequence Analysis-All Coding Exons)	C3AR1	C3AR1, C3AR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C2orf88 geni (Tüm Gen Dizi Analizi)	C2orf88 gene (Sequence Analysis-All Coding Exons)	C2orf88	C2orf88, SMAKAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C2orf80 geni (Tüm Gen Dizi Analizi)	C2orf80 gene (Sequence Analysis-All Coding Exons)	C2orf80	C2orf80, GONDA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C2orf40 geni (Tüm Gen Dizi Analizi)	C2orf40 gene (Sequence Analysis-All Coding Exons)	C2orf40	C2orf40, ECRG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

C2CD4C geni (Tüm Gen Dizi Analizi)	C2CD4C gene (Sequence Analysis-All Coding Exons)	C2CD4C	C2CD4C, NLF3, KIAA1957	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C2CD4B geni (Tüm Gen Dizi Analizi)	C2CD4B gene (Sequence Analysis-All Coding Exons)	C2CD4B	C2CD4B, NLF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C2CD4A geni (Tüm Gen Dizi Analizi)	C2CD4A gene (Sequence Analysis-All Coding Exons)	C2CD4A	C2CD4A, NLF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C21orf33 geni (Tüm Gen Dizi Analizi)	C21orf33 gene (Sequence Analysis-All Coding Exons)	C21orf33	ES1, HES1, GT335, C21orf33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C21orf2 geni (Tüm Gen Dizi Analizi)	C21orf2 gene (Sequence Analysis-All Coding Exons)	C21orf2	C21orf2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C20orf194 geni (Tüm Gen Dizi Analizi)	C20orf194 gene (Sequence Analysis-All Coding Exons)	C20orf194	C20orf194	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1RL geni (Tüm Gen Dizi Analizi)	C1RL gene (Sequence Analysis-All Coding Exons)	C1RL	C1RL, CLSPA, C1RLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF9B geni (Tüm Gen Dizi Analizi)	C1QTNF9B gene (Sequence Analysis-All Coding Exons)	C1QTNF9B	C1QTNF9B, CTRP9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
INA geni (Tüm Gen Dizi Analizi)	INA gene (Sequence Analysis-All Coding Exons)	INA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF9 geni (Tüm Gen Dizi Analizi)	C1QTNF9 gene (Sequence Analysis-All Coding Exons)	C1QTNF9	C1QTNF9A, CTRP9, C1QTNF9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF8 geni (Tüm Gen Dizi Analizi)	C1QTNF8 gene (Sequence Analysis-All Coding Exons)	C1QTNF8	C1QTNF8, CTRP8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF6 geni (Tüm Gen Dizi Analizi)	C1QTNF6 gene (Sequence Analysis-All Coding Exons)	C1QTNF6	C1QTNF6, CTFP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF4 geni (Tüm Gen Dizi Analizi)	C1QTNF4 gene (Sequence Analysis-All Coding Exons)	C1QTNF4	C1QTNF4, CTRP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF3 geni (Tüm Gen Dizi Analizi)	C1QTNF3 gene (Sequence Analysis-All Coding Exons)	C1QTNF3	C1ATNF3, CTRP3, CORS26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF12 geni (Tüm Gen Dizi Analizi)	C1QTNF12 gene (Sequence Analysis-All Coding Exons)	C1QTNF12	FAM132A, C1QDC2, C1QTNF12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QTNF1 geni (Tüm Gen Dizi Analizi)	C1QTNF1 gene (Sequence Analysis-All Coding Exons)	C1QTNF1	C1QTNF1, GIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QL4 geni (Tüm Gen Dizi Analizi)	C1QL4 gene (Sequence Analysis-All Coding Exons)	C1QL4	C1QL4, CTRP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QL3 geni (Tüm Gen Dizi Analizi)	C1QL3 gene (Sequence Analysis-All Coding Exons)	C1QL3	C1QL3, CTRP13, K100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QL2 geni (Tüm Gen Dizi Analizi)	C1QL2 gene (Sequence Analysis-All Coding Exons)	C1QL2	C1QL2, CTRP10, C1QTNF10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QL1 geni (Tüm Gen Dizi Analizi)	C1QL1 gene (Sequence Analysis-All Coding Exons)	C1QL1	C1QL1, CRF, C1QRF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1QBP geni (Tüm Gen Dizi Analizi)	C1QBP gene (Sequence Analysis-All Coding Exons)	C1QBP	C1QBP, HABP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1orf186 geni (Tüm Gen Dizi Analizi)	C1orf186 gene (Sequence Analysis-All Coding Exons)	C1orf186	C1orf186, RHEX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1orf116 geni (Tüm Gen Dizi Analizi)	C1orf116 gene (Sequence Analysis-All Coding Exons)	C1orf116	SARG, C1orf116	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1orf109 geni (Tüm Gen Dizi Analizi)	C1orf109 gene (Sequence Analysis-All Coding Exons)	C1orf109	C1orf109	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1GALT1 geni (Tüm Gen Dizi Analizi)	C1GALT1 gene (Sequence Analysis-All Coding Exons)	C1GALT1	C1GALT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C1D geni (Tüm Gen Dizi Analizi)	C1D gene (Sequence Analysis-All Coding Exons)	C1D	C1D, SUNCOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C19orf70 geni (Tüm Gen Dizi Analizi)	C19orf70 gene (Sequence Analysis-All Coding Exons)	C19orf70	C19orf70, MIC13, QIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
ILF2 geni (Tüm Gen Dizi Analizi)	ILF2 gene (Sequence Analysis-All Coding Exons)	ILF2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C19orf66 geni (Tüm Gen Dizi Analizi)	C19orf66 gene (Sequence Analysis-All Coding Exons)	C19orf66	C19orf66, RYDEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C18orf54 geni (Tüm Gen Dizi Analizi)	C18orf54 gene (Sequence Analysis-All Coding Exons)	C18orf54	C18orf54, LAS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C17orf49 geni (Tüm Gen Dizi Analizi)	C17orf49 gene (Sequence Analysis-All Coding Exons)	C17orf49	C17orf49, BAP18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C15orf59 geni (Tüm Gen Dizi Analizi)	C15orf59 gene (Sequence Analysis-All Coding Exons)	C15orf59	C15orf59, INSYN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C15orf48 geni (Tüm Gen Dizi Analizi)	C15orf48 gene (Sequence Analysis-All Coding Exons)	C15orf48	NMES1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C14orf39 geni (Tüm Gen Dizi Analizi)	C14orf39 gene (Sequence Analysis-All Coding Exons)	C14orf39	C14orf39, SIX6OS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C14orf2 geni (Tüm Gen Dizi Analizi)	C14orf2 gene (Sequence Analysis-All Coding Exons)	C14orf2	C14orf2, MP68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C14orf166 geni (Tüm Gen Dizi Analizi)	C14orf166 gene (Sequence Analysis-All Coding Exons)	C14orf166	C14orf166, CGI99	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C14orf1 geni (Tüm Gen Dizi Analizi)	C14orf1 gene (Sequence Analysis-All Coding Exons)	C14orf1	C14orf1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C12orf4 geni (Tüm Gen Dizi Analizi)	C12orf4 gene (Sequence Analysis-All Coding Exons)	C12orf4	C12orf4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C12orf10 geni (Tüm Gen Dizi Analizi)	C12orf10 gene (Sequence Analysis-All Coding Exons)	C12orf10	C12orf10, MYG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C11orf95 geni (Tüm Gen Dizi Analizi)	C11orf95 gene (Sequence Analysis-All Coding Exons)	C11orf95	C11orf95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C11orf80 geni (Tüm Gen Dizi Analizi)	C11orf80 gene (Sequence Analysis-All Coding Exons)	C11orf80	C11orf80	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C11orf54 geni (Tüm Gen Dizi Analizi)	C11orf54 gene (Sequence Analysis-All Coding Exons)	C11orf54	C11orf54, PTOD012	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C11orf24 geni (Tüm Gen Dizi Analizi)	C11orf24 gene (Sequence Analysis-All Coding Exons)	C11orf24	C11orf24	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C11orf21 geni (Tüm Gen Dizi Analizi)	C11orf21 gene (Sequence Analysis-All Coding Exons)	C11orf21	C11orf21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
C10orf10 geni (Tüm Gen Dizi Analizi)	C10orf10 gene (Sequence Analysis-All Coding Exons)	C10orf10	C10orf10, DEPP, FIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BYSL geni (Tüm Gen Dizi Analizi)	BYSL gene (Sequence Analysis-All Coding Exons)	BYSL	BYSL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BUD31 geni (Tüm Gen Dizi Analizi)	BUD31 gene (Sequence Analysis-All Coding Exons)	BUD31	BUD31, G10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BUD23 geni (Tüm Gen Dizi Analizi)	BUD23 gene (Sequence Analysis-All Coding Exons)	BUD23	WBSCR22, MERM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BUB3 geni (Tüm Gen Dizi Analizi)	BUB3 gene (Sequence Analysis-All Coding Exons)	BUB3	BUB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTRC geni (Tüm Gen Dizi Analizi)	BTRC gene (Sequence Analysis-All Coding Exons)	BTRC	BTRC, BTRCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTNL8 geni (Tüm Gen Dizi Analizi)	BTNL8 gene (Sequence Analysis-All Coding Exons)	BTNL8	BTNL8, BTN9.2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTNL3 geni (Tüm Gen Dizi Analizi)	BTNL3 gene (Sequence Analysis-All Coding Exons)	BTNL3	BTNL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN3A3 geni (Tüm Gen Dizi Analizi)	BTN3A3 gene (Sequence Analysis-All Coding Exons)	BTN3A3	BTN3A3, BTF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN3A2 geni (Tüm Gen Dizi Analizi)	BTN3A2 gene (Sequence Analysis-All Coding Exons)	BTN3A2	BTN3A2, BTF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN3A1 geni (Tüm Gen Dizi Analizi)	BTN3A1 gene (Sequence Analysis-All Coding Exons)	BTN3A1	BTN3A1, BTF5, CD277	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN2A3P geni (Tüm Gen Dizi Analizi)	BTN2A3P gene (Sequence Analysis-All Coding Exons)	BTN2A3P	BTN2A3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN2A2 geni (Tüm Gen Dizi Analizi)	BTN2A2 gene (Sequence Analysis-All Coding Exons)	BTN2A2	BTN2A2, BTF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN2A1 geni (Tüm Gen Dizi Analizi)	BTN2A1 gene (Sequence Analysis-All Coding Exons)	BTN2A1	BTN2A1, BTF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTN1A1 geni (Tüm Gen Dizi Analizi)	BTN1A1 gene (Sequence Analysis-All Coding Exons)	BTN1A1	BTN1A1, BTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTLA geni (Tüm Gen Dizi Analizi)	BTLA gene (Sequence Analysis-All Coding Exons)	BTLA	BTLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTG4 geni (Tüm Gen Dizi Analizi)	BTG4 gene (Sequence Analysis-All Coding Exons)	BTG4	BTG4, PC3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTG3 geni (Tüm Gen Dizi Analizi)	BTG3 gene (Sequence Analysis-All Coding Exons)	BTG3	BTG3, ANA, TOB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTG2 geni (Tüm Gen Dizi Analizi)	BTG2 gene (Sequence Analysis-All Coding Exons)	BTG2	BTG2, PC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTG1 geni (Tüm Gen Dizi Analizi)	BTG1 gene (Sequence Analysis-All Coding Exons)	BTG1	BTG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTC geni (Tüm Gen Dizi Analizi)	BTC gene (Sequence Analysis-All Coding Exons)	BTC	BTC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTBD9 geni (Tüm Gen Dizi Analizi)	BTBD9 gene (Sequence Analysis-All Coding Exons)	BTBD9	BTBD9, KIAA1880	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTBD7 geni (Tüm Gen Dizi Analizi)	BTBD7 gene (Sequence Analysis-All Coding Exons)	BTBD7	BTBD7, FUP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTBD3 geni (Tüm Gen Dizi Analizi)	BTBD3 gene (Sequence Analysis-All Coding Exons)	BTBD3	BTBD3, KIAA0952	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTBD10 geni (Tüm Gen Dizi Analizi)	BTBD10 gene (Sequence Analysis-All Coding Exons)	BTBD10	BTBD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BTAF1 geni (Tüm Gen Dizi Analizi)	BTAF1 gene (Sequence Analysis-All Coding Exons)	BTAF1	BTAF1, TAFII170, TAF172, MOT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BSX geni (Tüm Gen Dizi Analizi)	BSX gene (Sequence Analysis-All Coding Exons)	BSX	BSX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BST2 geni (Tüm Gen Dizi Analizi)	BST2 gene (Sequence Analysis-All Coding Exons)	BST2	BST2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BST1 geni (Tüm Gen Dizi Analizi)	BST1 gene (Sequence Analysis-All Coding Exons)	BST1	BST1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BSPH1 geni (Tüm Gen Dizi Analizi)	BSPH1 gene (Sequence Analysis-All Coding Exons)	BSPH1	BSPH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BSN geni (Tüm Gen Dizi Analizi)	BSN gene (Sequence Analysis-All Coding Exons)	BSN	BSN, ZNF231	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
IL22RA1 geni (Tüm Gen Dizi Analizi)	IL22RA1 gene (Sequence Analysis-All Coding Exons)	IL22RA1	IL22RA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRSK2 geni (Tüm Gen Dizi Analizi)	BRSK2 gene (Sequence Analysis-All Coding Exons)	BRSK2	BRSK2, SAD1, PEN1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRSK1 geni (Tüm Gen Dizi Analizi)	BRSK1 gene (Sequence Analysis-All Coding Exons)	BRSK1	BRSK1, KIAA1811	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRS3 geni (Tüm Gen Dizi Analizi)	BRS3 gene (Sequence Analysis-All Coding Exons)	BRS3	BRS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRPF3 geni (Tüm Gen Dizi Analizi)	BRPF3 gene (Sequence Analysis-All Coding Exons)	BRPF3	BRPF3, KIAA1286	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
IL20RB geni (Tüm Gen Dizi Analizi)	IL20RB gene (Sequence Analysis-All Coding Exons)	IL20RB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRMS1 geni (Tüm Gen Dizi Analizi)	BRMS1 gene (Sequence Analysis-All Coding Exons)	BRMS1	BRMS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRK1 geni (Tüm Gen Dizi Analizi)	BRK1 gene (Sequence Analysis-All Coding Exons)	BRK1	BRK1, C3orf10, HSPC300	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRINP1 geni (Tüm Gen Dizi Analizi)	BRINP1 gene (Sequence Analysis-All Coding Exons)	BRINP1	BRINP1, DBC1, DBCCR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRI3BP geni (Tüm Gen Dizi Analizi)	BRI3BP gene (Sequence Analysis-All Coding Exons)	BRI3BP	BRI3BP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRI3 geni (Tüm Gen Dizi Analizi)	BRI3 gene (Sequence Analysis-All Coding Exons)	BRI3	BRI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRF2 geni (Tüm Gen Dizi Analizi)	BRF2 gene (Sequence Analysis-All Coding Exons)	BRF2	BRF2, BRFU, TFIIIB50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRE geni (Tüm Gen Dizi Analizi)	BRE gene (Sequence Analysis-All Coding Exons)	BRE	BRE, BRCC4, BRCC45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRDT geni (Tüm Gen Dizi Analizi)	BRDT gene (Sequence Analysis-All Coding Exons)	BRDT	BRDT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRD4 geni (Tüm Gen Dizi Analizi)	BRD4 gene (Sequence Analysis-All Coding Exons)	BRD4	BRD4, CAP, HUNK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRD3 geni (Tüm Gen Dizi Analizi)	BRD3 gene (Sequence Analysis-All Coding Exons)	BRD3	RING3L, ORFX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRD2 geni (Tüm Gen Dizi Analizi)	BRD2 gene (Sequence Analysis-All Coding Exons)	BRD2	BRD2, RING3, FSRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample
BRD1 geni (Tüm Gen Dizi Analizi)	BRD1 gene (Sequence Analysis-All Coding Exons)	BRD1	BRL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	-	-	60	Postnatal	-	EDTA Blood Sample

BRCC3 geni (Tüm Gen Dizi Analizi)	BRCC3 gene (Sequence Analysis-All Coding Exons)	BRCC3	BRCC3, BRCC36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BRAP geni (Tüm Gen Dizi Analizi)	BRAP gene (Sequence Analysis-All Coding Exons)	BRAP	BRAP, BRAP2, IMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPY2 geni (Tüm Gen Dizi Analizi)	BPY2 gene (Sequence Analysis-All Coding Exons)	BPY2	BPY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPTF geni (Tüm Gen Dizi Analizi)	BPTF gene (Sequence Analysis-All Coding Exons)	BPTF	BPTF, FALZ, FAC1, NURF301	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPNT1 geni (Tüm Gen Dizi Analizi)	BPNT1 gene (Sequence Analysis-All Coding Exons)	BPNT1	BPNT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFC geni (Tüm Gen Dizi Analizi)	BPIFC gene (Sequence Analysis-All Coding Exons)	BPIFC	BPIFC, BPIL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFB6 geni (Tüm Gen Dizi Analizi)	BPIFB6 gene (Sequence Analysis-All Coding Exons)	BPIFB6	BPIFB6, BPIL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFB4 geni (Tüm Gen Dizi Analizi)	BPIFB4 gene (Sequence Analysis-All Coding Exons)	BPIFB4	BPIFB4, RY2G5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFB3 geni (Tüm Gen Dizi Analizi)	BPIFB3 gene (Sequence Analysis-All Coding Exons)	BPIFB3	BPIFBP3, RYA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFB2 geni (Tüm Gen Dizi Analizi)	BPIFB2 gene (Sequence Analysis-All Coding Exons)	BPIFB2	BPIFB2, BPIL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFA4P geni (Tüm Gen Dizi Analizi)	BPIFA4P gene (Sequence Analysis-All Coding Exons)	BPIFA4P	BASE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPIFA1 geni (Tüm Gen Dizi Analizi)	BPIFA1 gene (Sequence Analysis-All Coding Exons)	BPIFA1	BPIFA1, PLUNC, SPLUNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPI geni (Tüm Gen Dizi Analizi)	BPI gene (Sequence Analysis-All Coding Exons)	BPI	BPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BPHL geni (Tüm Gen Dizi Analizi)	BPHL gene (Sequence Analysis-All Coding Exons)	BPHL	BPHL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BORCS8 geni (Tüm Gen Dizi Analizi)	BORCS8 gene (Sequence Analysis-All Coding Exons)	BORCS8	BORCS8, MEF2BNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BORCS7 geni (Tüm Gen Dizi Analizi)	BORCS7 gene (Sequence Analysis-All Coding Exons)	BORCS7	BORCS7, C10orf32	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BORCS6 geni (Tüm Gen Dizi Analizi)	BORCS6 gene (Sequence Analysis-All Coding Exons)	BORCS6	BORCS6, C17orf59, LYSPD8/39L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BORCS5 geni (Tüm Gen Dizi Analizi)	BORCS5 gene (Sequence Analysis-All Coding Exons)	BORCS5	BORCS5, LOH12CR1, MYBLYS/IN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BORA geni (Tüm Gen Dizi Analizi)	BORA gene (Sequence Analysis-All Coding Exons)	BORA	BORA, C13orf34	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOP1 geni (Tüm Gen Dizi Analizi)	BOP1 gene (Sequence Analysis-All Coding Exons)	BOP1	BOP1, KIAA0124	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOLL geni (Tüm Gen Dizi Analizi)	BOLL gene (Sequence Analysis-All Coding Exons)	BOLL	BOLL, BOULE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOLA2 geni (Tüm Gen Dizi Analizi)	BOLA2 gene (Sequence Analysis-All Coding Exons)	BOLA2	BOLA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOLA1 geni (Tüm Gen Dizi Analizi)	BOLA1 gene (Sequence Analysis-All Coding Exons)	BOLA1	BOLA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOD1L1 geni (Tüm Gen Dizi Analizi)	BOD1L1 gene (Sequence Analysis-All Coding Exons)	BOD1L1	BOD1L1, FAM44A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOD1 geni (Tüm Gen Dizi Analizi)	BOD1 gene (Sequence Analysis-All Coding Exons)	BOD1	BOD1, FAM44B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNIPL geni (Tüm Gen Dizi Analizi)	BNIPL gene (Sequence Analysis-All Coding Exons)	BNIPL	BNIPL, BNIP5, BNIPL1, BNIP-2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNIP3L geni (Tüm Gen Dizi Analizi)	BNIP3L gene (Sequence Analysis-All Coding Exons)	BNIP3L	BNIP3L, NIX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNIP3 geni (Tüm Gen Dizi Analizi)	BNIP3 gene (Sequence Analysis-All Coding Exons)	BNIP3	BNIP3, NIP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNIP2 geni (Tüm Gen Dizi Analizi)	BNIP2 gene (Sequence Analysis-All Coding Exons)	BNIP2	BNIP2, NIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNIP1 geni (Tüm Gen Dizi Analizi)	BNIP1 gene (Sequence Analysis-All Coding Exons)	BNIP1	BNIP1, NIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNC2 geni (Tüm Gen Dizi Analizi)	BNC2 gene (Sequence Analysis-All Coding Exons)	BNC2	BNC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BNC1 geni (Tüm Gen Dizi Analizi)	BNC1 gene (Sequence Analysis-All Coding Exons)	BNC1	BNC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMX geni (Tüm Gen Dizi Analizi)	BMX gene (Sequence Analysis-All Coding Exons)	BMX	BMX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP8B geni (Tüm Gen Dizi Analizi)	BMP8B gene (Sequence Analysis-All Coding Exons)	BMP8B	BMP8B, OP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP7 geni (Tüm Gen Dizi Analizi)	BMP7 gene (Sequence Analysis-All Coding Exons)	BMP7	BMP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP6 geni (Tüm Gen Dizi Analizi)	BMP6 gene (Sequence Analysis-All Coding Exons)	BMP6	BMP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP5 geni (Tüm Gen Dizi Analizi)	BMP5 gene (Sequence Analysis-All Coding Exons)	BMP5	BMP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP3 geni (Tüm Gen Dizi Analizi)	BMP3 gene (Sequence Analysis-All Coding Exons)	BMP3	BMP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMP10 geni (Tüm Gen Dizi Analizi)	BMP10 gene (Sequence Analysis-All Coding Exons)	BMP10	BMP10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMI1 geni (Tüm Gen Dizi Analizi)	BMI1 gene (Sequence Analysis-All Coding Exons)	BMI1	BMI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BMF geni (Tüm Gen Dizi Analizi)	BMF gene (Sequence Analysis-All Coding Exons)	BMF	BMF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IKZF5 geni (Tüm Gen Dizi Analizi)	IKZF5 gene (Sequence Analysis-All Coding Exons)	IKZF5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLVRB geni (Tüm Gen Dizi Analizi)	BLVRB gene (Sequence Analysis-All Coding Exons)	BLVRB	BLVRB, BVRB, FLR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLOC1S5 geni (Tüm Gen Dizi Analizi)	BLOC1S5 gene (Sequence Analysis-All Coding Exons)	BLOC1S5	BLOC1S5, BLOS5, MUTED	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLOC1S4 geni (Tüm Gen Dizi Analizi)	BLOC1S4 gene (Sequence Analysis-All Coding Exons)	BLOC1S4	BLOC1S4, BLOS4, CNO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLOC1S2 geni (Tüm Gen Dizi Analizi)	BLOC1S2 gene (Sequence Analysis-All Coding Exons)	BLOC1S2	BLOC1S2, BLOS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLOC1S1 geni (Tüm Gen Dizi Analizi)	BLOC1S1 gene (Sequence Analysis-All Coding Exons)	BLOC1S1	GCN5L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLMH geni (Tüm Gen Dizi Analizi)	BLMH gene (Sequence Analysis-All Coding Exons)	BLMH	BLMH, BMH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLID geni (Tüm Gen Dizi Analizi)	BLID gene (Sequence Analysis-All Coding Exons)	BLID	BLID, BRCC@	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLCAP geni (Tüm Gen Dizi Analizi)	BLCAP gene (Sequence Analysis-All Coding Exons)	BLCAP	BLCAP, BC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIRC7 geni (Tüm Gen Dizi Analizi)	BIRC7 gene (Sequence Analysis-All Coding Exons)	BIRC7	BIRC7, MLIAP, KIAP, LIVIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIRC6 geni (Tüm Gen Dizi Analizi)	BIRC6 gene (Sequence Analysis-All Coding Exons)	BIRC6	BIRC6, KIAA1289	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIRC5 geni (Tüm Gen Dizi Analizi)	BIRC5 gene (Sequence Analysis-All Coding Exons)	BIRC5	API4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIRC3 geni (Tüm Gen Dizi Analizi)	BIRC3 gene (Sequence Analysis-All Coding Exons)	BIRC3	API2, HAIP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIRC2 geni (Tüm Gen Dizi Analizi)	BIRC2 gene (Sequence Analysis-All Coding Exons)	BIRC2	API1, HAIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIN3 geni (Tüm Gen Dizi Analizi)	BIN3 gene (Sequence Analysis-All Coding Exons)	BIN3	BIN3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIN2 geni (Tüm Gen Dizi Analizi)	BIN2 gene (Sequence Analysis-All Coding Exons)	BIN2	BIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BIK geni (Tüm Gen Dizi Analizi)	BIK gene (Sequence Analysis-All Coding Exons)	BIK	BIK, NBK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BID geni (Tüm Gen Dizi Analizi)	BID gene (Sequence Analysis-All Coding Exons)	BID	BID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BICDL2 geni (Tüm Gen Dizi Analizi)	BICDL2 gene (Sequence Analysis-All Coding Exons)	BICDL2	BICDL2, BICDR2, CCDC64B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BICDL1 geni (Tüm Gen Dizi Analizi)	BICDL1 gene (Sequence Analysis-All Coding Exons)	BICDL1	BICDL1, BICDR1, CCDC64	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BICD1 geni (Tüm Gen Dizi Analizi)	BICD1 gene (Sequence Analysis-All Coding Exons)	BICD1	BICD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHMT2 geni (Tüm Gen Dizi Analizi)	BHMT2 gene (Sequence Analysis-All Coding Exons)	BHMT2	BHMT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHMT geni (Tüm Gen Dizi Analizi)	BHMT gene (Sequence Analysis-All Coding Exons)	BHMT	BHMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHLHE40 geni (Tüm Gen Dizi Analizi)	BHLHE40 gene (Sequence Analysis-All Coding Exons)	BHLHE40	BHLHE40, HLHB2, STRA13, DEC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGSF11 geni (Tüm Gen Dizi Analizi)	IGSF11 gene (Sequence Analysis-All Coding Exons)	IGSF11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHLHE23 geni (Tüm Gen Dizi Analizi)	BHLHE23 gene (Sequence Analysis-All Coding Exons)	BHLHE23	BHLHE23, BHLHB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHLHE22 geni (Tüm Gen Dizi Analizi)	BHLHE22 gene (Sequence Analysis-All Coding Exons)	BHLHE22	BHLHE22, BHLHB5, BETA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHLHB9 geni (Tüm Gen Dizi Analizi)	BHLHB9 gene (Sequence Analysis-All Coding Exons)	BHLHB9	BHLHB9, P60TRP, KIAA1701	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGLC1 geni (Tüm Gen Dizi Analizi)	IGLC1 gene (Sequence Analysis-All Coding Exons)	IGLC1	IGLC1, IGLC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGKV1OR2-108 geni (Tüm Gen Dizi Analizi)	IGKV1OR2-108 gene (Sequence Analysis-All Coding Exons)	IGKV1OR2-108	IGKV1OR2-108, IGO1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHV3-23 geni (Tüm Gen Dizi Analizi)	IGHV3-23 gene (Sequence Analysis-All Coding Exons)	IGHV3-23	IGHV3/23, IGHV323, DP47, VH26	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BGLT3 geni (Tüm Gen Dizi Analizi)	BGLT3 gene (Sequence Analysis-All Coding Exons)	BGLT3	BGLT3, LINC01083	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BGLAP geni (Tüm Gen Dizi Analizi)	BGLAP gene (Sequence Analysis-All Coding Exons)	BGLAP	BGLAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHG4 geni (Tüm Gen Dizi Analizi)	IGHG4 gene (Sequence Analysis-All Coding Exons)	IGHG4	IGHG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHG3 geni (Tüm Gen Dizi Analizi)	IGHG3 gene (Sequence Analysis-All Coding Exons)	IGHG3	IGHG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHG1 geni (Tüm Gen Dizi Analizi)	IGHG1 gene (Sequence Analysis-All Coding Exons)	IGHG1	IGHG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHE geni (Tüm Gen Dizi Analizi)	IGHE gene (Sequence Analysis-All Coding Exons)	IGHE	IGHE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHD3-3 geni (Tüm Gen Dizi Analizi)	IGHD3-3 gene (Sequence Analysis-All Coding Exons)	IGHD3-3	IGHD3-3, DXP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHD geni (Tüm Gen Dizi Analizi)	IGHD gene (Sequence Analysis-All Coding Exons)	IGHD	IGHD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHA2 geni (Tüm Gen Dizi Analizi)	IGHA2 gene (Sequence Analysis-All Coding Exons)	IGHA2	IGHA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IGHA1 geni (Tüm Gen Dizi Analizi)	IGHA1 gene (Sequence Analysis-All Coding Exons)	IGHA1	IGHA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEX5 geni (Tüm Gen Dizi Analizi)	BEX5 gene (Sequence Analysis-All Coding Exons)	BEX5	BEX5, NGFRAP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEX4 geni (Tüm Gen Dizi Analizi)	BEX4 gene (Sequence Analysis-All Coding Exons)	BEX4	BEX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEX3 geni (Tüm Gen Dizi Analizi)	BEX3 gene (Sequence Analysis-All Coding Exons)	BEX3	NGFRAP1, BEX3, NADE, DXS6984E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEX2 geni (Tüm Gen Dizi Analizi)	BEX2 gene (Sequence Analysis-All Coding Exons)	BEX2	BEX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEX1 geni (Tüm Gen Dizi Analizi)	BEX1 gene (Sequence Analysis-All Coding Exons)	BEX1	BEX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BET1L geni (Tüm Gen Dizi Analizi)	BET1L gene (Sequence Analysis-All Coding Exons)	BET1L	BET1L, GS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BET1 geni (Tüm Gen Dizi Analizi)	BET1 gene (Sequence Analysis-All Coding Exons)	BET1	BET1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEST4 geni (Tüm Gen Dizi Analizi)	BEST4 gene (Sequence Analysis-All Coding Exons)	BEST4	BEST4, VMD2L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEST3 geni (Tüm Gen Dizi Analizi)	BEST3 gene (Sequence Analysis-All Coding Exons)	BEST3	BEST3, VMD2L3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEST2 geni (Tüm Gen Dizi Analizi)	BEST2 gene (Sequence Analysis-All Coding Exons)	BEST2	BEST2, VMD2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BEND3 geni (Tüm Gen Dizi Analizi)	BEND3 gene (Sequence Analysis-All Coding Exons)	BEND3	BEND3, KIAA1553	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

BECN2 geni (Tüm Gen Dizi Analizi)	BECN2 gene (Sequence Analysis-All Coding Exons)	BECN2	BECN2, BECN1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BECN1 geni (Tüm Gen Dizi Analizi)	BECN1 gene (Sequence Analysis-All Coding Exons)	BECN1	BECN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BDP1 geni (Tüm Gen Dizi Analizi)	BDP1 gene (Sequence Analysis-All Coding Exons)	BDP1	BDP1, TFNR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BDNF-AS geni (Tüm Gen Dizi Analizi)	BDNF-AS gene (Sequence Analysis-All Coding Exons)	BDNF-AS	BDNFAS, BDNFOS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BDKRB2 geni (Tüm Gen Dizi Analizi)	BDKRB2 gene (Sequence Analysis-All Coding Exons)	BDKRB2	BDKRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BDKRB1 geni (Tüm Gen Dizi Analizi)	BDKRB1 gene (Sequence Analysis-All Coding Exons)	BDKRB1	BDKRB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BDH1 geni (Tüm Gen Dizi Analizi)	BDH1 gene (Sequence Analysis-All Coding Exons)	BDH1	BDH1, BDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCORL1 geni (Tüm Gen Dizi Analizi)	BCORL1 gene (Sequence Analysis-All Coding Exons)	BCORL1	BCORL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCO2 geni (Tüm Gen Dizi Analizi)	BCO2 gene (Sequence Analysis-All Coding Exons)	BCO2	BCDO2, BCO2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCLAF1 geni (Tüm Gen Dizi Analizi)	BCLAF1 gene (Sequence Analysis-All Coding Exons)	BCLAF1	BCLAF1, BTF, KIAA0164	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL9L geni (Tüm Gen Dizi Analizi)	BCL9L gene (Sequence Analysis-All Coding Exons)	BCL9L	BCL9L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL9 geni (Tüm Gen Dizi Analizi)	BCL9 gene (Sequence Analysis-All Coding Exons)	BCL9	BCL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL7C geni (Tüm Gen Dizi Analizi)	BCL7C gene (Sequence Analysis-All Coding Exons)	BCL7C	BCL7C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL7B geni (Tüm Gen Dizi Analizi)	BCL7B gene (Sequence Analysis-All Coding Exons)	BCL7B	BCL7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL6B geni (Tüm Gen Dizi Analizi)	BCL6B gene (Sequence Analysis-All Coding Exons)	BCL6B	BCL6B, BAZF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L2 geni (Tüm Gen Dizi Analizi)	BCL2L2 gene (Sequence Analysis-All Coding Exons)	BCL2L2	BCL2L2, BCLW	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L14 geni (Tüm Gen Dizi Analizi)	BCL2L14 gene (Sequence Analysis-All Coding Exons)	BCL2L14	BCLG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L12 geni (Tüm Gen Dizi Analizi)	BCL2L12 gene (Sequence Analysis-All Coding Exons)	BCL2L12	BCL2L12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L11 geni (Tüm Gen Dizi Analizi)	BCL2L11 gene (Sequence Analysis-All Coding Exons)	BCL2L11	BCL2L11, BIM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L10 geni (Tüm Gen Dizi Analizi)	BCL2L10 gene (Sequence Analysis-All Coding Exons)	BCL2L10	BCL2L10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2L1 geni (Tüm Gen Dizi Analizi)	BCL2L1 gene (Sequence Analysis-All Coding Exons)	BCL2L1	BCL2L1, BCLX, BCLXL, BCLX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCL2A1 geni (Tüm Gen Dizi Analizi)	BCL2A1 gene (Sequence Analysis-All Coding Exons)	BCL2A1	BCL2A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCCIP geni (Tüm Gen Dizi Analizi)	BCCIP gene (Sequence Analysis-All Coding Exons)	BCCIP	BCCIP, TOK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAS4 geni (Tüm Gen Dizi Analizi)	BCAS4 gene (Sequence Analysis-All Coding Exons)	BCAS4	BCAS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAS2 geni (Tüm Gen Dizi Analizi)	BCAS2 gene (Sequence Analysis-All Coding Exons)	BCAS2	BCAS2, DAM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAS1 geni (Tüm Gen Dizi Analizi)	BCAS1 gene (Sequence Analysis-All Coding Exons)	BCAS1	BCAS1, NABC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAR4 geni (Tüm Gen Dizi Analizi)	BCAR4 gene (Sequence Analysis-All Coding Exons)	BCAR4	BCAR4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAR3 geni (Tüm Gen Dizi Analizi)	BCAR3 gene (Sequence Analysis-All Coding Exons)	BCAR3	BCAR3, SH2D3B, NSP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAR1 geni (Tüm Gen Dizi Analizi)	BCAR1 gene (Sequence Analysis-All Coding Exons)	BCAR1	BCAR1, CRKAS, CAS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BBOX1 geni (Tüm Gen Dizi Analizi)	BBOX1 gene (Sequence Analysis-All Coding Exons)	BBOX1	BBOX1, BBOX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BBC3 geni (Tüm Gen Dizi Analizi)	BBC3 gene (Sequence Analysis-All Coding Exons)	BBC3	PUMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAZ2B geni (Tüm Gen Dizi Analizi)	BAZ2B gene (Sequence Analysis-All Coding Exons)	BAZ2B	BAZ2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAZ2A geni (Tüm Gen Dizi Analizi)	BAZ2A gene (Sequence Analysis-All Coding Exons)	BAZ2A	BAZ2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAZ1B geni (Tüm Gen Dizi Analizi)	BAZ1B gene (Sequence Analysis-All Coding Exons)	BAZ1B	BAZ1B, WSTF, WBSCR9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAZ1A geni (Tüm Gen Dizi Analizi)	BAZ1A gene (Sequence Analysis-All Coding Exons)	BAZ1A	BAZ1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BATF3 geni (Tüm Gen Dizi Analizi)	BATF3 gene (Sequence Analysis-All Coding Exons)	BATF3	BATF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BATF2 geni (Tüm Gen Dizi Analizi)	BATF2 gene (Sequence Analysis-All Coding Exons)	BATF2	BATF2, SARI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BATF geni (Tüm Gen Dizi Analizi)	BATF gene (Sequence Analysis-All Coding Exons)	BATF	BATF, BATF1, SFA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BASP1 geni (Tüm Gen Dizi Analizi)	BASP1 gene (Sequence Analysis-All Coding Exons)	BASP1	BASP1, CAP23, NAP22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BARX2 geni (Tüm Gen Dizi Analizi)	BARX2 gene (Sequence Analysis-All Coding Exons)	BARX2	BARX2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BARX1 geni (Tüm Gen Dizi Analizi)	BARX1 gene (Sequence Analysis-All Coding Exons)	BARX1	BARX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BARHL2 geni (Tüm Gen Dizi Analizi)	BARHL2 gene (Sequence Analysis-All Coding Exons)	BARHL2	BARHL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BARHL1 geni (Tüm Gen Dizi Analizi)	BARHL1 gene (Sequence Analysis-All Coding Exons)	BARHL1	BARHL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BANP geni (Tüm Gen Dizi Analizi)	BANP gene (Sequence Analysis-All Coding Exons)	BANP	BANP, SMAR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BANK1 geni (Tüm Gen Dizi Analizi)	BANK1 gene (Sequence Analysis-All Coding Exons)	BANK1	BANK1, FLJ20706, BANK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAMBI geni (Tüm Gen Dizi Analizi)	BAMBI gene (Sequence Analysis-All Coding Exons)	BAMBI	BAMBI, NMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAK1 geni (Tüm Gen Dizi Analizi)	BAK1 gene (Sequence Analysis-All Coding Exons)	BAK1	BAK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAIAP3 geni (Tüm Gen Dizi Analizi)	BAIAP3 gene (Sequence Analysis-All Coding Exons)	BAIAP3	BAIAP3, BAP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAIAP2L1 geni (Tüm Gen Dizi Analizi)	BAIAP2L1 gene (Sequence Analysis-All Coding Exons)	BAIAP2L1	BAIAP2L1, IRTKS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAIAP2 geni (Tüm Gen Dizi Analizi)	BAIAP2 gene (Sequence Analysis-All Coding Exons)	BAIAP2	BAIAP2, IRSP53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAHD1 geni (Tüm Gen Dizi Analizi)	BAHD1 gene (Sequence Analysis-All Coding Exons)	BAHD1	BAHD1, KIAA0945	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAG6 geni (Tüm Gen Dizi Analizi)	BAG6 gene (Sequence Analysis-All Coding Exons)	BAG6	BAG6, BAT3, D6S52E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAG4 geni (Tüm Gen Dizi Analizi)	BAG4 gene (Sequence Analysis-All Coding Exons)	BAG4	BAG4, SODD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAG2 geni (Tüm Gen Dizi Analizi)	BAG2 gene (Sequence Analysis-All Coding Exons)	BAG2	BAG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAG1 geni (Tüm Gen Dizi Analizi)	BAG1 gene (Sequence Analysis-All Coding Exons)	BAG1	BAG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAD geni (Tüm Gen Dizi Analizi)	BAD gene (Sequence Analysis-All Coding Exons)	BAD	BAD, BCL2L8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BACH2 geni (Tüm Gen Dizi Analizi)	BACH2 gene (Sequence Analysis-All Coding Exons)	BACH2	BACH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BACH1 geni (Tüm Gen Dizi Analizi)	BACH1 gene (Sequence Analysis-All Coding Exons)	BACH1	BACH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BACE2 geni (Tüm Gen Dizi Analizi)	BACE2 gene (Sequence Analysis-All Coding Exons)	BACE2	BACE2, ALP56, DRAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BACE1 geni (Tüm Gen Dizi Analizi)	BACE1 gene (Sequence Analysis-All Coding Exons)	BACE1	BACE1, BACE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BABAM1 geni (Tüm Gen Dizi Analizi)	BABAM1 gene (Sequence Analysis-All Coding Exons)	BABAM1	C10orf62, MERIT40, NBA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAALC geni (Tüm Gen Dizi Analizi)	BAALC gene (Sequence Analysis-All Coding Exons)	BAALC	BAALC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALT6 geni (Tüm Gen Dizi Analizi)	B4GALT6 gene (Sequence Analysis-All Coding Exons)	B4GALT6	B4GALT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALT5 geni (Tüm Gen Dizi Analizi)	B4GALT5 gene (Sequence Analysis-All Coding Exons)	B4GALT5	B4GALT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALT4 geni (Tüm Gen Dizi Analizi)	B4GALT4 gene (Sequence Analysis-All Coding Exons)	B4GALT4	B4GALT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALT3 geni (Tüm Gen Dizi Analizi)	B4GALT3 gene (Sequence Analysis-All Coding Exons)	B4GALT3	B4GALT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALT2 geni (Tüm Gen Dizi Analizi)	B4GALT2 gene (Sequence Analysis-All Coding Exons)	B4GALT2	B4GALT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALNT3 geni (Tüm Gen Dizi Analizi)	B4GALNT3 gene (Sequence Analysis-All Coding Exons)	B4GALNT3	B4GALNT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B4GALNT2 geni (Tüm Gen Dizi Analizi)	B4GALNT2 gene (Sequence Analysis-All Coding Exons)	B4GALNT2	B4GALGT2, GALGT2, SD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNTL1 geni (Tüm Gen Dizi Analizi)	B3GNTL1 gene (Sequence Analysis-All Coding Exons)	B3GNTL1	B3GNTL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT8 geni (Tüm Gen Dizi Analizi)	B3GNT8 gene (Sequence Analysis-All Coding Exons)	B3GNT8	B3GNT8, BGALT15, B3GALT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT7 geni (Tüm Gen Dizi Analizi)	B3GNT7 gene (Sequence Analysis-All Coding Exons)	B3GNT7	B3GNT7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT6 geni (Tüm Gen Dizi Analizi)	B3GNT6 gene (Sequence Analysis-All Coding Exons)	B3GNT6	B3GNT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT5 geni (Tüm Gen Dizi Analizi)	B3GNT5 gene (Sequence Analysis-All Coding Exons)	B3GNT5	B3GNT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT4 geni (Tüm Gen Dizi Analizi)	B3GNT4 gene (Sequence Analysis-All Coding Exons)	B3GNT4	B3GNT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT3 geni (Tüm Gen Dizi Analizi)	B3GNT3 gene (Sequence Analysis-All Coding Exons)	B3GNT3	TMEM3, B3GNT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GNT2 geni (Tüm Gen Dizi Analizi)	B3GNT2 gene (Sequence Analysis-All Coding Exons)	B3GNT2	B3GNT2, B3GNT1, B3GNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GAT2 geni (Tüm Gen Dizi Analizi)	B3GAT2 gene (Sequence Analysis-All Coding Exons)	B3GAT2	GLCAT5, KIAA1963	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GAT1 geni (Tüm Gen Dizi Analizi)	B3GAT1 gene (Sequence Analysis-All Coding Exons)	B3GAT1	B3GAT1, GLCATP, CD57, HNK1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GALT5 geni (Tüm Gen Dizi Analizi)	B3GALT5 gene (Sequence Analysis-All Coding Exons)	B3GALT5	B3GALT5, GLCT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GALT4 geni (Tüm Gen Dizi Analizi)	B3GALT4 gene (Sequence Analysis-All Coding Exons)	B3GALT4	B3GALT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GALT2 geni (Tüm Gen Dizi Analizi)	B3GALT2 gene (Sequence Analysis-All Coding Exons)	B3GALT2	B3GALT2, GLCT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
B3GALT1 geni (Tüm Gen Dizi Analizi)	B3GALT1 gene (Sequence Analysis-All Coding Exons)	B3GALT1	B3GALT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
IFI27L1 geni (Tüm Gen Dizi Analizi)	IFI27L1 gene (Sequence Analysis-All Coding Exons)	IFI27L1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AZU1 geni (Tüm Gen Dizi Analizi)	AZU1 gene (Sequence Analysis-All Coding Exons)	AZU1	AZU1, CAP37	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AZIN2 geni (Tüm Gen Dizi Analizi)	AZIN2 gene (Sequence Analysis-All Coding Exons)	AZIN2	AZIN2, ODC1L, KIAA1945	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AZIN1 geni (Tüm Gen Dizi Analizi)	AZIN1 gene (Sequence Analysis-All Coding Exons)	AZIN1	AZIN1, OAZIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AZI2 geni (Tüm Gen Dizi Analizi)	AZI2 gene (Sequence Analysis-All Coding Exons)	AZI2	AZI2, AZ2, NAP1, TILP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AZGP1 geni (Tüm Gen Dizi Analizi)	AZGP1 gene (Sequence Analysis-All Coding Exons)	AZGP1	AZGP1, ZAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AXL geni (Tüm Gen Dizi Analizi)	AXL gene (Sequence Analysis-All Coding Exons)	AXL	AXL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AWAT2 geni (Tüm Gen Dizi Analizi)	AWAT2 gene (Sequence Analysis-All Coding Exons)	AWAT2	AWAT2, DC4, MFAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AWAT1 geni (Tüm Gen Dizi Analizi)	AWAT1 gene (Sequence Analysis-All Coding Exons)	AWAT1	AWAT1, DGA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AVPR1B geni (Tüm Gen Dizi Analizi)	AVPR1B gene (Sequence Analysis-All Coding Exons)	AVPR1B	AVPR1B, AVPR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

AVPR1A geni (Tüm Gen Dizi Analizi)	AVPR1A gene (Sequence Analysis-All Coding Exons)	AVPR1A	AVPR1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AVL9 geni (Tüm Gen Dizi Analizi)	AVL9 gene (Sequence Analysis-All Coding Exons)	AVL9	AVL9, KIAA0241	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AVIL geni (Tüm Gen Dizi Analizi)	AVIL gene (Sequence Analysis-All Coding Exons)	AVIL	AVIL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AVEN geni (Tüm Gen Dizi Analizi)	AVEN gene (Sequence Analysis-All Coding Exons)	AVEN	AVEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AURKB geni (Tüm Gen Dizi Analizi)	AURKB gene (Sequence Analysis-All Coding Exons)	AURKB	STK12, ARK2, AIK2, AIM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AURKAIP1 geni (Tüm Gen Dizi Analizi)	AURKAIP1 gene (Sequence Analysis-All Coding Exons)	AURKAIP1	AURKAIP1, AKIP, AIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AUP1 geni (Tüm Gen Dizi Analizi)	AUP1 gene (Sequence Analysis-All Coding Exons)	AUP1	AUP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATXN7L3B geni (Tüm Gen Dizi Analizi)	ATXN7L3B gene (Sequence Analysis-All Coding Exons)	ATXN7L3B	ATXN7L3B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATXN3L geni (Tüm Gen Dizi Analizi)	ATXN3L gene (Sequence Analysis-All Coding Exons)	ATXN3L	ATXN3L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATXN2L geni (Tüm Gen Dizi Analizi)	ATXN2L gene (Sequence Analysis-All Coding Exons)	ATXN2L	ATXN2L, A2D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATXN1L geni (Tüm Gen Dizi Analizi)	ATXN1L gene (Sequence Analysis-All Coding Exons)	ATXN1L	ATXN1L, BOAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATRNL1 geni (Tüm Gen Dizi Analizi)	ATRNL1 gene (Sequence Analysis-All Coding Exons)	ATRNL1	ATRNL1, KIAA0534, ALP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATRN geni (Tüm Gen Dizi Analizi)	ATRN gene (Sequence Analysis-All Coding Exons)	ATRN	ATRN, MGCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATRIP geni (Tüm Gen Dizi Analizi)	ATRIP gene (Sequence Analysis-All Coding Exons)	ATRIP	ATRIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATPIF1 geni (Tüm Gen Dizi Analizi)	ATPIF1 gene (Sequence Analysis-All Coding Exons)	ATPIF1	ATP1F1, IF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP9B geni (Tüm Gen Dizi Analizi)	ATP9B gene (Sequence Analysis-All Coding Exons)	ATP9B	ATP9B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP8B3 geni (Tüm Gen Dizi Analizi)	ATP8B3 gene (Sequence Analysis-All Coding Exons)	ATP8B3	ATP8B3, ATPIK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP8B2 geni (Tüm Gen Dizi Analizi)	ATP8B2 gene (Sequence Analysis-All Coding Exons)	ATP8B2	ATP8B2, ATPID	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1G2 geni (Tüm Gen Dizi Analizi)	ATP6V1G2 gene (Sequence Analysis-All Coding Exons)	ATP6V1G2	ATP6V1G2, ATP6G	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1G1 geni (Tüm Gen Dizi Analizi)	ATP6V1G1 gene (Sequence Analysis-All Coding Exons)	ATP6V1G1	ATP6V1G1, ATP6G1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1E2 geni (Tüm Gen Dizi Analizi)	ATP6V1E2 gene (Sequence Analysis-All Coding Exons)	ATP6V1E2	ATP6V1E2, ATP6E1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1E1 geni (Tüm Gen Dizi Analizi)	ATP6V1E1 gene (Sequence Analysis-All Coding Exons)	ATP6V1E1	ATP6V1E1, ATP6E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1D geni (Tüm Gen Dizi Analizi)	ATP6V1D gene (Sequence Analysis-All Coding Exons)	ATP6V1D	ATP6V1D, ATP6M	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1C1 geni (Tüm Gen Dizi Analizi)	ATP6V1C1 gene (Sequence Analysis-All Coding Exons)	ATP6V1C1	ATP6V1C1, ATP6C, ATP6D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1A geni (Tüm Gen Dizi Analizi)	ATP6V1A gene (Sequence Analysis-All Coding Exons)	ATP6V1A	ATP6V1A1, HO68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0E2 geni (Tüm Gen Dizi Analizi)	ATP6V0E2 gene (Sequence Analysis-All Coding Exons)	ATP6V0E2	ATP6V0E2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0E1 geni (Tüm Gen Dizi Analizi)	ATP6V0E1 gene (Sequence Analysis-All Coding Exons)	ATP6V0E1	ATP6V0E, ATP6H	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0D1 geni (Tüm Gen Dizi Analizi)	ATP6V0D1 gene (Sequence Analysis-All Coding Exons)	ATP6V0D1	ATP6V0D1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0C geni (Tüm Gen Dizi Analizi)	ATP6V0C gene (Sequence Analysis-All Coding Exons)	ATP6V0C	ATP6V0C, ATP6L, ATP6C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0B geni (Tüm Gen Dizi Analizi)	ATP6V0B gene (Sequence Analysis-All Coding Exons)	ATP6V0B	ATP6V0B, ATP6F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V0A1 geni (Tüm Gen Dizi Analizi)	ATP6V0A1 gene (Sequence Analysis-All Coding Exons)	ATP6V0A1	ATP6V0A1, ATP6N1A, VPP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5SL geni (Tüm Gen Dizi Analizi)	ATP5SL gene (Sequence Analysis-All Coding Exons)	ATP5SL	ATP5SL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5O geni (Tüm Gen Dizi Analizi)	ATP5O gene (Sequence Analysis-All Coding Exons)	ATP5O	ATP5O	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5J geni (Tüm Gen Dizi Analizi)	ATP5J gene (Sequence Analysis-All Coding Exons)	ATP5J	ATP5J, ATP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5I geni (Tüm Gen Dizi Analizi)	ATP5I gene (Sequence Analysis-All Coding Exons)	ATP5I	ATP5I, ATP5K	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5G3 geni (Tüm Gen Dizi Analizi)	ATP5G3 gene (Sequence Analysis-All Coding Exons)	ATP5G3	ATP5G3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5G2 geni (Tüm Gen Dizi Analizi)	ATP5G2 gene (Sequence Analysis-All Coding Exons)	ATP5G2	ATP5G2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5G1 geni (Tüm Gen Dizi Analizi)	ATP5G1 gene (Sequence Analysis-All Coding Exons)	ATP5G1	ATP5G1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5F1 geni (Tüm Gen Dizi Analizi)	ATP5F1 gene (Sequence Analysis-All Coding Exons)	ATP5F1	ATP5F1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5D geni (Tüm Gen Dizi Analizi)	ATP5D gene (Sequence Analysis-All Coding Exons)	ATP5D	ATP5D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5C1 geni (Tüm Gen Dizi Analizi)	ATP5C1 gene (Sequence Analysis-All Coding Exons)	ATP5C1	ATP5C1, ATP5CL1, ATP5C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP5B geni (Tüm Gen Dizi Analizi)	ATP5B gene (Sequence Analysis-All Coding Exons)	ATP5B	ATP5B, ATPSB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP4B geni (Tüm Gen Dizi Analizi)	ATP4B gene (Sequence Analysis-All Coding Exons)	ATP4B	ATP4B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP4A geni (Tüm Gen Dizi Analizi)	ATP4A gene (Sequence Analysis-All Coding Exons)	ATP4A	ATP4A, ATP6A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP2C2 geni (Tüm Gen Dizi Analizi)	ATP2C2 gene (Sequence Analysis-All Coding Exons)	ATP2C2	ATP2C2, SPCA2, KIAA0703	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP2B4 geni (Tüm Gen Dizi Analizi)	ATP2B4 gene (Sequence Analysis-All Coding Exons)	ATP2B4	ATP2B4, ATP2B2, PMCA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP2B1 geni (Tüm Gen Dizi Analizi)	ATP2B1 gene (Sequence Analysis-All Coding Exons)	ATP2B1	ATP2B1, PMCA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP2A3 geni (Tüm Gen Dizi Analizi)	ATP2A3 gene (Sequence Analysis-All Coding Exons)	ATP2A3	ATP2A3, SERCA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP1B3 geni (Tüm Gen Dizi Analizi)	ATP1B3 gene (Sequence Analysis-All Coding Exons)	ATP1B3	ATP1B3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP1B2 geni (Tüm Gen Dizi Analizi)	ATP1B2 gene (Sequence Analysis-All Coding Exons)	ATP1B2	ATP1B2, AMOG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP1A4 geni (Tüm Gen Dizi Analizi)	ATP1A4 gene (Sequence Analysis-All Coding Exons)	ATP1A4	ATP1A4, ATP1AL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP1A1 geni (Tüm Gen Dizi Analizi)	ATP1A1 gene (Sequence Analysis-All Coding Exons)	ATP1A1	ATP1A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP13A4 geni (Tüm Gen Dizi Analizi)	ATP13A4 gene (Sequence Analysis-All Coding Exons)	ATP13A4	ATP13A4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP13A3 geni (Tüm Gen Dizi Analizi)	ATP13A3 gene (Sequence Analysis-All Coding Exons)	ATP13A3	ATP13A3, AFURS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP12A geni (Tüm Gen Dizi Analizi)	ATP12A gene (Sequence Analysis-All Coding Exons)	ATP12A	ATP12A, ATP1AL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP11C geni (Tüm Gen Dizi Analizi)	ATP11C gene (Sequence Analysis-All Coding Exons)	ATP11C	ATP11C, ATPIQ, ATPIG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP11B geni (Tüm Gen Dizi Analizi)	ATP11B gene (Sequence Analysis-All Coding Exons)	ATP11B	ATP11B, ATPIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP11A geni (Tüm Gen Dizi Analizi)	ATP11A gene (Sequence Analysis-All Coding Exons)	ATP11A	ATP11A, ATPIS, ATPIH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP10A geni (Tüm Gen Dizi Analizi)	ATP10A gene (Sequence Analysis-All Coding Exons)	ATP10A	ATP10A, ATP10C, ATPVC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATOX1 geni (Tüm Gen Dizi Analizi)	ATOX1 gene (Sequence Analysis-All Coding Exons)	ATOX1	ATOX1, HAH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATOH1 geni (Tüm Gen Dizi Analizi)	ATOH1 gene (Sequence Analysis-All Coding Exons)	ATOH1	ATOH1, ATH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATMIN geni (Tüm Gen Dizi Analizi)	ATMIN gene (Sequence Analysis-All Coding Exons)	ATMIN	ATMIN, ASCIZ, KIAA0431	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATL2 geni (Tüm Gen Dizi Analizi)	ATL2 gene (Sequence Analysis-All Coding Exons)	ATL2	ARL3IP2, ATL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG9B geni (Tüm Gen Dizi Analizi)	ATG9B gene (Sequence Analysis-All Coding Exons)	ATG9B	ATG9B, NOS3AS, APG9L2, SONE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG9A geni (Tüm Gen Dizi Analizi)	ATG9A gene (Sequence Analysis-All Coding Exons)	ATG9A	ATG9A, APG9L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG7 geni (Tüm Gen Dizi Analizi)	ATG7 gene (Sequence Analysis-All Coding Exons)	ATG7	ATG7, APG7L, GSA7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG5 geni (Tüm Gen Dizi Analizi)	ATG5 gene (Sequence Analysis-All Coding Exons)	ATG5	APG5L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG4D geni (Tüm Gen Dizi Analizi)	ATG4D gene (Sequence Analysis-All Coding Exons)	ATG4D	ATG4D, APG4D, AUTL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG4C geni (Tüm Gen Dizi Analizi)	ATG4C gene (Sequence Analysis-All Coding Exons)	ATG4C	ATG4C, APG4C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG4B geni (Tüm Gen Dizi Analizi)	ATG4B gene (Sequence Analysis-All Coding Exons)	ATG4B	ATG4B, APG4B, KIAA0943	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG4A geni (Tüm Gen Dizi Analizi)	ATG4A gene (Sequence Analysis-All Coding Exons)	ATG4A	ATG4A, APG4A, AUTL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG2B geni (Tüm Gen Dizi Analizi)	ATG2B gene (Sequence Analysis-All Coding Exons)	ATG2B	ATG2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG2A geni (Tüm Gen Dizi Analizi)	ATG2A gene (Sequence Analysis-All Coding Exons)	ATG2A	ATG2A, KIAA0404	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG14 geni (Tüm Gen Dizi Analizi)	ATG14 gene (Sequence Analysis-All Coding Exons)	ATG14	ATG14, ATG14L, KIAA0831, BARKOR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG13 geni (Tüm Gen Dizi Analizi)	ATG13 gene (Sequence Analysis-All Coding Exons)	ATG13	ATG13, KIAA0652, PARATARG8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG101 geni (Tüm Gen Dizi Analizi)	ATG101 gene (Sequence Analysis-All Coding Exons)	ATG101	ATG101, C12orf44	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG10 geni (Tüm Gen Dizi Analizi)	ATG10 gene (Sequence Analysis-All Coding Exons)	ATG10	ATG10, APG10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF7IP2 geni (Tüm Gen Dizi Analizi)	ATF7IP2 gene (Sequence Analysis-All Coding Exons)	ATF7IP2	ATF7IP2, MCAF2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF7IP geni (Tüm Gen Dizi Analizi)	ATF7IP gene (Sequence Analysis-All Coding Exons)	ATF7IP	ATF7IP, AM, MCAF, MCAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF7 geni (Tüm Gen Dizi Analizi)	ATF7 gene (Sequence Analysis-All Coding Exons)	ATF7	ATF7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF6B geni (Tüm Gen Dizi Analizi)	ATF6B gene (Sequence Analysis-All Coding Exons)	ATF6B	ATF6B, CREBL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF5 geni (Tüm Gen Dizi Analizi)	ATF5 gene (Sequence Analysis-All Coding Exons)	ATF5	ATF5, ATFX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF4 geni (Tüm Gen Dizi Analizi)	ATF4 gene (Sequence Analysis-All Coding Exons)	ATF4	ATF4, CREB2, TAXREB67	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF3 geni (Tüm Gen Dizi Analizi)	ATF3 gene (Sequence Analysis-All Coding Exons)	ATF3	ATF3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF2 geni (Tüm Gen Dizi Analizi)	ATF2 gene (Sequence Analysis-All Coding Exons)	ATF2	ATF2, CREB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATF1 geni (Tüm Gen Dizi Analizi)	ATF1 gene (Sequence Analysis-All Coding Exons)	ATF1	ATF1, TREB36	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAT1 geni (Tüm Gen Dizi Analizi)	ATAT1 gene (Sequence Analysis-All Coding Exons)	ATAT1	ATAT1, MEC17, C6orf134	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAD5 geni (Tüm Gen Dizi Analizi)	ATAD5 gene (Sequence Analysis-All Coding Exons)	ATAD5	ATAD5, C17orf41, FRAG1, FLJ12735	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAD3C geni (Tüm Gen Dizi Analizi)	ATAD3C gene (Sequence Analysis-All Coding Exons)	ATAD3C	ATAD3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAD3B geni (Tüm Gen Dizi Analizi)	ATAD3B gene (Sequence Analysis-All Coding Exons)	ATAD3B	ATAD3B, TOB3, KIAA1273	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSPBAP1 geni (Tüm Gen Dizi Analizi)	HSPBAP1 gene (Sequence Analysis-All Coding Exons)	HSPBAP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAD2B geni (Tüm Gen Dizi Analizi)	ATAD2B gene (Sequence Analysis-All Coding Exons)	ATAD2B	ATAD2B, KIAA1240	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATAD2 geni (Tüm Gen Dizi Analizi)	ATAD2 gene (Sequence Analysis-All Coding Exons)	ATAD2	ATAD2, ANCCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ATAD1 geni (Tüm Gen Dizi Analizi)	ATAD1 gene (Sequence Analysis-All Coding Exons)	ATAD1	ATAD1, THORASE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASZ1 geni (Tüm Gen Dizi Analizi)	ASZ1 gene (Sequence Analysis-All Coding Exons)	ASZ1	ASZ1, ALP1, ANKL1, GASZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASXL2 geni (Tüm Gen Dizi Analizi)	ASXL2 gene (Sequence Analysis-All Coding Exons)	ASXL2	ASXL2, KIAA1685, SHAPNS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Note=A chromosomal aberration involving ASXL2 is a cause of therapy-related myelodysplastic syndrome. Translocation t(2;8)(q23;p11.2) with KAT6A generates a KAT6A-ASXL2 fusion protein.	EDTA Blood Sample
ASTN2 geni (Tüm Gen Dizi Analizi)	ASTN2 gene (Sequence Analysis-All Coding Exons)	ASTN2	ASTN2, KIAA0634	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASTN1 geni (Tüm Gen Dizi Analizi)	ASTN1 gene (Sequence Analysis-All Coding Exons)	ASTN1	ASTN1, ASTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASPRV1 geni (Tüm Gen Dizi Analizi)	ASPRV1 gene (Sequence Analysis-All Coding Exons)	ASPRV1	ASPRV1, SASP, TAPS, MUNO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASNA1 geni (Tüm Gen Dizi Analizi)	ASNA1 gene (Sequence Analysis-All Coding Exons)	ASNA1	ASNA1, ARSA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASMTL geni (Tüm Gen Dizi Analizi)	ASMTL gene (Sequence Analysis-All Coding Exons)	ASMTL	ASMTLX, ASTML	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASMTL geni (Tüm Gen Dizi Analizi)	ASMTL gene (Sequence Analysis-All Coding Exons)	ASMTL	ASMTLY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASMT geni (Tüm Gen Dizi Analizi)	ASMT gene (Sequence Analysis-All Coding Exons)	ASMT	ASMT, HIOMT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASMT geni (Tüm Gen Dizi Analizi)	ASMT gene (Sequence Analysis-All Coding Exons)	ASMT	ASMTY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASIC5 geni (Tüm Gen Dizi Analizi)	ASIC5 gene (Sequence Analysis-All Coding Exons)	ASIC5	ASIC5, HINAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASIC4 geni (Tüm Gen Dizi Analizi)	ASIC4 gene (Sequence Analysis-All Coding Exons)	ASIC4	ACCN4, ASIC4, BNAC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASIC3 geni (Tüm Gen Dizi Analizi)	ASIC3 gene (Sequence Analysis-All Coding Exons)	ASIC3	ACCN3, ASIC3, TNAC1, hINaC6"	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASIC2 geni (Tüm Gen Dizi Analizi)	ASIC2 gene (Sequence Analysis-All Coding Exons)	ASIC2	ACCN1, BNC1, MDEG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASIC1 geni (Tüm Gen Dizi Analizi)	ASIC1 gene (Sequence Analysis-All Coding Exons)	ASIC1	ACCN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASH2L geni (Tüm Gen Dizi Analizi)	ASH2L gene (Sequence Analysis-All Coding Exons)	ASH2L	ASH2L, ASH2L2, ASH2L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASH1L geni (Tüm Gen Dizi Analizi)	ASH1L gene (Sequence Analysis-All Coding Exons)	ASH1L	ASH1L, KIAA1420, ASH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASGR2 geni (Tüm Gen Dizi Analizi)	ASGR2 gene (Sequence Analysis-All Coding Exons)	ASGR2	ASGR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASGR1 geni (Tüm Gen Dizi Analizi)	ASGR1 gene (Sequence Analysis-All Coding Exons)	ASGR1	ASGR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASF1B geni (Tüm Gen Dizi Analizi)	ASF1B gene (Sequence Analysis-All Coding Exons)	ASF1B	ASF1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASF1A geni (Tüm Gen Dizi Analizi)	ASF1A gene (Sequence Analysis-All Coding Exons)	ASF1A	ASF1A, CIA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASCL2 geni (Tüm Gen Dizi Analizi)	ASCL2 gene (Sequence Analysis-All Coding Exons)	ASCL2	ASCL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASCC3 geni (Tüm Gen Dizi Analizi)	ASCC3 gene (Sequence Analysis-All Coding Exons)	ASCC3	ASCC3, p200	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSH2D geni (Tüm Gen Dizi Analizi)	HSH2D gene (Sequence Analysis-All Coding Exons)	HSH2D		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASCC2 geni (Tüm Gen Dizi Analizi)	ASCC2 gene (Sequence Analysis-All Coding Exons)	ASCC2	ASCC2, p100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB9 geni (Tüm Gen Dizi Analizi)	ASB9 gene (Sequence Analysis-All Coding Exons)	ASB9	ASB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB8 geni (Tüm Gen Dizi Analizi)	ASB8 gene (Sequence Analysis-All Coding Exons)	ASB8	ASB8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB7 geni (Tüm Gen Dizi Analizi)	ASB7 gene (Sequence Analysis-All Coding Exons)	ASB7	ASB7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB6 geni (Tüm Gen Dizi Analizi)	ASB6 gene (Sequence Analysis-All Coding Exons)	ASB6	ASB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB5 geni (Tüm Gen Dizi Analizi)	ASB5 gene (Sequence Analysis-All Coding Exons)	ASB5	ASB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB4 geni (Tüm Gen Dizi Analizi)	ASB4 gene (Sequence Analysis-All Coding Exons)	ASB4	ASB4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB3 geni (Tüm Gen Dizi Analizi)	ASB3 gene (Sequence Analysis-All Coding Exons)	ASB3	ASB3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB2 geni (Tüm Gen Dizi Analizi)	ASB2 gene (Sequence Analysis-All Coding Exons)	ASB2	ASB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB16 geni (Tüm Gen Dizi Analizi)	ASB16 gene (Sequence Analysis-All Coding Exons)	ASB16	ASB16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB13 geni (Tüm Gen Dizi Analizi)	ASB13 gene (Sequence Analysis-All Coding Exons)	ASB13	ASB13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB12 geni (Tüm Gen Dizi Analizi)	ASB12 gene (Sequence Analysis-All Coding Exons)	ASB12	ASB12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB11 geni (Tüm Gen Dizi Analizi)	ASB11 gene (Sequence Analysis-All Coding Exons)	ASB11	ASB11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASB1 geni (Tüm Gen Dizi Analizi)	ASB1 gene (Sequence Analysis-All Coding Exons)	ASB1	ASB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASAP3 geni (Tüm Gen Dizi Analizi)	ASAP3 gene (Sequence Analysis-All Coding Exons)	ASAP3	ASAP3, UPLC1, CENTB6, DDEFL1, ACAP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASAP2 geni (Tüm Gen Dizi Analizi)	ASAP2 gene (Sequence Analysis-All Coding Exons)	ASAP2	DDEF2, PAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASAP1 geni (Tüm Gen Dizi Analizi)	ASAP1 gene (Sequence Analysis-All Coding Exons)	ASAP1	ASAP1, DDEF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HSD17B12 geni (Tüm Gen Dizi Analizi)	HSD17B12 gene (Sequence Analysis-All Coding Exons)	HSD17B12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASAH2 geni (Tüm Gen Dizi Analizi)	ASAH2 gene (Sequence Analysis-All Coding Exons)	ASAH2	ASAH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AS3MT geni (Tüm Gen Dizi Analizi)	AS3MT gene (Sequence Analysis-All Coding Exons)	AS3MT	AS3MT, CYT19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARVCF geni (Tüm Gen Dizi Analizi)	ARVCF gene (Sequence Analysis-All Coding Exons)	ARVCF	ARVCF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ART5 geni (Tüm Gen Dizi Analizi)	ART5 gene (Sequence Analysis-All Coding Exons)	ART5	ART5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ART3 geni (Tüm Gen Dizi Analizi)	ART3 gene (Sequence Analysis-All Coding Exons)	ART3	ART3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ART1 geni (Tüm Gen Dizi Analizi)	ART1 gene (Sequence Analysis-All Coding Exons)	ART1	ART1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSK geni (Tüm Gen Dizi Analizi)	ARSK gene (Sequence Analysis-All Coding Exons)	ARSK	ARSK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSJ geni (Tüm Gen Dizi Analizi)	ARSJ gene (Sequence Analysis-All Coding Exons)	ARSJ	ARSJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSI geni (Tüm Gen Dizi Analizi)	ARSI gene (Sequence Analysis-All Coding Exons)	ARSI	ARSI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSH geni (Tüm Gen Dizi Analizi)	ARSH gene (Sequence Analysis-All Coding Exons)	ARSH	ARSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSG geni (Tüm Gen Dizi Analizi)	ARSG gene (Sequence Analysis-All Coding Exons)	ARSG	ARSG, KIAA1001	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSF geni (Tüm Gen Dizi Analizi)	ARSF gene (Sequence Analysis-All Coding Exons)	ARSF	ARSF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARSD geni (Tüm Gen Dizi Analizi)	ARSD gene (Sequence Analysis-All Coding Exons)	ARSD	ARSD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARRDC3 geni (Tüm Gen Dizi Analizi)	ARRDC3 gene (Sequence Analysis-All Coding Exons)	ARRDC3	ARRDC3, TLIMP, KIAA1376	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARRB2 geni (Tüm Gen Dizi Analizi)	ARRB2 gene (Sequence Analysis-All Coding Exons)	ARRB2	ARRB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARRB1 geni (Tüm Gen Dizi Analizi)	ARRB1 gene (Sequence Analysis-All Coding Exons)	ARRB1	ARRB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARR3 geni (Tüm Gen Dizi Analizi)	ARR3 gene (Sequence Analysis-All Coding Exons)	ARR3	ARR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPP19 geni (Tüm Gen Dizi Analizi)	ARPP19 gene (Sequence Analysis-All Coding Exons)	ARPP19	ARPP19, ARPP16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPIN geni (Tüm Gen Dizi Analizi)	ARPIN gene (Sequence Analysis-All Coding Exons)	ARPIN	C15orf38, ARPIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPC5 geni (Tüm Gen Dizi Analizi)	ARPC5 gene (Sequence Analysis-All Coding Exons)	ARPC5	ARPC5, ARC16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPC4 geni (Tüm Gen Dizi Analizi)	ARPC4 gene (Sequence Analysis-All Coding Exons)	ARPC4	ARPC4, ARC20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPC3 geni (Tüm Gen Dizi Analizi)	ARPC3 gene (Sequence Analysis-All Coding Exons)	ARPC3	ARPC3, ARC21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPC1B geni (Tüm Gen Dizi Analizi)	ARPC1B gene (Sequence Analysis-All Coding Exons)	ARPC1B	ARPC1B, ARC41	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRH2 geni (Tüm Gen Dizi Analizi)	HRH2 gene (Sequence Analysis-All Coding Exons)	HRH2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPC1A geni (Tüm Gen Dizi Analizi)	ARPC1A gene (Sequence Analysis-All Coding Exons)	ARPC1A	ARPC1A, SOP2L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARNTL2 geni (Tüm Gen Dizi Analizi)	ARNTL2 gene (Sequence Analysis-All Coding Exons)	ARNTL2	ARNTL2, BMAL2, CLIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARNTL geni (Tüm Gen Dizi Analizi)	ARNTL gene (Sequence Analysis-All Coding Exons)	ARNTL	ARNTL, BMAL1, TIC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARNT geni (Tüm Gen Dizi Analizi)	ARNT gene (Sequence Analysis-All Coding Exons)	ARNT	ARNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HRASLS5 geni (Tüm Gen Dizi Analizi)	HRASLS5 gene (Sequence Analysis-All Coding Exons)	HRASLS5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMT1 geni (Tüm Gen Dizi Analizi)	ARMT1 gene (Sequence Analysis-All Coding Exons)	ARMT1	ARMT1, C5orf211	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMCX3 geni (Tüm Gen Dizi Analizi)	ARMCX3 gene (Sequence Analysis-All Coding Exons)	ARMCX3	ARMCX3, ALEX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMCX2 geni (Tüm Gen Dizi Analizi)	ARMCX2 gene (Sequence Analysis-All Coding Exons)	ARMCX2	ARMCX2, ALEX2, KIAA0512	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMCX1 geni (Tüm Gen Dizi Analizi)	ARMCX1 gene (Sequence Analysis-All Coding Exons)	ARMCX1	ARMCX1, ALEX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMC3 geni (Tüm Gen Dizi Analizi)	ARMC3 gene (Sequence Analysis-All Coding Exons)	ARMC3	ARMC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARMC10 geni (Tüm Gen Dizi Analizi)	ARMC10 gene (Sequence Analysis-All Coding Exons)	ARMC10	ARMC10, SVH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL9 geni (Tüm Gen Dizi Analizi)	ARL9 gene (Sequence Analysis-All Coding Exons)	ARL9	ARL9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL8B geni (Tüm Gen Dizi Analizi)	ARL8B gene (Sequence Analysis-All Coding Exons)	ARL8B	ARL8B, GIE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL8A geni (Tüm Gen Dizi Analizi)	ARL8A gene (Sequence Analysis-All Coding Exons)	ARL8A	ARL8A, GIE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL6IP6 geni (Tüm Gen Dizi Analizi)	ARL6IP6 gene (Sequence Analysis-All Coding Exons)	ARL6IP6	ARL6IP6, AIP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL6IP5 geni (Tüm Gen Dizi Analizi)	ARL6IP5 gene (Sequence Analysis-All Coding Exons)	ARL6IP5	ARL6IP5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL6IP4 geni (Tüm Gen Dizi Analizi)	ARL6IP4 gene (Sequence Analysis-All Coding Exons)	ARL6IP4	ARL6IP4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL5B geni (Tüm Gen Dizi Analizi)	ARL5B gene (Sequence Analysis-All Coding Exons)	ARL5B	ARL5B, ARL8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL5A geni (Tüm Gen Dizi Analizi)	ARL5A gene (Sequence Analysis-All Coding Exons)	ARL5A	ARL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL4D geni (Tüm Gen Dizi Analizi)	ARL4D gene (Sequence Analysis-All Coding Exons)	ARL4D	ARL4D, ARF4L	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL4C geni (Tüm Gen Dizi Analizi)	ARL4C gene (Sequence Analysis-All Coding Exons)	ARL4C	ARL4C, ARL7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL4A geni (Tüm Gen Dizi Analizi)	ARL4A gene (Sequence Analysis-All Coding Exons)	ARL4A	ARL4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL3 geni (Tüm Gen Dizi Analizi)	ARL3 gene (Sequence Analysis-All Coding Exons)	ARL3	ARL3, ARFL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL2 geni (Tüm Gen Dizi Analizi)	ARL2 gene (Sequence Analysis-All Coding Exons)	ARL2	ARL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL14EP geni (Tüm Gen Dizi Analizi)	ARL14EP gene (Sequence Analysis-All Coding Exons)	ARL14EP	C11orf46	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ARL14 geni (Tüm Gen Dizi Analizi)	ARL14 gene (Sequence Analysis-All Coding Exons)	ARL14	ARL14, ARF7	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL11 geni (Tüm Gen Dizi Analizi)	ARL11 gene (Sequence Analysis-All Coding Exons)	ARL11	ARL11, ARLTS1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARL1 geni (Tüm Gen Dizi Analizi)	ARL1 gene (Sequence Analysis-All Coding Exons)	ARL1	ARL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARIH2 geni (Tüm Gen Dizi Analizi)	ARIH2 gene (Sequence Analysis-All Coding Exons)	ARIH2	ARIH2, ARI2, TRIAD1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARIH1 geni (Tüm Gen Dizi Analizi)	ARIH1 gene (Sequence Analysis-All Coding Exons)	ARIH1	ARIH1, ARI, UBCH7BP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID5B geni (Tüm Gen Dizi Analizi)	ARID5B gene (Sequence Analysis-All Coding Exons)	ARID5B	ARID5B, MRF2, DESRT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID5A geni (Tüm Gen Dizi Analizi)	ARID5A gene (Sequence Analysis-All Coding Exons)	ARID5A	ARID5A, MRF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID4B geni (Tüm Gen Dizi Analizi)	ARID4B gene (Sequence Analysis-All Coding Exons)	ARID4B	ARID4B, SAP180, RBP1L1, RBBP1L1, BRCAA1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID4A geni (Tüm Gen Dizi Analizi)	ARID4A gene (Sequence Analysis-All Coding Exons)	ARID4A	ARID4A, RBP1, RBBP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID3B geni (Tüm Gen Dizi Analizi)	ARID3B gene (Sequence Analysis-All Coding Exons)	ARID3B	ARID3B, DRIL2, BDP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID3A geni (Tüm Gen Dizi Analizi)	ARID3A gene (Sequence Analysis-All Coding Exons)	ARID3A	DRIL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARID2 geni (Tüm Gen Dizi Analizi)	ARID2 gene (Sequence Analysis-All Coding Exons)	ARID2	ARID2, BAF200, KIAA1557	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF7 geni (Tüm Gen Dizi Analizi)	ARHGEF7 gene (Sequence Analysis-All Coding Exons)	ARHGEF7	ARHGEF7, PIXB, COOL1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF5 geni (Tüm Gen Dizi Analizi)	ARHGEF5 gene (Sequence Analysis-All Coding Exons)	ARHGEF5	ARHGEF5, TIM	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF40 geni (Tüm Gen Dizi Analizi)	ARHGEF40 gene (Sequence Analysis-All Coding Exons)	ARHGEF40	SOLO, FLJ10357	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF4 geni (Tüm Gen Dizi Analizi)	ARHGEF4 gene (Sequence Analysis-All Coding Exons)	ARHGEF4	ARHGEF4, ASEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF3 geni (Tüm Gen Dizi Analizi)	ARHGEF3 gene (Sequence Analysis-All Coding Exons)	ARHGEF3	ARHGEF3, XPLN	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF28 geni (Tüm Gen Dizi Analizi)	ARHGEF28 gene (Sequence Analysis-All Coding Exons)	ARHGEF28	RGNEF, p190RHOGEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF25 geni (Tüm Gen Dizi Analizi)	ARHGEF25 gene (Sequence Analysis-All Coding Exons)	ARHGEF25	ARHGEF25, GEFT	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF2 geni (Tüm Gen Dizi Analizi)	ARHGEF2 gene (Sequence Analysis-All Coding Exons)	ARHGEF2	ARHGEF2, GEFH1, KIAA0651	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF19 geni (Tüm Gen Dizi Analizi)	ARHGEF19 gene (Sequence Analysis-All Coding Exons)	ARHGEF19	ARHGEF19, WGEF	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF18 geni (Tüm Gen Dizi Analizi)	ARHGEF18 gene (Sequence Analysis-All Coding Exons)	ARHGEF18	SRHGEF18, KIAA0521	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF17 geni (Tüm Gen Dizi Analizi)	ARHGEF17 gene (Sequence Analysis-All Coding Exons)	ARHGEF17	ARHGEF17, RHOGEF17, TEM4, KIAA0237	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF15 geni (Tüm Gen Dizi Analizi)	ARHGEF15 gene (Sequence Analysis-All Coding Exons)	ARHGEF15	ARGEF15, KIAA0915	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF12 geni (Tüm Gen Dizi Analizi)	ARHGEF12 gene (Sequence Analysis-All Coding Exons)	ARHGEF12	ARHGEF12, LARG, KIAA0382	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF10L geni (Tüm Gen Dizi Analizi)	ARHGEF10L gene (Sequence Analysis-All Coding Exons)	ARHGEF10L	ARHGEF10L, GRINCH2EF, KIAA1626	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF1 geni (Tüm Gen Dizi Analizi)	ARHGEF1 gene (Sequence Analysis-All Coding Exons)	ARHGEF1	ARHGEF1, LBCL2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGDIB geni (Tüm Gen Dizi Analizi)	ARHGDIB gene (Sequence Analysis-All Coding Exons)	ARHGDIB	ARHGDIB, GDID4, D4	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP9 geni (Tüm Gen Dizi Analizi)	ARHGAP9 gene (Sequence Analysis-All Coding Exons)	ARHGAP9	ARHGAP9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP8 geni (Tüm Gen Dizi Analizi)	ARHGAP8 gene (Sequence Analysis-All Coding Exons)	ARHGAP8	ARHGAP8, BPGAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP6 geni (Tüm Gen Dizi Analizi)	ARHGAP6 gene (Sequence Analysis-All Coding Exons)	ARHGAP6	ARHGAP6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP5 geni (Tüm Gen Dizi Analizi)	ARHGAP5 gene (Sequence Analysis-All Coding Exons)	ARHGAP5	ARHGAP5, RHOGAP5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP45 geni (Tüm Gen Dizi Analizi)	ARHGAP45 gene (Sequence Analysis-All Coding Exons)	ARHGAP45	HMHA1, HLA-HA1, KIAA0223	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP42 geni (Tüm Gen Dizi Analizi)	ARHGAP42 gene (Sequence Analysis-All Coding Exons)	ARHGAP42	ARHGAP42, GRAF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP4 geni (Tüm Gen Dizi Analizi)	ARHGAP4 gene (Sequence Analysis-All Coding Exons)	ARHGAP4	RGC1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP39 geni (Tüm Gen Dizi Analizi)	ARHGAP39 gene (Sequence Analysis-All Coding Exons)	ARHGAP39	ARHGAP39, VILSE, KIAA1688	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP36 geni (Tüm Gen Dizi Analizi)	ARHGAP36 gene (Sequence Analysis-All Coding Exons)	ARHGAP36	ARHGAP36	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP35 geni (Tüm Gen Dizi Analizi)	ARHGAP35 gene (Sequence Analysis-All Coding Exons)	ARHGAP35	GRLF1, P190A	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP33 geni (Tüm Gen Dizi Analizi)	ARHGAP33 gene (Sequence Analysis-All Coding Exons)	ARHGAP33	ARHGAP33, TCGAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP32 geni (Tüm Gen Dizi Analizi)	ARHGAP32 gene (Sequence Analysis-All Coding Exons)	ARHGAP32	ARHGAP32, RICS, GRIT, p200RHOGAP, p200GAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP30 geni (Tüm Gen Dizi Analizi)	ARHGAP30 gene (Sequence Analysis-All Coding Exons)	ARHGAP30	ARHGAP30	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP29 geni (Tüm Gen Dizi Analizi)	ARHGAP29 gene (Sequence Analysis-All Coding Exons)	ARHGAP29	ARHGAP29, PARG1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP28 geni (Tüm Gen Dizi Analizi)	ARHGAP28 gene (Sequence Analysis-All Coding Exons)	ARHGAP28	ARHGAP28, KIAA1314	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP27 geni (Tüm Gen Dizi Analizi)	ARHGAP27 gene (Sequence Analysis-All Coding Exons)	ARHGAP27	ARHGAP27, CAMGAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP25 geni (Tüm Gen Dizi Analizi)	ARHGAP25 gene (Sequence Analysis-All Coding Exons)	ARHGAP25	ARHGAP25, KIAA0053	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP24 geni (Tüm Gen Dizi Analizi)	ARHGAP24 gene (Sequence Analysis-All Coding Exons)	ARHGAP24	ARHGAP24, RCGAP72, Eti GAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP23 geni (Tüm Gen Dizi Analizi)	ARHGAP23 gene (Sequence Analysis-All Coding Exons)	ARHGAP23	ARHGAP23, KIAA1501	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP22 geni (Tüm Gen Dizi Analizi)	ARHGAP22 gene (Sequence Analysis-All Coding Exons)	ARHGAP22	ARHGAP22	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP21 geni (Tüm Gen Dizi Analizi)	ARHGAP21 gene (Sequence Analysis-All Coding Exons)	ARHGAP21	ARHGAP21, KIAA1424	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP19 geni (Tüm Gen Dizi Analizi)	ARHGAP19 gene (Sequence Analysis-All Coding Exons)	ARHGAP19	ARHGAP19	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP18 geni (Tüm Gen Dizi Analizi)	ARHGAP18 gene (Sequence Analysis-All Coding Exons)	ARHGAP18	ARHGAP18	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP17 geni (Tüm Gen Dizi Analizi)	ARHGAP17 gene (Sequence Analysis-All Coding Exons)	ARHGAP17	ARHGAP17, RICH1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP15 geni (Tüm Gen Dizi Analizi)	ARHGAP15 gene (Sequence Analysis-All Coding Exons)	ARHGAP15	ARHGAP15, BM046	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP12 geni (Tüm Gen Dizi Analizi)	ARHGAP12 gene (Sequence Analysis-All Coding Exons)	ARHGAP12	ARHGAP12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP11B geni (Tüm Gen Dizi Analizi)	ARHGAP11B gene (Sequence Analysis-All Coding Exons)	ARHGAP11B	ARHGAP11B, FAM7B1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP11A geni (Tüm Gen Dizi Analizi)	ARHGAP11A gene (Sequence Analysis-All Coding Exons)	ARHGAP11A	ARHGAP11A, KIAA0013	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP10 geni (Tüm Gen Dizi Analizi)	ARHGAP10 gene (Sequence Analysis-All Coding Exons)	ARHGAP10	ARHGAP10, GRAF2, PSGAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP1 geni (Tüm Gen Dizi Analizi)	ARHGAP1 gene (Sequence Analysis-All Coding Exons)	ARHGAP1	ARHGAP1, RHOGAP1, CDC42GAP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARGLU1 geni (Tüm Gen Dizi Analizi)	ARGLU1 gene (Sequence Analysis-All Coding Exons)	ARGLU1	ARGLU1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARGFX geni (Tüm Gen Dizi Analizi)	ARGFX gene (Sequence Analysis-All Coding Exons)	ARGFX	ARGFX	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARG2 geni (Tüm Gen Dizi Analizi)	ARG2 gene (Sequence Analysis-All Coding Exons)	ARG2	ARG2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFRP1 geni (Tüm Gen Dizi Analizi)	ARFRP1 gene (Sequence Analysis-All Coding Exons)	ARFRP1	ARFRP1, ARP	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFIP2 geni (Tüm Gen Dizi Analizi)	ARFIP2 gene (Sequence Analysis-All Coding Exons)	ARFIP2	ARFIP2, POR1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFGEF1 geni (Tüm Gen Dizi Analizi)	ARFGEF1 gene (Sequence Analysis-All Coding Exons)	ARFGEF1	ARFGEF1, ARFGEP1, BIG1, P200	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFGAP3 geni (Tüm Gen Dizi Analizi)	ARFGAP3 gene (Sequence Analysis-All Coding Exons)	ARFGAP3	ARFGAP3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFGAP2 geni (Tüm Gen Dizi Analizi)	ARFGAP2 gene (Sequence Analysis-All Coding Exons)	ARFGAP2	ARFGAP2, ZNF289	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFGAP1 geni (Tüm Gen Dizi Analizi)	ARFGAP1 gene (Sequence Analysis-All Coding Exons)	ARFGAP1	ARFGAP1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARF6 geni (Tüm Gen Dizi Analizi)	ARF6 gene (Sequence Analysis-All Coding Exons)	ARF6	ARF6	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARF5 geni (Tüm Gen Dizi Analizi)	ARF5 gene (Sequence Analysis-All Coding Exons)	ARF5	ARF5	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARF4 geni (Tüm Gen Dizi Analizi)	ARF4 gene (Sequence Analysis-All Coding Exons)	ARF4	ARF4, ARF2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARF3 geni (Tüm Gen Dizi Analizi)	ARF3 gene (Sequence Analysis-All Coding Exons)	ARF3	ARF3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARF1 geni (Tüm Gen Dizi Analizi)	ARF1 gene (Sequence Analysis-All Coding Exons)	ARF1	ARF1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AREL1 geni (Tüm Gen Dizi Analizi)	AREL1 gene (Sequence Analysis-All Coding Exons)	AREL1	AREL1, KIAA0317	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AREG geni (Tüm Gen Dizi Analizi)	AREG gene (Sequence Analysis-All Coding Exons)	AREG	AREG	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARCN1 geni (Tüm Gen Dizi Analizi)	ARCN1 gene (Sequence Analysis-All Coding Exons)	ARCN1	ARCN1, SRMMD	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARC geni (Tüm Gen Dizi Analizi)	ARC gene (Sequence Analysis-All Coding Exons)	ARC	ARC, KIAA0278	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARAP3 geni (Tüm Gen Dizi Analizi)	ARAP3 gene (Sequence Analysis-All Coding Exons)	ARAP3	ARAP3, CENTD3	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARAP2 geni (Tüm Gen Dizi Analizi)	ARAP2 gene (Sequence Analysis-All Coding Exons)	ARAP2	ARAP2, CENTD1, KIAA0580	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARAP1 geni (Tüm Gen Dizi Analizi)	ARAP1 gene (Sequence Analysis-All Coding Exons)	ARAP1	ARAP1, CENTD2, ARAP1, KIAA0782	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARAF geni (Tüm Gen Dizi Analizi)	ARAF gene (Sequence Analysis-All Coding Exons)	ARAF	ARAF1, RAFA1, PKS2	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQR geni (Tüm Gen Dizi Analizi)	AQR gene (Sequence Analysis-All Coding Exons)	AQR	AQR, IBP160, KIAA0560	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP9 geni (Tüm Gen Dizi Analizi)	AQP9 gene (Sequence Analysis-All Coding Exons)	AQP9	AQP9	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP8 geni (Tüm Gen Dizi Analizi)	AQP8 gene (Sequence Analysis-All Coding Exons)	AQP8	AQP8	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP6 geni (Tüm Gen Dizi Analizi)	AQP6 gene (Sequence Analysis-All Coding Exons)	AQP6	AQP6, AQP2L	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP4 geni (Tüm Gen Dizi Analizi)	AQP4 gene (Sequence Analysis-All Coding Exons)	AQP4	AQP4, MWC	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP12A geni (Tüm Gen Dizi Analizi)	AQP12A gene (Sequence Analysis-All Coding Exons)	AQP12A	AQP12A, AQPX2, AQP12	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP11 geni (Tüm Gen Dizi Analizi)	AQP11 gene (Sequence Analysis-All Coding Exons)	AQP11	AQP11, AQPX1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APTR geni (Tüm Gen Dizi Analizi)	APTR gene (Sequence Analysis-All Coding Exons)	APTR	APTR	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APPL2 geni (Tüm Gen Dizi Analizi)	APPL2 gene (Sequence Analysis-All Coding Exons)	APPL2	APPL2, FLJ10659	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APPBP2 geni (Tüm Gen Dizi Analizi)	APPBP2 gene (Sequence Analysis-All Coding Exons)	APPBP2	APPBP2, PAT1	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HNRNPAB geni (Tüm Gen Dizi Analizi)	HNRNPAB gene (Sequence Analysis-All Coding Exons)	HNRNPAB		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOOL geni (Tüm Gen Dizi Analizi)	APOOL gene (Sequence Analysis-All Coding Exons)	APOOL	APOOL, FAM121A, Cxorf33	Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOO geni (Tüm Gen Dizi Analizi)	APOO gene (Sequence Analysis-All Coding Exons)	APOO		Tüm Gen Dizi Analizi' Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

APOLD1 geni (Tüm Gen Dizi Analizi)	APOLD1 gene (Sequence Analysis-All Coding Exons)	APOLD1	APOLD1, VERGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOL6 geni (Tüm Gen Dizi Analizi)	APOL6 gene (Sequence Analysis-All Coding Exons)	APOL6	APOL6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOL5 geni (Tüm Gen Dizi Analizi)	APOL5 gene (Sequence Analysis-All Coding Exons)	APOL5	APOL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOL3 geni (Tüm Gen Dizi Analizi)	APOL3 gene (Sequence Analysis-All Coding Exons)	APOL3	APOL3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOH geni (Tüm Gen Dizi Analizi)	APOH gene (Sequence Analysis-All Coding Exons)	APOH	APOH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOF geni (Tüm Gen Dizi Analizi)	APOF gene (Sequence Analysis-All Coding Exons)	APOF	APOF, LTIP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOD geni (Tüm Gen Dizi Analizi)	APOD gene (Sequence Analysis-All Coding Exons)	APOD	APOD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOC4 geni (Tüm Gen Dizi Analizi)	APOC4 gene (Sequence Analysis-All Coding Exons)	APOC4	APOC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOC1 geni (Tüm Gen Dizi Analizi)	APOC1 gene (Sequence Analysis-All Coding Exons)	APOC1	APOC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBR geni (Tüm Gen Dizi Analizi)	APOBR gene (Sequence Analysis-All Coding Exons)	APOBR	APOBR, APOB48R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC4 geni (Tüm Gen Dizi Analizi)	APOBEC4 gene (Sequence Analysis-All Coding Exons)	APOBEC4	APOBEC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3H geni (Tüm Gen Dizi Analizi)	APOBEC3H gene (Sequence Analysis-All Coding Exons)	APOBEC3H	APOBEC3H, A8P10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3G geni (Tüm Gen Dizi Analizi)	APOBEC3G gene (Sequence Analysis-All Coding Exons)	APOBEC3G	APOBEC3G, MDS019, CEM15, FLJ27240	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3F geni (Tüm Gen Dizi Analizi)	APOBEC3F gene (Sequence Analysis-All Coding Exons)	APOBEC3F	APOBEC3F	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3C geni (Tüm Gen Dizi Analizi)	APOBEC3C gene (Sequence Analysis-All Coding Exons)	APOBEC3C	APOBEC3C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3B geni (Tüm Gen Dizi Analizi)	APOBEC3B gene (Sequence Analysis-All Coding Exons)	APOBEC3B	APOBEC3B, PHRBNL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3A geni (Tüm Gen Dizi Analizi)	APOBEC3A gene (Sequence Analysis-All Coding Exons)	APOBEC3A	APOBEC3A, PHRBN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC2 geni (Tüm Gen Dizi Analizi)	APOBEC2 gene (Sequence Analysis-All Coding Exons)	APOBEC2	APOBEC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC1 geni (Tüm Gen Dizi Analizi)	APOBEC1 gene (Sequence Analysis-All Coding Exons)	APOBEC1	APOBEC1, BEDP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOA4 geni (Tüm Gen Dizi Analizi)	APOA4 gene (Sequence Analysis-All Coding Exons)	APOA4	APOA4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APMAP geni (Tüm Gen Dizi Analizi)	APMAP gene (Sequence Analysis-All Coding Exons)	APMAP	APMAP, C20orf3, BSCV	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APLP2 geni (Tüm Gen Dizi Analizi)	APLP2 gene (Sequence Analysis-All Coding Exons)	APLP2	APLP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APLP1 geni (Tüm Gen Dizi Analizi)	APLP1 gene (Sequence Analysis-All Coding Exons)	APLP1	APLP1, APLP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APLNR geni (Tüm Gen Dizi Analizi)	APLNR gene (Sequence Analysis-All Coding Exons)	APLNR	APLNR, AGTRL1, APJ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APLN geni (Tüm Gen Dizi Analizi)	APLN gene (Sequence Analysis-All Coding Exons)	APLN	APLN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APLF geni (Tüm Gen Dizi Analizi)	APLF gene (Sequence Analysis-All Coding Exons)	APLF	C2orf13, APFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APIP geni (Tüm Gen Dizi Analizi)	APIP gene (Sequence Analysis-All Coding Exons)	APIP	APIP, CGI29, MMRP19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
API5 geni (Tüm Gen Dizi Analizi)	API5 gene (Sequence Analysis-All Coding Exons)	API5	API5, AAC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APH1B geni (Tüm Gen Dizi Analizi)	APH1B gene (Sequence Analysis-All Coding Exons)	APH1B	APH1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APH1A geni (Tüm Gen Dizi Analizi)	APH1A gene (Sequence Analysis-All Coding Exons)	APH1A	APH1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APEX2 geni (Tüm Gen Dizi Analizi)	APEX2 gene (Sequence Analysis-All Coding Exons)	APEX2	APEX2, APE2, XTH2, APEXL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APEX1 geni (Tüm Gen Dizi Analizi)	APEX1 gene (Sequence Analysis-All Coding Exons)	APEX1	APEX, APE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APELA geni (Tüm Gen Dizi Analizi)	APELA gene (Sequence Analysis-All Coding Exons)	APELA	ELA, LOC100506013	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APEH geni (Tüm Gen Dizi Analizi)	APEH gene (Sequence Analysis-All Coding Exons)	APEH	APEH, D3S48E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBB3 geni (Tüm Gen Dizi Analizi)	APBB3 gene (Sequence Analysis-All Coding Exons)	APBB3	APBB3, FE65L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBB2 geni (Tüm Gen Dizi Analizi)	APBB2 gene (Sequence Analysis-All Coding Exons)	APBB2	APBB2, FE65L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBB1 geni (Tüm Gen Dizi Analizi)	APBB1 gene (Sequence Analysis-All Coding Exons)	APBB1	APBB1, FE65	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBA3 geni (Tüm Gen Dizi Analizi)	APBA3 gene (Sequence Analysis-All Coding Exons)	APBA3	APBA3, X11L2, MINT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBA2 geni (Tüm Gen Dizi Analizi)	APBA2 gene (Sequence Analysis-All Coding Exons)	APBA2	APBA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBA1 geni (Tüm Gen Dizi Analizi)	APBA1 gene (Sequence Analysis-All Coding Exons)	APBA1	APBA1, X11, D95411E, MINT1, LIN10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APAF1 geni (Tüm Gen Dizi Analizi)	APAF1 gene (Sequence Analysis-All Coding Exons)	APAF1	APAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APS1 geni (Tüm Gen Dizi Analizi)	APS1 gene (Sequence Analysis-All Coding Exons)	APS1	APS1, C20orf29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APM1 geni (Tüm Gen Dizi Analizi)	APM1 gene (Sequence Analysis-All Coding Exons)	APM1	APM1, MUDENG, MUD, C14orf108	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APB1 geni (Tüm Gen Dizi Analizi)	APB1 gene (Sequence Analysis-All Coding Exons)	APB1	APB1, DKFZp761E198	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP3S1 geni (Tüm Gen Dizi Analizi)	AP3S1 gene (Sequence Analysis-All Coding Exons)	AP3S1	AP3S1, CLAPS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP3M2 geni (Tüm Gen Dizi Analizi)	AP3M2 gene (Sequence Analysis-All Coding Exons)	AP3M2	AP3M2, CLA20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP3M1 geni (Tüm Gen Dizi Analizi)	AP3M1 gene (Sequence Analysis-All Coding Exons)	AP3M1	AP3M1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP2M1 geni (Tüm Gen Dizi Analizi)	AP2M1 gene (Sequence Analysis-All Coding Exons)	AP2M1	AP2M1, CLAPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP2B1 geni (Tüm Gen Dizi Analizi)	AP2B1 gene (Sequence Analysis-All Coding Exons)	AP2B1	AP2B1, CLAPB1, ADTB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP2A2 geni (Tüm Gen Dizi Analizi)	AP2A2 gene (Sequence Analysis-All Coding Exons)	AP2A2	AP2A2, KIAA0899	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP2A1 geni (Tüm Gen Dizi Analizi)	AP2A1 gene (Sequence Analysis-All Coding Exons)	AP2A1	AP2A1, CLAPA1, ADTAA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1M2 geni (Tüm Gen Dizi Analizi)	AP1M2 gene (Sequence Analysis-All Coding Exons)	AP1M2	AP1M2, MU1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1M1 geni (Tüm Gen Dizi Analizi)	AP1M1 gene (Sequence Analysis-All Coding Exons)	AP1M1	AP1M1, AP47, CLAPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1G1 geni (Tüm Gen Dizi Analizi)	AP1G1 gene (Sequence Analysis-All Coding Exons)	AP1G1	AP1G1, ADTG, CLAPG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1B1 geni (Tüm Gen Dizi Analizi)	AP1B1 gene (Sequence Analysis-All Coding Exons)	AP1B1	AP1B1, ADTB1, BAM22, CLAPB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1AR geni (Tüm Gen Dizi Analizi)	AP1AR gene (Sequence Analysis-All Coding Exons)	AP1AR	C4orf16, GBAR, 2C18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AOX1 geni (Tüm Gen Dizi Analizi)	AOX1 gene (Sequence Analysis-All Coding Exons)	AOX1	AOX1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HLA-DRB3 geni (Tüm Gen Dizi Analizi)	HLA-DRB3 gene (Sequence Analysis-All Coding Exons)	HLA-DRB3	HLA-DRB3, HLA-DRS2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AOC3 geni (Tüm Gen Dizi Analizi)	AOC3 gene (Sequence Analysis-All Coding Exons)	AOC3	AOC3, HPAO, VAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AOC2 geni (Tüm Gen Dizi Analizi)	AOC2 gene (Sequence Analysis-All Coding Exons)	AOC2	AOC2, RAO	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AOC1 geni (Tüm Gen Dizi Analizi)	AOC1 gene (Sequence Analysis-All Coding Exons)	AOC1	ABP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AOAH geni (Tüm Gen Dizi Analizi)	AOAH gene (Sequence Analysis-All Coding Exons)	AOAH	AOAH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA9 geni (Tüm Gen Dizi Analizi)	ANXA9 gene (Sequence Analysis-All Coding Exons)	ANXA9	ANXA9, ANX31	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA8 geni (Tüm Gen Dizi Analizi)	ANXA8 gene (Sequence Analysis-All Coding Exons)	ANXA8	ANXA8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA7 geni (Tüm Gen Dizi Analizi)	ANXA7 gene (Sequence Analysis-All Coding Exons)	ANXA7	ANXA7, SNX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA6 geni (Tüm Gen Dizi Analizi)	ANXA6 gene (Sequence Analysis-All Coding Exons)	ANXA6	ANXA6, CBP68	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA4 geni (Tüm Gen Dizi Analizi)	ANXA4 gene (Sequence Analysis-All Coding Exons)	ANXA4	ANXA4, ANX4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA3 geni (Tüm Gen Dizi Analizi)	ANXA3 gene (Sequence Analysis-All Coding Exons)	ANXA3	ANXA3, ANX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA2R geni (Tüm Gen Dizi Analizi)	ANXA2R gene (Sequence Analysis-All Coding Exons)	ANXA2R	ANXA2R, C5orf39, AX2R, AXIIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA2 geni (Tüm Gen Dizi Analizi)	ANXA2 gene (Sequence Analysis-All Coding Exons)	ANXA2	ANXA2, ANX2L4, LPC2D, LIP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA13 geni (Tüm Gen Dizi Analizi)	ANXA13 gene (Sequence Analysis-All Coding Exons)	ANXA13	ANX13, ISA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA11 geni (Tüm Gen Dizi Analizi)	ANXA11 gene (Sequence Analysis-All Coding Exons)	ANXA11	ANXA11, ANX11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA10 geni (Tüm Gen Dizi Analizi)	ANXA10 gene (Sequence Analysis-All Coding Exons)	ANXA10	ANXA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANXA1 geni (Tüm Gen Dizi Analizi)	ANXA1 gene (Sequence Analysis-All Coding Exons)	ANXA1	ANXA1, LPC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANPEP geni (Tüm Gen Dizi Analizi)	ANPEP gene (Sequence Analysis-All Coding Exons)	ANPEP	ANPEP, PEPN, CD13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANP32E geni (Tüm Gen Dizi Analizi)	ANP32E gene (Sequence Analysis-All Coding Exons)	ANP32E	ANP32E, LANPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANP32D geni (Tüm Gen Dizi Analizi)	ANP32D gene (Sequence Analysis-All Coding Exons)	ANP32D	ANP32D, PP32R2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANP32A geni (Tüm Gen Dizi Analizi)	ANP32A gene (Sequence Analysis-All Coding Exons)	ANP32A	ANP32A, PHAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANO8 geni (Tüm Gen Dizi Analizi)	ANO8 gene (Sequence Analysis-All Coding Exons)	ANO8	TMEM16H, KIAA1623	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANO7 geni (Tüm Gen Dizi Analizi)	ANO7 gene (Sequence Analysis-All Coding Exons)	ANO7	TMEM16G, PCANAP5, IPCAS, DTMPP, NGEP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANO4 geni (Tüm Gen Dizi Analizi)	ANO4 gene (Sequence Analysis-All Coding Exons)	ANO4	TMEM16D, FLJ34272	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANO2 geni (Tüm Gen Dizi Analizi)	ANO2 gene (Sequence Analysis-All Coding Exons)	ANO2	ANO2, TMEM16B, C12orf3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANO1 geni (Tüm Gen Dizi Analizi)	ANO1 gene (Sequence Analysis-All Coding Exons)	ANO1	ANO1, TMEM16A, FLJ10261	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKS4B geni (Tüm Gen Dizi Analizi)	ANKS4B gene (Sequence Analysis-All Coding Exons)	ANKS4B	ANKS4B, HARP, FLJ38819	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKS3 geni (Tüm Gen Dizi Analizi)	ANKS3 gene (Sequence Analysis-All Coding Exons)	ANKS3	ANKS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKS1B geni (Tüm Gen Dizi Analizi)	ANKS1B gene (Sequence Analysis-All Coding Exons)	ANKS1B	ANKS1B, EB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKS1A geni (Tüm Gen Dizi Analizi)	ANKS1A gene (Sequence Analysis-All Coding Exons)	ANKS1A	ANKS1, ODIN, KIAA0229	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD7 geni (Tüm Gen Dizi Analizi)	ANKRD7 gene (Sequence Analysis-All Coding Exons)	ANKRD7	ANKRD7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD6 geni (Tüm Gen Dizi Analizi)	ANKRD6 gene (Sequence Analysis-All Coding Exons)	ANKRD6	ANKRD6, KIAA0957	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD55 geni (Tüm Gen Dizi Analizi)	ANKRD55 gene (Sequence Analysis-All Coding Exons)	ANKRD55	ANKRD55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD55 geni (Tüm Gen Dizi Analizi)	ANKRD55 gene (Sequence Analysis-All Coding Exons)	ANKRD55	ANKRD55	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD54 geni (Tüm Gen Dizi Analizi)	ANKRD54 gene (Sequence Analysis-All Coding Exons)	ANKRD54	ANKRD54, LIAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD53 geni (Tüm Gen Dizi Analizi)	ANKRD53 gene (Sequence Analysis-All Coding Exons)	ANKRD53	ANKRD53	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ANKRD30B geni (Tüm Gen Dizi Analizi)	ANKRD30B gene (Sequence Analysis-All Coding Exons)	ANKRD30B	ANKRD30B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD30A geni (Tüm Gen Dizi Analizi)	ANKRD30A gene (Sequence Analysis-All Coding Exons)	ANKRD30A	ANKRD30A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD28 geni (Tüm Gen Dizi Analizi)	ANKRD28 gene (Sequence Analysis-All Coding Exons)	ANKRD28	ANKRD28, PITK, KIAA0379	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD23 geni (Tüm Gen Dizi Analizi)	ANKRD23 gene (Sequence Analysis-All Coding Exons)	ANKRD23	ANKRD23, DARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD2 geni (Tüm Gen Dizi Analizi)	ANKRD2 gene (Sequence Analysis-All Coding Exons)	ANKRD2	ANKRD2, ARPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD17 geni (Tüm Gen Dizi Analizi)	ANKRD17 gene (Sequence Analysis-All Coding Exons)	ANKRD17	ANKRD17, GTAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD13D geni (Tüm Gen Dizi Analizi)	ANKRD13D gene (Sequence Analysis-All Coding Exons)	ANKRD13D	ANKRD13D	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD13C geni (Tüm Gen Dizi Analizi)	ANKRD13C gene (Sequence Analysis-All Coding Exons)	ANKRD13C	ANKRD13C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD13B geni (Tüm Gen Dizi Analizi)	ANKRD13B gene (Sequence Analysis-All Coding Exons)	ANKRD13B	ANKRD13B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD13A geni (Tüm Gen Dizi Analizi)	ANKRD13A gene (Sequence Analysis-All Coding Exons)	ANKRD13A	ANKRD13A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD12 geni (Tüm Gen Dizi Analizi)	ANKRD12 gene (Sequence Analysis-All Coding Exons)	ANKRD12	ANKRD12, ANCO1, KIAA0874	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRD1 geni (Tüm Gen Dizi Analizi)	ANKRD1 gene (Sequence Analysis-All Coding Exons)	ANKRD1	ANKRD1, CARP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKRA2 geni (Tüm Gen Dizi Analizi)	ANKRA2 gene (Sequence Analysis-All Coding Exons)	ANKRA2	ANKRA2, ANKRA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKHD1 geni (Tüm Gen Dizi Analizi)	ANKHD1 gene (Sequence Analysis-All Coding Exons)	ANKHD1	ANKHD1, MASK, KIAA1085, VMAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKFY1 geni (Tüm Gen Dizi Analizi)	ANKFY1 gene (Sequence Analysis-All Coding Exons)	ANKFY1	ANKFY1, ANKHZN, KIAA1255	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANKAR geni (Tüm Gen Dizi Analizi)	ANKAR gene (Sequence Analysis-All Coding Exons)	ANKAR	FLJ25415	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPTL8 geni (Tüm Gen Dizi Analizi)	ANGPTL8 gene (Sequence Analysis-All Coding Exons)	ANGPTL8	ANGPTL8, C19orf80, LIPASIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPTL6 geni (Tüm Gen Dizi Analizi)	ANGPTL6 gene (Sequence Analysis-All Coding Exons)	ANGPTL6	ANGPTL6, AGF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPTL5 geni (Tüm Gen Dizi Analizi)	ANGPTL5 gene (Sequence Analysis-All Coding Exons)	ANGPTL5	ANGPTL5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPTL2 geni (Tüm Gen Dizi Analizi)	ANGPTL2 gene (Sequence Analysis-All Coding Exons)	ANGPTL2	ANGPTL2, ARP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPT4 geni (Tüm Gen Dizi Analizi)	ANGPT4 gene (Sequence Analysis-All Coding Exons)	ANGPT4	ANGPT4, ANG4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPT2 geni (Tüm Gen Dizi Analizi)	ANGPT2 gene (Sequence Analysis-All Coding Exons)	ANGPT2	ANGPT2, ANG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPT1 geni (Tüm Gen Dizi Analizi)	ANGPT1 gene (Sequence Analysis-All Coding Exons)	ANGPT1	ANGPT1, ANG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC7 geni (Tüm Gen Dizi Analizi)	ANAPC7 gene (Sequence Analysis-All Coding Exons)	ANAPC7	ANAPC7, APC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC5 geni (Tüm Gen Dizi Analizi)	ANAPC5 gene (Sequence Analysis-All Coding Exons)	ANAPC5	ANAPC5, APC5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC4 geni (Tüm Gen Dizi Analizi)	ANAPC4 gene (Sequence Analysis-All Coding Exons)	ANAPC4	ANAPC4, APC4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC2 geni (Tüm Gen Dizi Analizi)	ANAPC2 gene (Sequence Analysis-All Coding Exons)	ANAPC2	ANAPC2, APC2, KIAA1406	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC16 geni (Tüm Gen Dizi Analizi)	ANAPC16 gene (Sequence Analysis-All Coding Exons)	ANAPC16	ANAPC16, MSAG, C10orf104	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC15 geni (Tüm Gen Dizi Analizi)	ANAPC15 gene (Sequence Analysis-All Coding Exons)	ANAPC15	ANAPC15, APC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC13 geni (Tüm Gen Dizi Analizi)	ANAPC13 gene (Sequence Analysis-All Coding Exons)	ANAPC13	ANAPC13, APC13, SWM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC11 geni (Tüm Gen Dizi Analizi)	ANAPC11 gene (Sequence Analysis-All Coding Exons)	ANAPC11	ANAPC11, APC11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC10 geni (Tüm Gen Dizi Analizi)	ANAPC10 gene (Sequence Analysis-All Coding Exons)	ANAPC10	ANAPC10, DOC1, APC10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANAPC1 geni (Tüm Gen Dizi Analizi)	ANAPC1 gene (Sequence Analysis-All Coding Exons)	ANAPC1	ANAP1, APC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMZ2 geni (Tüm Gen Dizi Analizi)	AMZ2 gene (Sequence Analysis-All Coding Exons)	AMZ2	AMZ2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMZ1 geni (Tüm Gen Dizi Analizi)	AMZ1 gene (Sequence Analysis-All Coding Exons)	AMZ1	AMZ1, KIAA1950	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMY2B geni (Tüm Gen Dizi Analizi)	AMY2B gene (Sequence Analysis-All Coding Exons)	AMY2B	AMY2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMY2A geni (Tüm Gen Dizi Analizi)	AMY2A gene (Sequence Analysis-All Coding Exons)	AMY2A	AMY2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMY1C geni (Tüm Gen Dizi Analizi)	AMY1C gene (Sequence Analysis-All Coding Exons)	AMY1C	AMY1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMY1B geni (Tüm Gen Dizi Analizi)	AMY1B gene (Sequence Analysis-All Coding Exons)	AMY1B	AMY1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMY1A geni (Tüm Gen Dizi Analizi)	AMY1A gene (Sequence Analysis-All Coding Exons)	AMY1A	AMY1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMTN geni (Tüm Gen Dizi Analizi)	AMTN gene (Sequence Analysis-All Coding Exons)	AMTN	AMTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMPH geni (Tüm Gen Dizi Analizi)	AMPH gene (Sequence Analysis-All Coding Exons)	AMPH	AMPH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMOTL2 geni (Tüm Gen Dizi Analizi)	AMOTL2 gene (Sequence Analysis-All Coding Exons)	AMOTL2	AMOTL2, KIAA0989	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMOTL1 geni (Tüm Gen Dizi Analizi)	AMOTL1 gene (Sequence Analysis-All Coding Exons)	AMOTL1	AMOTL1, JEAP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMOT geni (Tüm Gen Dizi Analizi)	AMOT gene (Sequence Analysis-All Coding Exons)	AMOT	AMOT, KIAA1071	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMIGO3 geni (Tüm Gen Dizi Analizi)	AMIGO3 gene (Sequence Analysis-All Coding Exons)	AMIGO3	AMIGO3, ALI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMIGO2 geni (Tüm Gen Dizi Analizi)	AMIGO2 gene (Sequence Analysis-All Coding Exons)	AMIGO2	AMIGO2, ALI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMIGO1 geni (Tüm Gen Dizi Analizi)	AMIGO1 gene (Sequence Analysis-All Coding Exons)	AMIGO1	AMIGO1, ALI2, KIAA1163	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMFR geni (Tüm Gen Dizi Analizi)	AMFR gene (Sequence Analysis-All Coding Exons)	AMFR	AMFR, GP78	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMER2 geni (Tüm Gen Dizi Analizi)	AMER2 gene (Sequence Analysis-All Coding Exons)	AMER2	FAM123A, AMER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMELY geni (Tüm Gen Dizi Analizi)	AMELY gene (Sequence Analysis-All Coding Exons)	AMELY	AMELY, AMGL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMD1 geni (Tüm Gen Dizi Analizi)	AMD1 gene (Sequence Analysis-All Coding Exons)	AMD1	AMD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMBRA1 geni (Tüm Gen Dizi Analizi)	AMBRA1 gene (Sequence Analysis-All Coding Exons)	AMBRA1	AMBRA1, KIAA1736	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AMBP geni (Tüm Gen Dizi Analizi)	AMBP gene (Sequence Analysis-All Coding Exons)	AMBP	AMBP, ITIL, ITI, HCP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALYREF geni (Tüm Gen Dizi Analizi)	ALYREF gene (Sequence Analysis-All Coding Exons)	ALYREF	ALYREF, THOC4, ALY, BEF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALS2CL geni (Tüm Gen Dizi Analizi)	ALS2CL gene (Sequence Analysis-All Coding Exons)	ALS2CL	ALS2CL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALPPL2 geni (Tüm Gen Dizi Analizi)	ALPPL2 gene (Sequence Analysis-All Coding Exons)	ALPPL2	ALPPL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALPP geni (Tüm Gen Dizi Analizi)	ALPP gene (Sequence Analysis-All Coding Exons)	ALPP	ALPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALPK1 geni (Tüm Gen Dizi Analizi)	ALPK1 gene (Sequence Analysis-All Coding Exons)	ALPK1	ALPK1, LAK, KIAA1527	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALPI geni (Tüm Gen Dizi Analizi)	ALPI gene (Sequence Analysis-All Coding Exons)	ALPI	ALPI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALOX15B geni (Tüm Gen Dizi Analizi)	ALOX15B gene (Sequence Analysis-All Coding Exons)	ALOX15B	ALOX15B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALOX15 geni (Tüm Gen Dizi Analizi)	ALOX15 gene (Sequence Analysis-All Coding Exons)	ALOX15	ALOX15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALOX12 geni (Tüm Gen Dizi Analizi)	ALOX12 gene (Sequence Analysis-All Coding Exons)	ALOX12	ALOX12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH8 geni (Tüm Gen Dizi Analizi)	ALKBH8 gene (Sequence Analysis-All Coding Exons)	ALKBH8	ALKBH8, ABH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH7 geni (Tüm Gen Dizi Analizi)	ALKBH7 gene (Sequence Analysis-All Coding Exons)	ALKBH7	ALKBH7, ABH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH6 geni (Tüm Gen Dizi Analizi)	ALKBH6 gene (Sequence Analysis-All Coding Exons)	ALKBH6	ALKBH6, ABH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH5 geni (Tüm Gen Dizi Analizi)	ALKBH5 gene (Sequence Analysis-All Coding Exons)	ALKBH5	ALKBH5, ABH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH4 geni (Tüm Gen Dizi Analizi)	ALKBH4 gene (Sequence Analysis-All Coding Exons)	ALKBH4	ALKBH4, ABH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH3 geni (Tüm Gen Dizi Analizi)	ALKBH3 gene (Sequence Analysis-All Coding Exons)	ALKBH3	ALKBH3, PCA1, DEPC1, ABH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH2 geni (Tüm Gen Dizi Analizi)	ALKBH2 gene (Sequence Analysis-All Coding Exons)	ALKBH2	ALKBH2, ABH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALKBH1 geni (Tüm Gen Dizi Analizi)	ALKBH1 gene (Sequence Analysis-All Coding Exons)	ALKBH1	ALKBH1, ALKB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HIPK1 geni (Tüm Gen Dizi Analizi)	HIPK1 gene (Sequence Analysis-All Coding Exons)	HIPK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALG5 geni (Tüm Gen Dizi Analizi)	ALG5 gene (Sequence Analysis-All Coding Exons)	ALG5	ALG5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDOC geni (Tüm Gen Dizi Analizi)	ALDOC gene (Sequence Analysis-All Coding Exons)	ALDOC	ALDOC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH9A1 geni (Tüm Gen Dizi Analizi)	ALDH9A1 gene (Sequence Analysis-All Coding Exons)	ALDH9A1	ALDH9A1, ALDH9, E3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH8A1 geni (Tüm Gen Dizi Analizi)	ALDH8A1 gene (Sequence Analysis-All Coding Exons)	ALDH8A1	ALDH8A1, ALDH12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH3B2 geni (Tüm Gen Dizi Analizi)	ALDH3B2 gene (Sequence Analysis-All Coding Exons)	ALDH3B2	ALDH3B2, ALDH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH3B1 geni (Tüm Gen Dizi Analizi)	ALDH3B1 gene (Sequence Analysis-All Coding Exons)	ALDH3B1	ALDH3B1, ALDH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH3A1 geni (Tüm Gen Dizi Analizi)	ALDH3A1 gene (Sequence Analysis-All Coding Exons)	ALDH3A1	ALDH3A1, ALDH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH1L2 geni (Tüm Gen Dizi Analizi)	ALDH1L2 gene (Sequence Analysis-All Coding Exons)	ALDH1L2	ALDH1L2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH1L1 geni (Tüm Gen Dizi Analizi)	ALDH1L1 gene (Sequence Analysis-All Coding Exons)	ALDH1L1	ALDH1L1, FTHFD	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH1B1 geni (Tüm Gen Dizi Analizi)	ALDH1B1 gene (Sequence Analysis-All Coding Exons)	ALDH1B1	ALDH1B1, ALDH5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH1A2 geni (Tüm Gen Dizi Analizi)	ALDH1A2 gene (Sequence Analysis-All Coding Exons)	ALDH1A2	ALDH1A2, RALDH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH1A1 geni (Tüm Gen Dizi Analizi)	ALDH1A1 gene (Sequence Analysis-All Coding Exons)	ALDH1A1	ALDH1A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALDH16A1 geni (Tüm Gen Dizi Analizi)	ALDH16A1 gene (Sequence Analysis-All Coding Exons)	ALDH16A1	ALDH16A1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALCAM geni (Tüm Gen Dizi Analizi)	ALCAM gene (Sequence Analysis-All Coding Exons)	ALCAM	ALCAM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ALAS1 geni (Tüm Gen Dizi Analizi)	ALAS1 gene (Sequence Analysis-All Coding Exons)	ALAS1	ALAS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKTIP geni (Tüm Gen Dizi Analizi)	AKTIP gene (Sequence Analysis-All Coding Exons)	AKTIP	AKTIP, FTS, FT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR7A3 geni (Tüm Gen Dizi Analizi)	AKR7A3 gene (Sequence Analysis-All Coding Exons)	AKR7A3	AKR7A3, AFAR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR7A2 geni (Tüm Gen Dizi Analizi)	AKR7A2 gene (Sequence Analysis-All Coding Exons)	AKR7A2	AKR7A2, AKR7, AFAR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR1C3 geni (Tüm Gen Dizi Analizi)	AKR1C3 gene (Sequence Analysis-All Coding Exons)	AKR1C3	AKR1C3, HAKRB, DD3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR1C1 geni (Tüm Gen Dizi Analizi)	AKR1C1 gene (Sequence Analysis-All Coding Exons)	AKR1C1	AKR1C1, DDH1, DD1, HAKRC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR1B15 geni (Tüm Gen Dizi Analizi)	AKR1B15 gene (Sequence Analysis-All Coding Exons)	AKR1B15	AKR1B15, AKR1B10L, AKR1R1B7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

AKR1B10 geni (Tüm Gen Dizi Analizi)	AKR1B10 gene (Sequence Analysis-All Coding Exons)	AKR1B10	AKR1B10, ARL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR1B1 geni (Tüm Gen Dizi Analizi)	AKR1B1 gene (Sequence Analysis-All Coding Exons)	AKR1B1	AKR1B1, ALDR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR1A1 geni (Tüm Gen Dizi Analizi)	AKR1A1 gene (Sequence Analysis-All Coding Exons)	AKR1A1	AKR1A1, ALR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKIRIN2 geni (Tüm Gen Dizi Analizi)	AKIRIN2 gene (Sequence Analysis-All Coding Exons)	AKIRIN2	AKIRIN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKIRIN1 geni (Tüm Gen Dizi Analizi)	AKIRIN1 gene (Sequence Analysis-All Coding Exons)	AKIRIN1	AKIRIN1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKIP1 geni (Tüm Gen Dizi Analizi)	AKIP1 gene (Sequence Analysis-All Coding Exons)	AKIP1	AKIP1, BCA3, C11orf17	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP8L geni (Tüm Gen Dizi Analizi)	AKAP8L gene (Sequence Analysis-All Coding Exons)	AKAP8L	AKAP8L, HA95, NAKAP, HAP95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP8 geni (Tüm Gen Dizi Analizi)	AKAP8 gene (Sequence Analysis-All Coding Exons)	AKAP8	AKAP8, AKAP95	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP6 geni (Tüm Gen Dizi Analizi)	AKAP6 gene (Sequence Analysis-All Coding Exons)	AKAP6	ADAP6, ADAP100	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP5 geni (Tüm Gen Dizi Analizi)	AKAP5 gene (Sequence Analysis-All Coding Exons)	AKAP5	AKAP5, AKAP79	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP4 geni (Tüm Gen Dizi Analizi)	AKAP4 gene (Sequence Analysis-All Coding Exons)	AKAP4	AKAP4, AKAP82, HI, FSC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP2 geni (Tüm Gen Dizi Analizi)	AKAP2 gene (Sequence Analysis-All Coding Exons)	AKAP2	AKAP2, AKAPKL, KIAA0920, PALM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP17A geni (Tüm Gen Dizi Analizi)	AKAP17A gene (Sequence Analysis-All Coding Exons)	AKAP17A	AKAP17A, SFRS17A, DXYS155E, XE7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP17A geni (Tüm Gen Dizi Analizi)	AKAP17A gene (Sequence Analysis-All Coding Exons)	AKAP17A	AKAP17A, SFRS17A, XE7Y	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP14 geni (Tüm Gen Dizi Analizi)	AKAP14 gene (Sequence Analysis-All Coding Exons)	AKAP14	AKAP14, AKAP28	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP13 geni (Tüm Gen Dizi Analizi)	AKAP13 gene (Sequence Analysis-All Coding Exons)	AKAP13	AKAP13, HT31, LBC, BRX	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP11 geni (Tüm Gen Dizi Analizi)	AKAP11 gene (Sequence Analysis-All Coding Exons)	AKAP11	AKAP11, AKAP220	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP1 geni (Tüm Gen Dizi Analizi)	AKAP1 gene (Sequence Analysis-All Coding Exons)	AKAP1	AKAP1, AKAP149, SAKAP84	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEY1 geni (Tüm Gen Dizi Analizi)	HEY1 gene (Sequence Analysis-All Coding Exons)	HEY1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAIN1 geni (Tüm Gen Dizi Analizi)	AKAIN1 gene (Sequence Analysis-All Coding Exons)	AKAIN1	C18orf42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK9 geni (Tüm Gen Dizi Analizi)	AK9 gene (Sequence Analysis-All Coding Exons)	AK9	AK9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HEXDC geni (Tüm Gen Dizi Analizi)	HEXDC gene (Sequence Analysis-All Coding Exons)	HEXDC		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK8 geni (Tüm Gen Dizi Analizi)	AK8 gene (Sequence Analysis-All Coding Exons)	AK8	AK8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK7 geni (Tüm Gen Dizi Analizi)	AK7 gene (Sequence Analysis-All Coding Exons)	AK7	AK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK5 geni (Tüm Gen Dizi Analizi)	AK5 gene (Sequence Analysis-All Coding Exons)	AK5	AK5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK4 geni (Tüm Gen Dizi Analizi)	AK4 gene (Sequence Analysis-All Coding Exons)	AK4	AK3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AK3 geni (Tüm Gen Dizi Analizi)	AK3 gene (Sequence Analysis-All Coding Exons)	AK3	AK3, AK3L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AJAP1 geni (Tüm Gen Dizi Analizi)	AJAP1 gene (Sequence Analysis-All Coding Exons)	AJAP1	AJAP1, SHREW1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIRN geni (Tüm Gen Dizi Analizi)	AIRN gene (Sequence Analysis-All Coding Exons)	AIRN	AIRN, AIR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIMP2 geni (Tüm Gen Dizi Analizi)	AIMP2 gene (Sequence Analysis-All Coding Exons)	AIMP2	AIMP2, JTV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIM2 geni (Tüm Gen Dizi Analizi)	AIM2 gene (Sequence Analysis-All Coding Exons)	AIM2	AIM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIM1 geni (Tüm Gen Dizi Analizi)	AIM1 gene (Sequence Analysis-All Coding Exons)	AIM1	AIM1, ST4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIFM3 geni (Tüm Gen Dizi Analizi)	AIFM3 gene (Sequence Analysis-All Coding Exons)	AIFM3	AIFM3, AIFL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIF1 geni (Tüm Gen Dizi Analizi)	AIF1 gene (Sequence Analysis-All Coding Exons)	AIF1	AIF1, IRT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIDA geni (Tüm Gen Dizi Analizi)	AIDA gene (Sequence Analysis-All Coding Exons)	AIDA	AIDA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHSP geni (Tüm Gen Dizi Analizi)	AHSP gene (Sequence Analysis-All Coding Exons)	AHSP	ERAF, EDRF, AHSP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHSG geni (Tüm Gen Dizi Analizi)	AHSG gene (Sequence Analysis-All Coding Exons)	AHSG	AHSG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HERC6 geni (Tüm Gen Dizi Analizi)	HERC6 gene (Sequence Analysis-All Coding Exons)	HERC6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHSA1 geni (Tüm Gen Dizi Analizi)	AHSA1 gene (Sequence Analysis-All Coding Exons)	AHSA1	AHSA1, AHA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HERC4 geni (Tüm Gen Dizi Analizi)	HERC4 gene (Sequence Analysis-All Coding Exons)	HERC4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHRR geni (Tüm Gen Dizi Analizi)	AHRR gene (Sequence Analysis-All Coding Exons)	AHRR	AHRR, KIAA1234	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHR geni (Tüm Gen Dizi Analizi)	AHR gene (Sequence Analysis-All Coding Exons)	AHR	AHR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHNAK geni (Tüm Gen Dizi Analizi)	AHNAK gene (Sequence Analysis-All Coding Exons)	AHNAK	AHNAK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHCYL2 geni (Tüm Gen Dizi Analizi)	AHCYL2 gene (Sequence Analysis-All Coding Exons)	AHCYL2	AHCYL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHCYL1 geni (Tüm Gen Dizi Analizi)	AHCYL1 gene (Sequence Analysis-All Coding Exons)	AHCYL1	AHCYL1, DCAL, IRBIT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHCTF1 geni (Tüm Gen Dizi Analizi)	AHCTF1 gene (Sequence Analysis-All Coding Exons)	AHCTF1	AHCTF1, ELYS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGXT2 geni (Tüm Gen Dizi Analizi)	AGXT2 gene (Sequence Analysis-All Coding Exons)	AGXT2	AGXT2, AGT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGTR2 geni (Tüm Gen Dizi Analizi)	AGTR2 gene (Sequence Analysis-All Coding Exons)	AGTR2	AGTR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGTPBP1 geni (Tüm Gen Dizi Analizi)	AGTPBP1 gene (Sequence Analysis-All Coding Exons)	AGTPBP1	AGTPBP1, NNA1, KIAA1035, CCP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGR3 geni (Tüm Gen Dizi Analizi)	AGR3 gene (Sequence Analysis-All Coding Exons)	AGR3	AGR3, BCMP11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGR2 geni (Tüm Gen Dizi Analizi)	AGR2 gene (Sequence Analysis-All Coding Exons)	AGR2	AGR2, AG2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGPAT5 geni (Tüm Gen Dizi Analizi)	AGPAT5 gene (Sequence Analysis-All Coding Exons)	AGPAT5	AGPAT5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGPAT4 geni (Tüm Gen Dizi Analizi)	AGPAT4 gene (Sequence Analysis-All Coding Exons)	AGPAT4	AGPAT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGPAT3 geni (Tüm Gen Dizi Analizi)	AGPAT3 gene (Sequence Analysis-All Coding Exons)	AGPAT3	AGPAT3, LPAAT3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGPAT1 geni (Tüm Gen Dizi Analizi)	AGPAT1 gene (Sequence Analysis-All Coding Exons)	AGPAT1	AGPAT1, LPAATA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGO4 geni (Tüm Gen Dizi Analizi)	AGO4 gene (Sequence Analysis-All Coding Exons)	AGO4	EIF2C4, AGO4, KIAA1567	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGO3 geni (Tüm Gen Dizi Analizi)	AGO3 gene (Sequence Analysis-All Coding Exons)	AGO3	EIF2C3, AGO3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGO2 geni (Tüm Gen Dizi Analizi)	AGO2 gene (Sequence Analysis-All Coding Exons)	AGO2	AGO2, EIF2C2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGO1 geni (Tüm Gen Dizi Analizi)	AGO1 gene (Sequence Analysis-All Coding Exons)	AGO1	AGO1, EIF2C1, GERP95, EIF2C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGMO geni (Tüm Gen Dizi Analizi)	AGMO gene (Sequence Analysis-All Coding Exons)	AGMO	AGMO, TMEM195	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGGF1 geni (Tüm Gen Dizi Analizi)	AGGF1 gene (Sequence Analysis-All Coding Exons)	AGGF1	AGGF1, VG5Q, HUSI4971, FLJ10283	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGFG2 geni (Tüm Gen Dizi Analizi)	AGFG2 gene (Sequence Analysis-All Coding Exons)	AGFG2	AGFG2, HRBL, RABR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGFG1 geni (Tüm Gen Dizi Analizi)	AGFG1 gene (Sequence Analysis-All Coding Exons)	AGFG1	AGFG1, HRB, RIP, RAB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGER geni (Tüm Gen Dizi Analizi)	AGER gene (Sequence Analysis-All Coding Exons)	AGER	AGER, RAGE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGBL4 geni (Tüm Gen Dizi Analizi)	AGBL4 gene (Sequence Analysis-All Coding Exons)	AGBL4	AGBL4, CCP6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGBL3 geni (Tüm Gen Dizi Analizi)	AGBL3 gene (Sequence Analysis-All Coding Exons)	AGBL3	AGBL3, CCP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGBL2 geni (Tüm Gen Dizi Analizi)	AGBL2 gene (Sequence Analysis-All Coding Exons)	AGBL2	AGBL2, CCP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGAP3 geni (Tüm Gen Dizi Analizi)	AGAP3 gene (Sequence Analysis-All Coding Exons)	AGAP3	AGAP3, CENTG3, CRAG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGAP2 geni (Tüm Gen Dizi Analizi)	AGAP2 gene (Sequence Analysis-All Coding Exons)	AGAP2	AGAP2, CENTG1, PIKE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFM geni (Tüm Gen Dizi Analizi)	AFM gene (Sequence Analysis-All Coding Exons)	AFM	AFM, ALBA, ALB2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFG3L1P geni (Tüm Gen Dizi Analizi)	AFG3L1P gene (Sequence Analysis-All Coding Exons)	AFG3L1P	AFG3L1, AFG3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFF3 geni (Tüm Gen Dizi Analizi)	AFF3 gene (Sequence Analysis-All Coding Exons)	AFF3	LAF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFF1 geni (Tüm Gen Dizi Analizi)	AFF1 gene (Sequence Analysis-All Coding Exons)	AFF1	AFF1, MLLT2, AF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFDN geni (Tüm Gen Dizi Analizi)	AFDN gene (Sequence Analysis-All Coding Exons)	AFDN	MLLT4, AF6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFAP1L2 geni (Tüm Gen Dizi Analizi)	AFAP1L2 gene (Sequence Analysis-All Coding Exons)	AFAP1L2	AFAP1L2, XB130, KIAA1914	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC11 geni (Tüm Gen Dizi Analizi)	HDAC11 gene (Sequence Analysis-All Coding Exons)	HDAC11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HDAC10 geni (Tüm Gen Dizi Analizi)	HDAC10 gene (Sequence Analysis-All Coding Exons)	HDAC10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFAP1L1 geni (Tüm Gen Dizi Analizi)	AFAP1L1 gene (Sequence Analysis-All Coding Exons)	AFAP1L1	AFAP1L1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AFAP1 geni (Tüm Gen Dizi Analizi)	AFAP1 gene (Sequence Analysis-All Coding Exons)	AFAP1	AFAP1, AFAP, AFAP110	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HCRTR2 geni (Tüm Gen Dizi Analizi)	HCRTR2 gene (Sequence Analysis-All Coding Exons)	HCRTR2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AES geni (Tüm Gen Dizi Analizi)	AES gene (Sequence Analysis-All Coding Exons)	AES	AES	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AEN geni (Tüm Gen Dizi Analizi)	AEN gene (Sequence Analysis-All Coding Exons)	AEN	ISG20L1, AEN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADTRP geni (Tüm Gen Dizi Analizi)	ADTRP gene (Sequence Analysis-All Coding Exons)	ADTRP	C6orf105, ADTRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADSS geni (Tüm Gen Dizi Analizi)	ADSS gene (Sequence Analysis-All Coding Exons)	ADSS	ADSS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADRA2A geni (Tüm Gen Dizi Analizi)	ADRA2A gene (Sequence Analysis-All Coding Exons)	ADRA2A	ADRA2A, ADRA2R	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADRA1D geni (Tüm Gen Dizi Analizi)	ADRA1D gene (Sequence Analysis-All Coding Exons)	ADRA1D	ADRA1D, ADRA1A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADRA1B geni (Tüm Gen Dizi Analizi)	ADRA1B gene (Sequence Analysis-All Coding Exons)	ADRA1B	ADRA1B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADRA1A geni (Tüm Gen Dizi Analizi)	ADRA1A gene (Sequence Analysis-All Coding Exons)	ADRA1A	ADRA1C	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADPRHL2 geni (Tüm Gen Dizi Analizi)	ADPRHL2 gene (Sequence Analysis-All Coding Exons)	ADPRHL2	ADPRHL2, ARH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADPRHL1 geni (Tüm Gen Dizi Analizi)	ADPRHL1 gene (Sequence Analysis-All Coding Exons)	ADPRHL1	ADPRHL1, ARH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADPRH geni (Tüm Gen Dizi Analizi)	ADPRH gene (Sequence Analysis-All Coding Exons)	ADPRH	ADPRH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADPGK geni (Tüm Gen Dizi Analizi)	ADPGK gene (Sequence Analysis-All Coding Exons)	ADPGK	ADPGK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADORA3 geni (Tüm Gen Dizi Analizi)	ADORA3 gene (Sequence Analysis-All Coding Exons)	ADORA3	ADORA3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ADORA2B geni (Tüm Gen Dizi Analizi)	ADORA2B gene (Sequence Analysis-All Coding Exons)	ADORA2B	ADORA2B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADORA2A geni (Tüm Gen Dizi Analizi)	ADORA2A gene (Sequence Analysis-All Coding Exons)	ADORA2A	ADORA2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADORA1 geni (Tüm Gen Dizi Analizi)	ADORA1 gene (Sequence Analysis-All Coding Exons)	ADORA1	ADORA1, RDC7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADO geni (Tüm Gen Dizi Analizi)	ADO gene (Sequence Analysis-All Coding Exons)	ADO	ADO, C10orf22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADM geni (Tüm Gen Dizi Analizi)	ADM gene (Sequence Analysis-All Coding Exons)	ADM	ADM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADIPOR2 geni (Tüm Gen Dizi Analizi)	ADIPOR2 gene (Sequence Analysis-All Coding Exons)	ADIPOR2	ADIPOR2, FLJ21432	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADIPOR1 geni (Tüm Gen Dizi Analizi)	ADIPOR1 gene (Sequence Analysis-All Coding Exons)	ADIPOR1	ADIPOR1, CGI45	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADIG geni (Tüm Gen Dizi Analizi)	ADIG gene (Sequence Analysis-All Coding Exons)	ADIG	ADIG, SMAF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADI1 geni (Tüm Gen Dizi Analizi)	ADI1 gene (Sequence Analysis-All Coding Exons)	ADI1	ADI1, SIPL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADHFE1 geni (Tüm Gen Dizi Analizi)	ADHFE1 gene (Sequence Analysis-All Coding Exons)	ADHFE1	ADHFE1, HOT, ADH8	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADH7 geni (Tüm Gen Dizi Analizi)	ADH7 gene (Sequence Analysis-All Coding Exons)	ADH7	ADH7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADH6 geni (Tüm Gen Dizi Analizi)	ADH6 gene (Sequence Analysis-All Coding Exons)	ADH6	ADH6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADH5 geni (Tüm Gen Dizi Analizi)	ADH5 gene (Sequence Analysis-All Coding Exons)	ADH5	ADH5, FDH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADH4 geni (Tüm Gen Dizi Analizi)	ADH4 gene (Sequence Analysis-All Coding Exons)	ADH4	ADH4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADH1A geni (Tüm Gen Dizi Analizi)	ADH1A gene (Sequence Analysis-All Coding Exons)	ADH1A	ADH1A, ADH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRL4 geni (Tüm Gen Dizi Analizi)	ADGRL4 gene (Sequence Analysis-All Coding Exons)	ADGRL4	ADGRL4, ETL, ELTD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRL3 geni (Tüm Gen Dizi Analizi)	ADGRL3 gene (Sequence Analysis-All Coding Exons)	ADGRL3	ADGRL3, LPHN3, CIRL3, CL3, LEC3, KIAA0768	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRL2 geni (Tüm Gen Dizi Analizi)	ADGRL2 gene (Sequence Analysis-All Coding Exons)	ADGRL2	ADGRL2, LPHN2, LPH94, CIRL2, CL2, KIAA0786	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRL1 geni (Tüm Gen Dizi Analizi)	ADGRL1 gene (Sequence Analysis-All Coding Exons)	ADGRL1	ADGRL1, LPHN1, CIRL1, CL1, LEC2, KIAA0821	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRG7 geni (Tüm Gen Dizi Analizi)	ADGRG7 gene (Sequence Analysis-All Coding Exons)	ADGRG7	ADGRG7, GPR128	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRG5 geni (Tüm Gen Dizi Analizi)	ADGRG5 gene (Sequence Analysis-All Coding Exons)	ADGRG5	ADGRG5, GPR114	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRF4 geni (Tüm Gen Dizi Analizi)	ADGRF4 gene (Sequence Analysis-All Coding Exons)	ADGRF4	ADGRF4, GPR115	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRE5 geni (Tüm Gen Dizi Analizi)	ADGRE5 gene (Sequence Analysis-All Coding Exons)	ADGRE5	ADGRE5, CD97	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRE4P geni (Tüm Gen Dizi Analizi)	ADGRE4P gene (Sequence Analysis-All Coding Exons)	ADGRE4P	ADGRE4P, EMR4, GPR127, EMR4P, FIRE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRE3 geni (Tüm Gen Dizi Analizi)	ADGRE3 gene (Sequence Analysis-All Coding Exons)	ADGRE3	EMR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRE2 geni (Tüm Gen Dizi Analizi)	ADGRE2 gene (Sequence Analysis-All Coding Exons)	ADGRE2	ADGRE2, EMR2, VBU	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRE1 geni (Tüm Gen Dizi Analizi)	ADGRE1 gene (Sequence Analysis-All Coding Exons)	ADGRE1	ADGRE1, EMR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRD1 geni (Tüm Gen Dizi Analizi)	ADGRD1 gene (Sequence Analysis-All Coding Exons)	ADGRD1	ADGRD1, GPR133, PGR25	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRB3 geni (Tüm Gen Dizi Analizi)	ADGRB3 gene (Sequence Analysis-All Coding Exons)	ADGRB3	ADGRB3, BAI3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRB2 geni (Tüm Gen Dizi Analizi)	ADGRB2 gene (Sequence Analysis-All Coding Exons)	ADGRB2	ADGRB2, BAI2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRB1 geni (Tüm Gen Dizi Analizi)	ADGRB1 gene (Sequence Analysis-All Coding Exons)	ADGRB1	ADGRB1, BAI1, GDAIF	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRA3 geni (Tüm Gen Dizi Analizi)	ADGRA3 gene (Sequence Analysis-All Coding Exons)	ADGRA3	ADGRA3, GPR125	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRA2 geni (Tüm Gen Dizi Analizi)	ADGRA2 gene (Sequence Analysis-All Coding Exons)	ADGRA2	ADGRA2, GPR124, TEM5, KIAA1531	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGRA1 geni (Tüm Gen Dizi Analizi)	ADGRA1 gene (Sequence Analysis-All Coding Exons)	ADGRA1	ADGRA1, GPR123, KIAA1828	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADGB geni (Tüm Gen Dizi Analizi)	ADGB gene (Sequence Analysis-All Coding Exons)	ADGB	ADGB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADD2 geni (Tüm Gen Dizi Analizi)	ADD2 gene (Sequence Analysis-All Coding Exons)	ADD2	ADD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCYAP1R1 geni (Tüm Gen Dizi Analizi)	ADCYAP1R1 gene (Sequence Analysis-All Coding Exons)	ADCYAP1R1	ADCYAP1R1, PACAPR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCYAP1 geni (Tüm Gen Dizi Analizi)	ADCYAP1 gene (Sequence Analysis-All Coding Exons)	ADCYAP1	ADCYAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY9 geni (Tüm Gen Dizi Analizi)	ADCY9 gene (Sequence Analysis-All Coding Exons)	ADCY9	ADCY9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY8 geni (Tüm Gen Dizi Analizi)	ADCY8 gene (Sequence Analysis-All Coding Exons)	ADCY8	ADCY8, ADCY3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY7 geni (Tüm Gen Dizi Analizi)	ADCY7 gene (Sequence Analysis-All Coding Exons)	ADCY7	ADCY7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY4 geni (Tüm Gen Dizi Analizi)	ADCY4 gene (Sequence Analysis-All Coding Exons)	ADCY4	ADCY4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY3 geni (Tüm Gen Dizi Analizi)	ADCY3 gene (Sequence Analysis-All Coding Exons)	ADCY3	ADCY3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADCY2 geni (Tüm Gen Dizi Analizi)	ADCY2 gene (Sequence Analysis-All Coding Exons)	ADCY2	ADCY2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAT2 geni (Tüm Gen Dizi Analizi)	ADAT2 gene (Sequence Analysis-All Coding Exons)	ADAT2	ADAT2, TAD2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAT1 geni (Tüm Gen Dizi Analizi)	ADAT1 gene (Sequence Analysis-All Coding Exons)	ADAT1	ADAT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADARB2 geni (Tüm Gen Dizi Analizi)	ADARB2 gene (Sequence Analysis-All Coding Exons)	ADARB2	ADARB2, RED2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADARB1 geni (Tüm Gen Dizi Analizi)	ADARB1 gene (Sequence Analysis-All Coding Exons)	ADARB1	ADARB1, RED1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAP2 geni (Tüm Gen Dizi Analizi)	ADAP2 gene (Sequence Analysis-All Coding Exons)	ADAP2	ADAP2, CENTA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAP1 geni (Tüm Gen Dizi Analizi)	ADAP1 gene (Sequence Analysis-All Coding Exons)	ADAP1	CENTA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTSL3 geni (Tüm Gen Dizi Analizi)	ADAMTSL3 gene (Sequence Analysis-All Coding Exons)	ADAMTSL3	ADAMTSL3, KIAA1233	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTSL1 geni (Tüm Gen Dizi Analizi)	ADAMTSL1 gene (Sequence Analysis-All Coding Exons)	ADAMTSL1	ADAMTSL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS9 geni (Tüm Gen Dizi Analizi)	ADAMTS9 gene (Sequence Analysis-All Coding Exons)	ADAMTS9	ADAMTS9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS8 geni (Tüm Gen Dizi Analizi)	ADAMTS8 gene (Sequence Analysis-All Coding Exons)	ADAMTS8	ADAMTS8, METH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS7 geni (Tüm Gen Dizi Analizi)	ADAMTS7 gene (Sequence Analysis-All Coding Exons)	ADAMTS7	ADAMTS7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HAND1 geni (Tüm Gen Dizi Analizi)	HAND1 gene (Sequence Analysis-All Coding Exons)	HAND1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS6 geni (Tüm Gen Dizi Analizi)	ADAMTS6 gene (Sequence Analysis-All Coding Exons)	ADAMTS6	ADAMTS6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS5 geni (Tüm Gen Dizi Analizi)	ADAMTS5 gene (Sequence Analysis-All Coding Exons)	ADAMTS5	ADAMTS5, ADAMTS11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS4 geni (Tüm Gen Dizi Analizi)	ADAMTS4 gene (Sequence Analysis-All Coding Exons)	ADAMTS4	ADAMTS4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS3 geni (Tüm Gen Dizi Analizi)	ADAMTS3 gene (Sequence Analysis-All Coding Exons)	ADAMTS3	ADAMTS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS20 geni (Tüm Gen Dizi Analizi)	ADAMTS20 gene (Sequence Analysis-All Coding Exons)	ADAMTS20	ADAMTS20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS19 geni (Tüm Gen Dizi Analizi)	ADAMTS19 gene (Sequence Analysis-All Coding Exons)	ADAMTS19	ADAMTS19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS16 geni (Tüm Gen Dizi Analizi)	ADAMTS16 gene (Sequence Analysis-All Coding Exons)	ADAMTS16	ADAMTS16	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS15 geni (Tüm Gen Dizi Analizi)	ADAMTS15 gene (Sequence Analysis-All Coding Exons)	ADAMTS15	ADAMTS15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS14 geni (Tüm Gen Dizi Analizi)	ADAMTS14 gene (Sequence Analysis-All Coding Exons)	ADAMTS14	ADAMTS14	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS12 geni (Tüm Gen Dizi Analizi)	ADAMTS12 gene (Sequence Analysis-All Coding Exons)	ADAMTS12	ADAMTS12	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMTS1 geni (Tüm Gen Dizi Analizi)	ADAMTS1 gene (Sequence Analysis-All Coding Exons)	ADAMTS1	ADAMTS1, METH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAMDEC1 geni (Tüm Gen Dizi Analizi)	ADAMDEC1 gene (Sequence Analysis-All Coding Exons)	ADAMDEC1	ADAMDEC1, DECYSIN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM8 geni (Tüm Gen Dizi Analizi)	ADAM8 gene (Sequence Analysis-All Coding Exons)	ADAM8	ADAM8, CD156	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM7 geni (Tüm Gen Dizi Analizi)	ADAM7 gene (Sequence Analysis-All Coding Exons)	ADAM7	ADAM7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM33 geni (Tüm Gen Dizi Analizi)	ADAM33 gene (Sequence Analysis-All Coding Exons)	ADAM33	ADAM33	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM30 geni (Tüm Gen Dizi Analizi)	ADAM30 gene (Sequence Analysis-All Coding Exons)	ADAM30	ADAM30	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
HABP4 geni (Tüm Gen Dizi Analizi)	HABP4 gene (Sequence Analysis-All Coding Exons)	HABP4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM29 geni (Tüm Gen Dizi Analizi)	ADAM29 gene (Sequence Analysis-All Coding Exons)	ADAM29	ADAM29	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM28 geni (Tüm Gen Dizi Analizi)	ADAM28 gene (Sequence Analysis-All Coding Exons)	ADAM28	ADAM28, MDCL	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM23 geni (Tüm Gen Dizi Analizi)	ADAM23 gene (Sequence Analysis-All Coding Exons)	ADAM23	ADAM23, MDC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM22 geni (Tüm Gen Dizi Analizi)	ADAM22 gene (Sequence Analysis-All Coding Exons)	ADAM22	ADAM22	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM21 geni (Tüm Gen Dizi Analizi)	ADAM21 gene (Sequence Analysis-All Coding Exons)	ADAM21	ADAM21	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM20 geni (Tüm Gen Dizi Analizi)	ADAM20 gene (Sequence Analysis-All Coding Exons)	ADAM20	ADAM20	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM2 geni (Tüm Gen Dizi Analizi)	ADAM2 gene (Sequence Analysis-All Coding Exons)	ADAM2	ADAM2, FTNB, PH30, CRYN1, CRYN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM19 geni (Tüm Gen Dizi Analizi)	ADAM19 gene (Sequence Analysis-All Coding Exons)	ADAM19	ADAM19, MLTNB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM15 geni (Tüm Gen Dizi Analizi)	ADAM15 gene (Sequence Analysis-All Coding Exons)	ADAM15	ADAM15, MDC15	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM12 geni (Tüm Gen Dizi Analizi)	ADAM12 gene (Sequence Analysis-All Coding Exons)	ADAM12	ADAM12, MLTN	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAM11 geni (Tüm Gen Dizi Analizi)	ADAM11 gene (Sequence Analysis-All Coding Exons)	ADAM11	ADAM11, MDC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADAD1 geni (Tüm Gen Dizi Analizi)	ADAD1 gene (Sequence Analysis-All Coding Exons)	ADAD1	ADAD1, TENR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACYP1 geni (Tüm Gen Dizi Analizi)	ACYP1 gene (Sequence Analysis-All Coding Exons)	ACYP1	ACYP1, ACYPE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACY3 geni (Tüm Gen Dizi Analizi)	ACY3 gene (Sequence Analysis-All Coding Exons)	ACY3	ACY3, HCBP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACVR2A geni (Tüm Gen Dizi Analizi)	ACVR2A gene (Sequence Analysis-All Coding Exons)	ACVR2A	ACVR2A, ACVR2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACVR1C geni (Tüm Gen Dizi Analizi)	ACVR1C gene (Sequence Analysis-All Coding Exons)	ACVR1C	ACVR1C, ALK7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTRT3 geni (Tüm Gen Dizi Analizi)	ACTRT3 gene (Sequence Analysis-All Coding Exons)	ACTRT3	ARPM1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTRT2 geni (Tüm Gen Dizi Analizi)	ACTRT2 gene (Sequence Analysis-All Coding Exons)	ACTRT2	ACTRT2, ARPT2, ARPM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTRT1 geni (Tüm Gen Dizi Analizi)	ACTRT1 gene (Sequence Analysis-All Coding Exons)	ACTRT1	ACTRT1, ARPT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GZMK geni (Tüm Gen Dizi Analizi)	GZMK gene (Sequence Analysis-All Coding Exons)	GZMK		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ACTR3 geni (Tüm Gen Dizi Analizi)	ACTR3 gene (Sequence Analysis-All Coding Exons)	ACTR3	ACTR3, ARP3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTR2 geni (Tüm Gen Dizi Analizi)	ACTR2 gene (Sequence Analysis-All Coding Exons)	ACTR2	ACTR2, ARP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTR1B geni (Tüm Gen Dizi Analizi)	ACTR1B gene (Sequence Analysis-All Coding Exons)	ACTR1B	ACTR1B, ARP1B, CTRN2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTR1A geni (Tüm Gen Dizi Analizi)	ACTR1A gene (Sequence Analysis-All Coding Exons)	ACTR1A	ACTR1A, ARP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTL7B geni (Tüm Gen Dizi Analizi)	ACTL7B gene (Sequence Analysis-All Coding Exons)	ACTL7B	ACTL7B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTL7A geni (Tüm Gen Dizi Analizi)	ACTL7A gene (Sequence Analysis-All Coding Exons)	ACTL7A	ACTL7A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTL6B geni (Tüm Gen Dizi Analizi)	ACTL6B gene (Sequence Analysis-All Coding Exons)	ACTL6B	ACTL6B, BAF53B	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTL6A geni (Tüm Gen Dizi Analizi)	ACTL6A gene (Sequence Analysis-All Coding Exons)	ACTL6A	ACTL6A, BAF53, BAF53A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACTBL2 geni (Tüm Gen Dizi Analizi)	ACTBL2 gene (Sequence Analysis-All Coding Exons)	ACTBL2	ACTBL2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSS3 geni (Tüm Gen Dizi Analizi)	ACSS3 gene (Sequence Analysis-All Coding Exons)	ACSS3	ACSS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSS2 geni (Tüm Gen Dizi Analizi)	ACSS2 gene (Sequence Analysis-All Coding Exons)	ACSS2	ACSS2, ACS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSS1 geni (Tüm Gen Dizi Analizi)	ACSS1 gene (Sequence Analysis-All Coding Exons)	ACSS1	ACSS1, ACECS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSM5 geni (Tüm Gen Dizi Analizi)	ACSM5 gene (Sequence Analysis-All Coding Exons)	ACSM5	ACSM5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSM4 geni (Tüm Gen Dizi Analizi)	ACSM4 gene (Sequence Analysis-All Coding Exons)	ACSM4	ACSM4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSM2B geni (Tüm Gen Dizi Analizi)	ACSM2B gene (Sequence Analysis-All Coding Exons)	ACSM2B	ACSM2B, HXMA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSM2A geni (Tüm Gen Dizi Analizi)	ACSM2A gene (Sequence Analysis-All Coding Exons)	ACSM2A	ACSM2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSM1 geni (Tüm Gen Dizi Analizi)	ACSM1 gene (Sequence Analysis-All Coding Exons)	ACSM1	ACSM1, MACS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSL5 geni (Tüm Gen Dizi Analizi)	ACSL5 gene (Sequence Analysis-All Coding Exons)	ACSL5	ACSL5, FACL5, ACS5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSL3 geni (Tüm Gen Dizi Analizi)	ACSL3 gene (Sequence Analysis-All Coding Exons)	ACSL3	ACSL3, FACL3, ACS3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSL1 geni (Tüm Gen Dizi Analizi)	ACSL1 gene (Sequence Analysis-All Coding Exons)	ACSL1	ACSL1, FACL2, FACL1, LACS, ACS1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSF2 geni (Tüm Gen Dizi Analizi)	ACSF2 gene (Sequence Analysis-All Coding Exons)	ACSF2	ACSF2, FLJ20920	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSBG2 geni (Tüm Gen Dizi Analizi)	ACSBG2 gene (Sequence Analysis-All Coding Exons)	ACSBG2	ACSBG2, BGR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACSBG1 geni (Tüm Gen Dizi Analizi)	ACSBG1 gene (Sequence Analysis-All Coding Exons)	ACSBG1	ACSBG1, BG, BG1, KIAA0631	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACRV1 geni (Tüm Gen Dizi Analizi)	ACRV1 gene (Sequence Analysis-All Coding Exons)	ACRV1	ACRV1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACPP geni (Tüm Gen Dizi Analizi)	ACPP gene (Sequence Analysis-All Coding Exons)	ACPP	ACPP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACP7 geni (Tüm Gen Dizi Analizi)	ACP7 gene (Sequence Analysis-All Coding Exons)	ACP7	PAPL, FLJ16165	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACP6 geni (Tüm Gen Dizi Analizi)	ACP6 gene (Sequence Analysis-All Coding Exons)	ACP6	ACP6, LPAP, ACPL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACP1 geni (Tüm Gen Dizi Analizi)	ACP1 gene (Sequence Analysis-All Coding Exons)	ACP1	ACP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOX3 geni (Tüm Gen Dizi Analizi)	ACOX3 gene (Sequence Analysis-All Coding Exons)	ACOX3	ACOX3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT9 geni (Tüm Gen Dizi Analizi)	ACOT9 gene (Sequence Analysis-All Coding Exons)	ACOT9	ACOT9, MTACT48	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT8 geni (Tüm Gen Dizi Analizi)	ACOT8 gene (Sequence Analysis-All Coding Exons)	ACOT8	ACOT8, PTE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT7 geni (Tüm Gen Dizi Analizi)	ACOT7 gene (Sequence Analysis-All Coding Exons)	ACOT7	ACOT7, BACH, LACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT6 geni (Tüm Gen Dizi Analizi)	ACOT6 gene (Sequence Analysis-All Coding Exons)	ACOT6	ACOT6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT4 geni (Tüm Gen Dizi Analizi)	ACOT4 gene (Sequence Analysis-All Coding Exons)	ACOT4	ACOT4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT2 geni (Tüm Gen Dizi Analizi)	ACOT2 gene (Sequence Analysis-All Coding Exons)	ACOT2	ACOT2, PTE2, MTE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT13 geni (Tüm Gen Dizi Analizi)	ACOT13 gene (Sequence Analysis-All Coding Exons)	ACOT13	ACOT13, THEM2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTSE1 geni (Tüm Gen Dizi Analizi)	GTSE1 gene (Sequence Analysis-All Coding Exons)	GTSE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT12 geni (Tüm Gen Dizi Analizi)	ACOT12 gene (Sequence Analysis-All Coding Exons)	ACOT12	ACOT12, CACH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT11 geni (Tüm Gen Dizi Analizi)	ACOT11 gene (Sequence Analysis-All Coding Exons)	ACOT11	ACOT11, THEA, BFIT, BFIT1, BFIT2, KIAA0707	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOT1 geni (Tüm Gen Dizi Analizi)	ACOT1 gene (Sequence Analysis-All Coding Exons)	ACOT1	ACOT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACOD1 geni (Tüm Gen Dizi Analizi)	ACOD1 gene (Sequence Analysis-All Coding Exons)	ACOD1	IRG1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACO1 geni (Tüm Gen Dizi Analizi)	ACO1 gene (Sequence Analysis-All Coding Exons)	ACO1	ACO1, IREB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACLY geni (Tüm Gen Dizi Analizi)	ACLY gene (Sequence Analysis-All Coding Exons)	ACLY	ACLY	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACKR4 geni (Tüm Gen Dizi Analizi)	ACKR4 gene (Sequence Analysis-All Coding Exons)	ACKR4	ACKR4, CCRL1, PPR1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C4 geni (Tüm Gen Dizi Analizi)	GTF3C4 gene (Sequence Analysis-All Coding Exons)	GTF3C4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACKR3 geni (Tüm Gen Dizi Analizi)	ACKR3 gene (Sequence Analysis-All Coding Exons)	ACKR3	ACKR3, CXCR7, CMKOR1, GPR159, RDC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF3C2 geni (Tüm Gen Dizi Analizi)	GTF3C2 gene (Sequence Analysis-All Coding Exons)	GTF3C2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACKR2 geni (Tüm Gen Dizi Analizi)	ACKR2 gene (Sequence Analysis-All Coding Exons)	ACKR2	ACKR2, CCBP2, CMKBR9, D6, CCR9, CCR10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACIN1 geni (Tüm Gen Dizi Analizi)	ACIN1 gene (Sequence Analysis-All Coding Exons)	ACIN1	ACIN1, KIAA0670	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2IRD2B geni (Tüm Gen Dizi Analizi)	GTF2IRD2B gene (Sequence Analysis-All Coding Exons)	GTF2IRD2B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2IRD2 geni (Tüm Gen Dizi Analizi)	GTF2IRD2 gene (Sequence Analysis-All Coding Exons)	GTF2IRD2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACER3 geni (Tüm Gen Dizi Analizi)	ACER3 gene (Sequence Analysis-All Coding Exons)	ACER3	ACER3, APHC, PHCA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACER2 geni (Tüm Gen Dizi Analizi)	ACER2 gene (Sequence Analysis-All Coding Exons)	ACER2	ACER2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACER1 geni (Tüm Gen Dizi Analizi)	ACER1 gene (Sequence Analysis-All Coding Exons)	ACER1	ACER1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACE2 geni (Tüm Gen Dizi Analizi)	ACE2 gene (Sequence Analysis-All Coding Exons)	ACE2	ACE2, ACEH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GTF2H3 geni (Tüm Gen Dizi Analizi)	GTF2H3 gene (Sequence Analysis-All Coding Exons)	GTF2H3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACCS geni (Tüm Gen Dizi Analizi)	ACCS gene (Sequence Analysis-All Coding Exons)	ACCS	ACCS, ACS, PHACS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACBD6 geni (Tüm Gen Dizi Analizi)	ACBD6 gene (Sequence Analysis-All Coding Exons)	ACBD6	ACBD6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACBD5 geni (Tüm Gen Dizi Analizi)	ACBD5 gene (Sequence Analysis-All Coding Exons)	ACBD5	ACBD5, KIAA1996	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACBD3 geni (Tüm Gen Dizi Analizi)	ACBD3 gene (Sequence Analysis-All Coding Exons)	ACBD3	ACBD3, GOCAP1, GOLPH1, GCP60	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAP2 geni (Tüm Gen Dizi Analizi)	ACAP2 gene (Sequence Analysis-All Coding Exons)	ACAP2	CENTB2, KIAA0041, ACAP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAP1 geni (Tüm Gen Dizi Analizi)	ACAP1 gene (Sequence Analysis-All Coding Exons)	ACAP1	CENTB1, KIAA0050, ACAP1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAD11 geni (Tüm Gen Dizi Analizi)	ACAD11 gene (Sequence Analysis-All Coding Exons)	ACAD11	ACAD11	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAD10 geni (Tüm Gen Dizi Analizi)	ACAD10 gene (Sequence Analysis-All Coding Exons)	ACAD10	ACAD10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACACB geni (Tüm Gen Dizi Analizi)	ACACB gene (Sequence Analysis-All Coding Exons)	ACACB	ACACB, ACCB, ACC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAA2 geni (Tüm Gen Dizi Analizi)	ACAA2 gene (Sequence Analysis-All Coding Exons)	ACAA2	ACAA2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACAA1 geni (Tüm Gen Dizi Analizi)	ACAA1 gene (Sequence Analysis-All Coding Exons)	ACAA1	ACAA1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABTB1 geni (Tüm Gen Dizi Analizi)	ABTB1 gene (Sequence Analysis-All Coding Exons)	ABTB1	ABTB1, BPOZ	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABRA geni (Tüm Gen Dizi Analizi)	ABRA gene (Sequence Analysis-All Coding Exons)	ABRA	ABRA, STARS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABR geni (Tüm Gen Dizi Analizi)	ABR gene (Sequence Analysis-All Coding Exons)	ABR	ABR	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABLIM3 geni (Tüm Gen Dizi Analizi)	ABLIM3 gene (Sequence Analysis-All Coding Exons)	ABLIM3	ABLIM3, KIAA0843	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABLIM2 geni (Tüm Gen Dizi Analizi)	ABLIM2 gene (Sequence Analysis-All Coding Exons)	ABLIM2	ABLIM2, KIAA1808	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSTT1 geni (Tüm Gen Dizi Analizi)	GSTT1 gene (Sequence Analysis-All Coding Exons)	GSTT1	GSTT1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABLIM1 geni (Tüm Gen Dizi Analizi)	ABLIM1 gene (Sequence Analysis-All Coding Exons)	ABLIM1	LIMAB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABI3BP geni (Tüm Gen Dizi Analizi)	ABI3BP gene (Sequence Analysis-All Coding Exons)	ABI3BP	TARSH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABI3 geni (Tüm Gen Dizi Analizi)	ABI3 gene (Sequence Analysis-All Coding Exons)	ABI3	NESH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABI1 geni (Tüm Gen Dizi Analizi)	ABI1 gene (Sequence Analysis-All Coding Exons)	ABI1	ABI1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABHD6 geni (Tüm Gen Dizi Analizi)	ABHD6 gene (Sequence Analysis-All Coding Exons)	ABHD6	ABHB6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABHD3 geni (Tüm Gen Dizi Analizi)	ABHD3 gene (Sequence Analysis-All Coding Exons)	ABHD3	ABHD3, LABH3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABHD2 geni (Tüm Gen Dizi Analizi)	ABHD2 gene (Sequence Analysis-All Coding Exons)	ABHD2	ABHD2, LABH2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABHD16A geni (Tüm Gen Dizi Analizi)	ABHD16A gene (Sequence Analysis-All Coding Exons)	ABHD16A	ABHD16A, BAT5, D6S82E	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABHD1 geni (Tüm Gen Dizi Analizi)	ABHD1 gene (Sequence Analysis-All Coding Exons)	ABHD1	ABHD1, LABH1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCG4 geni (Tüm Gen Dizi Analizi)	ABCG4 gene (Sequence Analysis-All Coding Exons)	ABCG4	ABCG4, WHITE2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCG1 geni (Tüm Gen Dizi Analizi)	ABCG1 gene (Sequence Analysis-All Coding Exons)	ABCG1	ABCG1, ABC8, WHITE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCF2 geni (Tüm Gen Dizi Analizi)	ABCF2 gene (Sequence Analysis-All Coding Exons)	ABCF2	ABCF2, HUSSY18	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCF1 geni (Tüm Gen Dizi Analizi)	ABCF1 gene (Sequence Analysis-All Coding Exons)	ABCF1	ABCF1, ABC50	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCE1 geni (Tüm Gen Dizi Analizi)	ABCE1 gene (Sequence Analysis-All Coding Exons)	ABCE1	ABCE1, RNASEL1, RNS4I	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCD2 geni (Tüm Gen Dizi Analizi)	ABCD2 gene (Sequence Analysis-All Coding Exons)	ABCD2	ABCD2, ALDR, ALDL1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC5 geni (Tüm Gen Dizi Analizi)	ABCC5 gene (Sequence Analysis-All Coding Exons)	ABCC5	ABCC5, MRP5, MOATC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC4 geni (Tüm Gen Dizi Analizi)	ABCC4 gene (Sequence Analysis-All Coding Exons)	ABCC4	ABCC4, MRP4, MOATB	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC3 geni (Tüm Gen Dizi Analizi)	ABCC3 gene (Sequence Analysis-All Coding Exons)	ABCC3	ABCC3, CMOAT2, MRP3, MLP2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC13 geni (Tüm Gen Dizi Analizi)	ABCC13 gene (Sequence Analysis-All Coding Exons)	ABCC13	ABCC13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC12 geni (Tüm Gen Dizi Analizi)	ABCC12 gene (Sequence Analysis-All Coding Exons)	ABCC12	ABCC12, MRP9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ABCC10 geni (Tüm Gen Dizi Analizi)	ABCC10 gene (Sequence Analysis-All Coding Exons)	ABCC10	ABCC10, MRP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCC1 geni (Tüm Gen Dizi Analizi)	ABCC1 gene (Sequence Analysis-All Coding Exons)	ABCC1	ABCC1, MRP1, MRP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCB9 geni (Tüm Gen Dizi Analizi)	ABCB9 gene (Sequence Analysis-All Coding Exons)	ABCB9	ABCB9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSK3A geni (Tüm Gen Dizi Analizi)	GSK3A gene (Sequence Analysis-All Coding Exons)	GSK3A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GSG2 geni (Tüm Gen Dizi Analizi)	GSG2 gene (Sequence Analysis-All Coding Exons)	GSG2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCB8 geni (Tüm Gen Dizi Analizi)	ABCB8 gene (Sequence Analysis-All Coding Exons)	ABCB8	ABCB8, MABC1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCB5 geni (Tüm Gen Dizi Analizi)	ABCB5 gene (Sequence Analysis-All Coding Exons)	ABCB5	ABCB5	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCB10 geni (Tüm Gen Dizi Analizi)	ABCB10 gene (Sequence Analysis-All Coding Exons)	ABCB10	ABCB10, MTABC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA9 geni (Tüm Gen Dizi Analizi)	ABCA9 gene (Sequence Analysis-All Coding Exons)	ABCA9	ABCA9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA8 geni (Tüm Gen Dizi Analizi)	ABCA8 gene (Sequence Analysis-All Coding Exons)	ABCA8	ABCA8, KIAA0822	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA6 geni (Tüm Gen Dizi Analizi)	ABCA6 gene (Sequence Analysis-All Coding Exons)	ABCA6	ABCA6	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA5 geni (Tüm Gen Dizi Analizi)	ABCA5 gene (Sequence Analysis-All Coding Exons)	ABCA5	ABCA5, KIAA1888	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA2 geni (Tüm Gen Dizi Analizi)	ABCA2 gene (Sequence Analysis-All Coding Exons)	ABCA2	ABCA2, ABC2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA13 geni (Tüm Gen Dizi Analizi)	ABCA13 gene (Sequence Analysis-All Coding Exons)	ABCA13	ABCA13	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABCA10 geni (Tüm Gen Dizi Analizi)	ABCA10 gene (Sequence Analysis-All Coding Exons)	ABCA10	ABCA10	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABALON geni (Tüm Gen Dizi Analizi)	ABALON gene (Sequence Analysis-All Coding Exons)	ABALON	ABALON, INXS	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AATK geni (Tüm Gen Dizi Analizi)	AATK gene (Sequence Analysis-All Coding Exons)	AATK	AATK, AATYK	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AATF geni (Tüm Gen Dizi Analizi)	AATF gene (Sequence Analysis-All Coding Exons)	AATF	AATF, CHE1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AASDHPPT geni (Tüm Gen Dizi Analizi)	AASDHPPT gene (Sequence Analysis-All Coding Exons)	AASDHPPT	AASDHPPT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AASDH geni (Tüm Gen Dizi Analizi)	AASDH gene (Sequence Analysis-All Coding Exons)	AASDH	AASDH, ACSF4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AARSD1 geni (Tüm Gen Dizi Analizi)	AARSD1 gene (Sequence Analysis-All Coding Exons)	AARSD1	AARSD1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AAR2 geni (Tüm Gen Dizi Analizi)	AAR2 gene (Sequence Analysis-All Coding Exons)	AAR2	AAR2, C20orf4	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AANAT geni (Tüm Gen Dizi Analizi)	AANAT gene (Sequence Analysis-All Coding Exons)	AANAT	AANAT, SNAT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AAMP geni (Tüm Gen Dizi Analizi)	AAMP gene (Sequence Analysis-All Coding Exons)	AAMP	AAMP	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AAK1 geni (Tüm Gen Dizi Analizi)	AAK1 gene (Sequence Analysis-All Coding Exons)	AAK1	AAK1, KIAA1048	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AADAT geni (Tüm Gen Dizi Analizi)	AADAT gene (Sequence Analysis-All Coding Exons)	AADAT	AADAT, KAT2	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AADAC geni (Tüm Gen Dizi Analizi)	AADAC gene (Sequence Analysis-All Coding Exons)	AADAC	AADAC, DAC	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AACS geni (Tüm Gen Dizi Analizi)	AACS gene (Sequence Analysis-All Coding Exons)	AACS	AACS, ACSF1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
A4GNT geni (Tüm Gen Dizi Analizi)	A4GNT gene (Sequence Analysis-All Coding Exons)	A4GNT	A4GNT	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
A2ML1 geni (Tüm Gen Dizi Analizi)	A2ML1 gene (Sequence Analysis-All Coding Exons)	A2ML1	A2ML1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
A1BG geni (Tüm Gen Dizi Analizi)	A1BG gene (Sequence Analysis-All Coding Exons)	A1BG	A1BG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRIN3B geni (Tüm Gen Dizi Analizi)	GRIN3B gene (Sequence Analysis-All Coding Exons)	GRIN3B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GRID2IP geni (Tüm Gen Dizi Analizi)	GRID2IP gene (Sequence Analysis-All Coding Exons)	GRID2IP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ZIC4 (MLPA)	ZIC4 (MLPA)	ZIC4	ZIC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
SLC2A1 (MLPA)	SLC2A1 (MLPA)	SLC2A1	HTLVR	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	DISEASE: Dystonia 9 (DYT9) [MIM:601042]: An autosomal dominant neurologic disorder characterized by childhood onset of paroxysmal choreoathetosis and progressive spastic paraplegia. Most patients show some degree of cognitive impairment. Other variable features may include seizures, migraine headaches, and ataxia. {ECO:0000269	PubMed:21832227}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	EDTA Blood Sample
NTNG1 (MLPA)	NTNG1 (MLPA)	NTNG1	NTNG1, LMNT1, KIAA0976	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
IGFBP3 (MLPA)	IGFBP3 (MLPA)	IGFBP3	IGFBP3	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
GRAP geni (Tüm Gen Dizi Analizi)	GRAP gene (Sequence Analysis-All Coding Exons)	GRAP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPC4 (MLPA)	GPC4 (MLPA)	GPC4	GPC4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CYP3A4 (MLPA)	CYP3A4 (MLPA)	CYP3A4	CYP3A4	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CYP2E1 (MLPA)	CYP2E1 (MLPA)	CYP2E1	CYP2E, CYP2E1, P450C2E	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CYP1A1 (MLPA)	CYP1A1 (MLPA)	CYP1A1	CYP1A1, CYP1	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
CFHR2 (MLPA)	CFHR2 (MLPA)	CFHR2	CFHR2, FHR2, HFL3, CFHL2	MLPA (CNV)	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
GPSM1 geni (Tüm Gen Dizi Analizi)	GPSM1 gene (Sequence Analysis-All Coding Exons)	GPSM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPS2 geni (Tüm Gen Dizi Analizi)	GPS2 gene (Sequence Analysis-All Coding Exons)	GPS2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPRC6A geni (Tüm Gen Dizi Analizi)	GPRC6A gene (Sequence Analysis-All Coding Exons)	GPRC6A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR42 geni (Tüm Gen Dizi Analizi)	GPR42 gene (Sequence Analysis-All Coding Exons)	GPR42	GPR42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPR182 geni (Tüm Gen Dizi Analizi)	GPR182 gene (Sequence Analysis-All Coding Exons)	GPR182		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPNMB geni (Tüm Gen Dizi Analizi)	GPNMB gene (Sequence Analysis-All Coding Exons)	GPNMB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPHB5 geni (Tüm Gen Dizi Analizi)	GPHB5 gene (Sequence Analysis-All Coding Exons)	GPHB5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPHA2 geni (Tüm Gen Dizi Analizi)	GPHA2 gene (Sequence Analysis-All Coding Exons)	GPHA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GPA33 geni (Tüm Gen Dizi Analizi)	GPA33 gene (Sequence Analysis-All Coding Exons)	GPA33		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GORASP2 geni (Tüm Gen Dizi Analizi)	GORASP2 gene (Sequence Analysis-All Coding Exons)	GORASP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GOLGA7 geni (Tüm Gen Dizi Analizi)	GOLGA7 gene (Sequence Analysis-All Coding Exons)	GOLGA7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG7 geni (Tüm Gen Dizi Analizi)	GNG7 gene (Sequence Analysis-All Coding Exons)	GNG7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GNG3 geni (Tüm Gen Dizi Analizi)	GNG3 gene (Sequence Analysis-All Coding Exons)	GNG3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GMFB geni (Tüm Gen Dizi Analizi)	GMFB gene (Sequence Analysis-All Coding Exons)	GMFB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GJC1 geni (Tüm Gen Dizi Analizi)	GJC1 gene (Sequence Analysis-All Coding Exons)	GJC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GIT2 geni (Tüm Gen Dizi Analizi)	GIT2 gene (Sequence Analysis-All Coding Exons)	GIT2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GHDC geni (Tüm Gen Dizi Analizi)	GHDC gene (Sequence Analysis-All Coding Exons)	GHDC		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGTLC3 geni (Tüm Gen Dizi Analizi)	GGTLC3 gene (Sequence Analysis-All Coding Exons)	GGTLC3	GGTLC3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GGA2 geni (Tüm Gen Dizi Analizi)	GGA2 gene (Sequence Analysis-All Coding Exons)	GGA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GEMIN4 geni (Tüm Gen Dizi Analizi)	GEMIN4 gene (Sequence Analysis-All Coding Exons)	GEMIN4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GDF10 geni (Tüm Gen Dizi Analizi)	GDF10 gene (Sequence Analysis-All Coding Exons)	GDF10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
GABBR1 geni (Tüm Gen Dizi Analizi)	GABBR1 gene (Sequence Analysis-All Coding Exons)	GABBR1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUT11 geni (Tüm Gen Dizi Analizi)	FUT11 gene (Sequence Analysis-All Coding Exons)	FUT11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FUK geni (Tüm Gen Dizi Analizi)	FUK gene (Sequence Analysis-All Coding Exons)	FUK		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FTCDNL1 geni (Tüm Gen Dizi Analizi)	FTCDNL1 gene (Sequence Analysis-All Coding Exons)	FTCDNL1	FTCDNL1, FONG	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRS3 geni (Tüm Gen Dizi Analizi)	FRS3 gene (Sequence Analysis-All Coding Exons)	FRS3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FRAT1 geni (Tüm Gen Dizi Analizi)	FRAT1 gene (Sequence Analysis-All Coding Exons)	FRAT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FOLR3 geni (Tüm Gen Dizi Analizi)	FOLR3 gene (Sequence Analysis-All Coding Exons)	FOLR3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FLT3LG geni (Tüm Gen Dizi Analizi)	FLT3LG gene (Sequence Analysis-All Coding Exons)	FLT3LG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FGFR1OP2 geni (Tüm Gen Dizi Analizi)	FGFR1OP2 gene (Sequence Analysis-All Coding Exons)	FGFR1OP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FEM1C geni (Tüm Gen Dizi Analizi)	FEM1C gene (Sequence Analysis-All Coding Exons)	FEM1C		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FDCSP geni (Tüm Gen Dizi Analizi)	FDCSP gene (Sequence Analysis-All Coding Exons)	FDCSP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FCRL6 geni (Tüm Gen Dizi Analizi)	FCRL6 gene (Sequence Analysis-All Coding Exons)	FCRL6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FBN3 geni (Tüm Gen Dizi Analizi)	FBN3 gene (Sequence Analysis-All Coding Exons)	FBN3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FARSA geni (Tüm Gen Dizi Analizi)	FARSA gene (Sequence Analysis-All Coding Exons)	FARSA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM83G geni (Tüm Gen Dizi Analizi)	FAM83G gene (Sequence Analysis-All Coding Exons)	FAM83G		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM46A geni (Tüm Gen Dizi Analizi)	FAM46A gene (Sequence Analysis-All Coding Exons)	FAM46A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
FAM122A geni (Tüm Gen Dizi Analizi)	FAM122A gene (Sequence Analysis-All Coding Exons)	FAM122A	FAM122A, C9orf42	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EXOC3L2 geni (Tüm Gen Dizi Analizi)	EXOC3L2 gene (Sequence Analysis-All Coding Exons)	EXOC3L2	EXOC3L2, XTP7	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERP27 geni (Tüm Gen Dizi Analizi)	ERP27 gene (Sequence Analysis-All Coding Exons)	ERP27		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERI1 geni (Tüm Gen Dizi Analizi)	ERI1 gene (Sequence Analysis-All Coding Exons)	ERI1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EREG geni (Tüm Gen Dizi Analizi)	EREG gene (Sequence Analysis-All Coding Exons)	EREG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERC1 geni (Tüm Gen Dizi Analizi)	ERC1 gene (Sequence Analysis-All Coding Exons)	ERC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ERAL1 geni (Tüm Gen Dizi Analizi)	ERAL1 gene (Sequence Analysis-All Coding Exons)	ERAL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EPHA4 geni (Tüm Gen Dizi Analizi)	EPHA4 gene (Sequence Analysis-All Coding Exons)	EPHA4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMILIN3 geni (Tüm Gen Dizi Analizi)	EMILIN3 gene (Sequence Analysis-All Coding Exons)	EMILIN3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMILIN2 geni (Tüm Gen Dizi Analizi)	EMILIN2 gene (Sequence Analysis-All Coding Exons)	EMILIN2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMID1 geni (Tüm Gen Dizi Analizi)	EMID1 gene (Sequence Analysis-All Coding Exons)	EMID1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EMCN geni (Tüm Gen Dizi Analizi)	EMCN gene (Sequence Analysis-All Coding Exons)	EMCN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

ELOB geni (Tüm Gen Dizi Analizi)	ELOB gene (Sequence Analysis-All Coding Exons)	ELOB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ELOA2 geni (Tüm Gen Dizi Analizi)	ELOA2 gene (Sequence Analysis-All Coding Exons)	ELOA2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF5B geni (Tüm Gen Dizi Analizi)	EIF5B gene (Sequence Analysis-All Coding Exons)	EIF5B		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF4EBP3 geni (Tüm Gen Dizi Analizi)	EIF4EBP3 gene (Sequence Analysis-All Coding Exons)	EIF4EBP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2S2 geni (Tüm Gen Dizi Analizi)	EIF2S2 gene (Sequence Analysis-All Coding Exons)	EIF2S2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EIF2S1 geni (Tüm Gen Dizi Analizi)	EIF2S1 gene (Sequence Analysis-All Coding Exons)	EIF2S1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EFNA5 geni (Tüm Gen Dizi Analizi)	EFNA5 gene (Sequence Analysis-All Coding Exons)	EFNA5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDIL3 geni (Tüm Gen Dizi Analizi)	EDIL3 gene (Sequence Analysis-All Coding Exons)	EDIL3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDF1 geni (Tüm Gen Dizi Analizi)	EDF1 gene (Sequence Analysis-All Coding Exons)	EDF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EDEM3 geni (Tüm Gen Dizi Analizi)	EDEM3 gene (Sequence Analysis-All Coding Exons)	EDEM3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
EAPP geni (Tüm Gen Dizi Analizi)	EAPP gene (Sequence Analysis-All Coding Exons)	EAPP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
E2F6 geni (Tüm Gen Dizi Analizi)	E2F6 gene (Sequence Analysis-All Coding Exons)	E2F6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DZIP3 geni (Tüm Gen Dizi Analizi)	DZIP3 gene (Sequence Analysis-All Coding Exons)	DZIP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DZIP1 geni (Tüm Gen Dizi Analizi)	DZIP1 gene (Sequence Analysis-All Coding Exons)	DZIP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DYNC1LI2 geni (Tüm Gen Dizi Analizi)	DYNC1LI2 gene (Sequence Analysis-All Coding Exons)	DYNC1LI2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DVL2 geni (Tüm Gen Dizi Analizi)	DVL2 gene (Sequence Analysis-All Coding Exons)	DVL2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP19 geni (Tüm Gen Dizi Analizi)	DUSP19 gene (Sequence Analysis-All Coding Exons)	DUSP19		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DUSP18 geni (Tüm Gen Dizi Analizi)	DUSP18 gene (Sequence Analysis-All Coding Exons)	DUSP18		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DRG1 geni (Tüm Gen Dizi Analizi)	DRG1 gene (Sequence Analysis-All Coding Exons)	DRG1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DPP9 geni (Tüm Gen Dizi Analizi)	DPP9 gene (Sequence Analysis-All Coding Exons)	DPP9		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DONSON geni (Tüm Gen Dizi Analizi)	DONSON gene (Sequence Analysis-All Coding Exons)	DONSON		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DOK6 geni (Tüm Gen Dizi Analizi)	DOK6 gene (Sequence Analysis-All Coding Exons)	DOK6		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNASE1L2 geni (Tüm Gen Dizi Analizi)	DNASE1L2 gene (Sequence Analysis-All Coding Exons)	DNASE1L2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DNAJA1 geni (Tüm Gen Dizi Analizi)	DNAJA1 gene (Sequence Analysis-All Coding Exons)	DNAJA1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLL1 geni (Tüm Gen Dizi Analizi)	DLL1 gene (Sequence Analysis-All Coding Exons)	DLL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLGAP3 geni (Tüm Gen Dizi Analizi)	DLGAP3 gene (Sequence Analysis-All Coding Exons)	DLGAP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DLEU1 geni (Tüm Gen Dizi Analizi)	DLEU1 gene (Sequence Analysis-All Coding Exons)	DLEU1	DLEU1, LEU1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DKK2 geni (Tüm Gen Dizi Analizi)	DKK2 gene (Sequence Analysis-All Coding Exons)	DKK2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DIRC3 geni (Tüm Gen Dizi Analizi)	DIRC3 gene (Sequence Analysis-All Coding Exons)	DIRC3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DHDH geni (Tüm Gen Dizi Analizi)	DHDH gene (Sequence Analysis-All Coding Exons)	DHDH		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DEFA3 geni (Tüm Gen Dizi Analizi)	DEFA3 gene (Sequence Analysis-All Coding Exons)	DEFA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DCAF7 geni (Tüm Gen Dizi Analizi)	DCAF7 gene (Sequence Analysis-All Coding Exons)	DCAF7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAZAP2 geni (Tüm Gen Dizi Analizi)	DAZAP2 gene (Sequence Analysis-All Coding Exons)	DAZAP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
DAZAP1 geni (Tüm Gen Dizi Analizi)	DAZAP1 gene (Sequence Analysis-All Coding Exons)	DAZAP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYBRD1 geni (Tüm Gen Dizi Analizi)	CYBRD1 gene (Sequence Analysis-All Coding Exons)	CYBRD1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CXXC1 geni (Tüm Gen Dizi Analizi)	CXXC1 gene (Sequence Analysis-All Coding Exons)	CXXC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CUX2 geni (Tüm Gen Dizi Analizi)	CUX2 gene (Sequence Analysis-All Coding Exons)	CUX2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTU2 geni (Tüm Gen Dizi Analizi)	CTU2 gene (Sequence Analysis-All Coding Exons)	CTU2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTDSPL geni (Tüm Gen Dizi Analizi)	CTDSPL gene (Sequence Analysis-All Coding Exons)	CTDSPL		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTDSP2 geni (Tüm Gen Dizi Analizi)	CTDSP2 gene (Sequence Analysis-All Coding Exons)	CTDSP2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTAGE4 geni (Tüm Gen Dizi Analizi)	CTAGE4 gene (Sequence Analysis-All Coding Exons)	CTAGE4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CTAGE1 geni (Tüm Gen Dizi Analizi)	CTAGE1 gene (Sequence Analysis-All Coding Exons)	CTAGE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CST8 geni (Tüm Gen Dizi Analizi)	CST8 gene (Sequence Analysis-All Coding Exons)	CST8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CSPG5 geni (Tüm Gen Dizi Analizi)	CSPG5 gene (Sequence Analysis-All Coding Exons)	CSPG5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRYGN geni (Tüm Gen Dizi Analizi)	CRYGN gene (Sequence Analysis-All Coding Exons)	CRYGN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRTAC1 geni (Tüm Gen Dizi Analizi)	CRTAC1 gene (Sequence Analysis-All Coding Exons)	CRTAC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRNN geni (Tüm Gen Dizi Analizi)	CRNN gene (Sequence Analysis-All Coding Exons)	CRNN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CRELD2 geni (Tüm Gen Dizi Analizi)	CRELD2 gene (Sequence Analysis-All Coding Exons)	CRELD2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CREBZF geni (Tüm Gen Dizi Analizi)	CREBZF gene (Sequence Analysis-All Coding Exons)	CREBZF		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPXM1 geni (Tüm Gen Dizi Analizi)	CPXM1 gene (Sequence Analysis-All Coding Exons)	CPXM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPSF3 geni (Tüm Gen Dizi Analizi)	CPSF3 gene (Sequence Analysis-All Coding Exons)	CPSF3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPSF2 geni (Tüm Gen Dizi Analizi)	CPSF2 gene (Sequence Analysis-All Coding Exons)	CPSF2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPSF1 geni (Tüm Gen Dizi Analizi)	CPSF1 gene (Sequence Analysis-All Coding Exons)	CPSF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPO geni (Tüm Gen Dizi Analizi)	CPO gene (Sequence Analysis-All Coding Exons)	CPO		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPLX4 geni (Tüm Gen Dizi Analizi)	CPLX4 gene (Sequence Analysis-All Coding Exons)	CPLX4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPLX3 geni (Tüm Gen Dizi Analizi)	CPLX3 gene (Sequence Analysis-All Coding Exons)	CPLX3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CPA5 geni (Tüm Gen Dizi Analizi)	CPA5 gene (Sequence Analysis-All Coding Exons)	CPA5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COX6A2 geni (Tüm Gen Dizi Analizi)	COX6A2 gene (Sequence Analysis-All Coding Exons)	COX6A2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COQ3 geni (Tüm Gen Dizi Analizi)	COQ3 gene (Sequence Analysis-All Coding Exons)	COQ3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
COL26A1 geni (Tüm Gen Dizi Analizi)	COL26A1 gene (Sequence Analysis-All Coding Exons)	COL26A1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CNGA4 geni (Tüm Gen Dizi Analizi)	CNGA4 gene (Sequence Analysis-All Coding Exons)	CNGA4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIC4 geni (Tüm Gen Dizi Analizi)	CLIC4 gene (Sequence Analysis-All Coding Exons)	CLIC4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLIC1 geni (Tüm Gen Dizi Analizi)	CLIC1 gene (Sequence Analysis-All Coding Exons)	CLIC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN7 geni (Tüm Gen Dizi Analizi)	CLDN7 gene (Sequence Analysis-All Coding Exons)	CLDN7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CLDN18 geni (Tüm Gen Dizi Analizi)	CLDN18 gene (Sequence Analysis-All Coding Exons)	CLDN18		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIDEB geni (Tüm Gen Dizi Analizi)	CIDEB gene (Sequence Analysis-All Coding Exons)	CIDEB		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIB4 geni (Tüm Gen Dizi Analizi)	CIB4 gene (Sequence Analysis-All Coding Exons)	CIB4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CIB3 geni (Tüm Gen Dizi Analizi)	CIB3 gene (Sequence Analysis-All Coding Exons)	CIB3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHRFAM7A geni (Tüm Gen Dizi Analizi)	CHRFAM7A gene (Sequence Analysis-All Coding Exons)	CHRFAM7A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHORDC1 geni (Tüm Gen Dizi Analizi)	CHORDC1 gene (Sequence Analysis-All Coding Exons)	CHORDC1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHCHD1 geni (Tüm Gen Dizi Analizi)	CHCHD1 gene (Sequence Analysis-All Coding Exons)	CHCHD1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CHAF1A geni (Tüm Gen Dizi Analizi)	CHAF1A gene (Sequence Analysis-All Coding Exons)	CHAF1A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGN geni (Tüm Gen Dizi Analizi)	CGN gene (Sequence Analysis-All Coding Exons)	CGN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB8 geni (Tüm Gen Dizi Analizi)	CGB8 gene (Sequence Analysis-All Coding Exons)	CGB8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB5 geni (Tüm Gen Dizi Analizi)	CGB5 gene (Sequence Analysis-All Coding Exons)	CGB5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CGB2 geni (Tüm Gen Dizi Analizi)	CGB2 gene (Sequence Analysis-All Coding Exons)	CGB2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM3 geni (Tüm Gen Dizi Analizi)	CEACAM3 gene (Sequence Analysis-All Coding Exons)	CEACAM3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CEACAM19 geni (Tüm Gen Dizi Analizi)	CEACAM19 gene (Sequence Analysis-All Coding Exons)	CEACAM19		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDRT1 geni (Tüm Gen Dizi Analizi)	CDRT1 gene (Sequence Analysis-All Coding Exons)	CDRT1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDIPT geni (Tüm Gen Dizi Analizi)	CDIPT gene (Sequence Analysis-All Coding Exons)	CDIPT		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CDCA3 geni (Tüm Gen Dizi Analizi)	CDCA3 gene (Sequence Analysis-All Coding Exons)	CDCA3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD84 geni (Tüm Gen Dizi Analizi)	CD84 gene (Sequence Analysis-All Coding Exons)	CD84		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CD101 geni (Tüm Gen Dizi Analizi)	CD101 gene (Sequence Analysis-All Coding Exons)	CD101		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CCL3L3 geni (Tüm Gen Dizi Analizi)	CCL3L3 gene (Sequence Analysis-All Coding Exons)	CCL3L3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CBX1 geni (Tüm Gen Dizi Analizi)	CBX1 gene (Sequence Analysis-All Coding Exons)	CBX1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPNS2 geni (Tüm Gen Dizi Analizi)	CAPNS2 gene (Sequence Analysis-All Coding Exons)	CAPNS2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN14 geni (Tüm Gen Dizi Analizi)	CAPN14 gene (Sequence Analysis-All Coding Exons)	CAPN14		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAPN13 geni (Tüm Gen Dizi Analizi)	CAPN13 gene (Sequence Analysis-All Coding Exons)	CAPN13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CAMKK1 geni (Tüm Gen Dizi Analizi)	CAMKK1 gene (Sequence Analysis-All Coding Exons)	CAMKK1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALN1 geni (Tüm Gen Dizi Analizi)	CALN1 gene (Sequence Analysis-All Coding Exons)	CALN1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALML5 geni (Tüm Gen Dizi Analizi)	CALML5 gene (Sequence Analysis-All Coding Exons)	CALML5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CALCOCO2 geni (Tüm Gen Dizi Analizi)	CALCOCO2 gene (Sequence Analysis-All Coding Exons)	CALCOCO2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CACNG4 geni (Tüm Gen Dizi Analizi)	CACNG4 gene (Sequence Analysis-All Coding Exons)	CACNG4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample

CABP1 geni (Tüm Gen Dizi Analizi)	CABP1 gene (Sequence Analysis-All Coding Exons)	CABP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CABLES1 geni (Tüm Gen Dizi Analizi)	CABLES1 gene (Sequence Analysis-All Coding Exons)	CABLES1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CA13 geni (Tüm Gen Dizi Analizi)	CA13 gene (Sequence Analysis-All Coding Exons)	CA13		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C6orf15 geni (Tüm Gen Dizi Analizi)	C6orf15 gene (Sequence Analysis-All Coding Exons)	C6orf15		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C3orf35 geni (Tüm Gen Dizi Analizi)	C3orf35 gene (Sequence Analysis-All Coding Exons)	C3orf35		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
C1orf27 geni (Tüm Gen Dizi Analizi)	C1orf27 gene (Sequence Analysis-All Coding Exons)	C1orf27		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BTF3 geni (Tüm Gen Dizi Analizi)	BTF3 gene (Sequence Analysis-All Coding Exons)	BTF3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BTBD2 geni (Tüm Gen Dizi Analizi)	BTBD2 gene (Sequence Analysis-All Coding Exons)	BTBD2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BTBD1 geni (Tüm Gen Dizi Analizi)	BTBD1 gene (Sequence Analysis-All Coding Exons)	BTBD1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BRD8 geni (Tüm Gen Dizi Analizi)	BRD8 gene (Sequence Analysis-All Coding Exons)	BRD8		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOK geni (Tüm Gen Dizi Analizi)	BOK gene (Sequence Analysis-All Coding Exons)	BOK		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BOC geni (Tüm Gen Dizi Analizi)	BOC gene (Sequence Analysis-All Coding Exons)	BOC		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLZF1 geni (Tüm Gen Dizi Analizi)	BLZF1 gene (Sequence Analysis-All Coding Exons)	BLZF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BLACE geni (Tüm Gen Dizi Analizi)	BLACE gene (Sequence Analysis-All Coding Exons)	BLACE	BLACE	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BHLHA15 geni (Tüm Gen Dizi Analizi)	BHLHA15 gene (Sequence Analysis-All Coding Exons)	BHLHA15		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BCAS3 geni (Tüm Gen Dizi Analizi)	BCAS3 gene (Sequence Analysis-All Coding Exons)	BCAS3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
BAG5 geni (Tüm Gen Dizi Analizi)	BAG5 gene (Sequence Analysis-All Coding Exons)	BAG5		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATPAF1 geni (Tüm Gen Dizi Analizi)	ATPAF1 gene (Sequence Analysis-All Coding Exons)	ATPAF1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP9A geni (Tüm Gen Dizi Analizi)	ATP9A gene (Sequence Analysis-All Coding Exons)	ATP9A		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP8B4 geni (Tüm Gen Dizi Analizi)	ATP8B4 gene (Sequence Analysis-All Coding Exons)	ATP8B4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP8A1 geni (Tüm Gen Dizi Analizi)	ATP8A1 gene (Sequence Analysis-All Coding Exons)	ATP8A1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1H geni (Tüm Gen Dizi Analizi)	ATP6V1H gene (Sequence Analysis-All Coding Exons)	ATP6V1H		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATP6V1F geni (Tüm Gen Dizi Analizi)	ATP6V1F gene (Sequence Analysis-All Coding Exons)	ATP6V1F		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG3 geni (Tüm Gen Dizi Analizi)	ATG3 gene (Sequence Analysis-All Coding Exons)	ATG3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATG12 geni (Tüm Gen Dizi Analizi)	ATG12 gene (Sequence Analysis-All Coding Exons)	ATG12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ATE1 geni (Tüm Gen Dizi Analizi)	ATE1 gene (Sequence Analysis-All Coding Exons)	ATE1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASTL geni (Tüm Gen Dizi Analizi)	ASTL gene (Sequence Analysis-All Coding Exons)	ASTL		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASRGL1 geni (Tüm Gen Dizi Analizi)	ASRGL1 gene (Sequence Analysis-All Coding Exons)	ASRGL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASCL4 geni (Tüm Gen Dizi Analizi)	ASCL4 gene (Sequence Analysis-All Coding Exons)	ASCL4		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ASCL3 geni (Tüm Gen Dizi Analizi)	ASCL3 gene (Sequence Analysis-All Coding Exons)	ASCL3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARTN geni (Tüm Gen Dizi Analizi)	ARTN gene (Sequence Analysis-All Coding Exons)	ARTN		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARPP21 geni (Tüm Gen Dizi Analizi)	ARPP21 gene (Sequence Analysis-All Coding Exons)	ARPP21		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGEF11 geni (Tüm Gen Dizi Analizi)	ARHGEF11 gene (Sequence Analysis-All Coding Exons)	ARHGEF11		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGDIG geni (Tüm Gen Dizi Analizi)	ARHGDIG gene (Sequence Analysis-All Coding Exons)	ARHGDIG		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARHGAP20 geni (Tüm Gen Dizi Analizi)	ARHGAP20 gene (Sequence Analysis-All Coding Exons)	ARHGAP20		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ARFIP1 geni (Tüm Gen Dizi Analizi)	ARFIP1 gene (Sequence Analysis-All Coding Exons)	ARFIP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AQP10 geni (Tüm Gen Dizi Analizi)	AQP10 gene (Sequence Analysis-All Coding Exons)	AQP10		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOM geni (Tüm Gen Dizi Analizi)	APOM gene (Sequence Analysis-All Coding Exons)	APOM		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APOBEC3D geni (Tüm Gen Dizi Analizi)	APOBEC3D gene (Sequence Analysis-All Coding Exons)	APOBEC3D		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
APBB1IP geni (Tüm Gen Dizi Analizi)	APBB1IP gene (Sequence Analysis-All Coding Exons)	APBB1IP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP3S2 geni (Tüm Gen Dizi Analizi)	AP3S2 gene (Sequence Analysis-All Coding Exons)	AP3S2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AP1G2 geni (Tüm Gen Dizi Analizi)	AP1G2 gene (Sequence Analysis-All Coding Exons)	AP1G2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ANGPTL1 geni (Tüm Gen Dizi Analizi)	ANGPTL1 gene (Sequence Analysis-All Coding Exons)	ANGPTL1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKT1S1 geni (Tüm Gen Dizi Analizi)	AKT1S1 gene (Sequence Analysis-All Coding Exons)	AKT1S1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKR7L geni (Tüm Gen Dizi Analizi)	AKR7L gene (Sequence Analysis-All Coding Exons)	AKR7L	AKR7A4, AFAR3	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKNA geni (Tüm Gen Dizi Analizi)	AKNA gene (Sequence Analysis-All Coding Exons)	AKNA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP7 geni (Tüm Gen Dizi Analizi)	AKAP7 gene (Sequence Analysis-All Coding Exons)	AKAP7		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP3 geni (Tüm Gen Dizi Analizi)	AKAP3 gene (Sequence Analysis-All Coding Exons)	AKAP3		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AKAP12 geni (Tüm Gen Dizi Analizi)	AKAP12 gene (Sequence Analysis-All Coding Exons)	AKAP12		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AJUBA geni (Tüm Gen Dizi Analizi)	AJUBA gene (Sequence Analysis-All Coding Exons)	AJUBA		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIG1 geni (Tüm Gen Dizi Analizi)	AIG1 gene (Sequence Analysis-All Coding Exons)	AIG1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AIFM2 geni (Tüm Gen Dizi Analizi)	AIFM2 gene (Sequence Analysis-All Coding Exons)	AIFM2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AHNAK2 geni (Tüm Gen Dizi Analizi)	AHNAK2 gene (Sequence Analysis-All Coding Exons)	AHNAK2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGTRAP geni (Tüm Gen Dizi Analizi)	AGTRAP gene (Sequence Analysis-All Coding Exons)	AGTRAP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AGAP1 geni (Tüm Gen Dizi Analizi)	AGAP1 gene (Sequence Analysis-All Coding Exons)	AGAP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
AEBP1 geni (Tüm Gen Dizi Analizi)	AEBP1 gene (Sequence Analysis-All Coding Exons)	AEBP1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADRM1 geni (Tüm Gen Dizi Analizi)	ADRM1 gene (Sequence Analysis-All Coding Exons)	ADRM1		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ADM2 geni (Tüm Gen Dizi Analizi)	ADM2 gene (Sequence Analysis-All Coding Exons)	ADM2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACRBP geni (Tüm Gen Dizi Analizi)	ACRBP gene (Sequence Analysis-All Coding Exons)	ACRBP		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ACMSD geni (Tüm Gen Dizi Analizi)	ACMSD gene (Sequence Analysis-All Coding Exons)	ACMSD		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
ABI2 geni (Tüm Gen Dizi Analizi)	ABI2 gene (Sequence Analysis-All Coding Exons)	ABI2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzom Sekanslama	Exome Sequencing	.	.	Ekzom/ Exome	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
Fabry Hastalığı, 301500, X'e Bağlı (Fabry Hastalığı)-TARAMA (GLA geni) (Tüm Gen Dizi Analizi) (Postnatal)	Fabry disease, 301500, X-linked (Fabry disease)-SCREENING (GLA gene) (Sequence Analysis-All Coding Exons) (Postnatal)	GLA	GLA	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Fabry disease (FD) [MIM:301500]: Rare X-linked sphingolipidosis disease where glycolipid accumulates in many tissues. The disease consists of an inborn error of glycosphingolipid catabolism. FD patients show systemic accumulation of globotriaosylceramide (Gb3) and related glycosphingolipids in the plasma and cellular lysosomes throughout the body. Clinical recognition in males results from characteristic skin lesions (angiokeratomas) over the lower trunk. Patients may show ocular deposits, febrile episodes, and burning pain in the extremities. Death results from renal failure, cardiac or cerebral complications of hypertension or other vascular disease. Heterozygous females may exhibit the disorder in an attenuated form, they are more likely to show corneal opacities. (ECO:0000269|PubMed:10090526, ECO:0000269|PubMed:10208848, ECO:0000269|PubMed:10666480, ECO:0000269|PubMed:10916280, ECO:0000269|PubMed:11076046, ECO:0000269|PubMed:11295840, ECO:0000269|PubMed:11688641, ECO:0000269|PubMed:11889412, ECO:0000269|PubMed:12694230, ECO:0000269|PubMed:1315715, ECO:0000269|PubMed:15162124, ECO:0000269|PubMed:15712228, ECO:0000269|PubMed:16533976, ECO:0000269|PubMed:1846223, ECO:0000269|PubMed:19621417, ECO:0000269|PubMed:2152885, ECO:0000269|PubMed:2171331, ECO:0000269|PubMed:2539398, ECO:0000269|PubMed:7504405.	EDTA Blood Sample
DNA izolasyonu	DNA isolation	.	.	.	EDTA'lı Kan (2-4 ml)	*	1	3	.	.	2	Postnatal	.	EDTA Blood Sample
RNA izolasyonu	RNA isolation	.	.	.	EDTA'lı Kan (2-4 ml)	*	1	3	.	.	2	Postnatal	.	EDTA Blood Sample
cDNA sentezi	cDNA synthesis	.	.	.	EDTA'lı Kan (2-4 ml)		1	3	.	.	2	Postnatal	.	EDTA Blood Sample
Ekzom Konfirmasyonu	Exome Confirmation	.	.	.	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Mikrosatellit İnstabilitesi (STR Analizi) (Kan)	Mikrosatellit Instability (STR Analysis) (Blood)	.	.	STR analizi	Parafinize Tümör Dokusu + EDTA'lı Kan (2-4 ml)		10	45	.	.	30	Postnatal	.	Parafinized Tumor Tissue + EDTA Blood Sample
Multipl Myeloma Paneli-2 (Del 1q21) (FISH)	Multiple Myeloma Panel-2 (Del 1q21) (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Sperm/ Gonad Dondurma Öncesi DNA Kimliklendirme Testi (STR Analizi)	DNA Fingerprint testing before Sperm and Gonad Sampling-Storing	.	.	STR analizi	EDTA'lı Kan (2-4 ml)		10	30	.	.	20	Postnatal	Türkiye'de yönetmelikler gereği Sperm yada gonad dondurulmadan önce kişinin DNA kimliklendirme testlerinin yapılması zorunlu değildir.	EDTA Blood Sample
Periferik Kandan Kromozom Eldesi- Analiz Edilmeden	Chromosome preparation without Karyotype Analysis	.	.	Kromozom analizi/ Karyotype analysis	Heparinli Kan (2-4 ml)		15	45	.	.	21	Postnatal	.	Heparinized blood sample
Trizomi 7 (FISH)	Tesomy 7 (FISH)	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)	*	10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Spinoserebellar Ataksi-Tekrar Sayısı Paneli (SCA1-SCA2-SCA3-SCA6-SCA7-SCA8-SCA10-SCA17)	Spinocerebellar Ataxia- Repeat Analysis Panel (SCA1-SCA2-SCA3-SCA6-SCA7-SCA8-SCA10-SCA17)	.	.	Tekrar sayısı/ Repeat Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal	.	EDTA Blood Sample
Array - CGH (PRENATAL)	Array - CGH (PRENATAL)	.	.	Array-CGH	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample

EGFR (Ekzon 18 - 19 - 20 - 21) (EGFR geni) (Dizi Analizi) (Postnatal)	EGFR (Exons 18 - 19 - 20 - 21) (EGFR gene) (Dizi Analizi) (Postnatal)	EGFR	.	Dizi Analizi/ Sequence Analysis	Tümör dokusu (Taze ve ya etanol ile fikse) / Parafinize doku bloğu		10	30	.	.	20	Postnatal	DISEASE: Leukemia, acute myelogenous (AML) [MIM:601626]: A subtype of acute leukemia, a cancer of the white blood cells. AML is a malignant disease of bone marrow characterized by maturational arrest of hematopoietic precursors at an early stage of development. Clonal expansion of myeloid blasts occurs in bone marrow, blood, and other tissue. Myelogenous leukemias develop from changes in cells that normally produce neutrophils, basophils, eosinophils and monocytes. {ECO:0000269	PubMed:8955068}. Note=The disease is caused by mutations affecting the gene represented in this entry.	Tumor tissue (Fresh or etanol fixed) /Parafinized tissue sample
Maternal kontaminasyon Testi (STR Analizi)	Maternal Contamination Test (STR analysis)	.	.	STR analizi	EDTA'lı Kan (2-4 ml) / Amniyon sıvısı /CVS/ EDTA'lı kordon kanı		.	.	15	30	20	Prenatal	.	EDTA Blood Sample / Amniotic fluid / CVS/ EDTA blood (BOTH) fetal blood	
Talasemi, Alfa-, 604131 (Heinz Cisimciği Anemisi) (HBA1-HBA2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	Thalassemia, alpha-, 604131 (Heinz body anemia) (HBA1-HBA2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	HBA1-HBA2	HBA2, HBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	EDTA Blood Sample
Talasemi, Alfa-, 604131 (Heinz Cisimciği Anemisi) (Prenatal) (HBA1-HBA2 gen) (Tüm Gen Dizi Analizi) (Prenatal)	Thalassemia, alpha-, 604131 (Heinz body anemia) (HBA1-HBA2 gene) (Sequence Analysis-All Coding Exons) (Prenatal)	HBA1-HBA2	HBA2, HBH	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Heinz body anemias (HEIBAN) [MIM:140700]: Form of non- spherocytic hemolytic anemia of Dacie type 1. After splenectomy, which has little benefit, basophilic inclusions called Heinz bodies are demonstrable in the erythrocytes. Before splenectomy, diffuse or punctate basophilia may be evident. Most of these cases are probably instances of hemoglobinopathy. The hemoglobin demonstrates heat lability. Heinz bodies are observed also with the Ivemark syndrome (asplenia with cardiovascular anomalies) and with glutathione peroxidase deficiency. {ECO:0000269	PubMed:2833478}. Note=The disease may be caused by mutations affecting the gene represented in this entry.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis X'e Bağlı, 308100, X'e Bağlı; XLI (Resesif X'e Bağlı İktiyozis) (Prenatal) (STS gen) (Tüm Gen Dizi Analizi) (Prenatal)	Ichthyosis, X-linked, 308100, X-linked recessive; XLI (Recessive X-linked ichthyosis) (STS gene) (Sequence Analysis-All Coding Exons) (Prenatal)	STS	STS, ARSC1, ARSC, SSDD, XLI	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, X-linked (IXL) [MIM:308100]: A keratinization disorder manifesting with mild erythroderma and generalized exfoliation of the skin within a few weeks after birth. Affected boys later develop large, polygonal, dark brown scales, especially on the neck, extremities, trunk, and buttocks. {ECO:0000269|PubMed:10679952, ECO:0000269|PubMed:10844566, ECO:0000269|PubMed:1539590, ECO:0000269|PubMed:9252398}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
İktiyozis X'e Bağlı, 308100, X'e Bağlı; XLI (Resesif X'e Bağlı İktiyozis) (Prenatal) (MLPA)	Ichthyosis, X-linked, 308100, X-linked recessive; XLI (Recessive X-linked ichthyosis) (Prenatal) (MLPA)	STS	STS, ARSC1, ARSC, SSDD, XLI	MLPA (CNV)	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal	DISEASE: Ichthyosis, X-linked (IXL) [MIM:308100]: A keratinization disorder manifesting with mild erythroderma and generalized exfoliation of the skin within a few weeks after birth. Affected boys later develop large, polygonal, dark brown scales, especially on the neck, extremities, trunk, and buttocks. {ECO:0000269|PubMed:10679952, ECO:0000269|PubMed:10844566, ECO:0000269|PubMed:1539590, ECO:0000269|PubMed:9252398}. Note=The disease is caused by mutations affecting the gene represented in this entry.;	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
CYP2C19 (CYP2C19 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP2C19 (CYP2C19 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C19	CYP2C, CYP2C19	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2D6 (CYP2D6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP2D6 (CYP2D6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2D6	CYP2D6, CPD6, P450DB1	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2A6 (CYP2A6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP2A6 (CYP2A6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2A6	CYP2A6, CYP2A3, CYP2A, P450C2A	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP1A2 (CYP1A2 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP1A2 (CYP1A2 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP1A2		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2B6 (CYP2B6 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP2B6 (CYP2B6 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2B6	CYP2B6, CYP2B, EFVM	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
CYP2C9 (CYP2C9 gen) (Tüm Gen Dizi Analizi) (Postnatal)	CYP2C9 (CYP2C9 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	CYP2C9	CYP2C9	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
t(4;11) (q21;q23) (MLL/ AF4) (FISH)	t(4;11) (q21;q23) (MLL/ AF4) (FISH)	4q21-11q23	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddei), 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B3	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddei), 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B1	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddei), 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B3 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B3 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B3	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Leukoencephalopathy With Vanishing White Matter; VWM (Lökoensefalopati Kaybolan Beyaz Maddei), 603896, Otozomal Resesif (Geç İnfantil CACH Sendromu) (EIF2B1 geni) (Tüm Gen Dizi Analizi) (Postnatal)	Leukoencephalopathy with vanishing white matter, 603896, Autosomal recessive; VWM (Late infantile CACH syndrome) (EIF2B1 gene) (Sequence Analysis-All Coding Exons) (Postnatal)	EIF2B1	.	Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzom Sekanslama (Prenatal)	Exome Sequencing (Prenatal)	.	.	Ekzom/ Exome	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	.	.	.	Prenatal	.	Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Trombofili Panel 3 (Tüm trombofili testleri) (. geni) (Dizi Analizi) (Postnatal)	Thrombophilia Panel 3 (All Thrombophilia tests) (Panel 1+ Panel 2) (. gene) (Sequence Analysis) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Trombofili Panel 1 (Sık Görülenler) (Protrombin G20210A Mutasyonu, Faktor V Leiden Mutasyonu, Faktör V R2 Mutasyonu, Faktör XIII V34L Mutasyonu, MTHFR 677 Polimorfizmi, MTHFR 1298 Polimorfizmi, PAI - 1 4G/ 5G Polimorfizm) (. geni) (Dizi Analizi) (Postnatal)	Thrombophilia Panel 1 (Frequent causes) (Protrombin G20210A mutation, Factor V Leiden mutation, Factor V R2 mutation, Faktör XIII V34L Mutasyonu, MTHFR 677 polymorphism, MTHFR 1298 polymorphism, PAI - 1 4G/ 5G polymorphism) (. gene) (Sequence Analysis) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
Trombofili Panel 2 (Diğer testler) (Faktör V Cambridge Mutasyonu, Faktör V HongKong Mutasyonu, ACE gen Polimorfizmi, EPCR A4600G Mutasyonu, EPCR G4678C Mutasyonu) (. gen) (Dizi Analizi) (Postnatal)	Thrombophilia Panel 2 (other tests) (Factor V Cambridge mutation, Factor V HongKong mutation, ACE gen polymorphism, EPCR A4600G mutation, EPCR G4678C mutation) (. gene) (Sequence Analysis) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		20	60	.	.	30	Postnatal	.	EDTA Blood Sample
(Multipl Myeloma Panel) t(4;14)(p16.3;q32.3) (IGH/FGFR3) t(11;14)(q13.3;q32) (IGH/MYEOV) t(11;14)(q13;q32) (IGH/CCND1) t(14;20) (IGH/MAFB) t(14;16) (IGH/MAF) PS3 13q14.3 (FISH)	Multiple Myeloma Panel t(4;14)(p16.3;q32.3) (IGH/FGFR3) t(11;14)(q13.3;q32) (IGH/MYEOV) t(11;14)(q13;q32) (IGH/CCND1) t(14;20) (IGH/MAFB) t(14;16) (IGH/MAF) PS3 13q14.3	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
KLL Paneli (Del 17p/ p53 mutasyonu t(4;14)(p16.3;q32.3)(FGFR3;IGH) t(11;14)(q13;q32) (CCND1;IGH) t(14;20) (q32.q12)(IGH/MAFB) t(14;16)(q32, q23.1-q23.2) IGH/MAF ATM del Trizomi 12 (TEL;AML1 PROBU) Del13q14.3 (FISH)	CLL Panel Del 17p/ p53 mutation t(4;14)(p16.3;q32.3)(FGFR3;IGH) t(11;14)(q13;q32) (CCND1;IGH) t(14;20) (q32.q12)(IGH/MAFB) t(14;16)(q32, q23.1-q23.2) IGH/MAF ATM del Trisomy 12 (TEL;AML1 PROBE) Del13q14.3	.	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Tüm Genom Sekanslama	Whole Genome Sequencing			Ekzom/ Exome	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
BCL6 Breakapart (FISH)	BCL6 Breakapart (FISH)	3q27.3	.	FISH	Heparinli Kan (2-4 ml) / Heparinli Kemik İliği (2-3 ml)		10	30	.	.	20	Postnatal	.	Heparinized blood sample / Heparinized bone marrow
Ekzom Sekanslama Data Analizi	Exome Sequencing Data Analysis			Ekzom/ Exome	Dosya		.	.	.	.	.	Postnatal	.	Documents
Pregnarisk Panel 1 (Alfa ve Beta Talasemi ile Orak Hücre anemisi - Fragil X - Spinal Müskiler Atrofi (SMA) - Duchenne Müskiler Distrofi (Dizi analizi+MLPA) - Fenilketonüri - Biotinidaz Eksikliği - 21 Hidroksilaz Eksikliği (Dizi analizi+MLPA) - Kistik Fibrozis)				Tüm Gen Dizi Analizi / MLPA / Tekrar sayısı	EDTA'lı Kan (2-4 ml)							Postnatal		
Pregnarisk Panel 2 (Geniş tarama (Klinik ekzom) - Fragil X - Spinal Müskiler Atrofi (SMA) - Duchenne Müskiler Distrofi (MLPA) - 21 Hidroksilaz Eksikliği (MLPA) - Alfa Talasemi (MLPA))				Tüm Gen Dizi Analizi / MLPA / Tekrar sayısı	EDTA'lı Kan (2-4 ml)							Postnatal		
Pregnarisk Panel 3 (Geniş tarama (Ekzom Sekanslama) - Fragil X - Spinal Müskiler Atrofi (SMA) - Duchenne Müskiler Distrofi (MLPA) - 21 Hidroksilaz Eksikliği (MLPA) - Alfa Talasemi (MLPA))				Tüm Gen Dizi Analizi / MLPA / Tekrar sayısı	EDTA'lı Kan (2-4 ml)							Postnatal		
Babyseq-TEMEL PANEL				Dizi Analizi/ Sequence Analysis	Özl Tüpte Kan (Merkezimizden Temin Ediniz)							Prenatal		
Babyseq-GENİŞ PANEL				Dizi Analizi/ Sequence Analysis	Özl Tüpte Kan (Merkezimizden Temin Ediniz)							Prenatal		
Klinik Ekzom				Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	Edta'lı kan							Postnatal		
Aneuploidi tarama (Düşük materyalinden) (NGS)	Aneuploidy screening (Abortus) (NGS)			Dizi Analizi/ Sequence Analysis	Merkezimizden temin edilen transport besiyeri içinde							Prenatal		Transport medium supplie/from our center
Aneuploidi tarama (CVS materyalinden) (NGS)	Aneuploidy screening (CVS) (NGS)			Dizi Analizi/ Sequence Analysis	Merkezimizden temin edilen transport besiyeri içinde							Prenatal		Transport medium supplie/from our center
t(9;22) (q34;q11.2) (BCR/ ABL) (Philadelphia Kromozomu) (p190) (REAL - TIME PCR)	t (9;22) (q34;q11.2) (BCR/ABL) (Philadelphia chromosome) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)	*	5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
t(9;22) (q34;q11.2) (BCR/ ABL) (Philadelphia Kromozomu) (p210) (REAL - TIME PCR)	t (9;22) (q34;q11.2) (BCR/ABL) (Philadelphia chromosome) (REAL-TIME PCR)	.	.	Real-Time PCR	EDTA'lı Kan (2-4 ml) (Soğuk zincir)	*	5	30	.	.	15	Postnatal	.	EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
RNAseq				Dizi Analizi/ Sequence Analysis	Edta'lı kan							Postnatal		EDTA Blood Sample/ EDTA Bone Marrow (Cold transfer)
CYP2C18 geni (Tüm Gen Dizi Analizi)	CYP2C18 gene (Sequence Analysis-All Coding Exons)	CYP2C18		Tüm Gen Dizi Analizi/ Sequence Analysis-All Coding Exons	EDTA'lı Kan (2-4 ml)		30	120	.	.	60	Postnatal	.	EDTA Blood Sample
Ekzom sekanslama (Ayrı alleden birden fazla bireyi)	Exome Sequencing (additional sample from same family)	.	.	Ekzom/ Exome	EDTA'lı Kan (2-4 ml)		.	.	.	.	.	Postnatal	.	EDTA Blood Sample
21 Hidroksilaz Eksikliği Paneli (CYP21A2) (index olgu) (CYP21A2 geni) (Tüm Gen Dizi Analizi + MLPA) (Postnatal) Anne - Baba - Çocuk	21 Hydroxylase Deficiency Panel (CYP21A2) (index) (Sequence Analysis-All Coding Exons + MLPA)	CYP21A2		Tüm Gen Dizi Analizi + MLPA	EDTA Blood Sample (2-4 ml)/ Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample		30	120	20	90				

21 Hidroksilaz Eksikliği Paneli (CYP21A2) (Ek bireyi) (CYP21A2 geni) (Tüm Gen Dizi Analizi + MLPA) (Postnatal) Anne - Baba - Çocuk	21 Hydroxylase Deficiency Panel (CYP21A2) (family members) (Sequence Analysis-All Coding Exons + MLPA)	CYP21A2		Tüm Gen Dizi Analizi + MLPA	EDTA Blood Sample (2-4 ml)/ Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample		30	120	20	90				
21 Hidroksilaz Eksikliği Paneli (CYP21A2) (Tekil çalışma) (CYP21A2 geni) (Tüm Gen Dizi Analizi + MLPA) (Postnatal)	21 Hydroxylase Deficiency Panel (CYP21A2) (one person) (Sequence Analysis-All Coding Exons + MLPA)	CYP21A2		Tüm Gen Dizi Analizi + MLPA	EDTA Blood Sample (2-4 ml)/ Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample		30	120	20	90				
Mikrosatellit İnstabilitesi (STR Analizi) (Doku)	Mikrosatellit Instability (STR Analysis) (Tissue)	.	.	STR analizi	Parafinize Tümör Dokusu + EDTA'lı Kan (2-4 ml)		10	45	.	.	30	Postnatal	.		Parafinized Tumor Tissue + EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (1 Disease)(CONTACT WITH OUR CENTER)	.	.	PGT	EDTA'lı Kan (2-4 ml)		15	45	.	.	28	Preimplantasyon	.		EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (2 Diseases)(CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	45			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (3 Diseases)(CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	45			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (4 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (4 Diseases)(CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	45			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (5 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (5 Diseases)(CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	45			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Kurulum (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Set-up (6 Diseases)(CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	45			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT)(1Embryo) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(2 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(3 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(4 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(5 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(6 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(7 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(8 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(9 Embryos)(1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(10 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(11 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(12 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (13 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(13 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (14 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(14 Embryo) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(15 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(16 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(17 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(18 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(19 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (1 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(20 Embryos) (1 Disease) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT)(1Embryo) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(2 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(3 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(4 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(5 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(6 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(7 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(8 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(9 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(10 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(11 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(12 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (13 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(13 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (14 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(14 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(15 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(16 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(17 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(18 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(19 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (2 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(20 Embryos) (2 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT)(1Embryo) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(2 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(3 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(4 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(5 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(6 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(7 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(8 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(9 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(10 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(11 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (3 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(12 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon			Embryo Biopsy

Preimplantasyon Genetik Test (PGT) (13 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(13 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (14 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(14 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(15 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(16 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(17 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(18 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(19 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (3 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(20 Embryos) (3 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing (PGT)(1Embryo) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(2 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(3 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(4 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(5 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(6 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(7 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(8 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(9 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(10 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(11 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(12 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (13 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(13 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (14 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(14 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(15 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(16 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(17 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(18 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(19 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (4 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(20 Embryos) (4 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing (PGT)(1Embryo) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(2 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(3 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(4 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(5 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(6 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(7 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(8 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(9 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(10 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(11 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(12 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (13 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(13 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (14 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(14 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(15 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(16 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(17 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(18 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(19 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (5 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(20 Embryos) (5 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (1 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing (PGT)(1Embryo) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (2 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(2 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (3 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(3 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (4 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(4 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (5 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(5 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (6 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(6 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (7 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(7 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (8 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(8 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (9 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(9 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (10 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(10 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (11 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(11 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (12 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(12 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (13 Embriyo) (6 Hastalık) (MEREZİMİZE DANIŞIN).	Preimplantation Genetic Testing(PGT)(13 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER).			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy

Preimplantasyon Genetik Test (PGT) (14 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(14 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (15 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(15 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (16 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(16 Embryo) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (17 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(17 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (18 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(18 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (19 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(19 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) (20 Embriyo) (6 Hastalık) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing(PGT)(20 Embryos) (6 Diseases) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) Kurulum (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) Set-up (CONTACT WITH OUR CENTER)			PGT	EDTA'lı Kan (2-4 ml)		15	4S			28	Preimplantasyon			EDTA Blood Sample
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (1 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (1 Embryo)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (2 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (2 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (3 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (3 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (4 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (4 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (5 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (5 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (6 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (6 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (7 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (7 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (8 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (8 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (9 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (9 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (10 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (10 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (11 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (11 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (12 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (12 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (13 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (13 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (14 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (14 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (15 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (15 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (16 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (16 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (17 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (17 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (18 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (18 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (19 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (19 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test (PGT) Doku Uyumu (HLA Analizi) (20 Embriyo) (MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing (PGT) Tissue Matching (HLA Analysis) (20 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (1 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(1 Embryo) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (2 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(2 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (3 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(3 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (4 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(4 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (5 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(5 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (6 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(6 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (7 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(7 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (8 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(8 Embryo) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (9 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(9 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (10 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(10 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (11 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(11 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (12 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(12 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (13 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(13 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (14 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(14 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (15 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(15 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (16 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(16 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (17 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(17 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (18 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(18 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (19 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(19 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (PGT-M Hastalanmıza) (20 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A)(For our PGT-M Patients)(20 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (1 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (1 Embryo)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (2 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (2 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (3 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (3 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (4 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (4 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (5 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (5 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy
Preimplantasyon Genetik Test Aneploidi için (PGT-A) (6 Embriyo)(MERKEZİMİZE DANIŞIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (6 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	4S			28	Preimplantasyon			Embryo Biopsy

Preimplantasyon Genetik Test Anöploidi için (PGT-A) (7Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (7 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (8 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (8 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (9 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (9 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (10 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (10 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (11 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (11 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (12 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (12 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (13 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (13 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (14 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (14 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (15 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (15 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (16 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (16 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (17 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (17 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (18 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (18 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A)(19 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (19 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Anöploidi için (PGT-A) (20 Embriyo)(MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing for Aneuploidies (PGT-A) (20 Embryos)(CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(1 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (1 Embryo) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(2 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (2 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(3 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (3 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(4 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (4 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(5 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (5 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(6 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (6 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(7 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (7 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(8 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (8 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(9 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (9 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(10 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (10 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(11 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (11 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(12 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (12 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(13 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (13 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(14 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (14 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(15 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (15 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(16 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (16 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(17 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (17 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(18 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (18 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(19 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (19 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Preimplantasyon Genetik Test Yapısal Kromozomların Yeniden Düzenlenmeleri (PGT-SR)(20 Embriyo) (MERKEZİMİZE DANISIN)	Preimplantation Genetic Testing Structural Chromosomal Rearengement (PGT-SR) (20 Embryos) (CONTACT WITH OUR CENTER)			PGT	Embriyo biyopsisi		15	45			28	Preimplantasyon		Embryo Biopsy
Bilinen Mutasyon Analizi-Dizi Analizi (1 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (1 mutation) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (2 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (2 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (3 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (3 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (4 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (4 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (5 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (5 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (6 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (6 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (7 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (7 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (8 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (8 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (9 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (9 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (10 Mutasyon) (. geni) (Dizi Analizi) (Postnatal)	Known Familial Mutation Analysis-Sequence Analysis (10 mutations) (Dizi Analizi) (Postnatal)	.	.	Dizi Analizi/ Sequence Analysis	EDTA'lı Kan (2-4 ml)		30	60	.	.	45	Postnatal		EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (1 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Familial Mutation Analysis-Sequence Analysis (1 mutation) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (2 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (2 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (3 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (3 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (4 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (4 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (5 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (5 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (6 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (6 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (7 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (7 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (8 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (8 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (9 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (9 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample
Bilinen Mutasyon Analizi-Dizi Analizi (10 Mutasyon) (. geni) (Dizi Analizi) (Prenatal)	Known Mutation Analysis (Multiple mutations) (10 mutations) (Sequence Analysis) (Prenatal)	.	.	Dizi Analizi/ Sequence Analysis	Amniyon sıvısı / CVS / Heparinli ve EDTA'lı (İKİSİ BİRDEN) kordon kanı VE ANNEDEN EDTA'LI KAN		.	.	15	30	20	Prenatal		Amniotic Fluid / CVS / Heparinized and EDTA (BOTH) Fetal Blood and Maternal EDTA Blood Sample